UNIVERSITY OF KWAZULU-NATAL
COLLEGE OF LAW AND MANAGEMENT STUDIES
SCHOOL OF LAW, HOWARD COLLEGE

Unregulated or not? A legal analysis of South Africa's legislative framework relevant to direct-to-consumer genetic testing

Amy Elizabeth Gooden
215031300

This dissertation is submitted in fulfilment of the requirements for the degree of Master of Laws

Professor Donrich W Thaldar
2021

DECLARATION REGARDING ORIGINALITY

I, Amy Elizabeth Gooden (215031300) declare that:

A. The research reported in this dissertation, except where otherwise indicated, is my original research.
B. This dissertation has not been submitted for any degree or examination at any other university.
C. This dissertation does not contain other persons' data, pictures, graphs or other information, unless specifically acknowledged as being sourced from other persons.
D. This dissertation does not contain other persons' writing, unless specifically acknowledged as being sourced from other researchers. Where other written sources have been quoted, then:
    a. their words have been re-written, but the general information attributed to them has been referenced;
    b. where their exact words have been used, their writing has been placed inside quotation marks, and referenced.
E. Where I have reproduced a publication of which I am an author, co-author or editor, I have indicated in detail which part of the publication was written by myself alone and have fully referenced such publications.
F. This dissertation does not contain text, graphics or tables copied and pasted from the Internet, unless specifically acknowledged, and the sources being detailed in the dissertation and in the References sections.

Signed: 

Date: 2 November 2021

PHOTOCOPYING AND INTER-LIBRARY LOAN

This dissertation is an original piece of work which is made available for photocopying and for inter-library loan.

Signed: 

Date: 2 November 2021

ACKNOWLEDGMENTS

To my supervisor, Professor Donrich Thaldar. Thank you for all the support, motivation, and guidance that you have given me over the past two years. Thank you for constantly encouraging me to strive further, for broadening my knowledge, and for sparking my interest in this field. It is because of you that I am where I am today, and I could not think of a better supervisor. You go the extra mile for your students, and I will never forget all the invaluable lessons that you have taught me – something which I know will hold me in good stead for my future career.

To the members of the Health Law and Ethics Research Group, thank you for all the assistance with my topic and for pointing me in the right direction. I appreciate all the guidance and insight that you gave me regarding my dissertation. Thank you especially to Bonginkosi Shozi for reading through my dissertation and for your helpful comments. I would also like to express my gratitude to Dr Marietjie Botes and Vaughn Harrison for their input on my chapters.

To my family. Mom and Dad, thank you for everything that you have done for me, not just over these past two years but throughout my studies. Thank you for supporting and encouraging me, and allowing me to further my education. Thank you for all the tea and snacks that you brought me while I was working and for paying for my expenses when I could not – I will forever be grateful to have parents like you. Katherine, thank you for all the help and support that you gave me, even though you were far away.

Importantly, to my person, Meshandren. I cannot put into words how grateful I am for you. Thank you for being there with me on this journey, and for the endless hours that you spent reading through my work. Thank you for listening to me while I tried to formulate arguments and for discussing ideas with me until they made sense. Thank you for bringing me back when I lost focus, and for constantly motivating me. I could not have done this without you.

ABSTRACT

Recent advances in science and technology have enabled genetic testing to be conducted inexpensively, expeditiously, and directly by consumers, therefore allowing individuals access to their genetic information without the intervention of healthcare practitioners. This technology can assist individuals to better manage their wellbeing and conserve healthcare funds. Yet, direct-to-consumer genetic testing is not free from controversy primarily due to potential human rights infringements and a perceived lack of regulation. While direct-to-consumer genetic testing may provide consumers with autonomy, involvement in healthcare decisions, convenience, and enhanced genetic literacy, the field remains contentious. The questionable validity, accuracy, and utility of tests, the absence of professional oversight and lack of suitable genetic counselling, potential result misinterpretation, consent processes, follow-up costs which burden healthcare systems, and privacy concerns surrounding the usage and confidentiality of genetic data for research, have brought direct-to-consumer genetic testing to the fore.

Despite its growing prevalence, direct-to-consumer genetic testing remains greatly under-investigated in South Africa and, while the need for regulation has been highlighted, it is yet to be fully examined. Therefore, in this dissertation, I map the current legal landscape relating to direct-to-consumer genetic testing in South Africa. This is done through a comprehensive legal analysis of South Africa's extant law relevant to the industry, and the issues associated therewith – with the intention of determining if, and how, direct-to-consumer genetic testing is legally governed in South Africa and how its various aspects and processes function within the current legislative framework.

Through this analysis, I find that the legal landscape in South Africa relating to direct-to-consumer genetic testing is multi-layered and the industry is, in fact, governed by a variety of, sometimes overlapping, statutes and regulations. Clarifying South Africa's current legal landscape regarding direct-to-consumer genetic testing enables local, as well as foreign, direct-to-consumer genetic testing companies operating in South Africa to better understand the parameters within which they may legally function, in terms of offering genetic tests directly to the public and subsequent genetic research conducted using the genetic data obtained from the samples of consumers.

Keywords: direct-to-consumer genetic testing; regulation; legal analysis; South Africa

TABLE OF CONTENTS

LIST OF ACRONYMS

A	Adenine
AB	*AB v Minister of Social Development* 2016 (2) SA 27 (GP)
ANVISA	Brazilian Health Regulatory Agency
ARB	Advertising Regulatory Board
ASA	Advertising Standards Authority of South Africa
ASSAf	Academy of Science of South Africa
BRCA	Breast Cancer Gene
C	Cytosine
CDRH	Center for Devices and Radiological Health
CFS	Certificate of Free Sale
CJEU	Court of Justice of the European Union
CPA	Consumer Protection Act 68 of 2008
DNA	Deoxyribonucleic Acid
DoH	Department of Health
DTA	Data Transfer Agreement
DTC	Direct-to-consumer
ECTA	Electronic Communications and Transactions Act 25 of 2002
EU	European Union
FDA	Food and Drug Administration
FDCA	Federal Food, Drug, and Cosmetic Act, 21 USC § 321(h)
G	Guanine
G6PD	Glucose-6-Phosphate Dehydrogenase
GDPR	General Data Protection Regulation 2016/679
GG	Government Gazette
GHR	Genetic Health Risk
GINA	Genetic Information Non-discrimination Act of 2008 122 Stat. 881

GMDN	Global Medical Device Nomenclature
GN	Government Notice
HCP	Health Care Practitioner
HIPAA	Health Insurance Portability and Accountability Act of 1996 110 Stat. 1936
HPCSA	Health Professions Council of South Africa
HREC	Health Research Ethics Committee
IP	Internet Protocol
IVD	In Vitro Diagnostic Device
LDT	Laboratory Developed Test
MAH	Marketing Authorization Holder
MCA	Marketing Code Authority
MCC	Medicines Control Council
MTA	Material Transfer Agreement
NCC	National Consumer Commission
NCT	National Consumer Tribunal
NFL	National Football League
NHA	National Health Act 61 of 2003
NHRC	National Health Research Committee
NHREC	National Health Research Ethics Council
NM	*NM v Smith* 2007 (5) SA 250 (CC)
PDPC	Personal Data Protection Commissioner
PGS	Personal Genome Service
PIPEDA	Personal Information Protection and Electronic Documents Act SC 2000
POPIA	Protection of Personal Information Act 4 of 2013
REC	Research Ethics Committee
RNA	Ribonucleic Acid
SA MTA	Material Transfer Agreement for Human Biological Materials GN R719 GG 41781 of 20 July 2018

SADC	Southern African Development Community
SAHPRA	South African Health Products Regulatory Authority
SALDA	Southern African Laboratory Diagnostic Association
SAMA	South African Medical Association
SAMED	South African Medical Device Industry Association
SARS	South African Revenue Service
SASHG	Southern African Society for Human Genetics
SCC	Standard Contractual Clause
SNP	Single Nucleotide Polymorphism
T	Thymine
TGA	Therapeutic Goods Administration
TOS	Terms of Service
UK	United Kingdom
US	United States of America
WES	Whole-Exome Sequencing
WHO	World Health Organisation

LIST OF DEFINITIONS

Analytical validity	Analytical validity refers to a test's accuracy in identifying the presence or absence of a specific genetic variant, and is often dependent on the quality of the laboratory conducting the test.
Anonymisation	Commonly involves the permanent removal of personal identifiers from data, so that it cannot be re-linked to an individual.
Big data	Substantial data sets which, when analysed, uncover links, patterns, and trends, and are used to for predictions and discoveries.
Biological material	Defined in the Human Biological Material Regulations as 'material from a human being *including* DNA, RNA, blastomeres, polar bodies, cultured cells, embryos, gametes, progenitor stem cells, small tissue biopsies and growth factors from the same'.
Biometric information	The process of personal identification predicated on the categorisation of persons – including DNA analysis.
Blanket consent	Neither defines a study nor restricts the type of research that may be undertaken.
Body fluid	A fluid or fluid secretion (such as blood, lymph, saliva, semen, or urine) of the body.
Broad consent	Defines a wide variety of studies and entails an individual permitting the use of their sample for current research, storage, and potential future research.
Buccal sample	Cellular material inside the mouth.

Chromosomes	Located in the nucleus of a cell and contain genetic information encoded in DNA.
Clinical utility	Clinical utility concerns the application of such tests and examines whether they can offer information regarding diagnosis, management, treatment, or prevention of disease.
Clinical validity	A test's ability to differentiate between individuals who possess, or will develop, a condition and those who will not. It measures the accuracy of a test in detecting or predicting disease risk.
Consumers	Those individuals who purchase and undergo direct-to-consumer genetic testing.
Data	Defined as 'any information, including personal information in any form derived directly or indirectly during the conduct of research or clinical care'.
Data subject	The person to whom personal information relates which, in this instance, is the consumer.
De-identification	Entails removing personally identifying information, which has the potential to be re-linked to the data.
Direct-to-consumer genetic testing	Defined as 'DNA-based testing…ordered by the consumer outside of established health-care delivery systems by for-profit vendors'.
DNA	The Human Biological Material Regulations define DNA as a 'a nucleic acid, composed of building blocks called nucleotides'.
DNA analysis	Interpreting genetic sequences, and is used to determine predisposition to disease.

DNA extraction	A method of purifying DNA by isolating it from the nucleus of cells, which can then be used for DNA sequencing and analysis.
DNA sequencing	A method for determining the sequence of nucleotide bases in DNA, which differs amongst individuals.
Eukaryote	Eukaryotes refer to organisms that have a defined nucleus and internal membranes. Examples include humans, animals, plants, and fungi.
Genes	Made up of a sequence of nucleotides and are the 'basic unit of inheritance'. Genes are examined when determining if an individual is a carrier of, or is predisposed to, a disease.
Genetic counsellor	Health professionals with specialised education and training in medical genetics and counselling, who assist individuals in understanding the implications of the role of genetics in disease. Genetic counsellors practice client-centred, non-directive counselling and prepare individuals and their families for test results, ensuring that they receive the necessary support and treatment.
Genetic data	Either: (1) information occurring naturally in the human body that is carried and stored by DNA; or (2) information relating to an individual's inherited or acquired genetic characteristics derived from sequencing, processing, and analysing DNA samples, and interpreting data.
Genetic determinism	The view that DNA and genetic makeup defines an individual's qualities and is the sole determining factor in the development of a disease.
Genetic literacy	An individual's basic knowledge of genetic science. It includes central genetic concepts like gene expression,

	transmission, and elementary awareness of how genes affect health.
Health-related genetic tests	Tests that aim ‘to predict risk of disease, screen for disease, direct clinical management, identify carriers, or establish prenatal diagnoses, clinical diagnoses, or prognoses in individual people or families’.
Healthcare professional	Generally used to refer to both medical or healthcare practitioners as well as genetic counsellors.
Human biological material	Defined as ‘any biological material removed from a human being, or directly or indirectly derived from such removed biological material, including sub-cellular components (such as genetic material), cells, blood, tissues, organs, gametes (sperm and ova), embryos, foetal tissues, and waste (hair, nail clippings, urine, faeces, and sweat)’.
In vitro	Outside of the body.
Informed consent	Suggests that information has been provided to participants concerning the nature and purpose of the research as well as other options. Informed consent entails free choice by individuals regarding research participation based on information allowing an informed decision.
Monogenic diseases	Heritable and arise as a result of a single genetic mutation.
Nutrigenetic tests	Offer tailored information about lifestyle and nutrition to consumers, based on their genetic profile.
Pharmacogenetic tests	Offer information regarding the effectiveness and suitability of certain drugs.

Polygenic diseases	Do not follow Mendelian inheritance patterns and include conditions such as hypertension, Alzheimer's disease, prostate cancer, and cardiovascular diseases.
Single nucleotide polymorphisms	A change at a sole point in a DNA sequence and is the most prevalent form of genetic variation.
Specific consent	Defines a particular study, limiting sample and/or data use to that purpose.
Testing companies	This refers to those companies that offer direct-to-consumer genetic testing.
Tiered consent	Combination of specific and broad consent and allows for consent individually to different aspects of a study. Tiered consent allows an individual to decide to partake in the main study and to authorise their sample to be stored and used in the future. Tiered consent also requires the provision of further information.
Tissue	Defined as 'human tissue, and includes flesh, bone, a gland, an organ, skin, bone marrow or body fluid, but excludes blood or a gamete'.
Whole exome sequencing	This targets the protein-coding regions of the human genome, allowing the identification of differences in the protein-coding region of any gene, as opposed to in a few chosen genes.

TABLE OF CASES

I SOUTH AFRICA

Gelderma Laboratories South Africa (Pty) Ltd v Medicines Control Council [2014] ZAGPPHC 360

Hayne & Co v Kaffrarian Steam Mill Co Ltd 1914 AD 363

HMBMP Properties (*Pty*) *Ltd v King* 1981 (1) SA 906 (N)

In Re Estate Cullingworth 1936 NPD 251 (NPD)

Independent Institute of Education (Pty) Limited v Kwazulu-Natal Law Society 2020 (2) SA 325 (CC)

Jaga v Dönges NO; Bhana v Dönges NO 1950 (4) SA 653 (A)

Khumalo v Holomisa 2002 (5) SA 401 (CC)

Loryan (Pty) Ltd v Solarsh Tea and Coffee (Pty) Ltd 1984 (3) SA (WLD)

Member of the Executive Council for Education: KwaZulu-Natal v Pillay 2008 (1) SA 474 (CC)

Minister of Safety and Security v Xaba 2003 (2) SA 703 (D)

Minister of the Interior v Machadodorp Investments (*Pty*) *Ltd* 1957 (2) SA 395 (A)

Mpisi v Trebble 1994 (2) SA 136 (AD)

Natal Joint Municipal Pension Fund v Endumeni Municipality 2012 (4) SA 593 (SCA)

National Coalition for Gay and Lesbian Equality v Minister of Justice 1999 (1) SA 6 (CC)

New Modderfontein Gold Mining Company v Transvaal Provincial Administration 1919 AD 367

NM v Smith 2007 (5) SA 250 (CC)

Norden v Bhanki 1974 (4) SA 647 (AD)

Nyembezi v Law Society, Natal 1981 (2) SA 752 (A)

Omegalabs (Pty) Ltd v Medicines Control Council [2016] ZAGPPHC 1157

Poswa v MEC for Economic Affairs, Environment and Tourism, Eastern Cape 2001 (3) SA 582 (SCA)

R v Debele 1956 (4) SA 570 (AD)

R v Hugo 1926 AD 268

Rex v Ah Tong 1919 AD 186

Robb NO v Standard Bank Ltd 1979 (2) SA 420 (R)

Roodepoort City Council v Shepherd 1981 (2) SA 720 (A)

Rosen v Rand Townships Registrar 1939 WLD 5 (WLD)

Ruta v Minister of Home Affairs 2019 (2) SA 329 (CC)

S v Crawford 1979 2 SA 48 (A)

S v Dane 1957 (2) SA 472 (N)

S v Kgogong 1980 (3) SA 600 (AD)

S v Maqhina 2001 (1) SACR 241 (T)

S v Mhlungu 1995 (3) SA 867 (CC)

S v Nyembe 2014 (1) SACR 105 (GSJ)

S v Orrie [2003] ZAWCHC 63

S v Weinberg 1979 (3) SA 89 (A)

S v Williams [2005] ZAWCHC 35

S v Wood 1976 (1) SA 703 (A)

Secretary for Inland Revenue v Charkay Properties (Pty) Ltd 1976 (4) SA 872 (A)

Shaik v Minister of Justice and Constitutional Development 2004 (3) SA 599 (CC)

South African Airways (Pty) Ltd v Aviation Union of South Africa 2011 (3) SA 148 (SCA)

Southern Life Association Ltd v Commissioner for Inland Revenue 1985 (2) SA 267 (C)

Stellenbosch University Law Clinic v Lifestyle Direct Group International (Pty) Ltd [2021] 4 All SA 219 (WCC)

Teddy Bear Clinic for Abused Children v Minister of Justice and Constitutional Development 2014 (2) SA 168 (CC)

Transvaal Consolidated Land and Exploration Co Ltd v Johannesburg City Council 1972 (1) SA 88 (W)

Union Government (*Minister of Finance*) *v Mack* 1917 (AD)

Venter v R 1907 TS 910 (TS)

Volschenk v Volschenk 1946 TPD 486

Waylite Diary CC v First National Bank Ltd 1995 (1) SA 645 (A)

II FOREIGN

Beckford v Wade 17 Ves

Carlill v Carbolic Smoke Ball Co [1892] 2 QB 484

Data Protection Commissioner v Facebook Ireland and Maximillian Schrems (C-311/18)

Duck v Bates 12 QBD 79

Schrems v Data Protection Commissioner (C-362/14) (6 October 2015)

TABLE OF STATUTES

I SOUTH AFRICA

Constitution of the Republic of South Africa Act 108 of 1996

Consumer Protection Act 68 of 2008

Draft Regulations Regarding the use of Human DNA, RNA Cultured Cells, Stem Cells, Blastomeres, Polar Bodies, Embryos, Embryonic Tissue and Small Tissue Biopsies for Diagnostic Testing, Health Research and Therapeutics GN R7 GG 29526 of 5 January 2007

Electronic Communications and Transactions Act 25 of 2002

General Regulations made in terms of the Medicines and Related Substances Act, 1965, as Amended GN R510 GG 24727 of 10 April 2003

Health Professions Act 56 of 1974

Interpretation Act 33 of 1957

Material Transfer Agreement for Human Biological Materials GN R719 GG 41781 of 20 July 2018

Medicines and Related Substances Act 101 of 1965 (as amended)

National Health Act 61 of 2003

Nursing Act 33 of 2005

Protection of Personal Information Act 4 of 2013

Regulations Regarding the General Control of Human Bodies, Tissue, Blood, Blood Products and Gametes GN R180 GG 35099 of 2 March 2012

Regulations Relating to Medical Devices and In Vitro Diagnostic Medical Devices (IVDs) GN R1515 GG 40480 of 9 December 2016

Regulations Relating to Research with Human Participants GN R719 GG 38000 of 19 September 2014

Regulations Relating to the Import and Export of Human Tissue, Blood, Blood Products, Cultured Cells, Stem Cells, Embryos, Fetal Tissue, Zygotes and Gametes GN R181 GG 35099 of 2 March 2012

Regulations Relating to the Taking of Buccal Sample or Withdrawal of Blood from a Living Person for Testing (Amendment) GN R944 GG 34750 of 11 November 2011

Regulations Relating to the Use of Human Biological Material GN R177 GG 35099 of 2 March 2012

Regulations Relating to Tissue Banks GN R182 GG 35099 of 2 March 2012

II FOREIGN

Australian Privacy Act 1988

Austrian Gene Technology Act of 1995

Canadian Personal Information Protection and Electronic Documents Act SC 2000 c. 5

European Union General Data Protection Regulations 2016/679

European Union Medical Devices Directive 93/42/EEC

French Loi n°. 2004-800 rélative à la bioéthique

German Human Genetic Examination Act 2009 BGBL I at 2529

Hungarian Genetic Act 2008

Portuguese Law 12/2005 of 26 January 2005

Swiss Federal Act on Human Genetic Testing 2007

United Kingdom Data Protection Act 1998

US Code: Federal Food, Drug, and Cosmetic Act 21 USC §§ 301-392

US Genetic Information Non-discrimination Act of 2008 122 Stat. 881

US Health Insurance Portability and Accountability Act of 1996 110 Stat. 1936

TABLE OF BOOKS AND ARTICLES

Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018).

Adams, R, F Adeleke, D Anderson et al 'POPIA Code of Conduct for research' (2021) 117(5/6) *S Afr J Sci* 1–12.

Adams, R, S Veldsman, M Ramsay et al 'Drafting a Code of Conduct for research under the Protection of Personal Information Act No. 4 of 2013' (2021) 117(5/6) *S Afr J Sci* 1–3.

Advertising Regulatory Board *Code of Advertising Practice* (2021).

Advertising Standards Authority of South Africa *Advertising Code of Practice*.

Allyse, MA, DH Robinson, MJ Ferber et al 'Direct-to-consumer testing 2.0: Emerging models of direct-to-consumer genetic testing' (2018) 93(1) *Symposium on Precision Medicine* 113–120.

Altman, R 'Direct-to-consumer genetic testing: Failure is not an option' (2009) 86(1) *Clin Pharmacol Ther* 15–17.

Annes, JP, MA Giovanni & MF Murray 'Risks of presymptomatic direct-to-consumer genetic testing' (2010) 363(12) *N Engl J Med* 1100–1101.

Apathy, NC, T Menser, LM Keeran et al 'Trends and gaps in awareness of direct-to-consumer genetic tests from 2007 to 2014' (2018) 54(6) *Am J Prev Med* 806–813.

Armstrong Holland, CM, EH Arbe-Barnes, EJ McGivern et al 'The 10th Oxbridge varsity medical ethics debate – Should we fear the rise of direct-to-consumer genetic testing?' (2018) 13(14) *Philos Ethics Humanit Med* 1–7.

Association for the Accreditation of Human Research Protections 'Privacy vs. confidentiality – What's the difference?' (2009) *University of Kentucky* 1–4.

Bair, S 'Direct-to-consumer genetic testing: Learning from the past and looking toward the future' (2012) 67 *Food & Drug LJ* 413–433.

Berg, C & K Fryer-Edwards 'The ethical challenges of direct-to-consumer genetic testing' (2008) 77(1) *J Bus Ethics* 17–31.

Bloss, CS, BF Darst, EJ Topol et al 'Direct-to-consumer personalized genomic testing' (2011) 20(2) *Hum Mol Genet* 132–141.

Borry, P, MC Cornel & HC Howard 'Where are you going, where have you been: A recent history of the direct-to-consumer genetic testing market' (2010) 1(3) *J Community Genet* 101–106.

Botes, M, A Olckers & M Labuschaigne 'Data commercialisation in the South African health care context' (2021) 24 *PER/PELJ* 1–35.

Botha, CJ *Statutory Interpretation* 5 ed Cape Town: Juta, (2012).

Brett, GR, SA Metcalfe, DJ Amor et al 'An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice' (2012) 20 *Eur J Hum Genet* 825–830.

Bunnik, EM, A Cecile, JW Janssens et al 'Informed consent in direct-to-consumer personal genome testing: The outline of a model between specific and generic consent' (2012) 28(7) *Bioethics* 343–351.

Cacchio, J 'What you don't know can hurt you: The legal risk of peering into the gene pool with direct-to-consumer genetic testing' (2018) 87(1) *UMKC L Rev* 219–244.

Campbell, N & S Logan *Consumer Protection Guide for Lawyers* (2011).

Caulfield, T, NM Ries, PN Ray et al 'Direct-to-consumer genetic testing: Good, bad or benign?' (2010) 77(2) *Clin Genet* 101–105.

Chandros Hull, S & K Prasad 'Reading between the lines: Direct-to-consumer advertising of genetic testing' (2001) 31(3) *Hastings Center Rep* 33–35.

Chokoshvili, D, DF Vears & P Borry 'Growing complexity of (expanded) carrier screening: Direct-to-consumer, physician-mediated, and clinic-based offers' (2017) 44 *Best Prac Res Cl Ob* 57–67.

Chow-White, PA, M MacAulay, A Charters et al 'From the bench to the bedside in the big data age: Ethics and practices of consent and privacy for clinical genomics and personalized medicine' (2015) 17(3) *Ethics Inf Technol* 189–200.

Consumer Reports *Direct-to-consumer Genetic Testing: The Law Must Protect Consumers' Genetic Privacy* (2020).

Cornel, MC, CG van El & P Borry 'The challenge of implementing genetic tests with clinical utility while avoiding unsound applications' (2014) 5(1) *J Community Genet* 7–12.

Covolo, L, S Rubinelli, E Ceretti et al 'Internet-based direct-to-consumer genetic testing: A systematic review' (2015) 17(12) *J Med Internet Res* 1–19.

Curnutte, M 'Challenging the boundaries of medicine: Consumer culture and genetic testing' (2012) 20(2) *Medic* 138–144.

Curnutte, M & G Testa 'Consuming genomes: Scientific and social innovation in direct-to-consumer genetic testing' (2012) 31(2) *New Genet Soc* 159–181.

Currie, I & J De Waal *The Bill of Rights Handbook* 6 ed Cape Town: Juta, (2013).

da Veiga, A, R Vorster, C Pilkington et al 'Compliance with the Protection of Personal Information Act and consumer privacy expectations: A comparison between the retail and medical aid industry' *16th International Information Security South Africa Conference* (August 2017 Johannesburg, South Africa) 16–23.

Daemmrich, AA '23andMe: The business and ethics of personal genetics testing' (2015) *University of Kansas School of Medicine* 1–15.

Dandara, C, J Greenberg, L Lambie et al 'Direct-to-consumer genetic testing: To test or not to test, that is the question' (2013) 103(8) *SAMJ* 510–512.

de Wet, S, H Oosthuizen & J Visser 'DNA profiling and the law in South Africa' (2011) 14(4) *PER/PELJ* 171–207.

Delaney, SK & MF Christman 'Direct-to-consumer genetic testing: Perspectives on its value in healthcare' (2016) 99(2) *Clin Pharmacol Ther* 146–148.

Department of Health *Ethics in Health Research: Principles, Processes and Structures* 2 ed Pretoria: Department of Health (2015).

Department of Health *Guidelines for Good Practice in the Conduct of Clinical Trials with Human Participants in South Africa* 2 ed (2006).

Department of Health *South Africa Frequently Asked Questions (FAQs) Import/Export Permit Programme* (2015).

Devenish, GE *Interpretation of Statutes* Cape Town: Juta, (1992).

Dickenson, D 'Testing times for the consumer genetics revolution' (2014) 221(2951) *New Scientist* 26–27.

Donnelly, DL 'Privacy by (re)Design: A Comparative Study of the Protection of Personal Information in the Mobile Applications Ecosystem under United States, European Union and South African Law' (unpublished Doctor of Laws thesis, University of KwaZulu-Natal, 2020).

Dorfman, R, R Khan & G Mukerjee 'Proposed regulatory framework for direct-to-consumer genetic testing: Diagnostics vs genetic screening' (2014) 60(11) *Clin Chem* 1455–1456.

Du Plessis, L 'Interpretation' in S Woolman, M Bishop & J Brickhill (eds) 2 ed *Constitutional Law of South Africa* Juta: Cape Town, (2008) 32–161.

Du Plessis, LM 'Statute law and interpretation' in LM Du Plessis (ed) 2 ed *The Law of South Africa* 25(1) Durban: Butterworths, (2011).

Eissenberg, JC 'Direct-to-consumer genomics: Harmful or empowering?' (2017) 114(1) *Mo Med* 26–32.

Erlich, Y, JB Williams, D Glazer et al 'Redefining genomic privacy: Trust and empowerment' (2014) 12(11) *PLoS Biol* 1–5.

Field, A, A Krokosky & SF Terry 'Direct-to-consumer marketing of genetic tests: Access does not reflect clinical utility' (2010) 14(6) *Genetic Alliance* 731–732.

Fukuda, R & F Takada 'Legal regulations on health-related direct-to-consumer genetic testing in 11 countries' (2018) 48 *Kitasato Med J* 52–59.

Geransar, R & E Einsiedel 'Evaluating online direct-to-consumer marketing of genetic tests: Informed choices or buyers beware?' (2008) 12(1) *Genet Test* 13–23.

Giovanni, MA, MR Fickie, LS Lehmann et al 'Health-care referrals from direct-to-consumer genetic testing' (2010) 14(6) *Genet Test Mol Biomarkers* 817–819.

GMDN *GMDN User Guide: A Comprehensive Guide to the Global Medical Device Nomenclature* (2018).

Gniady, JA 'Regulating direct-to-consumer genetic testing: Protecting the consumer without quashing a medical revolution' (2008) 76(5) *Fordham L Rev* 2429–2475.

Goddard, KAB, J Robitaille, NF Dowling et al 'Health-related direct-to-consumer genetic tests: A public health assessment and analysis of practices related to internet-based tests for risk of thrombosis' (2009) 12(2) *Public Health Genomics* 92–104.

Goemans, B & R McLaughlin *Medical Devices: An Innovation Guide from Lab to Commercialisation* (2018).

Goldswain, GK 'The purposive approach to the interpretation of fiscal legislation – The winds of change' (2008) 16(2) *Meditari Accountancy Research* 107–121.

Green, RC & NA Farahany 'Regulation: The FDA is overcautious on consumer genomics' (2014) 505 *Nature* 286–287.

Greenleaf, G '2014-2017 update to Graham Greenleaf's *Asia Data Privacy Laws – Trade and Human Rights Perspectives*' (2017) 47 *UNSWLRS* 1–40.

Gupta, N 'DNA extraction and Polymerase Chain Reaction' (2019) 36(2) *J Cytol* 116–117.

Gutmann Koch, V & K Todd 'Research revolution or status quo: The new common rule and research arising from direct-to-consumer genetic testing' (2018) 56(1) *Hous L Rev* 81–121.

Gutmann Koch, V 'PGTandMe: Social networking-based genetic testing and the evolving research model' (2012) 22(1) *Health Matrix* 33–74.

Gymrek, M, AL McGuire, D Golan et al 'Identifying personal genomes by surname inference' (2013) 339(6117) *Science* 321–324.

Hawkins, AK & A Ho 'Genetic counseling and the ethical issues around direct to consumer genetic testing' (2012) 21(3) *J Genet Counsel* 367–373.

Hazel, JW & C Slobogin 'Who knows what, and when? A survey of the privacy policies proffered by U.S. direct-to-consumer genetic testing companies' (2018) 28(1) *Cornell J Law Public Policy* 35–66.

Health Professions Council of South Africa *Guidelines for Good Practice in the Health Care Professions: General Ethical Guidelines for Health Researchers (Booklet 13)* (2016).

Health Professions Council of South Africa *Standards of Practice for Genetic Counsellors* (2013).

Helgason, A & K Stefánsson 'The past, present, and future of direct-to-consumer genetic tests' (2010) 12(1) *Dialogues Clin Neurosci* 61–68.

Hendricks-Sturrup, RM & CY Lu 'Direct-to-consumer genetic testing data privacy: Key concerns and recommendations based on consumer perspectives' (2019) 9(2) *J Pers Med* 1–7.

Hogarth S & D Melzer *The IVD Directive and Genetic Testing Problems and Proposals: A Briefing Presented to the 20th Meeting of Competent Authorities* (2007) 1–22.

Hogarth, S & P Saukko 'A market in the making: The past, present and future of direct-to-consumer genomics' (2017) 36(3) *New Genet Soc* 197–208.

Hogarth, S, G Javitt & D Melzer 'The current landscape for direct-to-consumer genetic testing: Legal, ethical, and policy issues' (2008) 9 *Annu Rev Genom Hum Genet* 161–182.

Hoglund-Shen, A 'Direct-to-consumer genetic testing, gamete donation, and the law' (2017) 55(3) *Fam Court Rev* 472–484.

Howard, HC & P Borry 'Direct-to-consumer genetic testing: More questions than benefits?' (2008) 5(4) *Per Med* 317–320.

Howard, HC & P Borry 'Europe and direct-to-consumer genetic tests' (2011) 13 *Nat Rev Genet* 146.

Howard, HC & P Borry 'Survey of European clinical geneticists on awareness, experiences and attitudes towards direct-to-consumer genetic testing' (2013) 5(45) *Genome Med* 1–11.

Howard, HC, BM Knoppers & P Borry 'Blurring lines: The research activities of direct-to-consumer genetic testing companies raise questions about consumers as research subjects' (2010) 11(8) *EMBO Rep* 579–582.

Howard, HC, S Sterckx, J Cockbain et al 'The convergence of direct-to-consumer genetic testing companies and biobanking activities: The example of 23andMe' in M Wienroth & E Rodrigues (eds) 1 ed *Knowing New Biotechnologies: Social Aspects of Technological Convergence* London: Routledge, (2015) 59–74.

Howes, R 'SALDA In Vitro Diagnostics in South Africa' (2014) *SALDA* 1–18.

Hudson, K, G Javitt, W Burke et al 'ASHG statement on direct-to-consumer genetic testing in the United States' (2007) 110(6) *Obstet Gynecol* 1392–1395.

Hulshizer, R ‘The impending impact of direct-to-consumer personal genomic services’ (2010) 22(3) *ECRI Institute* 1–4.

Huml, AM, C Sullivan, M Figueroa et al ‘Consistency of direct-to-consumer genetic testing results among identical twins’ (2020) 133(1) *Am J Med* 143–146.

Jackson, L, L Goldsmith & H Skirton ‘Guidance for patients considering direct-to-consumer genetic testing and health professionals involved in their care: Development of a practical decision tool’ (2014) 31(3) *Fam Pract* 341–348.

Jeong, G ‘Estimating the effects of consumer characteristics on the intention to use direct-to-consumer genetic testing’ (2018) 7(3) *Health Policy Technol* 233–238.

Jordaan, DW ‘Autonomy as an element of human dignity in South African case law’ (2009) 9(3) *Journal of Philosophy, Science & Law* 1–15.

Jordaan, DW ‘Social justice and research using human biological material: A response to Mahomed, Nöthling-Slabbert and Pepper’ (2016) 106(7) *SAMJ* 678–680.

Jordaan, DW ‘The boy and his microscope: Interpreting section 56(1) of the National Health Act’ (2009) 2(1) *SAJBL* 12–19.

Kalokairinou, L, HC Howard, S Slokenberga et al ‘Legislation of direct-to-consumer genetic testing in Europe: A fragmented regulatory landscape’ (2018) 9(2) *J Community Genet* 117–132.

Kaye, J ‘The regulation of direct-to-consumer genetic tests’ (2008) 17(2) *Hum Mol Genet* 180–183.

Kaye, J ‘The tension between data sharing and the protection of privacy in genomics research’ (2012) 13 *Annu Rev Genomics Hum Genet* 415–431.

Kim, J ‘Health technology update’ (2017) 18 *CADTH* 1–17.

Klug, WS, MR Cummings, CA Spencer et al *Concepts of Genetics* 12 ed New York: Pearson, (2018).

Knoppers, BM, D Avard & HC Howard ‘Direct-to-consumer genetic testing: Driving choice?’ (2010) 10(8) *Expert Rev Mol Diagn* 965–968.

Krause, A ‘New genetic testing technologies: Advantages and limitations’ (2019) 109(4) *SAMJ* 207–209.

Laestadius, LI, JR Rich & PL Auer 'All your data (effectively) belong to us: Data practices among direct-to-consumer genetic testing firms' (2016) 19(5) *Genet Med* 513–520.

Lee, SSJ & L Crawley 'Research 2.0: Social networking and DTC genomics' (2009) 9(6-7) *Am J Bioeth* 35–44.

Lippi, G, EJ Favaloro & M Plebani 'Direct-to-consumer testing: More risks than opportunities' (2011) 65(12) *Int J Clin Pract* 1221–1229.

Lombard, Z, F Baine, A Krause et al 'Implications of direct-to-consumer whole-exome sequencing in South Africa' (2016) 106(2) *SAMJ* 139–140.

Lucivero, F & B Prainsack 'The lifestylisation of healthcare? "Consumer genomics" and mobile health as technologies for healthy lifestyle' (2015) 4 *Appl Transl Genom* 44–49.

Lynch, J, A Parrott, RJ Hopkin et al 'Media coverage of direct-to-consumer genetic testing' (2011) 20(5) *J Genet Counsel* 486–494.

Magnus, D, MK Cho & R Cook-Deegan 'Direct-to-consumer genetic tests: Beyond medical regulation?' (2009) 1(2) *Genome Med* 1–4.

Mahmoud-Davis, SA 'Direct-to-consumer genetic testing: Empowering EU consumers and giving meaning to the informed consent process within the IVDR and GDPR frameworks' (2020) 19(1) *Wash U Global Stud L Rev* 1–52.

Mahomed, S, M Nöthling-Slabbert & MS Pepper 'The legal position on the classification of human tissue in South Africa: Can tissues be owned?' (2013) 6(1) *SAJBL* 16–20.

Mainetti, R, S Oliveri, A Gorini et al 'Usability testing of two mini-games and one serious game to educate people about genetics' *pHealth 16th International Conference on Wearable, Micro & Nano Technologies for Personalized Health* (10-12 June 2019 Genoa, Italy) 82–87.

Majumder, MA, CJ Guerrini & AL McGuire 'Direct-to-consumer genetic testing: Value and risk' (2021) 72 *Annu Rev Med* 155–166.

Malin, B, D Karp & RH Scheuermann 'Technical and policy approaches to balancing patient privacy and data sharing in clinical and translational research' (2010) 58(1) *J Investig Med* 11–18.

Marketing Code Authority *The South African Code of Marketing Practice for Health Products Code & Guideline, Version 14* (2021).

Mathews, CK, KE van Holde, DR Appling et al *Biochemistry* 4 ed Toronto: Pearson, (2012).

McGuire, AL, BJ Evans, T Caulfield et al 'Regulating direct-to-consumer personal genome testing' (2010) 330(6001) *Science* 181–182.

Middleton, A, A Mendes, CM Benjamin et al 'Direct-to-consumer genetic testing: Where and how does genetic counseling fit?' (2017) 14(3) *Per Med* 249–257.

Mykitiuk, R 'Caveat emptor: Direct-to-consumer supply and advertising of genetic testing' (2004) 27(1) *Clin Invest Med* 23–32.

Naidoo, M 'The CRISPR Patent Landscape: A South African Perspective' (Degree of Master of Laws in Business Law, University of KwaZulu-Natal 2020).

Nicol, D, M Hagger, N Ries et al 'Time to get serious about privacy policies: The special case of genetic privacy' (2014) 42(1) *Fed L Rev* 149–179.

Nienaber, A 'Consent to and authorisation of the export and use of human biological specimens for future research – Perspectives from three African countries' (2011) 44(2) *CILSA* 225–254.

Nienaber, A & KN Bailey 'The right to physical integrity and informed refusal: Just how far does a patient's right to refuse medical treatment go?' (2016) 9(2) *SAJBL* 73–77.

Nöthling Slabbert, M & M Pepper '"A room of our own?" Legal lacunae regarding genomic sovereignty in South Africa' (2010) 73 *THRHR* 432–450.

Norrgard, K 'DTC genetic testing for diabetes, breast cancer, heart disease and paternity' (2008) 1(1) *Nat Education* 86.

Novy, MC 'Privacy at a price: Direct-to-consumer genetic testing & the need for regulation' (2010) 1 *JL Tech & Pol'y* 157–180.

Nuffield Council on Bioethics *Medical Profiling and Online Medicine: The Ethics of 'Personalised Healthcare' in a Consumer Age* (2010).

Oh, B 'Direct-to-consumer genetic testing: Advantages and pitfalls' (2019) 17(3) *Genomics Inform* 1–3.

Oliveri, S & G Pravettoni 'The disclosure of direct-to-consumer genetic testing: Sounding out the psychological perspective of consumers' (2016) 8(5) *Biol Med* 1–4.

Pàmpols Ros, T, JM García Sagredo, A Pérez Aytés et al 'Directed to consumer genetic testing: Perspective from the Ethics Commission of the Spanish Society for Human Genetics' (2019) 153(1) *Med Clin (Barc)* 35–40.

Pearson, YE & Y Liu-Thompkins 'Consuming direct-to-consumer genetic tests: The role of genetic literacy and knowledge calibration' (2012) 31(1) *J Public Policy Mark* 42–57.

Pepper, MS 'The human genome and molecular medicine – Promises and pitfalls' (2010) 100(11) *SAMJ* 722–724.

Pepper, MS, C Dandara, J de Vries et al 'An optimistic vision for biosciences in South Africa: Reply to Thaldar et al. (2019)' (2019) 115(7/8) *S Afr J Sci* 1.

Pepper, MS, C Dandara, J de Vries et al 'ASSAf consensus study on the ethical, legal and social implications of genetics and genomics in South Africa' (2018) 114(11/12) *S Afr J Sci* 1–3.

Phillips, AM 'Genomic privacy and direct-to-consumer genetics: Big consumer genetic data – What's in that contract?' (2015) *IEEE Symposium on Security and Privacy Workshops (SPW)* (21-22 May 2015 San Jose, California) 60–64.

Pike, ER 'Securing sequences: Ensuring adequate protections for genetic samples in the age of big data' (2015) 37 *Cardozo L Rev* 1977–2033.

Popovsky, M 'Exaggerated benefits and underestimated harms: The direct-to-market consumer genetic test market and how to manage it going forward' (2010) 8(2) *Dartmouth LJ* 65–87.

Powell, KP, WA Cogswell, CA Christianson et al 'Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing' (2011) 21(1) *J Genet Counsel* 113–126.

Prainsack, B 'The power of prediction: How personal genomics became a policy challenge' (2011) 40(4) *ÖZP* 401–415.

Resnik, DB 'Direct-to-consumer genomics, social networking, and confidentiality' (2009) 9(6-7) *Am J Bioeth* 45–46.

Richter, M, WDF Venter & A Gray 'Home self-testing for HIV: AIDS exceptionalism gone wrong' (2010) 100(10) *SAMJ* 636–642.

Robertson, AS ‘Taking responsibility: Regulations and protections in direct-to-consumer genetic testing’ (2009) 24(1) *Berkeley Tech LJ* 213–243.

Ruhl, GL, JW Hazel, E Wright Clayton et al ‘Public attitudes toward direct to consumer genetic testing’ (2019) *AMIA Annu Symp Proc* 774–783.

Saidi, T & T Douglas ‘Medical device regulation in South Africa: The Medicines and Related Substances Amendment Act 14 of 2015’ (2018) 108(3) *SAMJ* 168–170.

Sandroff, R ‘Direct-to-consumer genetic tests and the right to know’ (2010) 40(5) *Hastings Center Rep* 24–25.

Sanfilippo, PG, LS Kearns, P Wright et al ‘Current landscape of direct-to-consumer genetic testing and its role in ophthalmology: A review’ (2015) 43(6) *Clin Exp Ophthalmol* 578–590.

Sarata, AK ‘FDA regulation of Laboratory-Developed Tests (LDTs)’ (2019) *Congressional Research Service* 1–3.

Sarin, R ‘Ethics and clinical utility of direct-to-consumer genetic tests’ (2015) 11(1) *J Can Res Ther* 1–2.

Saukko, P ‘State of play in direct-to-consumer genetic testing for lifestyle-related diseases: Market, marketing content, user experiences and regulation’ (2013) 72(1) *Proc Nutr Soc* 53–60.

Saunders, R ‘Legal implications of direct-to-consumer genetic testing for common diseases’ (2010) 1 *QMLJ* 71–79.

Savard, J, C Hickerton, SA Metcalfe et al ‘From expectations to experiences: Consumer autonomy and choice in personal genomic testing’ (2020) 11(1) *AJOB Empir Bioeth* 63–76.

Schickedanz, AD & R Herdman ‘Direct-to-consumer genetic testing: The need to get retail genomics right’ (2009) 86(1) *Clin Pharmacol Ther* 17–20.

Shabani, M & P Borry ‘Rules for processing genetic data for research purposes in view of the new EU General Data Protection Regulation’ (2018) 26(2) *Eur J Hum Genet* 149–156.

Sharkey, CM ‘Direct-to-consumer genetic testing: The FDA’s dual role as safety and health information regulator’ (2019) 68(2) *Depaul L Rev* 343–384.

Sheehan, M ‘The right to know and genetic testing’ (2015) 41(4) *J Med Ethics* 287–288.

Shi, X & X Wu ‘An overview of human genetic privacy’ (2017) 1387(1) *Ann NY Acad Sci* 61–72.

Sieber, JE ‘Privacy and confidentiality: As related to human research in social and behavioral science’ (2001) *Online Ethics Center at the National Academy of Engineering* 1–50.

Singh, A ‘The Impact of the Constitution on Transforming the Process of Statutory Interpretation in South Africa’ (Doctor of Philosophy, University of KwaZulu-Natal, 2014).

Singleton, A, LH Erby, KV Foisie et al ‘Informed choice in direct-to-consumer genetic testing (DTCGT) websites: A content analysis of benefits, risks, and limitations’ (2012) 21(3) *J Genet Counsel* 1–12.

South African Health Products Regulatory Agency (SAHPRA) *Classification of Medical Devices and IVDs* (2019).

South African Health Products Regulatory Authority (SAHPRA) *Guideline for a Licence to Manufacture, Import, Export or Distribute Medical Devices & IVDs* (2019).

South African Law Reform Commission *Discussion Paper 112 Statutory Revision: Review of the Interpretation Act 33 of 1957 (Project 25)* (2006).

South African Medical Device Industry Association *An Overview of the Medical Device Code of Ethical Marketing and Business Practice* (2020).

South African Medical Device Industry Association *Medical Device Code of Ethical Marketing and Business Practice* (2017).

South African Medical Research Council *The South African Medical Research Council Guidelines on the Responsible Conduct of Research* (2017).

Staunton, C & K Moodley ‘Data mining and biological sample exportation from South Africa: A new wave of bioexploitation under the guise of clinical care?’ (2016) 106(2) *SAMJ* 136–138.

Staunton, C, R Adams, ES Dove et al ‘Ethical and practical issues to consider in the governance of genomic and human research data and data sharing in South Africa: A meeting report’ (2019) 2(15) *AAS Open Res* 1–11.

Staunton, C, R Adams, M Botes et al ‘Safeguarding the future of genomic research in South Africa: Broad consent and the Protection of Personal Information Act No. 4 of 2013’ (2019) 109(7) *SAMJ* 468–470.

Stoeklé HC, MF Mamzer-Bruneel, G Vogt et al ‘23andMe: A new two-sided data-banking market model’ (2016) 17(19) *BMC Med Ethics* 1–11.

Su, P ‘Direct-to-consumer genetic testing: A comprehensive view’ (2013) 86(3) *Yale J Biol Med* 359–365.

Swales, L ‘The Protection of Personal Information Act and data de-identification’ (2021) 117(7/8) *S Afr J Sci* 1–3.

Sweeny, K & AM Legg ‘Predictors of interest in direct-to-consumer genetic testing’ (2011) 26(10) *Psychol Health* 1259–1272.

Tamir, S ‘Direct-to-consumer genetic testing: Ethical-legal perspectives and practical considerations’ (2010) 18(2) *Med Law Rev* 213–238.

Tandy-Connor, S, J Guiltinan, K Krempely et al ‘False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care’ (2018) 20(12) *Genet Med* 1515–1521.

Teare, HJA, M Prictor & J Kaye ‘Reflections on dynamic consent in biomedical research: the story so far’ (2021) 29 *Eur J Hum Genet* 649–656.

Thaldar, D ‘One material transfer agreement to rule them all? A call for revising South Africa’s new standard material transfer agreement’ (2020) 7(105) *Humanities & Social Sciences Communications* 1–5.

Thaldar, D ‘Why POPIA does not apply to DNA’ (2021) 117(7/8) *S Afr J Sci* 1–2.

Thaldar, D & B Townsend ‘Protecting personal information in research: Is a code of conduct the solution?’ (2021) 117(3/4) *S Afr J Sci* 1–2.

Thaldar, D, J Kinderlerer & S Soni ‘An optimistic vision for biosciences in South Africa: A response to the ASSAf report on human genetics and genomics’ (2019) 115(7/8) *S Afr J Sci* 1.

Thaldar, D, M Botes, B Shozi et al ‘Human germline editing: Legal-ethical guidelines for South Africa’ (2020) 116(9/10) *S Afr J Sci* 1–7.

Thaldar, DW & B Shozi 'The legal status of human biological material used for research' *Forthcoming in SALJ*.

Thaldar, DW & B Townsend 'Genomic research and privacy: A response to Staunton *et al*' (2020) 110(3) *SAMJ* 172–174.

Thaldar, DW & BA Townsend 'Exempting health research from the consent provisions of POPIA' (2021) 24 *PER/PELJ* 1–30.

Thaldar, DW, M Botes & A Nienaber 'South Africa's new standard material transfer agreement: Proposals for improvement and pointers for implementation' (2020) 21(85) *BMC Med Ethics* 1–13.

Thaldar, DW & M Steytler 'Time for Cinderella to go to the ball: Reflections on the right to freedom of scientific research' (2020) 138(2) *SALJ* 260–288.

The British Medical Association *Complete Family Health Encyclopaedia* London: Colour Library Books, (1997).

The Lancet 'Direct-to-consumer genetic testing' (2012) 380(9837) *Lancet* 76.

The Specialist Forum 'Limitations of DIY genetic testing' (2019) 19(6) *The Specialist Forum* 43–44.

Thrush, SA & R McCaffrey 'Direct-to-consumer genetic testing: What the nurse practitioner should know' (2010) 6(4) *J Nurse Pract* 269–273.

Tiller, J & P Lacaze 'Regulation of internet-based genetic testing: Challenges for Australia and other jurisdictions' (2018) 6(24) *Front Public Health* 1–6.

Townsend, B 'The lawful sharing of health research data in South Africa and beyond' (2021) *Inf Commun Technol Law* 1–18.

Townsend, BA 'Software as a medical device: Critical rights issues regarding artificial intelligence software-based health technologies in South Africa' (2020) 4 *TSAR* 747–762.

Townsend, BA & DW Thaldar 'Navigating uncharted waters: Biobanks and informational privacy in South Africa' (2019) 35(4) *SAJHR* 329–350.

Turrini, M 'Online genomes: Problematizing the disruptiveness of direct-to-consumer genetic tests' (2018) 12(11) *Sociology Compass* 1–13.

Varga, EA ‘You want to do what? My mother’s choice to have direct-to-consumer genetic testing’ (2012) 21(3) *J Genet Counsel* 382–385.

Vayena, E ‘Direct-to-consumer genomics on the scales of autonomy’ (2015) 41(4) *J Med Ethics* 310–314.

Weiermiller, C ‘The future of direct-to-consumer genetic testing: Regulation and innovation’ (2015) 16(5) *NC JL & Tech* 137–171.

Weiss, MA & K Archick ‘U.S.-EU data privacy: From Safe Harbor to Privacy Shield’ (2016) *Congressional Research Service* 1–16.

Wright, CF, A Hall & RL Zimmern ‘Regulating direct-to-consumer genetic tests: What is all the fuss about?’ (2011) 13(4) *Genet Med* 295–300.

Yaneva-Deliverska, M ‘Legal aspects of direct-to-consumer genetic tests’ (2011) 17(1) *J of IMAB* 135–138.

Yim, SH & YJ Chung ‘Reflections on the US FDA’s warning on direct-to-consumer genetic testing’ (2014) 12(4) *Genomics Inform* 151–155.

Zettler, PJ, JS Sherkow & HT Greely ‘23andMe, the Food and Drug Administration, and the future of genetic testing’ (2014) 174(4) *JAMA Inter Med* 493–494.

TABLE OF ONLINE SOURCES

23andMe '23andMe for healthcare professionals' (2020) available at *https://medical.23andme.com/*, accessed on 5 October 2019.

23andMe 'Becoming part of something bigger' available at *https://www.23andme.com/research/*, accessed on 9 January 2021.

23andMe 'Biobanking consent document' (2020) available at *https://www.23andme.com/about/biobanking/*, accessed on 9 January 2021.

23andMe 'Difference between DNA genotyping & sequencing' available at *https://customercare.23andme.com/hc/en-us/articles/202904600-Difference-Between-DNA-Genotyping-Sequencing*, accessed on 17 January 2021.

23andMe 'Health + ancestry service' (2020) available at *https://www.23andme.com/dna-health-ancestry?mkpc=true*, accessed on 5 October 2019.

23andMe 'How it works' available at *https://www.23andme.com/howitworks/*, accessed on 20 November 2020.

23andMe 'Individual data sharing consent' (2020) available at *https://www.23andme.com/about/individual-data-consent/*, accessed on 9 January 2021.

23andMe 'Privacy' available at *https://www.23andme.com/privacy/*, accessed on 4 May 2020.

23andMe 'Providing saliva sample for DNA test kit' available at *https://customercare.23andme.com/hc/en-us/articles/202904530-Providing-Saliva-Sample-for-DNA-Test-Kit*, accessed on 9 January 2021.

23andMe 'Publications' available at *https://www.23andme.com/publications/*, accessed on 9 January 2021.

23andMe 'Research' available at *https://www.23andme.com/en-int/research/*, accessed on 4 May 2020.

23andMe 'Research consent document' (2020) available at *https://www.23andme.com/about/consent/*, accessed on 9 January 2021.

23andMe ‘Returning DNA sample to the lab’ available at *https://customercare.23andme.com/hc/en-us/articles/202904570-Returning-DNA-Sample-to-the-Lab*, accessed on 25 June 2020.

23andMe ‘Terms of service’ (30 September 2019) available at *https://www.23andme.com/about/tos/*, accessed on 9 January 2021.

23andMe ‘What countries do you ship to?’ available at *https://int.customercare.23andme.com/hc/en-us/articles/214806628-What-countries-do-you-ship-to-*, accessed on 2 January 2020.

Academy of Science of South Africa ‘POPIA: A code of conduct for research’ available at *https://www.assaf.org.za/index.php/2-uncategorised/798-popia-a-code-of-conduct-for-research*, accessed on 6 May 2021.

Alt Advisory ‘POPIA compliance’ available at *https://altadvisory.africa/popia/#applicationofpopia*, accessed on 9 October 2020.

Ancestry ‘Now your DNA reveals so much more with AncestryHealth’ available at *https://www.ancestry.com/health*, accessed on 5 October 2019.

AncestryDNA ‘Ancestry terms and conditions’ (23 September 2020) available at *https://www.ancestry.com/cs/legal/termsandconditions*, accessed on 9 January 2021.

AncestryDNA ‘AncestryDNA informed consent’ (24 July 2018) available at *https://www.ancestry.com/dna/lp/informedconsent-v4-en*, accessed on 9 January 2021.

AncestryDNA ‘Need to ship to another country?’ available at *https://www.ancestry.com/checkout/MLI?rtype=85&flow=3*, accessed on 2 January 2020.

AncestryDNA ‘Your privacy’ (23 September 2020) available at *https://www.ancestry.com/cs/legal/privacystatement*, accessed on 9 January 2021.

Angelica, N ‘FDA 23andMe Personal Genome Service (PGS) test – Evaluation of automatic class III designation – De novo request (memorandum)’ (24 October 2017) *Duke SciPol* available at *https://scipol.duke.edu/track/fda-23andme-personal-genome-service-pgs-test-%E2%80%93-evaluation-automatic-class-iii-designation-%E2%80%93-de*, accessed on 27 November 2020.

Australian Government Department of Health 'IVD medical devices: Definitions & links' (3 December 2010) available at *https://www.tga.gov.au/ivd-medical-devices-definitions-links*, accessed on 11 January 2021.

Bhagattjee, P 'South Africa – Data protection overview' (July 2020) *Data Guidance* available at *https://www.dataguidance.com/notes/south-africa-data-protection-overview*, accessed on 9 October 2020.

Biology Dictionary 'DNA sequencing' (4 October 2019) available at *https://biologydictionary.net/dna-sequencing/*, accessed on 4 February 2021.

Botes, M 'Direct-to-consumer genetic tests: A treacherous road' (2015) *Caveat Legal* available at *http://www.caveatlegal.com/direct-to-consumer-genetic-tests-a-treacherous-road/*, accessed on 2 June 2020.

Bottomley, EJ 'Less than 100 days before SA's strict new privacy rules. Here's how it will affect you' (25 March 2021) *Business Insider South Africa* available at *https://www.businessinsider.co.za/popia-what-your-business-needs-to-do-2021-3*, accessed on 7 April 2021.

Britannica 'Eukaryote' available at *https://www.britannica.com/science/eukaryote*, accessed on 18 February 2021.

Britannica 'Software' available at *https://www.britannica.com/technology/software*, accessed on 6 January 2021.

Buthelezi, T 'The Protection of Personal Information Act (POPIA) is here – A quick run down' (7 August 2020) *Shepstone & Wylie Attorneys* available at *https://www.wylie.co.za/articles/the-protection-of-personal-information-act-popia-is-here-a-quick-run-down/*, accessed on 9 October 2020.

Buxton, J 'Call to improve accuracy of predictive genetic tests' (12 October 2009) *BioNews* available at *https://www.bionews.org.uk/page_91911*, accessed on 17 October 2019.

Cambridge Dictionary 'Apparatus' available at *https://dictionary.cambridge.org/dictionary/english/apparatus*, accessed on 6 January 2021.

Cambridge Dictionary 'Appliance' available at *https://dictionary.cambridge.org/dictionary/english/appliance*, accessed on 6 January 2021.

Cambridge Dictionary ‘Machine’ available at *https://dictionary.cambridge.org/dictionary/english/machine*, accessed on 6 January 2021.

Cambridge Dictionary ‘Pathology’ available at *https://dictionary.cambridge.org/dictionary/english/pathology*, accessed on 20 August 2020.

Cambridge Dictionary ‘Reagent’ available at *https://dictionary.cambridge.org/dictionary/english/reagent*, accessed on 6 January 2021.

Christman, J ‘Autonomy in moral and political philosophy’ (29 June 2020) *Stanford Encyclopaedia of Philosophy* available at *https://plato.stanford.edu/entries/autonomy-moral/*, accessed on 12 July 2020.

Clark, D ‘Genetic exceptionalism and paternalism themes in new German legislation’ (2 September 2009) *The Privacy Report* available at *https://theprivacyreport.com/2009/09/02/genetic-exceptionalism-and-paternalism-themes-in-new-german-legislation/*, accessed on 19 October 2019.

Collins Dictionary ‘Body fluids’ available at *https://www.collinsdictionary.com/dictionary/english/body-fluids*, accessed on 3 August 2020.

Collins Dictionary ‘Instrument’ available at *https://www.collinsdictionary.com/dictionary/english/instrument*, accessed on 6 January 2021.

Collins Dictionary ‘Therapeutic’ available at *https://www.collinsdictionary.com/dictionary/english/therapeutic*, accessed on 6 May 2021.

Color ‘Color informed consent’ available at *https://www.color.com/informed-consent*, accessed on 9 January 2021.

CRIN ‘Bodily integrity’ (2018) available at *https://archive.crin.org/en/home/what-we-do/policy/bodily-integrity.html*, accessed on 14 April 2021.

Cussins, J ‘Direct-to-consumer genetic tests should come with a health warning’ (15 January 2015) *The Pharmaceutical Journal* available at *https://www.pharmaceutical-*

journal.com/news-and-analysis/opinion/comment/direct-to-consumer-genetic-tests-should-come-with-a-health-warning/20067564.article, accessed on 23 February 2021.

DNAlysis 'About' available at *https://dnalysis.co.za/about/*, accessed on 22 June 2020.

DNAlysis 'Privacy policy' (2020) available at *https://dnalysis.co.za/privacy-policy/*, accessed on 22 June 2020.

Dyer, L & C Kennedy 'Digital business in South Africa: Overview' (1 March 2021) *Thomson Reuters* available at *https://uk.practicallaw.thomsonreuters.com/w-007-8319?transitionType=Default&contextData=(sc.Default)&firstPage=true*, accessed on 5 April 2021.

EasyDNA 'About us' (2020) available at *https://www.easydna.co.za/about-us/*, accessed on 22 June 2020.

EasyDNA 'Home' available at *https://www.easydna.co.za/*, accessed on 22 June 2020.

EasyDNA 'Privacy policy' (20 May 2018) available at *https://www.easydna.co.za/privacy-policy/*, accessed on 9 January 2021.

EasyDNA 'Terms and conditions' (24 May 2018) available at *https://www.easydna.co.za/terms-of-service/*, accessed on 22 June 2020.

Educause 'Guidelines for data de-identification or anonymization' (July 2015) available at *https://www.educause.edu/focus-areas-and-initiatives/policy-and-security/cybersecurity-program/resources/information-security-guide/toolkits/guidelines-for-data-deidentification-or-anonymization*, accessed on 21 May 2021.

Eisenhart, S 'South African medical device regulatory system set for implementation' (22 August 2016) *Emergo* available at *https://www.emergobyul.com/blog/2016/08/south-african-medical-device-regulatory-system-set-implementation*, accessed on 18 June 2020.

Experian 'What is the Safe Harbour Agreement?' available at *https://www.experian.co.uk/business/glossary/safe-harbour-agreement/*, accessed on 13 May 2021.

Furnival, J 'The trouble with UNinformed consent in direct-to-consumer genetic testing' (19 May 2021) *SoundRocket* available at *https://soundrocket.com/trouble-informed-consent-dtc-genetic-testing/*, accessed on 8 June 2021.

GenebyGene 'Gene by Gene privacy policy' available at *https://genebygene.com/wp-content/uploads/2019/08/GenebyGene_PrivacyPolicy.pdf*, accessed on 9 January 2021.

GeneWay 'FAQ – Frequently asked questions' (2020) available at *https://www.geneway.co.za/faq-frequently-asked-questions*, accessed on 22 June 2020.

GeneWay 'Home' (2020) available at *https://geneway.co.za/*, accessed on 22 June 2020.

GeneWay 'How it works' available at *https://geneway.co.za/i-want-get-tested-how-does-it-work*, accessed on 21 November 2020.

GeneWay 'I want to get tested, how does it work?' (2020) available at *https://www.geneway.co.za/i-want-get-tested-how-does-it-work*, accessed on 25 June 2020.

Gibbs, S 'What is "safe harbour" and why did the EUCJ just declare it invalid?' (6 October 2015) *The Guardian* available at *https://www.theguardian.com/technology/2015/oct/06/safe-harbour-european-court-declare-invalid-data-protection*, accessed 13 May 2021.

Giles, J 'Must I comply with the POPI Act?' (12 February 2020) *Michalsons* available at *https://www.michalsons.com/blog/must-i-comply-with -the-popi-act/41827*, accessed on 3 March 2021.

GMDN Agency 'What is GMDN' available at *https://www.gmdnagency.org/services/gmdn*, accessed on 7 December 2020.

Griffiths, AJF 'DNA sequencing' *Britannica* available at *https://www.britannica.com/science/DNA-sequencing*, accessed on 4 February 2021.

Grothaus, M 'How 23andMe is monetizing your DNA' (5 January 2015) *Fast Company* available at *https://www.fastcompany.com/3040356/what-23andme-is-doing-with-all-that-dna*, accessed on 21 March 2020.

Gutmann Koch, V 'Recreational genetics or research enterprise? Cloudy consent issues arising from direct-to-consumer genetic testing' (2 August 2018) *Bill of Health* available at *https://blog.petrieflom.law.harvard.edu/2018/08/02/recreational-genetics-*

or-research-enterprise-cloudy-consent-issues-arising-from-direct-to-consumer-genetic-testing/, accessed on 26 June 2021.

Harvard Women's Health Watch 'Direct-to-consumer genetic testing kits' (September 2010) *Harvard Health Publishing* available at *https://www.health.harvard.edu/newsletter_article/direct-to-consumer-genetic-testing-kits*, accessed on 5 February 2021.

Helix 'Helix Laboratory Platform granted the first and only FDA authorization for a whole exome sequencing platform' (11 January 2021) available at *https://www.helix.com/pages/helix-laboratory-platform-granted-fda-authorization*, accessed on 7 April 2021.

Helix 'Helix platform consent' (10 February 2020) available at *https://www.helix.com/pages/platform-consent*, accessed on 9 January 2021.

Helix 'Helix privacy policy' (30 November 2020) available at *https://www.helix.com/pages/privacy-policy*, accessed on 9 January 2021.

Helix 'Helix terms of service' (30 November 2020) available at *https://www.helix.com/pages/terms-of-service*, accessed on 9 January 2021.

Hinkley, E 'What is DNA analysis?' (21 March 2017) *DNA Testing Choice* available at *https://dnatestingchoice.com/en-us/news/what-is-dna-analysis*, accessed on 4 February 2021.

Holpuch, A 'FDA orders genetics company 23andMe to cease marketing of screening service' (25 November 2013) *The Guardian* available at *https://www.theguardian.com/science/2013/nov/25/genetics-23andme-fda-marketing-pgs-screening*, accessed on 15 October 2019.

HomeDNADirect 'About us' available at *https://www.homednadirect.co.za/about-us/*, accessed on 22 June 2020.

HomeDNADirect 'Terms and conditions – General' (24 May 2018) available at *https://www.homednadirect.co.za/terms-and-conditions/*, accessed on 23 June 2020.

Jaslow, R 'FDA warns 23andMe, tells genetic testing firm to halt sales' (25 November 2013) *CBS News* available at *https://www.cbsnews.com/news/fda-warns-23andme-tells-genetic-testing-firm-to-halt-sales/*, accessed on 25 October 2020.

Kapdi, N, S Sulaiman & D Hofmeyr 'South Africa: Transferring personal information across borders' (2 February 2016) *Mondaq* available at *https://www.mondaq.com/southafrica/data-protection/462800/transferring-personal-information-across-borders*, accessed on 4 July 2020.

KnowU 'About us' available at *https://knowudna.com/about-us/#labs*, accessed on 21 November 2020.

KnowU 'Home' available at *https://knowudna.com/*, accessed on 26 July 2020.

Kurth, HA 'South Africa's Protection of Personal Information Act, 2013, goes into effect July 1' (29 June 2020) *The National Law Review* available at *https://www.natlawreview.com/article/south-africa-s-protection-personal-information-act-2013-goes-effect-july-1*, accessed on 9 October 2020.

Laher, I 'The Consumer Protection Act and cross-border transactions' (9 November 2010) *Polity* available at *https://www.polity.org.za/article/the-consumer-protection-act-and-cross-border-transactions-2010-11-09*, accessed on 8 March 2021.

LexisNexis 'De minimis definition' available at *https://www.lexisnexis.co.uk/legal/glossary/de-minimis*, accessed on 10 May 2021.

MapmyGenome 'Terms and conditions' available at *https://mapmygenome.in/terms-and-conditions/*, accessed on 9 January 2021.

Mayo Clinic 'Genetic testing' (14 April 2020) available at *https://www.mayoclinic.org/tests-procedures/genetic-testing/about/pac-20384827*, accessed on 5 January 2021.

Merriam-Webster 'Autonomy' available at *https://www.merriam-webster.com/dictionary/autonomy*, accessed on 3 March 2021.

Merriam-Webster 'Body fluid' available at *https://www.merriam-webster.com/medical/body%20fluid*, accessed on 3 August 2020.

Merriam-Webster 'Ejusdem generis rule' available at *https://www.merriam-webster.com/legal/ejusdem%20generis%20rule*, accessed on 6 May 2021.

Merriam-Webster 'Implement' available at *https://www.merriam-webster.com/dictionary/implement*, accessed on 6 January 2021.

Merriam-Webster 'Pathology' available at *https://www.merriam-webster.com/dictionary/pathology*, accessed on 20 August 2020.

Merriam-Webster ‘Reagent’ available at *https://www.merriam-webster.com/dictionary/reagent*, accessed on 6 January 2021.

Mitchell, C ‘IP address’ (23 June 2020) *Investopedia* available at *https://www.investopedia.com/terms/i/ip-address.asp*, accessed on 6 August 2020.

Molla, R ‘Why DNA tests are suddenly unpopular’ (13 February 2020) *Vox* available at *https://www.vox.com/recode/2020/2/13/21129177/consumer-dna-tests-23andme-ancestry-sales-decline*, accessed on 15 April 2020.

Monty, S ‘South Africa: Things to consider when packaging your products’ (7 November 2011) *Mondaq* available at *https://www.mondaq.com/southafrica/consumer-law/149662/things-to-consider-when-packaging-your-products*, accessed on 15 March 2021.

MyHeritage ‘How do I collect and send back my DNA sample to the lab?’ available at *https://faq.myheritage.com/en/article/how-do-i-collect-and-send-back-my-dna-sample-to-the-lab*, accessed on 25 June 2020.

MyHeritage ‘Privacy policy’ (17 December 2020) available at *https://www.myheritage.com/privacy-policy*, accessed on 9 January 2021.

MyHeritage ‘Terms and conditions’ (2020) available at *https://www.myheritage.com/terms-and-conditions*, accessed on 5 October 2019.

Myriad ‘Myriad informed consent policies’ available at *https://myriadwomenshealth.com/consent-policies/*, accessed on 9 January 2021.

National Human Genome Research Institute ‘Privacy in genomics’ (24 February 2020) *NIH* available at *https://www.genome.gov/about-genomics/policy-issues/Privacy*, accessed on 3 March 2020.

National Practitioner Data Bank ‘Definitions’ (October 2018) *US Department of Health and Human Services* available at *https://www.npdb.hrsa.gov/guidebook/CDefinitions.jsp*, accessed on 6 May 2021.

Nature Research ‘Genetics research’ available at *https://www.nature.com/subjects/genetics-research*, accessed on 7 January 2021.

Nordling, L ‘A new law was supposed to protect South Africans’ privacy. It may block important research instead’ (22 February 2019) *Science* available at

https://www.sciencemag.org/news/2019/02/new-law-was-supposed-protect-south-africans-privacy-it-may-block-important-research#:~:text=South%20Africa's%20current%2C%202015%20research,%E2%80%9Caware%20of%20the%20purpose.%E2%80%9D, accessed on 17 February 2020.

Oppenheim, J 'Medicines and Related Substances Amendment Acts come into force' (9 June 2017) *Bowmans* available at *https://www.bowmanslaw.com/insights/pharmaceuticals-healthcare/medicines-related-substances-amendment-acts-come-force/*, accessed on 22 July 2020.

Parsons, M & L Crawford 'Malaysia publishes draft "White List" for personal data exports' (27 April 2017) *Hogan Lovells* available at *https://www.hoganlovells.com/en/publications/malaysia-publishes-draft-white-list-for-personal-data-exports*, accessed on 6 May 2021.

Paxman, R 'Direct-to-consumer genetic test results are questionable, research suggests' (6 June 2011) *BioNews* available at *https://www.bionews.org.uk/page_93001*, accessed on 17 October 2019.

Picard, A 'Controversial genetic self-testing kits coming to Canada' (2 October 2014) *The Globe and Mail* available at *https://www.theglobeandmail.com/life/health-and-fitness/health/genetic-self-testing-kits-to-come-to-canada/article20885678/?click=sf_globefb*, accessed on 7 December 2020.

Pollack, A 'FDA orders genetic testing firm to stop selling DNA analysis service' (25 November 2013) *The New York Times* available at *https://www.nytimes.com/2013/11/26/business/fda-demands-a-halt-to-a-dna-test-kits-marketing.html*, accessed on 15 October 2019.

Privacy International 'DNA and genetic data' available at *https://privacyinternational.org/learn/dna-and-genetic-data#:~:text=DNA%20holds%20the%20key%20to,through%20DNA%20or%20RNA%20analysis.*, accessed on 8 January 2021.

Rakel, RE 'Therapeutics' (2021) *Britannica* available at *https://www.britannica.com/science/therapeutics*, accessed on 6 May 2021.

Ramirez, N 'The EU-US Privacy Shield invalidated: What this means for you' (1 June 2021) *Osano* available at *https://www.osano.com/articles/privacy-shield-invalidated*, accessed on 11 June 2021.

Reisenberger, T 'Does the Consumer Protection Act apply to foreign retailers selling goods online to South Africa?' (20 November 2018) *Legalese* available at *https://legalese.co.za/does-the-consumer-protection-act-apply-to-foreign-retailers-selling-goods-online-to-south-africa/*, accessed on 8 March 2021.

SAHPRA 'About us' available at *https://www.sahpra.org.za/who-we-are/*, accessed on 5 May 2020.

SAHPRA 'Medical devices' available at *https://www.sahpra.org.za/medical-devices/*, accessed on 5 May 2020.

SAPA 'Consumer Protection Act has international implications' (26 July 2010) *Sunday Times* available at *https://www.timeslive.co.za/sunday-times/business/2010-07-26-consumer-protection-act-has-international-implications/*, accessed on 8 March 2021.

Science Learning Hub 'DNA extraction' (18 June 2009) available at *https://www.sciencelearn.org.nz/resources/2036-dna-extraction#:~:text=DNA%20extraction%20is%20a%20routine,see%20a%20stringy%20white%20mass.*, accessed on 4 February 2021.

Servick, K 'Frustrated U.S. FDA issues warning to 23andMe' (25 November 2013) *Science* available at *https://www.sciencemag.org/news/2013/11/frustrated-us-fda-issues-warning-23andme*, accessed on 15 October 2019.

Singh, A 'Data without borders: How to manage cross-border data transfers in South Africa' (7 November 2017) *Alt Advisory* available at *https://altadvisory.africa/2017/11/07/data-without-borders-manage-cross-border-data-transfers-south-africa/*, accessed on 4 July 2020.

Smith, M 'Marketing, manufacturing, packaging & labelling, advertising' (12 October 2018) *Pharma Boardroom* available at *https://pharmaboardroom.com/legal-articles/marketing-manufacturing-packaging-labelling-advertising-south-africa/*, accessed on 16 January 2021.

South African Revenue Service 'FAQ: Do I need to register as an importer if I buy personal goods from abroad e.g. from Amazon?' (9 March 2021) available at

https://www.sars.gov.za/faq/faq-do-i-need-to-register-as-an-importer-if-i-buy-personal-goods-from-abroad-e-g-from-amazon/, accessed on 7 May 2021.

Southern African Development Community 'Member States' available at *https://www.sadc.int/member-states/*, accessed on 18 January 2021.

The Wellness Revolution 'FAQ' available at *https://thewellnessrevolution.co.za/faq/*, accessed on 22 June 2020.

The Wellness Revolution 'How it works' available at *https://thewellnessrevolution.co.za/how-it-works/*, accessed on 22 June 2020.

Tomlinson, C 'IN-DEPTH: The tangled web of medical device regulation in SA' (3 September 2020) *Spotlight* available at *https://www.spotlightnsp.co.za/2020/09/03/in-depth-the-tangled-web-of-medical-device-regulation-in-sa/*, accessed on 5 December 2020.

US Food and Drug Administration 'Review memorandum: 23andMe Personal Genome Service (PGS) test' (6 April 2017) *FDA* available at *https://www.accessdata.fda.gov/cdrh_docs/pdf16/den160026.pdf*, accessed on 3 October 2019.

van Harmelen, J, R Boda & R Dullabh 'Genetic information: A new resource to be mined?' (20 October 2020) *International Law Office* available at *https://www.internationallawoffice.com/Newsletters/Healthcare-Life-Sciences/South-Africa/ENSafrica/Genetic-information-a-new-resource-to-be-mined*, accessed on 16 February 2021.

Werksmans Attorneys 'Local consumer law has international ramifications' (7 July 2010) available at *https://www.werksmans.com/legal-updates-and-opinions/local-consumer-law-has-international-ramifications/*, accessed on 8 March 2021.

World Health Organization 'Genetic research' available at *https://www.who.int/genomics/research/en/*, accessed on 7 January 2021.

Young, S 'Privacy versus confidentiality' (8 December 2017) *Office of the Saskatchewan Information and Privacy Commissioner* available at *https://oipc.sk.ca/privacy-versus-confidentiality/#:~:text=In%20terms%20of%20information%2C%20privacy,used%2C%20and%2For%20disclosed.&text=Confidentiality%20is%20the%20duty%20to,only%20to%20the%20extent%20possible.*, accessed on 20 April 2020.

TABLE OF FIGURES

CHAPTER 1

INTRODUCTION: DIRECT-TO-CONSUMER GENETIC TESTING IN CONTEXT

I BACKGROUND

The human genome 'encodes a sensitive yet heritable signature of an individual that is marked by genetic variation reflecting one's ancestry and disclosing one's susceptibility to health and diseases'.[1] The completion of the Human Genome Project,[2] as well as recent advances in science and technology have enabled genetic testing to be conducted inexpensively,[3] expeditiously, and directly by consumers.[4] This allows individuals access to their genetic information without the intervention of a healthcare practitioner[5] or genetic counsellor.[6] Direct-to-consumer genetic testing is defined as 'DNA-based testing…ordered by the consumer outside of established health-care delivery systems by for-profit vendors'.[7] It refers to

[1] The human genome constitutes all the genetic information of an individual, necessary for its functioning. Genetic information is encoded by chromosomes, which consist of DNA strands containing genes. Genes, made up of a sequence of nucleotides, are the 'basic unit of inheritance' and are examined when determining if an individual is a carrier of, or is predisposed to, a disease. However, it is not just genes, but also environmental factors, which influence genetic predisposition and determine an individual's phenotype – the outward manifestation of the genotype – which is expressed differently amongst individuals. Xinghua Shi & Xintao Wu 'An overview of human genetic privacy' (2017) 1387(1) *Ann NY Acad Sci* 62; Michael S Pepper 'The human genome and molecular medicine – Promises and pitfalls' (2010) 100(11) *SAMJ* 722; Agnar Helgason & Kári Stefánsson 'The past, present, and future of direct-to-consumer genetic tests' (2010) 12(1) *Dialogues Clin Neurosci* 63.

[2] In 2003, the Human Genome Project published the complete sequence of the human genome, which was proclaimed to transform medicine and assist in understanding disease. Stephanie Bair 'Direct-to-consumer genetic testing: Learning from the past and looking toward the future' (2012) 67 *Food & Drug LJ* 413.

[3] Gicheol Jeong 'Estimating the effects of consumer characteristics on the intention to use direct-to-consumer genetic testing' (2018) 7(3) *Health Policy Technol* 233; Nate C Apathy, Terri Menser, Lindsay M Keeran et al 'Trends and gaps in awareness of direct-to-consumer genetic tests from 2007 to 2014' (2018) 54(6) *Am J Prev Med* 806.

[4] I use the term 'consumers' throughout this dissertation to refer to those individuals who purchase and undergo direct-to-consumer genetic testing.

[5] A 'healthcare practitioner' refers to 'an individual who is licensed or otherwise authorized by a state to provide health care services; or any individual who, without authority, holds himself or herself out to be so licensed or authorized'. Under the Health Professions Act 56 of 1974 (Health Professions Act), a 'health practitioner' is 'any person, including a student, registered with the council in a profession registrable' in terms of the Health Professions Act. National Practitioner Data Bank 'Definitions' *US Department of Health and Human Services* October 2018, available at *https://www.npdb.hrsa.gov/guidebook/CDefinitions.jsp*, accessed on 6 May 2021.

[6] Genetic counsellors are health professionals with specialised education and training in medical genetics and counselling, who assist individuals in understanding the implications of the role of genetics in disease. Genetic counsellors practice client-centred, non-directive counselling and prepare individuals and their families for test results, ensuring that they receive the necessary support and treatment. Rajiv Sarin 'Ethics and clinical utility of direct-to-consumer genetic tests' (2015) 11(1) *J Can Res Ther* 1; Health Professions Council of South Africa (HPCSA) *Standards of Practice for Genetic Counsellors* (2013) 4.

[7] Monica A Giovanni, Matthew R Fickie, Lisa S Lehmann et al 'Health-care referrals from direct-to-consumer genetic testing' (2010) 14(6) *Genet Test Mol Biomarkers* 817. For a further definition of direct-to-consumer genetic testing, see M Yaneva-Deliverska 'Legal aspects of direct-to-consumer genetic tests' (2011) 17(1) *J of IMAB* 136.

deoxyribonucleic acid (DNA) tests for traits – medical or otherwise – that provide communication and interpretation of results directly to consumers, thus bypassing healthcare professionals.[8] However, there has been a recent change in the direct-to-consumer genetic testing model, with an increase in the number of companies requiring physician involvement before and/or after testing.[9] This means that the unrestrained freedom which consumers had previously in choosing their tests, collecting and sending their samples, and receiving their own results directly, is now somewhat limited by the need for professional oversight.

The direct-to-consumer genetic testing industry is dynamic – new companies have emerged, others have ceased to operate, and existing ones have altered their business models or the types of tests offered.[10] Millions of individuals undergo direct-to-consumer genetic tests annually and for a variety of reasons that range from obtaining information on ancestry[11] and discovering miscellaneous traits,[12] to exploring carrier status, health risks, or predisposition to particular diseases and conditions such as various cancers, Alzheimer's disease, and diabetes.[13]

[8] I use the term 'healthcare professional' throughout this dissertation to generally refer to both medical or healthcare practitioners as well as genetic counsellors. Jane Tiller & Paul Lacaze 'Regulation of internet-based genetic testing: Challenges for Australia and other jurisdictions' (2018) 6(24) *Front Public Health* 1; Heidi Carmen Howard, Sigrid Sterckx, Julian Cockbain et al 'The convergence of direct-to-consumer genetic testing companies and biobanking activities: The example of 23andMe' in Matthias Wienroth & Eugénia Rodrigues (eds) *Knowing New Biotechnologies: Social Aspects of Technological Convergence* 1 ed (2015) 60; Sarin op cit note 6 at 1.

[9] Howard et al op cit note 8 at 60. See also, Mary A Majumder, Christi J Guerrini & Amy L McGuire 'Direct-to-consumer genetic testing: Value and risk' (2021) 72 *Annu Rev Med* 153.

[10] Howard et al op cit note 8 at 60.

[11] Amy L McGuire, Barbara J Evans, Timothy Caulfield et al 'Regulating direct-to-consumer personal genome testing' (2010) 330(6001) *Science* 181; Teresa Pàmpols Ros, José Miguel García Sagredo, Antonio Pérez Aytése et al 'Directed to consumer genetic testing: Perspective from the Ethics Commission of the Spanish Society for Human Genetics' (2019) 153(1) *Med Clin (Barc)* 35; Stuart Hogarth & Paula Saukko 'A market in the making: The past, present and future of direct-to-consumer genomics' (2017) *New Genet Soc* 198. See also, Valerie Gutmann Koch & Kelly Todd 'Research revolution or status quo: The new common rule and research arising from direct-to-consumer genetic testing' (2018) 56(1) *Hous L Rev* 83.

[12] This includes discovering paternity and family relationships, partner compatibility, behaviour and personality, talent and athletic skills, miscellaneous traits for physical characteristics, such as earwax type or hair colour, ability to metabolise certain foods and drugs like statins. Kathy Hudson, Gail Javitt, Wylie Burke et al 'ASHG statement on direct-to-consumer genetic testing in the United States' (2007) 110(6) *Obstet Gynecol* 1392; McGuire et al op cit note 11 at 181; Pàmpols Ros et al op cit note 11 at 35; T Caulfield, NM Ries, PN Ray et al 'Direct-to-consumer genetic testing: Good, bad or benign?' (2010) 77(2) *Clin Genet* 102; Hogarth & Saukko op cit note 11 at 198. See also, Gutmann Koch & Todd op cit note 11 at 83; Tiller & Lacaze op cit note 8 at 1; Pascal Borry, Martina C Cornel & Heidi C Howard 'Where are you going, where have you been: A recent history of the direct-to-consumer genetic testing market' (2010) 1 *J Community Genet* 102.

[13] Hudson et al op cit note 12 at 1392; Tiller & Lacaze op cit note 8 at 1; McGuire et al op cit note 11 at 181; Hogarth & Saukko op cit note 11 at 198; Borry et al op cit note 12 at 102. See also, Pàmpols Ros et al op cit note 11 at 35; Caulfield et al op cit note 12 at 102; Yaneva-Deliverska op cit note 7 at 136; Gutmann Koch & Todd op cit note 11 at 83.

Health-related genetic tests[14] commonly fall into three categories – pharmacogenetic tests,[15] nutrigenetic tests,[16] and predictive tests. Relevant to this dissertation are predictive genetic tests, which aim to obtain personal risk assessments for the development of a specific disease or group of diseases,[17] based on an individual's genetic profile.[18] Such tests can be separated into two broad groups – tests for monogenic (or 'Mendelian') diseases and tests for polygenic (or complex) diseases.[19] Monogenic diseases are heritable and arise as a result of a single genetic mutation, such as Huntington's disease, cystic fibrosis, and Duchenne muscular dystrophy. Certain monogenic diseases display complete penetrance, meaning that individuals with the mutation *will* exhibit symptoms.[20] Genetic testing for these types of diseases is simple as there is a link between a mutation and the likelihood of disease development.[21] On the other hand, most heritable conditions are complex and multifactorial. Polygenic diseases do not follow Mendelian inheritance patterns and include conditions such as hypertension, Alzheimer's disease, prostate cancer, and cardiovascular diseases.[22] Variation in numerous genes combined with environmental factors, such as diet and lifestyle, may increase or decrease the likelihood of developing a condition.[23] This means that even if an individual has the disease-causing gene, they may never develop the polygenic disease that it is linked to. Many predictive direct-to-consumer genetic tests assess an individual's predisposition to polygenic, rather than monogenic, diseases which is currently preliminary and unsupported by rigorous scientific evidence[24] – risk estimates are attained from population studies, comparing

[14] Health-related genetic tests may be defined as tests that aim 'to predict risk of disease, screen for disease, direct clinical management, identify carriers, or establish prenatal diagnoses, clinical diagnoses, or prognoses in individual people or families'. KAB Goddard, J Robitaille, NF Dowling et al 'Health-related direct-to-consumer genetic tests: A public health assessment and analysis of practices related to internet-based tests for risk of thrombosis' (2009) 12(2) *Public Health Genom* 93.
[15] Pharmacogenetic tests offer information regarding the effectiveness and suitability of certain drugs. Bair op cit note 2 at 414.
[16] Nutrigenetic tests offer tailored information about lifestyle and nutrition to consumers, based on their genetic profile. Ibid at 414.
[17] Ibid at 414.
[18] Ibid at 416.
[19] In these conditions, the relationship between cause and effect is evident. In other words, if a mutation is present, it is probable that the individual will develop the condition. C Dandara, J Greenberg, L Lambie et al 'Direct-to-consumer genetic testing: To test or not to test, that is the question' (2013) 103(8) *SAMJ* 510.
[20] Bair op cit note 2 at 416.
[21] Marietjie Botes 'Direct-to-consumer genetic tests: A treacherous road' *Caveat Legal* 2015, available at *http://www.caveatlegal.com/direct-to-consumer-genetic-tests-a-treacherous-road/*, accessed on 2 June 2020.
[22] Dandara et al op cit note 19 at 511; Bair op cit note 2 at 416.
[23] No sole genetic variant, or a few variants, can precisely determine disease risk. Dandara et al op cit note 19 at 510.
[24] Botes op cit note 21.

individuals who possess a particular genetic variant with those who do not[25] – thus such testing merely anticipates heightened risk.

Direct-to-consumer genetic testing differs from other forms of genetic testing, both in terms of its business model – which tends to focus on building large databases for research[26] – and its availability beyond the medical field.[27] Advancements in genetic testing technology, coupled with the falling cost of testing, have led to an increase in the emergence of direct-to-consumer genetic testing companies (testing companies).[28] The internet has made direct-to-consumer genetic testing significantly easier by making such tests publicly accessible[29] – anyone with internet access and a credit card can purchase a test.[30] Sample collection kits, containing a buccal swab or saliva collection tube, are mailed to consumers,[31] who then provide their saliva sample and return the sample via the post.[32] Certain testing companies offer a single test for one trait, others sell tests for a collection of traits, and some testing companies conduct genome-wide testing, which analyses multiple genetic variants, providing results for numerous traits.[33] In order to do so, DNA is extracted from the saliva sample and examined for many single nucleotide polymorphisms (SNPs), offering information about an individual's genetic makeup.[34] Test results can be viewed by consumers through their online account or on a mobile

[25] Ibid. See also, Dandara et al op cit note 19 at 511; Bair op cit note 2 at 416.
[26] Andrew S Robertson 'Taking responsibility: Regulations and protections in direct-to-consumer genetic testing' (2009) 24(1) *Berkeley Tech LJ* 218; Jennifer Cacchio 'What you don't know can hurt you: The legal risk of peering into the gene pool with direct-to-consumer genetic testing' (2018) 87 *UMKC L Rev* 224.
[27] Kate Sweeny & Angela M Legg 'Predictors of interest in direct-to-consumer genetic testing' (2011) 26(10) *Psychol Health* 1260.
[28] Although there are genetic testing companies that may not offer direct-to-consumer genetic testing, I use the abbreviated term 'testing companies' throughout this dissertation for the sake of brevity.
[29] Renato Mainetti, Serena Oliveri, Alessandra Gorini et al 'Usability testing of two mini-games and one serious game to educate people about genetics' *pHealth 16th International Conference on Wearable, Micro & Nano Technologies for Personalized Health* (10-12 June 2019 Genoa, Italy) 82.
[30] Barbara Prainsack 'The power of prediction: How personal genomics became a policy challenge' (2011) 40(4) *ÖZP* 401.
[31] Tiller & Lacaze op cit note 8 at 1.
[32] Borry et al op cit note 12 at 102.
[33] An example of a company who provides such testing is 23andMe. Howard et al op cit note 8 at 60.
[34] A SNP (pronounced 'snip') is a change at a sole point in a DNA sequence and is the most prevalent form of genetic variation. Testing companies utilise a 'SNP chip' to gather genetic information. Using research from relevant fields, SNPs are compared to the occurrence of conditions within the individual's gender, ethnicity, and age group in order to ascertain their predisposition to disease. Christian Michael Armstrong Holland, Edward Harry Arbe-Barnes, Euan Joseph McGivern et al 'The 10th Oxbridge varsity medical ethics debate – Should we fear the rise of direct-to-consumer genetic testing?' (2018) 13(14) *Philos Ethics Humanit Med* 1; Sharon A Thrush & Ruth McCaffrey 'Direct-to-consumer genetic testing: What the nurse practitioner should know' (2010) 6(4) *J Nurse Pract* 270; Chelsea Weiermiller 'The future of direct-to-consumer genetic testing: Regulation and innovation' (2015) 16(5) *NC JL & Tech* 138–9; Bair op cit note 2 at 415; Arthur A Daemmrich '23andMe: The business and ethics of personal genetics testing' (2015) *University of Kansas School of Medicine* 2.

app,[35] offering personalised details and risk estimates regarding their susceptibility to certain genetic conditions or traits.[36]

The proliferation of direct-to-consumer genetic testing has generated discussion on the ethical, legal, and social issues involved,[37] potential harms to consumers, as well as concerns regarding the commercialisation of genetic testing.[38] While direct-to-consumer genetic testing holds several advantages including autonomy,[39] involvement in healthcare decisions,[40] access to genetic information,[41] and greater privacy,[42] the field remains contentious. The questionable accuracy, utility, and legitimacy of tests, the absence of professional involvement, potential result misinterpretation, and privacy concerns surrounding genetic data[43] have brought direct-to-consumer genetic testing to the fore.

Prima facie, direct-to-consumer genetic testing may not appear to differ greatly from other medical tests in the clinical setting. However, three components make direct-to-consumer genetic tests distinct. Firstly, direct-to-consumer genetic tests can be accessed and undertaken by consumers without requiring a healthcare professional to conduct the test or interpret the results. Secondly, while traditional genetic testing in the clinical context is undertaken to obtain a conclusive diagnosis that enables treatment and management of disease, and addresses concerns regarding risks of genetic conditions,[44] direct-to-consumer genetic test results are generally not definitive, but rather show 'genetic propensity',[45] as additional factors such as

[35] Megan A Allyse, David H Robinson, Matthew J Ferber et al 'Direct-to-consumer testing 2.0: Emerging models of direct-to-consumer genetic testing' (2018) 93(1) *Symposium on Precision Medicine* 115; Prainsack op cit note 30 at 401.
[36] Margaret Curnutte 'Challenging the boundaries of medicine: Consumer culture and genetic testing' (2012) 20(2) *Medic* 140; Helgason & Stefánsson op cit note 1 at 65.
[37] Leigh Jackson, Lesley Goldsmith & Heather Skirton 'Guidance for patients considering direct-to-consumer genetic testing and health professionals involved in their care: Development of a practical decision tool' (2014) 31(3) *Family Pract* 348.
[38] Hogarth & Saukko op cit note 11 at 198.
[39] Serena Oliveri & Gabriella Pravettoni 'The disclosure of direct-to-consumer genetic testing: Sounding out the psychological perspective of consumers' (2016) 8(5) *Biol Med* 361.
[40] Dandara et al op cit note 19 at 511; Bermseok Oh 'Direct-to-consumer genetic testing: Advantages and pitfalls' (2019) 17(3) *Genomics Inform* 1.
[41] Jeong op cit note 3 at 233.
[42] Sweeny & Legg op cit note 27 at 1260; Cheryl Berg & Kelly Fryer-Edwards 'The ethical challenges of direct-to-consumer genetic testing' (2008) 77(1) *J Bus Ethics* 19.
[43] John Lynch, Ashley Parrott, Robert J Hopkin et al 'Media coverage of direct-to-consumer genetic testing' (2011) 20 *J Genet Counsel* 487; Peter A Chow-White, Maggie MacAulay, Anita Charters et al 'From the bench to the bedside in the big data age: Ethics and practices of consent and privacy for clinical genomics and personalized medicine' (2015) 17(3) *Ethics Inf Technol* 190; Linnea I Laestadius, Jennifer R Rich & Paul L Auer 'All your data (effectively) belong to us: Data practices among direct-to-consumer genetic testing firms' (2017) 19(5) *Genet Med* 513.
[44] A Krause 'New genetic testing technologies: Advantages and limitations' (2019) 109(4) *SAMJ* 207; Yaneva-Deliverska op cit note 7 at 135.
[45] Oh op cit note 40 at 2.

environment, lifestyle, and family history influence gene expression[46] – something which consumers may not fully appreciate.[47] Thirdly, besides its questionable accuracy, validity, and utility, direct-to-consumer genetic testing differs in the provision of informational support.[48] Not only does direct-to-consumer genetic testing provide vast quantities of data, but the interpretation of health-related tests remains ambiguous due to deficient information on environmental and other factors and uncertain gene-disease associations. While direct-to-consumer genetic tests may be valuable to consumers for purposes such as genealogy, it is unclear to what extent they offer medical information.[49]

One of the most prominent controversies surrounding direct-to-consumer genetic testing is the regulation of the industry.[50] The issue of regulating direct-to-consumer genetic testing has been a topic of discussion on numerous occasions and from a variety of different countries. Direct-to-consumer genetic testing has generated legal ambiguity as to which existing statutes may apply to the industry. Current legislation is often viewed as being applicable to genetic testing within the medical context, thus creating confusion when applying them to direct-to-consumer genetic testing[51] – which is offered outside of the traditional healthcare system, across multiple jurisdictions, and not only for health-related testing, but also for recreational purposes.[52] This has led many academics worldwide to assert that direct-to-consumer genetic

[46] Little is understood about the complexity of the human genome and the role of genetic (and non-genetic) factors in predisposition to disease. See, Ronnie Sandroff 'Direct-to-consumer genetic tests and the right to know' (2010) 40(5) *Hastings Center Rep* 25; Jess Buxton 'Call to improve accuracy of predictive genetic tests' *BioNews* 12 October 2009, available at *https://www.bionews.org.uk/page_91911*, accessed on 17 October 2019; Rosemary Paxman 'Direct-to-consumer genetic test results are questionable, research suggests' *BioNews* 6 June 2011, available at *https://www.bionews.org.uk/page_93001*, accessed on 17 October 2019; Oh op cit note 40 at 2.

[47] Jennifer A Gniady 'Regulating direct-to-consumer genetic testing: Protecting the consumer without quashing a medical revolution' (2008) 76(5) *Fordham L Rev* 2430–1. See also, Anne M Huml, Catherine Sullivan, Maria Figueroa et al 'Consistency of direct-to-consumer genetic testing results among identical twins' (2019) *Am J Med* 146.

[48] Yaneva-Deliverska op cit note 7 at 136.

[49] David Magnus, Mildred K Cho & Robert Cook-Deegan 'Direct-to-consumer genetic tests: Beyond medical regulation?' (2009) 1(2) *Genome Med* 1.

[50] Other controversies surrounding direct-to-consumer genetic testing include the questionable validity, accuracy, and utility of tests, the lack of genetic counselling, the inability of consumers to interpret their results, the scarce involvement of healthcare practitioners, and the impacts (both physical and psychological) that direct-to-consumer genetic testing will have on individuals.

[51] Pàmpols Ros et al op cit note 11 at 38; Paul G Sanfilippo, Lisa S Kearns, Philip Wright et al 'Current landscape of direct-to-consumer genetic testing and its role in ophthalmology: A review' (2015) 43 *Clin Exp Ophthalmol* 583.

[52] See, Mark Popovsky 'Exaggerated benefits and underestimated harms: The direct-to-market consumer genetic test market and how to manage it going forward' (2010) 8(2) *Dartmouth LJ* 79; Weiermiller op cit note 34 at 153; L Kalokairinou, HC Howard, S Slokenberga et al 'Legislation of direct-to-consumer genetic testing in Europe: A fragmented regulatory landscape' (2018) 9 *J Community Genet* 129–30; Sanfilippo et al op cit note 51 at 583.

testing is unregulated.[53] However, there is limited research on the regulation of direct-to-consumer genetic testing from a South African perspective.

II STATEMENT OF PURPOSE

This dissertation aims to map the current legal landscape relating to direct-to-consumer genetic testing in South Africa. This is done through a comprehensive legal analysis of South Africa's extant law relevant to the industry, and the issues associated therewith – with the intention of determining if direct-to-consumer genetic testing is legally governed in South Africa and how its various aspects function within the current legislative framework. The challenges and lacunas in South Africa's legal landscape are identified, and recommendations for improvement are made, where applicable. The following objectives assist in accomplishing the purpose of this dissertation –

1. To identify the relevant extant law in South Africa relating to direct-to-consumer genetic testing.
2. To determine if, and how, direct-to-consumer genetic testing is legally governed in South Africa – thus addressing whether direct-to-consumer genetic testing is unregulated (by legislation) in South Africa.
3. To analyse the application of current legislation and to make recommendations, where appropriate, for the possible improvement of South Africa's extant law that impacts on direct-to-consumer genetic testing.

This dissertation is necessary since one of the central debates surrounding direct-to-consumer genetic testing is the regulation of the industry. While direct-to-consumer genetic testing has become increasingly popular in the United States of America (US), Europe, the United Kingdom (UK), and Canada, it is yet to gain such traction in South Africa. Although most testing companies based in other jurisdictions do not ship their products to, or offer their services in, South Africa – meaning that individuals in this country are unable to order testing kits from overseas – several South African testing companies have begun to emerge in recent

[53] For further information on what has been said by certain academics, see Sivan Tamir 'Direct-to-consumer genetic testing: Ethical-legal perspectives and practical considerations' (2010) 18 *Med Law Rev* 219; Bair op cit note 2 at 413; Tiller & Lacaze op cit note 8 at 4; Popovsky op cit note 52 at 78–9; Bartha Maria Knoppers, Denise Avard & Heidi Carmen Howard 'Direct-to-consumer genetic testing: Driving choice?' (2010) 10(8) *Expert Rev Mol Diagn* 967.

years.[54] It is expected that this trend will only continue to grow. However, testing companies may operate without being aware of the law. Therefore, this dissertation examines South African legislation to ascertain whether they apply to direct-to-consumer genetic testing and, if so, how the provisions may be interpreted, and could affect the industry – and its implications for testing companies operating both in South Africa and abroad. This dissertation does not provide concrete recommendations for the regulation of direct-to-consumer genetic testing – it merely establishes which of South Africa's extant laws apply to direct-to-consumer genetic testing in order to form a foundation for further studies into the regulation of the industry, if necessary.[55]

While many different types of genetic tests are offered directly to consumers, this dissertation focuses solely on health-related direct-to-consumer genetic tests. This is because genetic tests focusing on health – disease risk in particular – have generated the most controversy and resulted in calls for the regulation of the direct-to-consumer genetic testing industry. Thus, this dissertation combines the inter-disciplinary fields of law and health, thereby enriching local academic knowledge in these areas. Clarifying the multi-layered, and sometimes overlapping, legal landscape relating to direct-to-consumer genetic testing in South Africa enables foreign, as well as local, testing companies operating in the country to be in a position to better understand the parameters within which they may legally function, in terms of offering genetic tests directly to the public and research conducted using the genetic data obtained from consumers. As the literature on direct-to-consumer genetic testing in South Africa is scarce, this dissertation aims to make a contribution to filling these knowledge gaps.

III RATIONALE

The field of medicine has transformed over the past decades. Services conventionally offered by healthcare professionals are now commercially available.[56] Medical devices and diagnostic

[54] These companies include: GeneWay, KnowU, DNAlysis, and The Wellness Revolution. HomeDNADirect and EasyDNA, and DNAlysis – although with offices in South Africa – are international companies. GeneWay 'Home' available at *https://geneway.co.za/*, accessed on 26 July 2020; KnowU 'Home' available at *https://knowudna.com/*, accessed on 26 July 2020; DNAlysis 'About' available at *https://dnalysis.co.za/about/*, accessed on 22 June 2020; The Wellness Revolution 'How it works' available at *https://thewellnessrevolution.co.za/how-it-works/*, accessed on 22 June 2020; HomeDNADirect 'About us' available at *https://www.homednadirect.co.za/about-us/*, accessed on 22 June 2020; EasyDNA 'About us' available at *https://www.easydna.co.za/about-us/*, accessed on 22 June 2020.

[55] Due to word restrictions and the focus of this dissertation, I do not undertake a specific comparative analysis of the regulation of direct-to-consumer genetic testing in foreign jurisdictions. But the regulatory landscape in other countries is mentioned where relevant.

[56] Curnutte op cit note 36 at 138.

tools such as pregnancy tests, thermometers, and sphygmomanometers, which could previously only be accessed through the healthcare system, can now be purchased by anyone who so desires. This is the route in which genetic testing appears to be moving – once only available in the clinical setting; such testing can now be accessed directly by consumers without the involvement of healthcare professionals. There is currently no data showing the interest in direct-to-consumer genetic testing in South Africa. However, it is expected that this industry will grow, and this technology will become more accessible and affordable in the country, as public awareness increases.[57]

Despite its growing prevalence, direct-to-consumer genetic testing remains greatly under-investigated in South Africa, illustrated by the fact that there has only been one article written on this topic.[58] Furthermore, direct-to-consumer genetic testing was raised as a problematic area in the Academy of Science of South Africa (ASSAf) Report on *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (ASSAf Report),[59] which committed several pages to this topic, and recommended that in order to protect the public, regulation is necessary.[60] However, no further guidance is provided for doing so and questions remain regarding how this should be put into practice.

There have also been multiple claims from various academics worldwide that direct-to-consumer genetic testing is unregulated.[61] This is because *nowhere is there any legislative instrument that deals only and comprehensively with direct-to-consumer genetic testing*[62] – what I shall refer to as 'dedicated' legislation. Yet, a lack of *dedicated* legislation governing

[57] Dandara et al op cit note 19 at 510.
[58] This article on direct-to-consumer genetic testing in the South African context is: Dandara et al op cit note 19 at 510–2. While another article has been written on direct-to-consumer genetic testing in South Africa, it focuses on whole-exome sequencing (WES) and is not directly relevant to this dissertation. This article is: Zané Lombard, Fiona Baine, Amanda Krause et al 'Implications of direct-to-consumer whole-exome sequencing in South Africa' (2016) 106(2) *SAMJ* 139–40. WES targets the protein-coding regions of the human genome, allowing the identification of differences in the protein-coding region of any gene, as opposed to in a few chosen genes. Testing companies typically utilise genotyping as opposed to sequencing to analyse DNA. Genotyping determines which genetic variants are possessed by an individual. Sequencing the whole genome of an individual is not yet feasible. 23andMe 'Difference between DNA genotyping & sequencing' available at *https://customercare.23andme.com/hc/en-us/articles/202904600-Difference-Between-DNA-Genotyping-Sequencing*, accessed on 17 January 2021.
[59] Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018).
[60] Ibid at 38.
[61] See, Tamir op cit note 53 at 219; Bair op cit note 2 at 413; Tiller & Lacaze op cit note 8 at 4; Popovsky op cit note 52 at 78–9; Knoppers et al op cit note 53 at 967.
[62] A 2017 study, which reviewed the regulation of health-related direct-to-consumer genetic testing in eleven countries (Austria, Belgium, Canada, France, Germany, Japan, Portugal, South Korea, Switzerland, the UK, and the US), found that none of them possess legislation specifically governing the industry. Rei Fukuda & Fumio Takada 'Legal regulations on health-related direct-to-consumer genetic testing in 11 countries' (2018) 48 *Kitasato Med J* 55.

direct-to-consumer genetic testing, does not mean that the industry is *unregulated*. On the contrary, direct-to-consumer genetic testing may fall within the ambits of a range of statutes. If the various aspects of direct-to-consumer genetic testing are sufficiently governed by extant laws, then *dedicated* legislation may not actually be necessary. At this point, the meaning of the term 'regulation' requires clarification. Regulation does not merely entail statutes and other legal instruments – this is termed 'statutory regulation'.[63] It includes additional legal mechanisms that influence direct-to-consumer genetic testing. Recommendations and position statements by various organisations may be deemed such an instrument. Clinical guidelines as well as physician and patient education can also be regulatory mechanisms.[64] But one cannot determine whether direct-to-consumer genetic testing is sufficiently regulated without considering the *legislation* that may be applicable. Although the term 'regulation' is broad, a comprehensive understanding of the direct-to-consumer genetic testing industry can only be obtained from examining the law. As it has not yet been done in South Africa, and given that it is imperative to clarify the legal landscape before proceeding with further research on direct-to-consumer genetic testing, this dissertation focuses on the applicability of South African legislation to direct-to-consumer genetic testing through a legal analysis of the current law relating to the industry – therefore concentrating on *statutory regulation*.

According to Thrush and McCaffrey, direct-to-consumer genetic testing gained momentum prior to the enactment of legislation or guidelines regarding its implications, accuracy, the manner in which such tests should be conducted, and the need for professional oversight.[65] It grew in popularity at such a rate that regulation was unable to keep pace – hence why its state of regulation remains largely unclear. Unlike many developed countries where direct-to-consumer genetic testing is prevalent, this is not the case in South Africa.[66] To avoid regulatory confusion in South Africa, now is the opportune time to examine the legal landscape relating to direct-to-consumer genetic testing. Furthermore, mapping South Africa's legal landscape prior to the direct-to-consumer genetic testing industry gaining momentum in this country will enable risks and challenges to be controlled in a fair manner that recognises the constitutional rights of citizens, whilst maintaining the integrity of the healthcare system and the medical profession.

---

[63] Caroline F Wright, Alison Hall & Ron L Zimmern 'Regulating direct-to-consumer genetic tests: What is all the fuss about?' (2011) 13(4) *Genet Med* 295–6.
[64] Ibid.
[65] Thrush & McCaffrey op cit note 34 at 273.
[66] Seon-Hee Yim & Yeun-Jun Chung 'Reflections on the US FDA's warning on direct-to-consumer genetic testing' (2014) 12(4) *Genomics Inform* 152.

IV RESEARCH QUESTIONS

This dissertation aims to answer the following main research question: *What is the current legal landscape in South Africa that impacts on direct-to-consumer genetic testing?* To answer this main research question, the following sub-questions are posed:

1. What statutory framework is relevant to direct-to-consumer genetic testing in South Africa?
2. Are direct-to-consumer genetic tests considered to be medical devices?
3. Can direct-to-consumer genetic testing be legally offered and undertaken in South Africa?
4. What is South Africa's legal position regarding the importing and exporting of direct-to-consumer genetic tests, as well as associated samples and data?
5. Can testing companies conduct research using genetic data gathered from consumers?

V LITERATURE REVIEW

The direct-to-consumer genetic testing landscape is complex and contentious, consisting of a variety of conflicting opinions and viewpoints regarding its benefits, drawbacks, usefulness in healthcare, and the manner in which this technology should be regulated, if at all. Most of the literature on direct-to-consumer genetic testing originates from the US and European contexts. As it is still a relatively new field, especially in South Africa, its full implications have not yet been established.[67] The following literature review involves a discussion of: (1) arguments in support of direct-to-consumer genetic testing; (2) concerns associated with direct-to-consumer genetic testing; (3) the issue of regulation; and (4) South Africa's legal landscape.

(a) Arguments in support of direct-to-consumer genetic testing

Although it has been widely criticised, direct-to-consumer genetic testing does possess several advantages which should not be overlooked.[68] Significantly, direct-to-consumer genetic testing promotes autonomy and consumer empowerment by allowing individuals to have greater

[67] See, Gemma R Brett, Sylvia A Metcalfe, David J Amor et al 'An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice' (2012) 20 *Eur J Hum Genet* 829.
[68] Elizabeth A Varga 'You want to do what? My mother's choice to have direct-to-consumer genetic testing' (2012) *J Genet Counsel* 385.

choice in terms of testing,[69] be better informed of genetic conditions,[70] actively involved in healthcare decisions,[71] and able to take proactive steps to improve their health.[72] Similarly, positive test results showing predisposition to disease may encourage individuals to undertake preventive measures and lifestyle changes, such as exercise or a healthy diet, which holds benefits for health generally.[73] Consumers do not need a medical reason to undergo direct-to-consumer genetic testing and can therefore choose to utilise such tests out of curiosity or interest.[74]

Direct-to-consumer genetic testing promotes convenience,[75] providing consumers with a private and easy means of obtaining genetic data[76] – whilst also democratising access to information.[77] An additional benefit is the affordability and speed of the tests compared to those conducted in the clinical context.[78] The saliva sample is collected non-invasively, unlike drawing blood, as is procedure at hospitals.[79] Direct-to-consumer genetic test results do not form part of an individual's medical record,[80] as would be the case with genetic testing in the clinical context[81] – allowing access to tests while avoiding the healthcare system and

[69] Direct-to-consumer genetic testing gives individuals the choice to decide which test they want to undergo, and what tests are valid, useful, and accurate based on their own view, rather than being told by a doctor. Dandara et al op cit note 19 at 511; Oh op cit note 40 at 1; Sanfilippo et al op cit note 51 at 579; Heidi C Howard & Pascal Borry 'Direct-to-consumer genetic testing: More questions than benefits?' (2008) 5(4) *Pers Med* 317.
[70] Dandara et al op cit note 19 at 511; Oh op cit note 40 at 1.
[71] Roxanne Mykitiuk 'Caveat emptor: Direct-to-consumer supply and advertising of genetic testing' (2004) 27(1) *Clin Invest Med* 25; The Lancet 'Direct-to-consumer genetic testing' (2012) 380 *The Lancet* 76.
[72] Dandara et al op cit note 19 at 511; Oh op cit note 40 at 1; Mykitiuk op cit note 71 at 25. For the positive impact that such tests may have on physicians, see Randy Hulshizer 'The impending impact of direct-to-consumer personal genomic services' (2010) 22(3) *ECRI Institute* 3; Majumder et al op cit note 9 at 157.
[73] Popovsky op cit note 52 at 68–9; Molly C Novy 'Privacy at a price: Direct-to-consumer genetic testing & the need for regulation' (2010) 2010(1) *U Ill JL Tech & Pol'y* 168.
[74] Paula Saukko 'State of play in direct-to-consumer genetic testing for lifestyle-related diseases: Market, marketing content, user experiences and regulation' (2013) 72 *Proc Nutr Soc* 57.
[75] Direct-to-consumer genetic testing can be accessed without doctor visits, medical prescriptions, or laboratory fees, and the saliva sample can be collected at home, thus minimising the costs and time associated with numerous appointments necessary to obtain a genetic test via the clinical route. G Lippi, EJ Favaloro & M Plebani 'Direct-to-consumer testing: More risks than opportunities' (2011) 65(12) *Int J Clin Pract* 1222; Berg & Fryer-Edwards op cit note 42 at 19; Borry et al op cit note 12 at 102.
[76] This is because individuals are not always required to access tests through healthcare professionals, meaning that test results do not form part of their medical record. Sweeny & Legg op cit note 27 at 1260; Berg & Fryer-Edwards op cit note 42 at 19.
[77] See, Pàmpols Ros et al op cit note 11 at 36; Sandra Soo-Jin Lee & LaVera Crawley 'Research 2.0: Social networking and DTC genomics' (2009) 9(6-7) *Am J Bioeth* 35–44 cited in David B Resnik 'Direct-to-consumer genomics, social networking, and confidentiality' (2009) 9(6-7) *Am J Bioeth* 45; Novy op cit note 73 at 168; Mykitiuk op cit note 71 at 25.
[78] Test results are delivered directly to consumers within a few weeks, generally without the need for a doctor's appointment. Dandara et al op cit note 19 at 511; Oh op cit note 40 at 1; Lippi et al op cit note 75 at 1222.
[79] Oh op cit note 40 at 1.
[80] It should be noted that this is so, unless a consumer chooses to approach their healthcare practitioner with their test results for interpretation or advice. Dianne Nicol, Meredith Hagger, Nola Ries et al 'Time to get serious about privacy policies: The special case of genetic privacy' (2014) 42 *Fed L Rev* 156.
[81] Howard & Borry op cit note 69 at 317–8.

maintaining privacy.[82] This also has the potential to lessen genetic discrimination as it prevents bodies such as insurance companies and employers from gaining access to an individual's personal genetic information.[83]

This technology can assist individuals to better manage their wellbeing and conserve much needed healthcare funds.[84] Through the collection and storage of genetic data,[85] which is invaluable for research,[86] direct-to-consumer genetic testing not only holds benefits for individuals, but it has the ability to advance science and medicine, leading to the future development of diagnostic and therapeutic modalities, and in understanding the role that genetic factors play in disease development.[87] Direct-to-consumer genetic testing has the potential to increase genetic literacy among the public,[88] heighten scientific discovery, and enhance knowledge regarding the correlation between genes and disease.[89]

(b) Concerns associated with direct-to-consumer genetic testing

Direct-to-consumer genetic testing is not free from controversy primarily due to potential human rights infringements and a perceived lack of regulation. While direct-to-consumer genetic testing may provide consumers with greater autonomy and responsibility,[90] the

[82] Berg & Fryer-Edwards op cit note 42 at 18–9.
[83] Ibid. See also, Alice K Hawkins & Anita Ho 'Genetic counseling and the ethical issues around direct to consumer genetic testing' (2012) 21 *J Genet Counsel* 370.
[84] If direct-to-consumer genetic testing is proven to successfully identify disease risk, it may result in preventative measures being taken and lessen demands for hospital-based testing – minimising strain on healthcare systems and reducing cost. Hulshizer op cit note 72 at 3.
[85] Oh op cit note 40 at 1; Howard et al op cit note 8 at 64.
[86] Oh op cit note 40 at 1.
[87] Lombard et al op cit note 58 at 139.
[88] Genetic literacy denotes an individual's basic knowledge of genetic science. It includes central genetic concepts like gene expression, transmission, and elementary awareness of how genes affect health. Given the vast amounts of information available on company websites, it is likely that individuals will learn about genetics. Yvette E Pearson & Yuping Liu-Thompkins 'Consuming direct-to-consumer genetic tests: The role of genetic literacy and knowledge calibration' (2012) 31(1) *J Public Policy Mark* 43; Amanda Field, Alyson Krokosky & Sharon F Terry 'Direct-to-consumer marketing of genetic tests: Access does not reflect clinical utility' (2010) 14(6) *Genetic Alliance* 731; Varga op cit note 68 at 385; Helgason & Stefánsson op cit note 1 at 67.
[89] Varga op cit note 68 at 385. See also, Dandara et al op cit note 19 at 511; Field et al op cit note 88 at 731; Oliveri & Pravettoni op cit note 39 at 361.
[90] Oliveri & Pravettoni op cit note 39 at 361.

analytical[91] and clinical validity,[92] and clinical utility[93] of such tests has been questioned.[94] Further, there is apprehension regarding the variability of test results,[95] consumer-patient protection,[96] unauthorised disclosure of information, genetic discrimination, the advancement of beliefs such as genetic determinism,[97] the overstated predictive ability of some tests,[98] consent processes,[99] burdening healthcare systems,[100] costs,[101] exaggerated advertising and

[91] Analytical validity refers to a test's accuracy in identifying the presence or absence of a specific genetic variant, and is often dependent on the quality of the laboratory conducting the test. Davit Chokoshvili, Danya F Vears & Pascal Borry 'Growing complexity of (expanded) carrier screening: Direct-to-consumer, physician-mediated, and clinic-based offers' (2017) *Best Prac Res Cl Ob* 2; Dandara et al op cit note 19 at 512; Margaret Curnutte & Giuseppe Testa 'Consuming genomes: Scientific and social innovation in direct-to-consumer genetic testing' (2012) 31(2) *New Genet Soc* 167. See also, Curnutte op cit note 36 at 141.

[92] Clinical validity denotes a test's ability to differentiate between individuals who possess, or will develop, a condition and those who will not. It measures the accuracy of a test in detecting or predicting disease risk. Chokoshvili et al op cit note 91 at 2; Dandara et al op cit note 19 at 512; Curnutte & Testa op cit note 91 at 167. See also, Popovsky op cit note 52 at 68; Helgason & Stefánsson op cit note 1 at 65; Curnutte op cit note 36 at 141.

[93] Clinical utility concerns the application of such tests and examines whether they are able to offer information regarding diagnosis, management, treatment, or prevention of disease. Clinical utility refers to the probability of test results improving patient outcomes. Dandara et al op cit note 19 at 512; Popovsky op cit note 52 at 68. See also, Curnutte op cit note 36 at 141.

[94] Concerns have been expressed regarding the lack of evidence supporting the value of direct-to-consumer genetic tests as they are commonly predicated on prefatory, unproven, or ambiguous scientific information. Yim & Chung op cit note 66 at 152; Ruth Saunders 'Legal implications of direct-to-consumer genetic testing for common diseases' (2010) 1 *QMLJ* 73; McGuire et al op cit note 11 at 181; Wright et al op cit note 63 at 299; Lippi et al op cit note 75 at 1224 & 1227; Anna Middleton, Álvaro Mendes, Caroline M Benjamin et al 'Direct-to-consumer genetic testing: Where and how does genetic counseling fit?' (2017) 14(3) *Per Med* 249; Kalokairinou et al op cit note 52 at 118. For further information regarding analytical and clinical validity and clinical utility, see Patricia J Zettler, Jacob S Sherkow & Henry T Greely '23andMe, the Food and Drug Administration, and the future of genetic testing' (2014) 174(4) *JAMA Internal Medicine* 493; Saukko op cit note 74 at 57.

[95] Justin P Annes, Monica A Giovanni & Michael F Murray 'Risks of presymptomatic direct-to-consumer genetic testing' (2010) 363(12) *N Engl J Med* 1101; Varga op cit note 68 at 385; Cinnamon S Bloss, Burcu F Darst, Eric J Topol et al 'Direct-to-consumer personalized genomic testing' (2011) 20(2) *Hum Mol Genet* 133; Daemmrich op cit note 34 at 5. See also, Stephany Tandy-Connor, Jenna Guiltinan, Kate Krempely et al 'False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care' (2018) *Genet Med* 5; Buxton op cit note 46; Paxman op cit note 46.

[96] Tamir op cit note 53 at 214.

[97] Genetic determinism refers to the view that DNA and genetic makeup defines an individual's qualities and is the sole determining factor in the development of a disease. Lynch et al op cit note 43 at 487; Berg & Fryer-Edwards op cit note 42 at 21. See also, Loredana Covolo, Sara Rubinelli, Elisabetta Ceretti et al 'Internet-based direct-to-consumer genetic testing: A systematic review' (2015) 17(12) *J Med Internet Res* 2; Karen P Powell, Whitney A Cogswell, Carol A Christianson et al 'Primary care physicians' awareness, experience and opinions of direct-to-consumer genetic testing' (2012) 21 *J Genet Counsel* 114.

[98] Jackson et al op cit note 37 at 341.

[99] Middleton et al op cit note 94 at 249; Jacqueline Savard, Chriselle Hickerton, Sylvia A Metcalfe et al 'From expectations to experiences: Consumer autonomy and choice in personal genomic testing' (2020) 11(1) *AJOB Empir Bioeth* 64.

[100] Direct-to-consumer genetic testing raises issues regarding downstream costs and burdens on already strained healthcare systems due to consumers approaching healthcare professionals for assistance in interpreting test results or requesting unnecessary follow-up visits or procedures. Kalokairinou et al op cit note 52 at 118; Caulfield et al op cit note 12 at 102; The Lancet op cit note 71 at 76. See also, Middleton et al op cit note 94 at 249.

[101] For example, wasting money on unnecessary medical procedures where test results were erroneous or misinterpreted by consumers, or spending money on inaccurate tests that lack analytical and clinical validity, thus yielding inconclusive information and leading to a non-use of results. Mainetti et al op cit note 29 at 82–3; Nuffield Council on Bioethics *Medical Profiling and Online Medicine: The Ethics of 'Personalised Healthcare' in a Consumer Age* (2010) 50.

marketing claims by testing companies,[102] psychological harm (such as misplaced anxiety or false reassurance) from erroneous test results,[103] the inability of consumers to interpret and understand test results,[104] the accuracy and usefulness of information provided to consumers, and the absence of adequate genetic counselling.[105] The abovementioned concerns have resulted in calls for the involvement of healthcare professionals and genetic counsellors in the testing process.

Further concerns include that because saliva samples are often collected by individuals with no professional assistance, contamination of the sample, and thus inaccurate results, are possible.[106] It also cannot be assured that the sample received by testing companies is that of the individual who purchased the test.[107]

Notwithstanding the above concerns, given that testing companies have turned their focus towards the collection of genetic data and the creation of research databases,[108] data privacy and control have become key issues related to direct-to-consumer genetic testing.[109] While increased privacy is viewed by some as a benefit of direct-to-consumer genetic testing, others question how testing companies will ensure the safety and privacy of genetic data.[110] There are real risks for consumers in direct-to-consumer genetic testing such as re-

[102] Advertisements by testing companies tend to focus solely on the benefits of such tests and overlook the risks and limitations associated therewith – thus preventing individuals from assessing their validity and ultimately impacting an individual's decision to undergo testing or not. Hogarth & Saukko op cit note 11 at 199; Oliveri & Pravettoni op cit note 39 at 361; Savard et al op cit note 99 at 64; Hawkins & Ho op cit note 83 at 368–9. See also, R Geransar & E Einsiedel 'Evaluating online direct-to-consumer marketing of genetic tests: Informed choices or buyers beware?' (2008) 12(1) *Genet Test* 14–5.

[103] Psychological impact may also occur where, for example, the test results are difficult to understand or where there is no treatment or cure available for a detected disease or condition. Varga op cit note 68 at 384; Nuffield Council on Bioethics op cit note 101 at 50.

[104] Without a healthcare professional, this may cause consumers to make unsuitable decisions regarding their health in terms of, for example, medications, treatments, lifestyle changes such as diet and exercise, future planning, and preventative measures like surgery to avoid breast cancer. Apathy et al op cit note 3 at 806.

[105] Traditionally, genetic testing entailed pre- and post-test counselling to minimise risks of result misinterpretation, misunderstanding the accuracy and limitations of a test, and potential harm to consumers. Testing companies differ in the provision of genetic counselling and whether it is undertaken by a certified genetic counsellor. Although certain companies may offer genetic counselling, others encourage consumers to refer to their personal physician, who may lack the time or knowledge to understand test results. Powell et al op cit note 97 at 113–4; Sweeny & Legg op cit note 27 at 1260; Dandara et al op cit note 19 at 512; Popovsky op cit note 52 at 72; Howard & Borry op cit note 69 at 319; Pàmpols Ros et al op cit note 11 at 36; Robertson op cit note 26 at 226.

[106] Thrush & McCaffrey op cit note 34 at 272.

[107] Ibid.

[108] Robertson op cit note 26 at 218; Heidi C Howard, Bartha Maria Knoppers & Pascal Borry 'Blurring lines: The research activities of direct-to-consumer genetic testing companies raise questions about consumers as research subjects' (2010) 11(8) *EMBO Rep* 581–2.

[109] See, Rachele M Hendricks-Sturrup & Christine Y Lu 'Direct-to-consumer genetic testing data privacy: Key concerns and recommendations based on consumer perspectives' (2019) 9(25) *J Pers Med* 1; Jackson et al op cit note 37 at 341.

[110] Lynch et al op cit note 43 at 487.

identification,[111] the storage and fate of data, future use and non-consensual sharing of data,[112] and withdrawing data from research.[113]

Since the emergence of direct-to-consumer genetic testing on mainstream markets, numerous contentions have been raised. Critics have called for extensive regulation of these services due to concerns alluded to above. However, proponents of direct-to-consumer genetic testing advocate for the positive impact that such tests have on consumers who utilise the service.[114]

(c) The issue of regulation

In the literature, there are divergent opinions regarding the type of regulation (if any) that is necessary for direct-to-consumer genetic testing, as well as the most suitable approach. Although there is agreement regarding the need for some form of regulatory review for direct-to-consumer genetic testing, there is disagreement as to how this should be achieved,[115] the extent of involvement by regulators, the appropriate minimum standards, and the need for legislation as opposed to self-governance or voluntary guidance.[116] Direct-to-consumer genetic testing is an international and internet-based industry, that is neither confined to a single jurisdiction nor to one set of legal norms, making potential regulation problematic.[117] Worldwide, legislation relating to genetic testing exists,[118] but there is no *dedicated* legislation for direct-to-consumer genetic testing.[119] Nonetheless, various facets of the industry may be governed by other distinct, yet overlapping, legal instruments.[120]

---

[111] Chow-White et al op cit note 43 at 190.
[112] See, Hendricks-Sturrup & Lu op cit note 109 at 2; Dandara et al op cit note 19 at 512; Knoppers et al op cit note 53 at 966.
[113] Laestadius et al op cit note 43 at 513.
[114] Karen Norrgard 'DTC genetic testing for diabetes, breast cancer, heart disease and paternity' (2008) 1(1) *Nat Education* 86.
[115] McGuire et al op cit note 11 at 181.
[116] Wright et al op cit note 63 at 295; Robertson op cit note 26 at 235.
[117] Although existing national legislation may apply to direct-to-consumer genetic testing in a specific country, given that direct-to-consumer genetic testing is offered to the public via the internet and across multiple jurisdictions (where regulation may be weaker) and where such testing may be governed in a different manner, such legislation cannot always be practically applied. For example, regarding data protection, electronic commerce, in vitro diagnostic medical devices, and consumer protection. Heidi Carmen Howard & Pascal Borry 'Europe and direct-to-consumer genetic tests' (2011) *Nat Rev Genet* 1; Kalokairinou et al op cit note 52 at 118.
[118] For example, the US Genetic Information Non-discrimination Act of 2008 122 Stat. 881 (GINA); Germany's Human Genetic Examination Act 2009, BGBL I at 2529; France's Loi n° 2004-800 rélative à la bioéthique; Austria's Gene Technology Act of 1995; Switzerland's Federal Act on Human Genetic Testing 2007; Hungary's Genetic Act 2008; and Portugal's Law n°12/2005 of 26 January 2005.
[119] Fukuda & Takada op cit note 62 at 55.
[120] Kalokairinou et al op cit note 52 at 118. For examples, see Howard & Borry op cit note 117 at 1; Pàmpols Ros et al op cit note 11 at 38.

While some academics focus on specific regulation of direct-to-consumer genetic testing,[121] a common logical mistake made in the literature is to describe direct-to-consumer genetic testing as 'unregulated' simply because there is no *dedicated* legislation that deals with it. As mentioned above, in the absence of *dedicated* legislation, direct-to-consumer genetic testing may be governed by other statutes. For instance, many countries have *general* consumer protection regulations, or *general* regulations regarding medical devices, which may include at least some aspects of direct-to-consumer genetic testing.

In this regard, Sheehan maintains that current laws protecting consumers are sufficient to regulate direct-to-consumer genetic testing, and that it should not be subject to additional types of regulation.[122] According to Sheehan, testing companies should be regulated in the same manner as other industries where the benefits of a product are promoted in order to increase sales and drive profit – claims must comply with consumer protection guidelines.[123] Issues such as misleading advertising and a lack of accurate information are not unique to direct-to-consumer genetic testing, and thus Vayena contends that the industry should follow consumer protection legislation.[124] However, Berg and Fryer-Edwards argue that direct-to-consumer genetic tests should be subject to more stringent oversight given that it comprises greater ethical concerns and may have potential health implications for consumers.[125] Authors in the literature have also recommended that safeguarding privacy requires improved consumer protection through well-founded policies, regulations, and laws.[126] Globally, various laws aim to protect the privacy of genetic information,[127] but the sufficiency of these measures has been questioned due to their limitations – especially when applied to the direct-to-consumer genetic testing context.

[121] Tamir op cit note 53 at 238.
[122] Mark Sheehan 'The right to know and genetic testing' (2015) 41(4) *J Med Ethics* 288.
[123] Ibid at 287.
[124] Effy Vayena 'Direct-to-consumer genomics on the scales of autonomy' (2015) 41(4) *J Med Ethics* 312.
[125] For example, while medical tests in the clinical context are expected to provide a definitive result, genetic tests merely offer probabilistic information regarding predisposition to disease. Berg & Fryer-Edwards op cit note 42 at 29.
[126] RB Altman 'Direct-to-consumer genetic testing: Failure is not an option' (2009) 86(1) *Clin Pharmacol Ther* 16; James W Hazel & Christopher Slobogin 'Who knows what, and when? A survey of the privacy policies proffered by U.S. direct-to-consumer genetic testing companies' (2018) 28 *Cornell J Law Public Policy* 66; Hendricks-Sturrup & Lu op cit note 109 at 5.
[127] For example, statutes such as the US's GINA and Health Insurance Portability and Accountability Act of 1996 110 Stat. 1936 (HIPAA); South Africa's Protection of Personal Information Act 4 of 2013 (POPIA); Canada's Personal Information Protection and Electronic Documents Act SC 2000 c. 5 (PIPEDA); Australia's Privacy Act 1988; the European Union's General Data Protection Regulations 2016/679 (GDPR); and the UK's Data Protection Act 1998 aim to protect individual privacy and genetic information.

From an examination of the literature, it is evident that the direct-to-consumer genetic testing landscape is currently fragmented. From a legal policy perspective, a balance must be struck between possible, although ambiguous, advantages, and potential harm that may be caused by direct-to-consumer genetic testing.[128]

(d) South Africa's legal landscape

As mentioned above, research on direct-to-consumer genetic testing has received little attention in South Africa.[129] Furthermore, the ASSAf Report, whilst emphasising that the regulation of direct-to-consumer genetic testing is needed, does not suggest how this should be approached or achieved.[130] Through the ASSAf Report's recommendation that direct-to-consumer genetic testing be regulated, the implication is that the industry is currently unregulated. The ASSAf Report does not consider South Africa's existing statutory framework that may be applicable to the direct-to-consumer genetic testing industry – something that this dissertation examines.

In their commentary on the ASSAf Report, Thaldar et al state that transforming South Africa into a 'vibrant bioscience research community' entails regulation,[131] in line with constitutional values.[132] Although Thaldar et al mention that developing the bioscience regulatory environment in South Africa entails an examination of existing legal authority, with respect being given to applicable constitutional rights,[133] direct-to-consumer genetic testing is not specifically mentioned. Additionally, the reply of Pepper et al to Thaldar et al does not explicitly discuss direct-to-consumer genetic testing.[134]

---

[128] The Lancet op cit note 71 at 76. See also, Bloss et al op cit note 95 at 138; Pascal Su 'Direct-to-consumer genetic testing: A comprehensive view' (2013) 86 *Yale J Biol Med* 363; Weiermiller op cit note 34 at 170.

[129] As already stated, there has only been one article written on this topic by Dandara et al. Dandara et al op cit note 19 at 512.

[130] The ASSAf Report notes that there is an absence of legislation, policy, regulations, or guidelines in the fields of genetics and genomics, but merely states that '[d]irect to consumer genetic marketing and testing must be regulated'. This shows that direct-to-consumer genetic testing is recognised as an industry in South Africa that requires regulation, but no further suggestion or approach to be followed for doing so effectively is provided. Another South African body that mentioned direct-to-consumer genetic testing is the Southern African Society for Human Genetics (SASHG), but they simply advised caution when using and interpreting such tests. ASSAf op cit note 59 at 34–5 & 38; Michael S Pepper, Collet Dandara, Jantina de Vries et al 'ASSAf consensus study on the ethical, legal and social implications of genetics and genomics in South Africa' (2018) 114(11/12) *S Afr J Sci* 1; The Specialist Forum 'Limitations of DIY genetic testing' (2019) 19(6) *The Specialist Forum* 43; Dandara et al op cit note 19 at 510.

[131] Donrich Thaldar, Julian Kinderlerer & Sheetal Soni 'An optimistic vision for biosciences in South Africa: A response to the ASSAf report on human genetics and genomics' (2019) 115(7/8) *S Afr J Sci* 1.

[132] Ibid.

[133] While the ASSAf report refers to the Constitution of the Republic of South Africa 1996, it does not undertake an in-depth legal analysis of the relevant rights and legal precedent. Thaldar et al op cit note 131 at 1.

[134] Michael S Pepper, Collet Dandara, Jantina de Vries et al 'An optimistic vision for biosciences in South Africa: Reply to Thaldar et al (2019)' (2019) 115(7/8) *S Afr J Sci* 1.

As there has only been one article written on direct-to-consumer genetic testing in South Africa, it is imperative to consider this. In this article, according to Dandara et al, there is an 'absence of regulation' of direct-to-consumer genetic testing in South Africa, and Africa as a whole.[135] Dandara et al mention how the issue of regulation has been approached in other jurisdictions, referring to guidelines, statements, principles, and recommendations in the US, Europe, and the UK; legislation in Germany that essentially prohibits direct-to-consumer genetic testing;[136] and the US Genetic Information Non-discrimination Act of 2008 (GINA),[137] which guards against genetic discrimination in the contexts of health insurance and employment.[138] Perhaps the lack of guidelines and regulation in South Africa is due to the fact that direct-to-consumer genetic testing is not as prominent in this country as it is abroad. This is evidenced by the fact that abroad, specifically in the US, direct-to-consumer genetic testing is advertised and marketed commercially.[139] Furthermore, two of the major testing companies globally, 23andMe and AncestryDNA, do not offer their products and services in South Africa.[140] Yet, Dandara et al do not consider fully South African legislation that may be applicable to the various stages of the direct-to-consumer genetic testing process. In their recommendations, Dandara et al only propose revising South Africa's laws to avoid stigmatisation and discrimination by employers or insurance companies based on genetic testing.[141] Although legislation relating specifically to genetic testing in South Africa is lacking,[142] other laws that relate to health, medical devices, advertising, importing and

[135] Dandara et al op cit note 19 at 512.

[136] In 2009, Germany passed the Human Genetic Examination Act. It restricts the usage of genetic tests. Diagnostic and predictive genetic tests require the involvement of a qualified medical specialist, genetic counselling, and written informed consent from patients, thereby outlawing direct-to-consumer genetic testing where professional oversight is not mandatory. Fukuda & Takada op cit note 62 at 54; Kalokairinou et al op cit note 52 at 121. See also, David Clark 'Genetic exceptionalism and paternalism themes in new German legislation' *The Privacy Report* 2 September 2009, available at *https://theprivacyreport.com/2009/09/02/genetic-exceptionalism-and-paternalism-themes-in-new-german-legislation/*, accessed on 19 October 2019.

[137] 122 Stat. 881.

[138] However, GINA does not prevent insurers from using such information in life, disability, or long-term care insurance. Moreover, GINA does not apply to epigenetic information or to non-medical uses of genetic information, like ancestry testing. Popovsky op cit note 52 at 75.

[139] For example, in the US direct-to-consumer genetic testing is advertised on television and in magazines. Yim & Chung op cit note 66 at 152; Gutmann Koch & Todd op cit note 11 at 83.

[140] 23andMe 'What countries do you ship to?' available at *https://int.customercare.23andme.com/hc/en-us/articles/214806628-What-countries-do-you-ship-to-*, accessed on 2 January 2020; AncestryDNA 'Need to ship to another country?' available at *https://www.ancestry.com/checkout/MLI?rtype=85&flow=3*, accessed on 2 January 2020.

[141] Dandara et al op cit note 19 at 512.

[142] While Chapter 3 of the National Health Act 61 of 2003 (NHA) instructs the Director-General to provide for genetic services, there is no *dedicated* legislation that deals with these areas in South Africa. Section 21(b)(*vii*) of the NHA states that '[t]he Director-General must issue and promote adherence to, norms and standards on health matters, including – genetic services'. ASSAf op cit note 59 at 20.

exporting, and research may govern the direct-to-consumer genetic testing industry in South Africa – which is analysed in this dissertation.

Although Dandara et al mention forms of regulation for direct-to-consumer genetic testing in foreign jurisdictions, they appear to overlook South Africa, specifically its statutory framework. Dandara et al highlight that direct-to-consumer genetic testing, due to its international reach and given that testing companies and consumers may be based in different jurisdictions, necessitates guidelines from international bodies, universal regulations, and an agreed 'code of practice'.[143] However, Dandara et al do not consider those testing companies based in South Africa, as well as foreign testing companies that offer their products and services in the country, and the existing legal framework that may be applicable to them. Dandara et al also seem to focus primarily on guidelines and recommendations and do not consider the extant laws in South Africa that may be relevant to the direct-to-consumer genetic testing industry. Just because direct-to-consumer genetic testing can be accessed across multiple jurisdictions, this does not mean that one set of universal regulations or a code of practice is necessarily a viable solution, nor does it mean that statutory regulation should be neglected – countries have different resources, capacities, systems, rules, and legal frameworks. Therefore, I suggest that prior to developing regulatory guidelines or codes, the direct-to-consumer genetic testing industry must be assessed in light of the legal framework of a specific country in order to determine how the industry is already governed by statute. From this, further (and possibly universal) regulation can be considered. I suggest that this is an oversight by Dandara et al that must be addressed, and is something that this dissertation intends to accomplish.

Given the above, there are several South African statutes (and their associated regulations) that may be relevant to direct-to-consumer genetic testing, and which are analysed throughout this dissertation, namely the National Health Act 61 of 2003 (NHA); the Protection of Personal Information Act 4 of 2013 (POPIA); the Medicines and Related Substances Act 101 of 1965 (as amended) (Medicines Act); the Consumer Protection Act 68 of 2008 (CPA); the Electronic Communications and Transactions Act 25 of 2002 (ECTA); the Health Professions Act 56 of 1974 (Health Professions Act); the Regulations Relating to the Use of Human Biological Material (Human Biological Material Regulations);[144] the Regulations Relating to Medical Devices and In Vitro Diagnostic Medical Devices (IVDs) (Medical Device

[143] Dandara et al op cit note 19 at 512.
[144] GN R177 GG 35099 of 2 March 2012.

Regulations);[145] the Regulations Regarding the General Control of Human Bodies, Tissue, Blood, Blood Products and Gametes (General Control Regulations);[146] the Regulations Relating to the Import and Export of Human Tissue, Blood, Blood Products, Cultured Cells, Stem Cells, Embryos, Foetal Tissue, Zygotes and Gametes (Import and Export Regulations);[147] the Material Transfer Agreement for Human Biological Materials (SA MTA);[148] the Draft Regulations Regarding the Use of Human DNA, RNA Cultured Cells, Stem Cells, Blastomeres, Polar Bodies, Embryos, Embryonic Tissue and Small Tissue Biopsies for Diagnostic Testing, Health Research and Therapeutics (Draft Testing and Research Regulations);[149] the Regulations Relating to Research with Human Participants (Human Research Participant Regulations);[150] and the Regulations Relating to the Taking of Buccal Sample or Withdrawal of Blood from a Living Person for Testing (Amendment) (Buccal Sample and Blood Withdrawal Regulations).[151] While there may be areas of overlap, these various pieces of legislation may cover different aspects of the direct-to-consumer genetic testing process and research. Therefore, it is necessary to examine if, how, and why these statutes apply to direct-to-consumer genetic testing in order to establish what stages in the process are covered and where gaps exist.

From a review of the literature, it is clear that direct-to-consumer genetic testing raises many controversial issues that cannot easily be overcome. This is partly because direct-to-consumer genetic testing does not fit into one clear category that can be easily defined. The entire process involves a range of actors from different fields,[152] numerous relevant areas,[153] and a variety of legal and ethical issues.[154] This is further exacerbated by the unclear legal landscape globally, as well as in South Africa, governing direct-to-consumer genetic testing.

[145] GN R1515 GG 40480 of 9 December 2016.
[146] GN R180 GG 35099 of 2 March 2012.
[147] GN R181 GG 35099 of 2 March 2012.
[148] GN R719 GG 41781 of 20 July 2018.
[149] GN R7 GG 29526 of 5 January 2007.
[150] GN R719 GG 38000 of 19 September 2014.
[151] GN R944 GG 34750 of 11 November 2011.
[152] This includes the consumers themselves, the testing companies who supply the tests to consumers, laboratories who extract and analyse the DNA, the genetic counsellors who are sometimes involved in assisting consumers with result interpretation and healthcare management, and the healthcare system that consumers may approach for help or require for further treatment or management of a condition.
[153] Such as the commercial consumer market, science, medicine, and the healthcare system. It involves a range of concepts including analytical validity, clinical validity, and clinical utility which each involve different actors. It blurs the boundaries between products and services, medicine and commercialisation, as well as the role of the healthcare practitioner and the individual.
[154] This includes the human rights implications of direct-to-consumer genetic testing such as autonomy, privacy, and informed consent as well as the issue of regulation, which seems to have dominated the direct-to-consumer genetic testing landscape.

Legislation exists to determine rights and duties, regulate certain industries, and protect individuals and society. Given the above concerns, by mapping the current statutory framework relating to direct-to-consumer genetic testing in South Africa, this dissertation aims to establish whether, how, and by which statutes direct-to-consumer genetic testing is legally governed in South Africa in order to determine the standards by which testing companies may function and be held accountable. Additionally, in the South African context and through this legal analysis, it must be ensured that direct-to-consumer genetic testing operates in a manner that gives effect to constitutional rights and values.

VI CONCEPTUAL FRAMEWORK

A positivist approach, based on an examination of South Africa's extant law in relation to direct-to-consumer genetic testing, is used as a conceptual framework for this dissertation. This, with the intention of clarifying the legal landscape and informing policy and legal development in South Africa regarding direct-to-consumer genetic testing.

VII RESEARCH METHODOLOGY

To address my research questions, this dissertation consists of desktop-based research examining primary and secondary sources of both print and electronic materials. While it would be beneficial to focus primarily on South African sources and academic research, such material is lacking in the field of direct-to-consumer genetic testing. Therefore, authority and information from other jurisdictions had to be consulted to obtain the requisite information.

Primary sources include, but are not limited to, national, foreign, and international statutes and regulations; international agreements and treaties; case law from South Africa and other jurisdictions; national guidelines, policies, government documents, and regulations; recommendations from various organisations and professional bodies; position statements and official reports; interviews, presentations, and panel discussions; statistical and empirical research data; and research reports.

Secondary sources largely comprise of scientific, medical, and legal academic textbooks and journals, as well as journal articles; dissertations and theses; internet articles and sources; editorials and opinion pieces; online magazine articles; books; and reports.

VIII CHAPTER OUTLINE

(a) Chapter 1 – Introduction: Direct-to-consumer genetic testing in context

Chapter 1 forms the introduction to the topic – providing an overview of direct-to-consumer genetic testing and the focus of this dissertation. It includes the statement of purpose, rationale, research questions, a review of the literature, conceptual framework, and research methodology. Placing direct-to-consumer genetic testing in context is done in preparation for the subsequent analyses, which separate the various aspects involved into different chapters – in line with my main research question.

(b) Chapter 2 – Where does direct-to-consumer genetic testing fit into South Africa's legislative scheme?

This Chapter investigates where direct-to-consumer genetic testing fits into South Africa's legislative scheme – this includes the things used, and stages involved, in the direct-to-consumer genetic testing process, and forms a basis upon which subsequent analyses follow. Chapter 2 examines various statutes and their associated regulations, including the NHA, POPIA, and the Human Biological Material Regulations.

(c) Chapter 3 – Overcoming the first hurdle: Can consumers lawfully collect their own saliva samples?

Chapter 3 assesses whether consumers are 'competent persons' in terms of legislation, and able to lawfully collect their own saliva samples for the purpose of a direct-to-consumer genetic test. In doing so, this Chapter examines inter alia the NHA, the Human Biological Material Regulations, the General Control Regulations, the Buccal Sample and Blood Withdrawal Regulations, and the Draft Testing and Research Regulations.

(d) Chapter 4 – Direct-to-consumer genetic tests as medical devices

This Chapter determines whether direct-to-consumer genetic tests are medical devices, in vitro diagnostic devices (IVDs), or both – and, if so, how they should be classified according to the Medicines Act and the Medical Devices Regulations.

(e) Chapter 5 – Offering direct-to-consumer genetic tests to the public in South Africa

Chapter 5 explores the offering of direct-to-consumer genetic tests to the public in South Africa. This includes the licensing, registering, and selling of direct-to-consumer genetic tests

as well as their advertising, marketing, labelling, and disclosure. This Chapter examines the legal requirements involved through analysing inter alia the Medicines Act, the Medical Device Regulations, POPIA, the CPA, and ECTA.

(f) Chapter 6 – The importing and exporting of direct-to-consumer genetic tests

This Chapter establishes the legal landscape related to the importing and exporting of direct-to-consumer genetic tests, as well as the associated samples and data. This is done through an analysis of the NHA, POPIA, the Medicines Act, the Medical Device Regulations, and the Import and Export Regulations.

(g) Chapter 7 – Can direct-to-consumer genetic testing companies conduct research using genetic data collected from consumers?

Chapter 7 considers the research conducted by testing companies. This Chapter looks at consent, the collection and processing of information, privacy issues, and data transfer to determine whether testing companies are legally permitted to conduct research utilising genetic data gathered from consumers. To do so, Chapter 7 examines legislation relevant to research including the NHA, POPIA, the Human Biological Material Regulations, the Human Research Participant Regulations, and the SA MTA.

(h) Chapter 8 – Where to from here? Conclusion and recommendations

This Chapter forms the conclusion and summarises the main points that have been discussed throughout – providing an overview of the analyses presented in this dissertation – with the ultimate aim of answering the main research question. From a legal analysis of South African legislation relating to direct-to-consumer genetic testing, Chapter 8 encapsulates which of the identified statutes are applicable to various aspects of the direct-to-consumer genetic testing process and outlines how they apply to the industry. This Chapter also makes recommendations to address problematic areas of South Africa's extant law related to direct-to-consumer genetic testing.

CHAPTER 2

WHERE DOES DIRECT-TO-CONSUMER GENETIC TESTING FIT INTO SOUTH AFRICA'S LEGISLATIVE SCHEME?

I INTRODUCTION AND CHAPTER OVERVIEW

Clarifying the various stages that form part of the direct-to-consumer genetic testing process assists in determining where the industry is positioned. In this Chapter, I investigate exactly how direct-to-consumer genetic testing fits into South Africa's legislative framework. This will determine which South African statutes are relevant to the industry and what activities direct-to-consumer genetic testing consumers and companies are legally permitted to perform.

As the primary source of South African law relating to human biological material is the NHA as well as its relevant regulations, this forms a central focus throughout this Chapter. The pertinent regulations that are evaluated include the Human Biological Material Regulations, the Human Research Participant Regulations, and the Draft Testing and Research Regulations. In addition, this Chapter considers POPIA.

II DECONSTRUCTING DIRECT-TO-CONSUMER GENETIC TESTING

Direct-to-consumer genetic testing entails particular stages which make use of certain things – namely saliva, DNA, and genetic data. I use the term 'thing' (as opposed to 'substance' or 'material' for example) in the legal sense, that is, to denote 'any object or entity, separate and distinct from any other object or entity',[1] which can be both tangible and intangible. At the outset, it is necessary to lay out the direct-to-consumer genetic testing process to assist in understanding how it works, what *things* are used at which *stage*, and how this is impacted by South Africa's current legal landscape.

In order to undergo a direct-to-consumer genetic test, the first *thing* required is a saliva sample – which consumers provide by spitting into a tube or swabbing the inside of their cheek. This collection is the first *stage*, that occurs using a kit shipped to consumers by the testing company. The saliva sample is then sent back to the testing company (or their associated laboratory) and used to extract DNA.[2] DNA is the second *thing* used in direct-to-consumer

[1] Donrich W Thaldar & Bonginkosi Shozi 'The legal status of human biological material used for research' *Forthcoming in SALJ*.
[2] Saliva contains cells which contain DNA.

genetic testing, with its extraction being the second *stage*.[3] DNA extraction is a simple process undertaken in laboratories, which results in a tangible biological material. As DNA carries genetic information,[4] the extracted DNA is then sequenced and analysed to produce genetic data – which is the third *thing* used in the direct-to-consumer genetic testing process, with sequencing and analysis being the third and final *stage*.[5]

As these things used in the different stages of the direct-to-consumer genetic testing process, as well as the legal rules governing them differ, the flow diagram below (Diagram 1) illustrates the process through which saliva results in DNA, and DNA is used to obtain genetic data more clearly,[6] and makes sense when viewed in conjunction with the paragraphs that follow.[7]

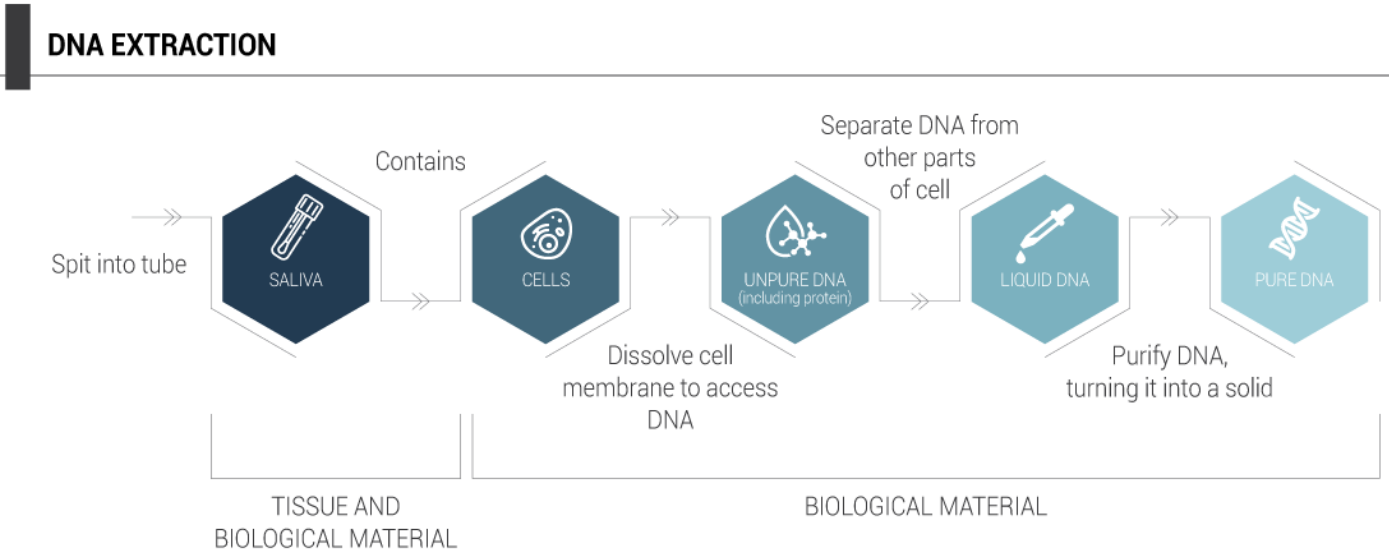
[3] DNA extraction is a method of purifying DNA by isolating it from the nucleus of cells. Tissues, including body fluids, are used in DNA extraction. The cells in a sample are split and the DNA released. The DNA is then separated from cellular debris. Once the DNA has been precipitated, it is cleaned. The extracted DNA can then be used for sequencing and analysis. Nalini Gupta 'DNA extraction and Polymerase Chain Reaction' (2019) 36(2) *J Cytol* 116; Science Learning Hub 'DNA extraction' 18 June 2009 available at *https://www.sciencelearn.org.nz/resources/2036-dna-extraction#:~:text=DNA%20extraction%20is%20a%20routine,see%20a%20stringy%20white%20mass.*, accessed on 4 February 2021.

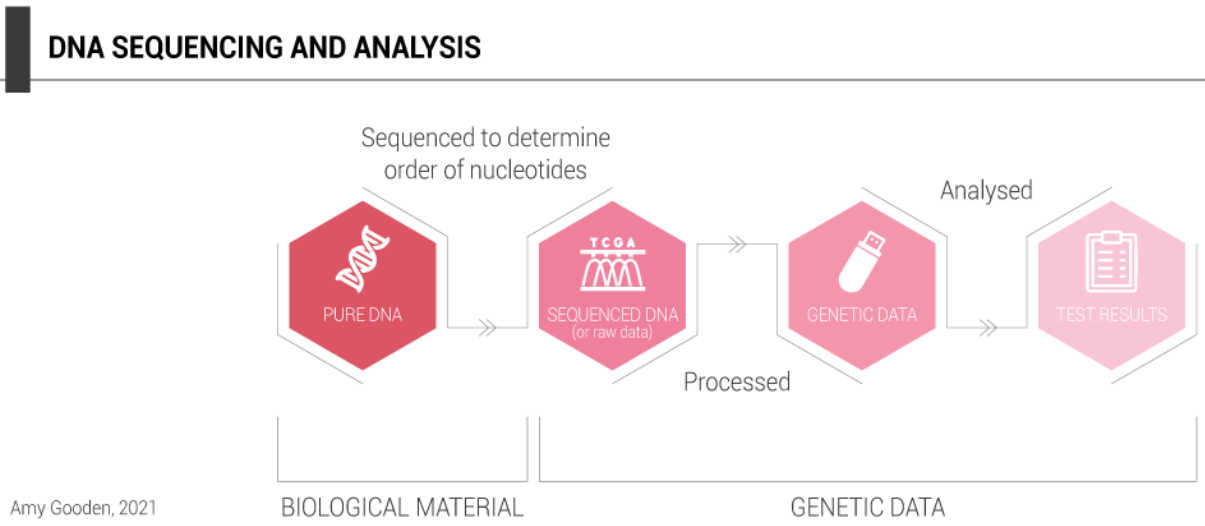
[4] William S Klug, Michael R Cummings, Charlotte A Spencer et al *Concepts of Genetics* 12 ed (2018) 6.

[5] DNA sequencing is a method for determining the sequence of nucleotide bases (adenine, guanine, cytosine, and thymine) in DNA, which differs amongst individuals. DNA analysis refers to interpreting genetic sequences, and is used to determine predisposition to disease. Biology Dictionary 'DNA sequencing' 4 October 2019 available at *https://biologydictionary.net/dna-sequencing/*, accessed on 4 February 2021; Anthony JF Griffiths 'DNA sequencing' *Britannica* available at *https://www.britannica.com/science/DNA-sequencing*, accessed on 4 February 2021; Ellen Hinkley 'What is DNA analysis?' *DNA Testing Choice* 21 March 2017, available at *https://dnatestingchoice.com/en-us/news/what-is-dna-analysis*, accessed on 4 February 2021.

[6] Extraction results in pure DNA; only once this DNA has been sequenced and analysed, is it genetic data.

[7] Adams et al refer to the 'exact point at which biological samples become personal information'. However, this should rather be thought of as a process than distinct stages, but rather as a process. As noted by Thaldar, referring to an 'exact point' is problematic. Firstly, DNA cannot become a genetic sequence – the sequence always existed in the DNA. Secondly, identifying an exact point is not in line with genetic research, which often involves the gradual gathering of information. Rachel Adams, Fola Adeleke, Dominique Anderson et al 'POPIA Code of Conduct for research' (2021) 117(5/6) *S Afr J Sci* 6; Donrich Thaldar 'Why POPIA does not apply to DNA' (2021) 117(7/8) *S Afr J Sci* 2.





III THINGS INVOLVED IN DIRECT-TO-CONSUMER GENETIC TESTING

Given that direct-to-consumer genetic testing involves the collection of saliva samples by consumers, the extraction and sequencing of DNA from this saliva, the analysis of the extracted and sequenced DNA by testing companies (or their associated laboratories), resulting in genetic data, which is used to determine predisposition to disease and for possible research, it is necessary to consider where these saliva samples and DNA, as well as genetic data, fit into South Africa's legislative scheme and, in doing so, human biological material becomes relevant.

Human biological material can either be understood in its general meaning, which is defined by Thaldar and Shozi as –

> '[A]ny biological material removed from a human being, or directly or indirectly derived from such removed biological material, including sub-cellular components (such as genetic material), cells, blood, tissues, organs, gametes (sperm and ova), embryos, foetal tissues, and waste (hair, nail clippings, urine, faeces, and sweat)'.[8]

Human biological material can also be understood in its legal technical meaning as defined in the Human Biological Material Regulations (discussed under heading (b) below). It should be noted that the definition given by Thaldar and Shozi is not provided for in any South African legislation and differs from that of 'biological material' in the Human Biological Material Regulations.[9] Yet, more than one definition of 'biological material' exists,[10] and differs somewhat based on the source one is looking at. Within this context, Thaldar and Shozi's definition can be seen as an attempt to provide a more comprehensive and complete definition of human biological material.

The primary source of law relating to human biological material (in its general meaning) is the NHA, as well as its various regulations. South Africa's legal framework makes use of two different lexicons to refer to human biological material (in its general meaning) and classes of such within the overarching concept.[11] The NHA does not refer to 'human biological material', but rather refers to numerous groups of things that meet the definition of human biological material – which are then used separately, and sometimes redefined, in the NHA's regulations – such as 'tissue', 'blood', 'blood products', and 'gametes'. To qualify as human biological material (in its general meaning), the thing must have derived, but be separated, from a human being.[12] Human biological material (in its general meaning) is relevant to direct-to-consumer genetic testing as saliva is the thing used in the first stage of the testing process, from which DNA is extracted and analysed. But are saliva, DNA, and/or genetic data, governed by South African legislation – in this context, specifically the NHA and its regulations? Under the sub-headings below, I consider tissue, biological material, and genetic data in turn in relation

[8] Thaldar & Shozi op cit note 1.
[9] Regulation 1 of the Human Biological Material Regulations.
[10] The Department of Health (DoH) *Ethics in Health Research: Principles, Processes and Structures* (DoH Ethics Guidelines) and the SA MTA define 'human biological materials' in the same manner as 'biological material' in the Human Biological Material Regulations. But the DoH Ethics Guidelines add 'blood and blood products' and the SA MTA includes 'and any modifications or derivatives thereof' to their definitions. Department of Health (DoH) *Ethics in Health Research: Principles, Processes and Structures* 2 ed (2015) 53; para 2.9 of the SA MTA.
[11] Thaldar & Shozi op cit note 1.
[12] This is implied by the words 'from a human being' in the definition of 'biological material'. Regulation 1 of the Human Biological Material Regulations.

to the South African legislation governing them, and determine whether saliva, DNA, or genetic data fall within these definitions.

(a) Tissue

Given its different lexicon, the NHA does not have a definition of human biological material, but it does contain a definition of 'tissue' which may be useful for present purposes. If any of the things at any stage of the direct-to-consumer genetic testing process meet the definition of 'tissue',[13] certain provisions in the NHA relevant to tissue may govern aspects of the direct-to-consumer genetic testing industry in South Africa.[14] For example, the NHA regulates the removal, use, and donation of inter alia tissue[15] – meaning that if a thing involved in direct-to-consumer genetic testing is not tissue, these activities are not covered by the NHA and, unless included in other legislation, such activities may be ungoverned. In section 1 of the NHA, 'tissue' is defined as –

> '[H]uman tissue, and includes flesh, bone, a gland, an organ, skin, bone marrow or body fluid, but excludes blood or a gamete'.[16]

Given the centrality of saliva in direct-to-consumer genetic testing,[17] I now determine whether saliva is included in the definition of 'tissue' in section 1 of the NHA. In order to do so, regard must be had to the ordinary meaning rule, which is closely linked to the literal approach to statutory interpretation, and entails understanding a statute's language based on its ordinary, grammatical meaning[18] – which is only departed from in order to avoid absurdity or when it is unreasonable and inconsistent with other provisions of a statute.[19] The rule that 'the language

[13] Section 1 of the NHA.
[14] For example, it may determine who can remove and use tissue and for which purposes this may be done.
[15] Sections 55, 56, and 63 of the NHA respectively. See also, section 59 of the NHA.
[16] Section 1 of the NHA. This is also followed by the General Control Regulations.
[17] Saliva contains the DNA that is extracted, sequenced, and analysed by testing companies in laboratories.
[18] *Union Government* (*Minister of Finance*) *v Mack* 1917 (AD) at 739 (*Mack*). In the case of *Mpisi v Trebble* 1994 (2) SA 136 (AD) at 9, it was held that '[t]he primary rule of statutory interpretation is to arrive at the intention of the Legislature having regard to the ordinary, grammatical meaning of the words of the enactment under consideration within their contextual setting'. This rule has been recognised in case law. See, *HMBMP Properties* (*Pty*) *Ltd v King* 1981 (1) SA 906 (N) at 909A; *Nyembezi v Law Society, Natal* 1981 (2) SA 752 (A) at 757H. LM Du Plessis 'Statute law and interpretation' in LM Du Plessis (ed) *The Law of South Africa* 2 ed 25(1) (2011) para 349.
[19] *Venter v R* 1907 TS 910 (TS) at 913. The position in South Africa that the plain meaning of words in a statute should be followed was reflected in the case of *Poswa v MEC for Economic Affairs, Environment and Tourism, Eastern Cape* 2001 (3) SA 582 (SCA) para 10 citing *Bhyat v Commissioner for Immigration* 1932 AD 125 at 129: 'In construing a provision of an Act of Parliament the plain meaning of its language must be adopted unless it leads to some absurdity, inconsistency, hardship or anomaly which from a consideration of the enactment as a whole a court of law is satisfied the Legislature could not have intended'. See also, *Volschenk v Volschenk* 1946 TPD 486 at 487; *Norden v Bhanki* 1974 (4) SA 647 (AD) at 510. Du Plessis op cit note 18 para 349.

of the Legislature should be read in its ordinary sense' assists in limiting the innumerable conceivable meanings that can be gleaned from an instrument's language.[20]

However, as legislation becomes increasingly technical, courts frequently utilise external aids, such as dictionaries, to determine the ordinary meaning of words.[21] In the field of medicine, 'tissue' is commonly understood as a group of cells designed for a specific function[22] – at an organisational level, tissue is between cells and organs.[23] However, the definition of 'tissue' in the NHA is both wider and narrower than its ordinary meaning, and is inclusive of glands, organs, and body fluid.[24] 'Body fluid' – as a likely category into which saliva falls – is not defined in the relevant legislation, hence regard must be had to its dictionary meaning. In this context, the definition of 'body fluid' generally refers to 'a fluid or fluid secretion (such as blood, lymph, saliva, semen, or urine) of the body'.[25] Adopting the ordinary meaning, the definition of 'body fluid' includes saliva. But dictionaries commonly provide several potential meanings that are context dependent.[26] While external aids are useful,[27] the court in *Loryan (Pty) Ltd v Solarsh Tea and Coffee (Pty) Ltd*[28] held that 'interpretation is not always fulfilled by recourse to a dictionary definition, for what must be ascertained is the meaning of the word in its particular context'.[29] In considering the context of this definition, it appears to suggest that 'human tissue' includes all human biological material, except specific categories that are

[20] *Mack* supra note 18 at 739; Lourens Du Plessis 'Interpretation' in Stuart Woolman, Michael Bishop & Jason Brickhill (eds) *Constitutional Law of South Africa* 2 ed (2008) 32–161.

[21] See, for example, *Secretary for Inland Revenue v Charkay Properties (Pty) Ltd* 1976 (4) SA 872 (A); *Roodepoort City Council v Shepherd* 1981 (2) SA 720 (A); *S v Weinberg* 1979 (3) SA 89 (A); *S v Crawford* 1979 2 SA 48 (A); *Waylite Diary CC v First National Bank Ltd* 1995 (1) SA 645 (A); *EMS Industries (Pty) Ltd v Inteletrack CC (Patent 2010/01326)* [2015] ZAGPPHC 696. In *Case v Minister of Safety and Security*; *Curtis v Minister of Safety & Security* 1996 (3) SA 617 (CC) para 58 (with reference to footnote 95 and *Minister of the Interior v Machadodorp Investments* (*Pty*) *Ltd* 1957 (2) SA 395 (A) at 360–1), the court held that 'resort to dictionaries is permissible in statutory interpretation'. Du Plessis op cit note 18 para 349; CJ Botha *Statutory Interpretation* 5 ed (2012) 123.

[22] Examples of tissues include epithelial tissue, muscle tissue, connective tissue, and nerve tissue. The British Medical Association *Complete Family Health Encyclopaedia* (1997) 995.

[23] Christopher K Mathews, KE van Holde, Dean R Appling et al *Biochemistry* 4 ed (2012) 67.

[24] Section 1 of the NHA.

[25] Merriam-Webster 'Body fluid' available at *https://www.merriam-webster.com/medical/body%20fluid*, accessed on 3 August 2020. See also, Collins Dictionary 'Body fluids' available at *https://www.collinsdictionary.com/dictionary/english/body-fluids*, accessed on 3 August 2020.

[26] Du Plessis op cit note 18 para 349.

[27] Botha op cit note 21 at 123.

[28] 1984 (3) SA (WLD).

[29] *Loryan (Pty) Ltd v Solarsh Tea and Coffee (Pty) Ltd* supra note 28 at 846G. Furthermore, in *Transvaal Consolidated Land and Exploration Co Ltd v Johannesburg City Council* 1972 (1) SA 88 (W) at 94G it was stated that '[d]ictionary definitions serve to mark out the scope of the meanings available for a word, but the task remains of ascertaining the particular meaning and sense of the language intended in the context of the statute'. In *De Beers Industrial Diamond Division (Pty) Ltd v Ishizuka* 1980 (2) SA 191 (T) at 452 it was emphasised that a 'dictionary meaning of a word cannot govern the interpretation. It can only afford a guide...The question is what is the meaning applicable in the context of the particular document under consideration'.

explicitly excluded. As such, I suggest that saliva is a 'body fluid' and thereby a type of 'human tissue', and thus meets the definition of 'tissue' in the NHA.[30] The implications of this are discussed below.

In the context of direct-to-consumer genetic testing and research (analysed in Chapter 7), a central question is: Does 'tissue' encompass individual cells or their sub-cellular components, such as DNA? The definition of 'tissue' in the NHA does not expressly mention DNA. But it is not a closed list, as alluded to by the word 'including' – which has generally been viewed by the courts as 'a word of addition, not of limitation'[31] – and indicates that it may encompass various other forms of 'tissue' not specifically listed in the definition.[32] Would DNA be one such example? In line with the conclusion reached by Thaldar and Shozi, I suggest not, as the laws relevant to tissue do not automatically apply to its derivatives.[33] As mentioned above, the definition of 'tissue' in the NHA is broader than its ordinary meaning. Yet, this does not open the floodgates to everything and there is no indication that this broadening covers DNA. A distinction must be made between: (1) things typically removed from individuals in the healthcare setting; and (2) things derived (directly or indirectly) from (1) in a laboratory, such as individual cells or their sub-cellular components.[34] To clarify, the legal rules relevant to saliva, apply to it as such. It does not mean that these rules also apply to components of saliva, like DNA.

[30] Although some direct-to-consumer genetic tests may use blood, urine, or hair samples, saliva is most common and is what this dissertation focuses on. Furthermore, as saliva samples sent by consumers to testing companies would generally be their own, rather than that of a non-human entity, it satisfies the 'human tissue' requirement under the definition of 'tissue' in section 1 of the NHA.

[31] *Attorney-General, Transvaal v Additional Magistrate for Johannesburg* 1924 AD 421 (A) at 430. See also, *Rosen v Rand Townships Registrar* 1939 WLD 5 (WLD) at 10; *Dibowitz v Commissioner for Inland Revenue* 1952 (1) SA 55 (AD) at 61; *Robb NO v Standard Bank Ltd* 1979 (2) SA 420 (R) at 428. See also, *Minister of Safety and Security v Xaba* 2003 (2) SA 703 (D) at 713 (*Xaba*).

[32] Section 1 of the NHA.

[33] Thaldar and Shozi illustrate this point with an example of a legal rule requiring vehicles to be licensed before being driven on public roads. Such a rule would apply to vehicles, not to components of the vehicle such as sparkplugs. Thaldar & Shozi op cit note 1.

[34] Ibid.

While the word 'including' is generally used to expand a defined term's meaning,[35] 'including' may also delimit a defined term's meaning in a narrower sense as its common meaning.[36] In our jurisprudence, the word 'include' can serve four functions[37] –

(1) The word 'include' is typically utilised in the definitions section of legislation to expand a defined word's meaning by including an object which the defined word's ordinary meaning does not encompass.[38]

(2) 'Include' may indicate that the defined word is defined exhaustively by the included objects, hence reducing the meaning of the defined word.[39] For example, in *Rex v Ah Tong*,[40] it was held that 'refreshment shops' were defined exhaustively by the included objects 'tea-rooms, cafés, restaurants, confectioners'.[41]

(3) Where there is difficulty in finding a suitable word that would, in its ordinary meaning, describe the group of objects that the legislature wishes to describe with one collective term, an unsuitable word may have been utilised by the legislature and defined through the inclusion of various objects.[42]

(4) The word 'include' may simply serve to clarify or illustrate the defined word with examples without reducing or expanding the defined word's meaning. The societal impact of a potential interpretation, and the broader legislative environment can inform this conclusion.[43]

The way that I suggest the word 'include' is used in the NHA's definition of 'tissue' is as a combination of functions (1) and (3) above. This is because the ordinary meaning of 'tissue'[44]

---

[35] *Rex v Ah Tong* 1919 AD 186 at 189. This was also cited in *R v Debele* 1956 (4) SA 570 (AD) at 575.
[36] This was the case in *Rex v Ah Tong* supra note 35 at 189–90, where it was held that '[t]he word "include" is often used in the definition of Acts of Parliament for the purpose of enlarging the meaning of a word or phrase by bringing it under something which is not comprehended under the ordinary meaning of that word or phrase. But…it is clear that this is not the sense in which the word is here used, for the shops enumerated are such as would ordinarily fall under the natural meaning of "refreshment shop". In this section the word is used not for the purpose of extending the meaning of the expression "refreshment shops", but for the purpose of enumerating the different kinds of shops which are intended to be comprehended under that denomination'. This was also quoted in *R v Debele* supra note 35 at 575 and *Xaba* supra note 31 at 713.
[37] The first three ways in which 'include' can be interpreted were provided by Southwood AJ in *Xaba* supra note 31 at 713. These were also mentioned in *Southern Life Association Ltd v Commissioner for Inland Revenue* 1985 (2) SA 267 (C) at 270 (*Southern Life Association*).
[38] *Xaba* supra note 31 at 713. See also, *Southern Life Association* supra note 37 at 270; *Rex v Ah Tong* supra note 35 at 189; *R v Debele* supra note 35 at 575.
[39] *Xaba* supra note 31 at 714. See also, *Southern Life Association* supra note 37 at 270.
[40] 1919 AD 186.
[41] *Rex v Ah Tong* supra note 35 at 189.
[42] *Xaba* supra note 31 at 714. See also, *Southern Life Association* supra note 37 at 270.
[43] *Southern Life Association* supra note 37.
[44] Section 1 of the NHA.

does seem relevant (hence (1)), but the ordinary meaning is both expanded to include other objects, such as organs, and reduced by the exclusion of blood.[45]

Applying the eiusdem generis rule[46] to the objects of the word 'include' under the definition of 'tissue' in the NHA,[47] I suggest that 'tissue'[48] is confined to its normal meaning and the list in the NHA's definition, and to things that are similar in nature as the examples listed – which DNA is not.[49] It does not go to the cellular level and precludes derivatives, such as individual cells or their sub-cellular components.[50]

An additional argument for DNA being excluded as a type of tissue, aided by the ordinary meaning rule, is that DNA is not tissue as the word is commonly understood. The ordinary meaning rule maintains that 'words should generally be given the meaning which the normal speaker of the English language would understand them to bear in the context'.[51] The Human Biological Material Regulations define DNA as a 'a nucleic acid, composed of building blocks called nucleotides'[52] – thus making DNA a chemical compound. Chemical compounds, due to their structure and nature, are not considered tissue in the ordinary sense of the word. The abovementioned definition of DNA views DNA as a chemical compound and not human tissue. In this respect, I suggest that DNA is not included in the NHA's definition of 'tissue'.[53]

To conclude this section, I have established that saliva, as a body fluid and based on its ordinary meaning, falls under the definition of 'tissue' in the NHA.[54] However, DNA – being a sub-cellular component and a derivative that is extracted from a saliva sample – extends

---

[45] Ibid.
[46] The eiusdem generis rule translates to 'of the same kind'. It is defined to mean that 'general words (as in a statute) that follow specific words in a list must be construed as referring only to the types of things identified by the specific words'. In terms of the eiusdem generis rule, the meaning of general words is limited by specific words in close proximity. Merriam-Webster 'Ejusdem generis rule' available at *https://www.merriam-webster.com/legal/ejusdem%20generis%20rule*, accessed on 6 May 2021; Annette Singh *The Impact of the Constitution on Transforming the Process of Statutory Interpretation in South Africa* (unpublished PhD thesis, University of KwaZulu-Natal, 2014) 171.
[47] Section 1 of the NHA.
[48] Ibid.
[49] Furthermore, the definition of 'tissue' as per section 1 of the NHA specifically refers to 'human tissue' and proceeds to list various examples of such. Therefore, in order to qualify as 'tissue' in accordance with the NHA's definition, DNA would need to be human tissue – is this the case? Although in this context the DNA used in direct-to-consumer genetic testing is that of a human, DNA is not tissue.
[50] Thaldar & Shozi op cit note 1.
[51] GE Devenish *Interpretation of Statutes* (1992) 26.
[52] Regulation 1 of the Human Biological Material Regulations. Regulation 1 of the Import and Export Regulations also provide the same definition of 'DNA'. Nucleic acid acts as a storehouse and transmitter of genetic information. Nucleic acid consists of nitrogenous bases, sugar, and phosphate. Mathews et al op cit note 23 at 91 & 109.
[53] Based on the definition of 'tissue' in section 1 of the NHA.
[54] Section 1 of the NHA.

beyond the normal meaning of 'tissue' as well as the NHA's definition.[55] Therefore, DNA is not considered to be 'tissue' according to the definition in the NHA.[56]

(b) Biological material

The Human Biological Material Regulations introduced the term 'biological material' into South African law, and thus shed some light on the purposes for which biological material may be removed and used – which may hold relevance for direct-to-consumer genetic testing. Given this, the question to be answered is whether DNA or saliva are deemed to be 'biological material'. The Human Biological Material Regulations define 'biological material' as –

> '[M]aterial from a human being *including* DNA, RNA, blastomeres, polar bodies, cultured cells, embryos, gametes, progenitor stem cells, small tissue biopsies and growth factors from the same' (own emphasis).[57]

Firstly, 'DNA' is a relevant term included in the definition of 'biological material'.[58] DNA is also defined separately in the Human Biological Material Regulations (see above).[59] The inclusion of DNA in the definition of 'biological material'[60] signifies two things: (1) DNA is a type of biological material; and (2) the definition of 'biological material' in the Human Biological Material Regulations,[61] unlike the definition of 'tissue' in the NHA,[62] is inclusive of derivatives and sub-cellular components, like DNA. In considering whether DNA is indeed a type of biological material, regard must be had to the principles of statutory interpretation.

The literal approach is not the only method of statutory interpretation in South Africa, and a purposive approach has been recognised by courts.[63] In *Bertie Van Zyl (Pty) Ltd v Minister for Safety and Security* (*Bertie Van Zyl*),[64] it was held that the Constitution of the

[55] Ibid.
[56] Ibid.
[57] Regulation 1 of the Human Biological Material Regulations. While 'tissue' is mentioned in this definition (although not defined), it refers specifically to 'small tissue biopsies' which have a different meaning to that of simple 'tissue'. It is interesting to note that regulation 1 of the Draft Testing and Research Regulations define 'biological material' as 'any material from a human being, including blood, cells, tissues, DNA, RNA, polar bodies, blastomeres, embryos and gametes'. Unlike the Human Biological Material Regulations, the Draft Testing and Research Regulations specifically include 'tissue' in the definition of 'biological material'. It must be questioned why 'tissue' was specifically included in this definition, but not in the Human Biological Material Regulations.
[58] Regulation 1 of the Human Biological Material Regulations.
[59] Ibid.
[60] Ibid.
[61] Ibid.
[62] Section 1 of the NHA.
[63] Singh op cit note 46 at 47.
[64] 2010 (2) SA 181 (CC).

Republic of South Africa, 1996 (the Constitution)[65] necessitates statutory interpretation to adopt a purposive approach,[66] particularly section 39(2) of the Constitution, which requires legislation to be interpreted in a way that advances the 'spirit, purport and objects of the Bill of Rights'.[67] *Jaga v Dönges NO; Bhana v Dönges NO*[68] expresses this as such –

> 'Certainly no less important than the oft repeated statement that the words and expressions used in a statute must be interpreted according to their ordinary meaning is the statement that they must be interpreted in the light of their context. But it may be useful to stress two points in relation to the application of this principle. The first is that 'the context', as here used, is not limited to the language of the rest of the statute regarded as throwing light of a dictionary kind on the part to be interpreted. Often of more importance is the matter of the statute, its apparent scope and purpose, and within limits, its background'.[69]

The Constitutional Court, in *Bato Star Fishing (Pty) Ltd v Minister of Environmental Affairs and Tourism* (*Bato Star*),[70] held that statutory interpretation requires one 'to have regard to the context within which the words occur, even where the words to be construed are clear and unambiguous'.[71] Therefore, despite the clear wording of the definition of 'biological material',[72] regard must be had to its purpose by considering its context. The Human Biological Material Regulations provide several purposes for which human biological material may be removed and used, including: (1) DNA-based genetic testing';[73] (2) health research;[74] and (3) studies on DNA 'obtained from human genetic material'.[75] As each of these purposes require DNA, it can be implied that if DNA were not a type of biological material, it would not be possible to achieve the above-mentioned purposes. Therefore, I suggest that, based on both a literal and purposive approach to statutory interpretation, DNA is a type of biological material.

Secondly, although saliva falls within the definition of 'tissue' in the NHA,[76] it must also be determined whether saliva meets the definition of 'biological material' in the Human Biological Material Regulations.[77] This is because the NHA stipulates who may remove and

---

[65] Act 108 of 1996.
[66] *Bertie Van Zyl* supra note 64 para 21.
[67] Ibid.
[68] 1950 (4) SA 653 (A).
[69] *Jaga v Dönges NO; Bhana v Dönges NO* supra note 68 at 662. Also cited in *Bertie Van Zyl* supra note 64 para 21.
[70] 2004 (4) SA 490 (CC).
[71] *Bato Star* supra note 70 para 91.
[72] Regulation 1 of the Human Biological Material Regulations.
[73] Regulation 5(a) of the Human Biological Material Regulations.
[74] Regulation 5(b) of the Human Biological Material Regulations.
[75] Regulation 5(c) of the Human Biological Material Regulations.
[76] Section 1 of the NHA.
[77] Regulation 1 of the Human Biological Material Regulations.

use *tissue* for certain purposes, while the Human Biological Material Regulations specify those permitted to remove and use *biological material* for particular purposes. If saliva is not included in the definition of 'biological material',[78] the Human Biological Material Regulations will not apply to saliva samples that are removed and used in direct-to-consumer genetic testing. But the Human Biological Material Regulations will apply to individual cells and sub-cellular components, such as DNA, that are extracted from such saliva samples, given that DNA is included in the definition of 'biological material'.[79]

The Department of Health (DoH) *Ethics in Health Research: Principles, Processes and Structures* (DoH Ethics Guidelines)[80] and the SA MTA,[81] although falling under research (dealt with in Chapter 7), use the definition of 'biological material' as it appears in the Human Biological Material Regulations (although these documents term it 'human biological materials'), but with the addition of their own examples. The DoH Ethics Guidelines add 'blood and blood products', and the SA MTA includes 'and any modifications or derivatives thereof' to the existing list of examples.[82] While these examples are not explicitly included in the definition of 'biological material' in the Human Biological Material Regulations,[83] they assist in clarifying its meaning, but do not alter the existing definition.[84] It can be seen that 'biological material' has a wider definition than that of 'tissue' in the NHA[85] – it is not confined to the list of examples in the definition.[86]

The question of whether tissue is a type of biological material has been debated by academics. Mahomed et al specifically state that the definition of 'biological material' in the Human Biological Material Regulations incorporates material from a human being – including tissue.[87] In his response, Jordaan uses the term 'human biological material' instead of 'human tissue' as used by Mahomed et al.[88] However, 'tissue'[89] and 'biological material'[90] fall under

[78] Ibid.
[79] Ibid.
[80] DoH op cit note 10. The DoH Ethics Guidelines are made legally binding through regulation 2(a) of the Human Research Participant Regulations, thus forming part of the NHA's subsidiary legislation.
[81] The SA MTA is given legal force by the NHA and mentions human biological material, making it relevant to consider in this Chapter.
[82] DoH op cit note 10 at 53; para 2.9 of the SA MTA.
[83] Regulation 1 of the Human Biological Material Regulations.
[84] Thaldar & Shozi op cit note 1.
[85] Section 1 of the NHA.
[86] Regulation 1 of the Human Biological Material Regulations.
[87] S Mahomed, M Nöthling-Slabbert & MS Pepper 'The legal position on the classification of human tissue in South Africa: Can tissues be owned?' (2013) 6(1) *SAJBL* 16.
[88] DW Jordaan 'Social justice and research using human biological material: A response to Mahomed, Nöthling-Slabbert and Pepper' (2016) 106(7) *SAMJ* 678.
[89] Section 1 of the NHA.
[90] Regulation 1 of the Human Biological Material Regulations.

two different lexicons and are distinct in terms of derivatives. The definition of 'biological material' expressly contains examples of such derivatives, like DNA.[91] This is strengthened by the definition of 'human biological materials' in the SA MTA, which includes 'any modifications or derivatives thereof' under the existing list of examples.[92]

While 'tissue' is not specifically defined in the Human Biological Material Regulations, the definition of 'biological material' requires such material to be 'from a human being'.[93] Furthermore, the word 'including' indicates that it is not a closed list and may incorporate other types of material from a human being.[94] In *Attorney-General, Transvaal v Additional Magistrate for Johannesburg*,[95] it was held that the word 'include' is 'generally a word of extension'.[96] I suggest that this widening includes tissue.[97] As tissue is material from a human being, as per the NHA,[98] I suggest that tissue (and therefore saliva) falls under the definition of 'biological material' in the Human Biological Material Regulations.[99] Adding to this suggestion is the fact that the NHA explicitly excludes blood and gametes from the definition of 'tissue'.[100] The definition of 'biological material' includes gametes, but does not mention blood.[101] However, blood is referred to under the definition of 'competent person' in the Human Biological Material Regulations in terms of the activities that such persons are permitted to undertake.[102] Because only a 'competent person' may remove and use biological material and, as blood is mentioned under the definition of 'competent person' in the Human

[91] Ibid.
[92] Para 2.9 of the SA MTA.
[93] Regulation 1 of the Human Biological Material Regulations.
[94] This is also in line with the principles of statutory interpretation. See, *Xaba* supra note 31 at 713–4.
[95] 1924 AD 421 (A).
[96] *Attorney-General, Transvaal v Additional Magistrate for Johannesburg* supra note 31 at 429 citing *Dilworth v Commissioner of Stamps* 1899 AC at 105. See also, *Rex v Ah Tong* supra note 35 at 189; *R v Debele* supra note 35 at 575; *Xaba* supra note 31 at 714. Moreover, in *Southern Life Association* supra note 37 at 270, the court held that where the general class encompasses the classes following the word 'include', it indicates that the object defines the general class 'exhaustively'.
[97] Although still in draft form, it is interesting to note that regulation 1 of the Draft Testing and Research Regulations define 'biological material' as 'any material from a human being, including blood, cells, tissues, DNA, RNA, polar bodies, blastomeres, embryos and gametes', therefore specifically including tissue in the types of material from a human being that qualify as 'biological material'. Unlike the Human Biological Material Regulations, the Draft Testing and Research Regulations provide that 'biological material' refers to *any* material from a human being.
[98] Section 1 of the NHA. The definition of 'tissue' in section 1 of the NHA mentions 'human tissue', meaning that tissue must be that of a human being. Similarly, the Human Biological Material Regulations require 'biological material' to be 'material from a human being' – as per the definition of 'biological material' in regulation 1 of the Human Biological Material Regulations.
[99] Regulation 1 of the Human Biological Material Regulations.
[100] Section 1 of the NHA.
[101] Regulation 1 of the Human Biological Material Regulations.
[102] Ibid.

Biological Material Regulations,[103] this implies that the withdrawal of blood is a type of withdrawal of biological material[104] – thereby including blood in the definition of 'biological material' in the Human Biological Material Regulations,[105] even though it is not specifically cited. I suggest that the same applies to tissue – although not explicitly mentioned in the definition of 'biological material',[106] it is included. This is further strengthened by the fact that regulation 4(1) of the Human Biological Material Regulations, although referring to deceased persons, explicitly mentions 'tissue'.[107] Thus, it can be inferred that tissue is included in the definition of biological material. Given the above, I suggest that tissue – and thus saliva – are types of biological material, in line with the definition in the Human Biological Material Regulations.[108] This means that because DNA is extracted from saliva samples of consumers – which is 'material from a human being',[109] as per the definition of 'biological material' in the Human Biological Material Regulations – and analysed in order to determine predisposition to disease, DNA is also a type of biological material.

In summary, in this section I established that DNA, due to it being included in the definition of 'biological material' in the Human Biological Material Regulations,[110] and given a purposive approach, is biological material as legally defined. Moreover, although neither tissue nor saliva are explicitly mentioned under the list of examples in the definition of 'biological material' in the Human Biological Material Regulations,[111] having regard to the principles of statutory interpretation, I suggest that tissue (and thereby saliva) is a type of biological material.

(c) Genetic data

The above sections have predominantly focused on saliva and DNA and whether they meet the definitions of 'tissue' in the NHA,[112] and/or 'biological material' in the Human Biological Material Regulations.[113] However, genetic data (also referred to as genetic information) is a

[103] Ibid.
[104] Regulation 1 of the Human Biological Material Regulations (definition of 'competent person') read with regulation 2(a) of the Human Biological Material Regulations.
[105] Regulation 1 of the Human Biological Material Regulations.
[106] Ibid.
[107] This appears under the heading 'removal of biological material from deceased persons'. Regulation 4(1) of the Human Biological Material Regulations.
[108] Regulation 1 of the Human Biological Material Regulations.
[109] Ibid.
[110] Ibid.
[111] Ibid.
[112] Section 1 of the NHA.
[113] Regulation 1 of the Human Biological Material Regulations.

further important component of the direct-to-consumer genetic testing process that warrants discussion. This is because genetic data is essential in providing consumers with the health-related information that they are paying for by undergoing testing, as well as in terms of the research activities of some testing companies. Therefore, in what follows I determine where genetic data falls in South Africa's legislative framework, and if genetic data is considered to be 'tissue' and/or 'biological material'.

Genetic data may refer to either: (1) information occurring naturally in the human body that is carried and stored by DNA;[114] or (2) information relating to an individual's inherited or acquired genetic characteristics derived from sequencing, processing, and analysing DNA samples,[115] and interpreting data.[116] The SA MTA defines 'data' as 'any information, including personal information in any form derived directly or indirectly during the conduct of research or clinical care'.[117] While genetic data is not specifically mentioned, the meaning of data is not limited as it includes '*any* information…in *any* form' (own emphasis).[118] Based on the rules of statutory interpretation and having regard to case law,[119] I suggest that this extends to genetic data. Adding to this is the fact that genetic data is derived from sequencing and analysing DNA, whether for the purposes of a direct-to-consumer genetic test or in research, thus also meeting the definition of 'data' in the SA MTA.[120]

As the meaning of genetic data has been clarified, it must firstly be established whether genetic data is 'tissue' in line with the definition in the NHA.[121] I have already concluded above

[114] Mathews et al op cit note 23 at 12; Klug et al op cit note 4 at 6.
[115] Genetic data contains both health and non-health-related information about an individual and their relatives. It is broader than the results of a direct-to-consumer genetic test and encompass numerous DNA markers, such as those linked to health and other conditions. Privacy International 'DNA and genetic data' available at *https://privacyinternational.org/learn/dna-and-genetic-data#:~:text=DNA%20holds%20the%20key%20to,through%20DNA%20or%20RNA%20analysis.*, accessed on 8 January 2021; Masha Shabani & Pascal Borry 'Rules for processing genetic data for research purposes in view of the new EU General Data Protection Regulation' (2018) 26 *Eur J Hum Genet* 149; 23andMe 'Terms of service' 30 September 2019 available at *https://www.23andme.com/about/tos/*, accessed on 9 January 2021; AncestryDNA 'AncestryDNA informed consent' 24 July 2018 available at *https://www.ancestry.com/dna/lp/informedconsent-v4-en*, accessed on 9 January 2021; Adams et al op cit note 7 at 6.
[116] Testing companies, such as 23andMe, who store information and perform research on the data of consumers, refer to this as 'genetic data'. 23andMe 'Research' available at *https://www.23andme.com/en-int/research/*, accessed on 4 May 2020; Helix 'Helix platform consent' 10 February 2020 available at *https://www.helix.com/pages/platform-consent*, accessed on 9 January 2021.
[117] Para 2.7 of the SA MTA.
[118] Ibid.
[119] In *R v Hugo* 1926 AD 268 at 271, the word 'any' was held to be a word of 'wide and unqualified generality', although it 'may be restricted by the subject matter or the context'. This was also cited in *Shaik v Minister of Justice and Constitutional Development* 2004 (3) SA 599 (CC) para 17 and *S v Williams* [2005] ZAWCHC 35 para 15.
[120] Para 2.7 of the SA MTA.
[121] Section 1 of the NHA.

that the meaning of 'tissue' excludes derivatives (such as DNA). Genetic data is obtained through DNA analysis and, as such, is a derivative of tissue. Further, given that 'tissue' is something tangible, which genetic data is not, I suggest that genetic data cannot be considered 'tissue' as per the NHA.[122]

The next question to answer is whether genetic data is biological material. The definition of 'biological material' in the Human Biological Material Regulations provides that such material must be '*from* a human being' (own emphasis).[123] Unlike the NHA's definition of 'tissue', this includes derivatives of material from a human being, such as DNA. However, incorporeal derivatives – for instance, an individual's genome sequence, extracted DNA, or genetic data – are not mentioned in the list in the definition of 'biological material'.[124] Also, in terms of its ordinary meaning, 'material' refers to a corporeal thing. Therefore, genetic data, which is incorporeal information that is attained through DNA analysis (see Diagram 1 above), is not 'biological material'. This is supported by the DoH Ethics Guidelines and the SA MTA, which both deal with biological material and data separately.[125] DNA is specifically included in the definition of 'biological material', suggesting that DNA is biological material, while genetic data is not.[126]

To complete this section: I suggest that genetic data is neither 'tissue' nor 'biological material' – rather, it may fall within the definitions of 'personal information' and 'special personal information' in terms of POPIA (to be determined below).

From the above analysis, I established that the things involved in direct-to-consumer genetic testing meet either the definition of 'tissue' in the NHA or the definition of 'biological material' in the Human Biological Material Regulations.[127] To conclude: Saliva falls within the definition of both 'tissue'[128] and 'biological material',[129] but not genetic data. DNA extracted from saliva meets the definition of 'biological material' *only*.[130] Once DNA has been extracted, sequenced, and analysed it results in genetic data. Genetic data is neither 'tissue' nor

[122] Ibid.
[123] Regulation 1 of the Human Biological Material Regulations.
[124] Ibid. However, the genetic information of an individual falls within POPIA's scope.
[125] While data may also be seen to be included under the SA MTA's definition of 'human biological materials', data is defined separately in the SA MTA. To add to this is the fact that the definition of 'materials' in para 2.13 of the SA MTA mentions human biological materials and data separately, indicating that they are two different things. DoH op cit note 10; para 2.7 of the SA MTA.
[126] Regulation 1 of the Human Biological Material Regulations.
[127] Section 1 of the NHA and regulation 1 of the Human Biological Material Regulations respectively.
[128] Section 1 of the NHA.
[129] Regulation 1 of the Human Biological Material Regulations.
[130] Ibid.

'biological material', but may meet the definition of 'personal information' under POPIA.[131] This means that any statutory provisions relating to either tissue, biological material, or genetic data are relevant to direct-to-consumer genetic testing and may shed some light on the purposes for which, and by whom, they can be used.

IV STAGES IN DIRECT-TO-CONSUMER GENETIC TESTING

Determining where direct-to-consumer genetic testing is positioned in South Africa's legal landscape not only requires establishing whether the various things used at different stages of the process fall within the definitions of 'tissue', 'biological material', or 'genetic data', but also determining the lawfulness of the collection and processing involved in direct-to-consumer genetic testing – and this is where POPIA becomes relevant. POPIA is South Africa's primary legal instrument dealing with the protection of, and access to, data.[132] POPIA safeguards personal information and thereby privacy, a right recognised under section 14 of the Constitution.[133] POPIA aims to give effect to this right, whilst balancing it against the right of access to information contained in section 32 of the Constitution.[134] POPIA places duties on those who request, collect, process, store, and use personal information.[135] POPIA is germane to direct-to-consumer genetic testing because not only may testing companies retain consumers' genetic data (obtained through DNA analyses), but they may also conduct research using it.

POPIA may be potentially relevant to direct-to-consumer genetic testing in three ways: (1) in terms of the collection of saliva samples by consumers; (2) in terms of the ensuing DNA

---

[131] Section 1 of POPIA.

[132] POPIA was promulgated on 26 November 2013. Since then, most of POPIA's provisions have been enforced. Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018) 56–7; Preeta Bhagattjee 'South Africa – Data protection overview' *Data Guidance* July 2020, available at *https://www.dataguidance.com/notes/south-africa-data-protection-overview*, accessed on 9 October 2020.

[133] C Staunton, R Adams, M Botes et al 'Safeguarding the future of genomic research in South Africa: Broad consent and the Protection of Personal Information Act No. 4 of 2013' (2019) 109(7) *SAMJ* 469.

[134] Section 32 of the Constitution states that:

'(1) Everyone has the right of access to –

(a) any information held by the state; and

(b) any information that is held by another person and that is required for the exercise or protection of any rights.

(2) National legislation must be enacted to give effect to this right, and may provide for reasonable measures to alleviate the administrative and financial burden on the state'.

Alt Advisory 'POPIA compliance' available at *https://altadvisory.africa/popia/#applicationofpopia*, accessed on 9 October 2020.

[135] Thulisile Buthelezi 'The Protection of Personal Information Act (POPIA) is here – A quick run down' *Shepstone & Wylie Attorneys* 7 August 2020, available at *https://www.wylie.co.za/articles/the-protection-of-personal-information-act-popia-is-here-a-quick-run-down/*, accessed on 9 October 2020.

extraction, sequencing, and analysis;[136] and (3) in terms of research conducted on genetic data. As research is dealt with in Chapter 7, it is necessary at this stage to establish whether, and how, POPIA applies to the DNA extraction, sequencing, and analysis that follows the collection of saliva samples by consumers.

(a) What constitutes 'personal information'?

To determine which things involved in direct-to-consumer genetic testing fall under the definition of 'personal information' in POPIA, the question to be answered is: Does POPIA cover saliva samples collected by consumers, extracted DNA, or genetic data?

Firstly, the meaning of 'personal information',[137] a term that is central to POPIA, must be established. If saliva, DNA, and genetic data are not 'personal information',[138] testing companies will not have to comply with the collection and processing provisions of POPIA. Under section 1 of POPIA, 'personal information' refers to 'information relating to an identifiable, living, natural person…including, but not limited to' inter alia an individual's biometric information.[139] POPIA includes a definition of 'biometrics', which is 'a technique of personal identification that is based on physical, physiological or behavioural characterisation including…DNA analysis'.[140] Unlike the European Union (EU) General Data Protection Regulation 2016/679 (GDPR), which defines 'genetic data',[141] POPIA does not specifically mention this term.[142] Given that DNA analysis produces genetic data, is genetic data deemed

[136] Undertaken by either testing companies or their associated laboratories.
[137] Section 1 of POPIA.
[138] Ibid.
[139] Ibid.
[140] Ibid.
[141] Article 4(13) of the GDPR defines 'genetic data' as 'personal data relating to the inherited or acquired genetic characteristics of a natural person which give unique information about the physiology or the health of that natural person and which result, in particular, from an analysis of a biological sample from the natural person in question'.
[142] Although the definition of 'genetic data' in the GDPR shares similarities with the definition of 'personal information' under section 1 of POPIA, genetic data is not specifically referred to.

to be 'personal information' in terms of POPIA?[143] In determining this, assistance may be sought from the meaning of 'biometrics' and, by extension, 'biometric information'.[144]

As mentioned above, 'biometrics' is the process of personal identification predicated on the categorisation of persons – including DNA analysis. While 'biometrics' refers to a technique, 'biometric information' is the result thereof.[145] As a brief excursus, Thaldar et al contend that in terms of health research, as 'personal information' in POPIA includes 'biometric information', and as 'biometrics' encompasses DNA analysis, it is likely that data will fall within POPIA's scope.[146] However, I suggest that DNA analysis cannot simply be equated with biometric information.[147] DNA analysis, included under the definition of

[143] It should be noted that there is other information that may fall within the definition of 'personal information' in POPIA. Section 1 of POPIA states that, 'personal information' also includes –

'(a) information relating to the race, gender, sex, pregnancy, marital status, national, ethnic or social origin, colour, sexual orientation, age, physical or mental health, well-being, disability, religion, conscience, belief, culture, language and birth of the person;

(b) information relating to the education or the medical, financial, criminal or employment history of the person;

(c) any identifying number, symbol, e-mail address, physical address, telephone number, location information, online identifier or other particular assignment to the person'.

When consumers undergo a direct-to-consumer genetic test, they generally register and create an account, and are required to fill out certain personal information about themselves, such as their name, email address, credit card information, physical address (for postage of the testing kit), and the website may gather their internet protocol address (IP address). An IP address is an identifying number linked to a specific computer or network. It enables computers to send and receive information when connected to the internet. This registration and personal information is stored by the testing company as part of their records and is generally assigned a randomised identification number and kept apart from genetic data to decrease the chances of identification. Moreover, consumers who consent to research may be required to provide additional information. For example, testing companies such as 23andMe encourage consumers to answer surveys that assist in the collection of lifestyle, phenotypic, and family information, that are then linked to their genetic data (with identifying information removed) and used for research. By providing information such as race, gender, sex, age, health, origin, email address, or location through the direct-to-consumer genetic testing process (either through registering, undergoing a test, or consenting to research), the above information falls within either subsection (a), (b), or (c) of the definition of 'personal information' under POPIA. Cory Mitchell 'IP address' *Investopedia* 23 June 2020, available at *https://www.investopedia.com/terms/i/ip-address.asp*, accessed on 6 August 2020; 23andMe 'Privacy' available at *https://www.23andme.com/privacy/*, accessed on 4 May 2020.

[144] Although not explicitly defined, 'biometric information' is a type of 'personal information' under section 1 of POPIA. If genetic data does not fall under 'biometric information' in the definition of 'personal information' in section 1 of POPIA, the following must be considered: Although the term 'genetic data' is specifically excluded from the definition of 'personal information' in section 1 of POPIA, when providing examples of 'personal information' POPIA states that it is 'including, but not limited to'. This suggests that it is not a closed list and the examples mentioned under the definition of 'personal information' in POPIA may extend to other types of information – one of which may be genetic data.

[145] DNA is extracted from saliva and converted into 'DNA data', a type of machine-readable biometric data. AncestryDNA 'Your privacy' 23 September 2020 available at *https://www.ancestry.com/cs/legal/privacystatement#genetic-information*, accessed on 9 January 2021.

[146] Donrich W Thaldar, Marietjie Botes & Annelize Nienaber 'South Africa's new standard material transfer agreement: Proposals for improvement and pointers for implementation' (2020) 21(85) *BMC Med Ethics* 4.

[147] The same way in which determining whether genetic data is included in POPIA, involves examining if it meets the definition of 'biometrics', as suggested by van Harmelen et al. Joanne van Harmelen, Ridwaan Boda & Rakhee Dullabh 'Genetic information: A new resource to be mined?' *International Law Office* 20 October 2020, available at *https://www.internationallawoffice.com/Newsletters/Healthcare-Life-Sciences/South-Africa/ENSafrica/Genetic-information-a-new-resource-to-be-mined*, accessed on 16 February 2021.

'biometrics' in POPIA,[148] refers to a technique; 'biometric information' under 'personal information' in POPIA is the result. Genetic data arises from biometrics; one cannot conflate genetic data with biometrics.

As such a technique is likely to alter the structure and composition of a thing as it occurs naturally (thereby excluding saliva and DNA),[149] it is the resulting information (biometric information), obtained from DNA analysis,[150] that is significant. But does biometric information include genetic data? As 'biometrics' includes DNA analysis,[151] and because the definition of 'personal information' in POPIA extends to 'biometric information'[152] – which is the information resulting from DNA analysis, namely genetic data – I suggest that POPIA applies to such and, in this regard, is relevant to direct-to-consumer genetic testing.[153]

Although noted above, it is worth briefly discussing that saliva is not 'personal information' in terms of POPIA. This is because inter alia POPIA requires personal information to relate to 'an identifiable, living, natural person'.[154] On its own, saliva is not innately identifiable. But when identifiable information relating to an individual (such as genetic data) is obtained from a saliva sample and recorded in some form, that information meets the definition of 'personal information' in POPIA.[155]

In terms of DNA, the strands of the double helix consist of nucleotides which,[156] when sequenced, provide health insights. DNA contains four different nucleotides – adenine (A), guanine (G), thymine (T), and cytosine (C) – each with a nitrogenous base. These bases on the two DNA strands form complementary pairs: A always pairs with T, and G always pairs with

[148] Section 1 of POPIA.
[149] Section 1 of POPIA requires that 'personal information' relates to an 'identifiable, living, natural person'. The usage of the words 'relating to' in the definition of 'personal information' in section 1 of POPIA indicates that such information does not need to come directly from such a person in its final form and can undergo various processes to alter its form, while still relating to a natural person. Therefore, while saliva and DNA may not qualify as 'personal information', it does not exclude things obtained through various extraction, sequencing, and analysis techniques that still relate to a natural person.
[150] As per the definition of 'biometrics' in section 1 of POPIA.
[151] Section 1 of POPIA.
[152] Ibid.
[153] However, as 'biometrics' excludes DNA, DNA is not regulated by POPIA. But the genetic data contained therein falls within POPIA's ambit. I disagree with Staunton and Moodley who, when recognising that POPIA includes biometric data, assert that this automatically extends to DNA. C Staunton & K Moodley 'Data mining and biological sample exportation from South Africa: A new wave of bioexploitation under the guise of clinical care?' (2016) 106(2) *SAMJ* 138.
[154] Section 1 of POPIA.
[155] Adams et al argue that identifiable biological samples (such as a fingerprint) create uncertainty regarding when biological samples become personal information. However, I disagree with this contention. In terms of direct-to-consumer genetic testing, the process is clear. Once DNA has been extracted, sequenced, analysed and recorded, it is 'personal information' in terms of POPIA. Adams et al op cit note 7 at 6.
[156] Klug et al op cit note 4 at 6 & 213.

C.[157] When paired in this specific way, hydrogen bonds are formed between the complementary nitrogenous bases on opposite DNA strands.[158] These four bases, in different sequence combinations, encode genetic information.[159]

Genetic mutations associated with certain conditions (such as cystic fibrosis, sickle cell anaemia, and Tay-Sachs disease) are due to changes in the nucleotide sequence at specific sites on the chromosome.[160] Depending on the point on the gene at which the mutation occurs, this can potentially result in a phenotypic presentation of a health disorder. However, some mutations change the DNA sequence of a gene without altering its function, meaning that it neither results in the development of health-related conditions, nor does it express itself in an individual. Given the above, DNA is a 'container' for genetic information,[161] that determines the 'physical characteristics' of an individual,[162] some of which relate to health. But DNA itself is precluded from regulation by POPIA.

POPIA applies to the processing of personal information 'entered in a record by or for a responsible party' through automated or non-automated means.[163] 'Record' is defined in POPIA as information recorded in any form or medium, including information produced, recorded, or stored using inter alia computer equipment (hardware, software, or both) or another device, as well as any material thereafter obtained from this information.[164] Information occurring naturally in the human body is not 'entered' or 'recorded' by a

[157] Mathews et al op cit note 23 at 101.
[158] Ibid at 98–9; Klug et al op cit note 4 at 6 & 213.
[159] Klug et al op cit note 4 at 6.
[160] Genes are DNA sequences existing at a specific site in the chromosome. Ibid at G–7.
[161] Mathews et al op cit note 23 at 4 & 12; Klug et al op cit note 4 at 1 & 6; S de Wet, H Oosthuizen & J Visser 'DNA profiling and the law in South Africa' (2011) 14(4) *PER/PELJ* 171. See also, *S v Maqhina* 2001 (1) SACR 241 (T).
[162] *S v Maqhina* supra note 161. Heredity and development are determined by genetic information located in genes, which are found in chromosomes – a structure made up of DNA and proteins called histones. *S v Orrie* [2003] ZAWCHC 63 para 18 held that chromosomes 'are made up of the complex chemical which is DNA'. In eukaryotes, chromosomes are located in the nucleus of a cell and contain genetic information encoded in DNA. Eukaryotes refer to organisms that have a defined nucleus and internal membranes. Examples include humans, animals, plants, and fungi. Klug et al op cit note 4 at 3, 30, 213 & G–3; Mathews et al op cit note 23 at 16; Britannica 'Eukaryote' available at *https://www.britannica.com/science/eukaryote*, accessed on 18 February 2021; Meshandren Naidoo *The CRISPR Patent Landscape: A South African Perspective* (unpublished LLM thesis, University of KwaZulu-Natal, 2020) 17–8.
[163] Section 3(1)(*a*) of POPIA. A 'responsible party' is defined in section 1 of POPIA as 'a public or private body or any other person which, alone or in conjunction with others, determines the purpose of and means for processing personal information'. In the present case, a responsible party refers to the testing company or their associated laboratory, where their services are outsourced – given that the phrase 'in conjunction with others' in the definition of 'responsible party' in section 1 of POPIA means that it is possible to have several responsible parties. Dusty-Lee Donnelly *Privacy by (re)Design: A Comparative Study of the Protection of Personal Information in the Mobile Applications Ecosystem under United States, European Union and South African Law* (Doctor of Laws thesis, University of KwaZulu-Natal, 2020) 228.
[164] Section 1 of POPIA.

responsible party in terms of POPIA.[165] Once extracted, sequenced, and analysed, the resulting genetic data is entered, recorded, and stored on computers or digital databases – either for the purpose of generating direct-to-consumer genetic test results or for research. Given that genetic data is contained within DNA, it already exists and such information is not 'produced' through processing.[166] In its natural state, DNA does not reveal anything of value – it is only useful once electronic machines and equipment extract, sequence, and process DNA,[167] thereby 'decoding' it to determine the order of the nucleotide base pairs (A, G, C, and T) and acquire genetic data. In this instance, I suggest that such genetic data is 'personal information' that falls within POPIA's ambit. This genetic data relates to an 'identifiable, living, natural person' in line with the definition of 'personal information' in POPIA – specifically falling under subsection (d), being 'biometric information'.[168] Although DNA contains 'personal information' (in the form of genetic data),[169] DNA itself is not 'personal information' and is thus not protected by POPIA. To consider DNA as 'personal information' under POPIA would be incorrect for the reasons given above. The same can be said of any human biological material – such as saliva. Although not considering DNA as 'personal information' under POPIA may seem counter intuitive, doing so would render almost all things containing DNA, such as a single skin cell, as 'personal information' worthy of protection under POPIA when it is only the DNA sequence obtained through sequencing and processing that is important – and even then, only specific parts of the sequence play a role in the development of certain health-related conditions.

POPIA contains another type of personal information which is relevant to consider – 'special personal information'. Section 1 of POPIA defines special personal information as 'personal information as referred to in section 26' of POPIA. Section 26(*a*) of POPIA refers to personal information concerning…health or sex life or biometric information of a data subject'.[170] As special personal information includes biometric information, and as genetic data is a type of biometric information, genetic data therefore also falls under 'special personal information' in POPIA.

---

[165] In terms of direct-to-consumer genetic testing, genetic data is also not collected directly from consumers – it is collected from a source, namely saliva. DNA is then extracted from saliva and sequenced in order to obtain genetic data. See also, Thaldar op cit note 7 at 1.
[166] 'Produced' as referred to in the definition of 'record' in section 1 of POPIA.
[167] For an explanation of what DNA is, see *S v Maqhina* supra note 161.
[168] Section 1 of POPIA.
[169] Ibid.
[170] Section 26(1)(*a*) of POPIA. A 'data subject' is defined in section 1 of POPIA as 'the person to whom personal information relates' which, in this instance, is the consumer.

In summation, where DNA is extracted from a consumer's saliva sample and then sequenced and analysed, neither the saliva nor the DNA qualify for protection under POPIA as 'personal information'.[171] This is because 'tissue' and 'biological material' are containers of genetic data – they themselves are not genetic data.[172] While saliva and DNA come from an 'identifiable, living, natural person' in line with the definition of 'personal information' in POPIA,[173] they are tangible specimens rather than 'information' and are not 'entered in a record' by a responsible party,[174] and thus do not fall within the scope of POPIA. However, the extracted and processed DNA sequence, which constitutes genetic data, qualifies as 'personal information' and a form of 'biometric information' under POPIA.

(b) The processing of personal information

Since the meaning of 'personal information' in POPIA,[175] in relation to direct-to-consumer genetic testing, has been dealt with, I now consider the processing of this personal information.[176] Understanding the meaning of processing in POPIA, and what it entails, is important to establishing whether the activities of testing companies fall within its ambit – whether it is the collection of genetic data; the extraction, sequencing, and analysing of DNA; the storage of saliva samples, DNA, or genetic data; or research. In terms of POPIA, 'processing' refers to a range of activities conducted on personal information, including –

> '(a) the collection, receipt, recording, organisation, collation, storage, updating or modification, retrieval, alteration, consultation or use;
>
> (b) dissemination by means of transmission, distribution or making available in any other form; or
>
> (c) merging, linking, as well as restriction, degradation, erasure or destruction of information'.[177]

[171] Section 1 of POPIA. Although saliva contains DNA and DNA holds genetic data, saliva, like DNA, does not meet the definition of 'personal information' in section 1 of POPIA.
[172] This is in line with the argument made by Thaldar et al that, where biological material is to be used for genetic health research, biological material can be viewed as a holder for biometric information. However, where I disagree with Thaldar et al is in the fact that biological material that contains biometric information will fall within POPIA's scope. This, I suggest, is not the case. Thaldar et al op cit note 146 at 4.
[173] Section 1 of POPIA.
[174] In terms of section 3(1)(*a*) of POPIA.
[175] Section 1 of POPIA.
[176] As mentioned above, section 13(1) of POPIA states that '[p]ersonal information must be collected for a specific, explicitly defined and lawful purpose related to a function or activity of the responsible party'.
[177] Section 1 of POPIA.

Firstly, only things that meet the definition of 'personal information' in POPIA may be processed.[178] Although saliva samples are used in DNA analyses to provide test results to consumers regarding their predisposition to certain diseases and conditions, or for research, because saliva and DNA are not 'personal information',[179] the processing provisions in POPIA do not apply. However, processing in POPIA is relevant to genetic data as a type of personal information. The activities of testing companies are not required to meet every type of processing in POPIA,[180] and even just one activity will suffice. As mentioned above, using DNA extraction and sequencing techniques, genetic data is recorded, organised, collated, and stored in databases, which is 'processing' in terms of POPIA.[181]

Collection, as a type of processing under POPIA, is important to consider. Section 13(1) of POPIA provides that personal information 'must be collected for a specific, explicitly defined and lawful purpose related to a function or activity of the responsible party'.[182] As saliva and DNA are not 'personal information',[183] POPIA is not applicable to such collection – collection takes place only once DNA has been sequenced, and such a sequence (information) is stored or recorded in some way by a scientist. It is important to determine whether section 13 of POPIA applies to direct-to-consumer genetic testing as this may affect what is necessary for testing companies to include in their consent forms and terms of service.

V DISCUSSION

Based on what has been established above, it may be useful to provide various scenarios involving the direct-to-consumer genetic testing process to demonstrate whether, and how, the relevant legislation regarding saliva qua tissue and biological material, as well as DNA and genetic data, apply.

---

[178] Ibid.
[179] Ibid.
[180] Ibid.
[181] Ibid. Where genetic data is shared for the purposes of research by testing companies, this meets 'dissemination' under the definition of 'processing' in POPIA. Also, in terms of research, genetic data may be destroyed or erased where the consumer requests it, or as required by law. Therefore, the range of activities undertaken by testing companies clearly meet the definition of 'processing' in section 1 of POPIA.
[182] Section 13(1) of POPIA. In terms of direct-to-consumer genetic testing, the collection of a saliva sample by consumers is for the specific purpose of undergoing a direct-to-consumer genetic test. As consumers will generally have agreed to undergo such testing, they will be aware of its purpose and the collection of personal information must be in line with POPIA.
[183] Under section 1 of POPIA.

(a) Scenario (1)

The saliva sample is collected by consumers in South Africa, and where the testing company is based, and the DNA extraction, sequencing, and analysis occur, in South Africa. In this scenario, as the entire process occurs within the country, South African legislation – namely the NHA and the Human Biological Material Regulations – apply. Saliva, which is collected by consumers, meets the definition of 'tissue' in the NHA as well as 'biological material' in the Human Biological Material Regulations. DNA extracted from the saliva sample and then sequenced and analysed falls under the definition of 'biological material' in the Human Biological Material Regulations. As genetic data is obtained from analysing DNA, and because it occurs within South Africa's borders, it is deemed to be 'personal information' and POPIA applies. POPIA does not apply to the collection of saliva samples, but rather to the DNA analysis which results in genetic data, and thus the requirements contained therein also apply.

(b) Scenario (2)

The collection of the saliva sample takes place in South Africa, but the testing company as well as the DNA extraction, sequencing, and analysis occur abroad. In this scenario, as the saliva sample is collected in South Africa, it falls under the definition of 'tissue' in the NHA and its relevant provisions apply in terms of collection and removal. Given that DNA extraction, sequencing, and analysis occur outside of South Africa, the Human Biological Material Regulations are not applicable as DNA, a type of 'biological material' as per the Human Biological Material Regulations, only exists within the saliva sample when it leaves the country. Moreover, as the saliva sample is not genetic data, it does not qualify as 'personal information' under POPIA.[184] Therefore, because the saliva sample leaves South Africa as an unsequenced thing, section 13 of POPIA does not apply. In this case, the laws of the jurisdiction in which the testing company is based are relevant.

(c) Scenario (3)

An overseas testing company is used by a consumer, but DNA extraction, sequencing, and analysis of the collected saliva sample occurs in South Africa. As the collection of the saliva sample as well as DNA extraction, sequencing, and analysis take place in the country, South African legislation is relevant. As with scenario (1), the saliva sample falls under the definition of 'tissue' in the NHA and 'biological material' in the Human Biological Material Regulations,

[184] Ibid.

while the sequenced DNA is only included in the definition of 'biological material' in the Human Biological Material Regulations. As the extraction and sequencing of DNA takes place within South Africa, section 13 of POPIA applies as, once the DNA (removed from the saliva sample) is extracted and sequenced, it results in genetic data – thus qualifying as 'personal information' and activating the provisions of POPIA.

(d) Conclusion on the scenarios

As the direct-to-consumer genetic testing industry is complex and operates inter-jurisdictionally, the application of South African legislation, specifically the NHA and the Human Biological Material Regulations to tissue and biological material, and POPIA to personal information, vary depending on where the testing company (or their associated laboratory) is based, as well as where the extraction, sequencing, and analysis of DNA occurs.

VI CONCLUSION

The different but overlapping definitions primarily of 'tissue' in the NHA[185] and 'biological material' in the Human Biological Material Regulations,[186] and the lack of clarity as to what things are included under each definition, make South Africa's legal landscape in relation to direct-to-consumer genetic testing challenging to traverse. This is further exacerbated by the complex direct-to-consumer genetic testing process and the numerous things that are used at various stages throughout. Although it would be beneficial for South Africa's statutory framework to contain a single, clear, and uniform definition encompassing the meanings of both tissue and biological material – amending this would be a monumental task.[187]

This Chapter explored the various things (saliva, DNA, and genetic data) as well as the stages, including collection and processing, involved in direct-to-consumer genetic testing. Through this analysis, I have found that the things involved in the direct-to-consumer genetic testing process – namely saliva, DNA, and genetic data – are indeed covered by South African legislation, albeit by different statutes and in varying ways.

[185] Section 1 of the NHA.
[186] Regulation 1 of the Human Biological Material Regulations.
[187] The analyses that follow in subsequent chapters are based on the definitions and processes in South Africa's extant law as it exists currently.

CHAPTER 3

OVERCOMING THE FIRST HURDLE: CAN CONSUMERS LAWFULLY COLLECT THEIR OWN SALIVA SAMPLES?

I INTRODUCTION AND CHAPTER OVERVIEW

One of the primary reasons for the growing popularity of direct-to-consumer genetic testing is that it allows individuals to avoid having to make appointments and approach the healthcare system to undergo a genetic test. It has thus far largely been assumed that consumers are legally permitted to take their own saliva sample for the purpose of a direct-to-consumer genetic test, but is this really the case in terms of South African legislation?

In this Chapter, I assess whether consumers are legally allowed to collect their own saliva samples for direct-to-consumer genetic testing. In order to do so, I examine the NHA and its relevant regulations, including the Human Biological Material Regulations, the General Control Regulations, the Buccal Sample and Blood Withdrawal Regulations, and the Draft Testing and Research Regulations. This Chapter also reviews the Health Professions Act.

II LEGAL LIMITS ON PERSONS WHO MAY WITHDRAW SALIVA

Generally, when dealing with tissue and biological material, there are defined categories of persons (usually qualified professionals) that are legally permitted to perform certain tasks related to these things. This is because tissue and biological material are typically removed and utilised for various purposes in the clinical context. Direct-to-consumer genetic testing differs in this regard as consumers (namely individual laypersons) are generally responsible for collecting their own saliva sample, with no professional oversight.[1] To minimise potential harm to individuals, due to the incorrect handling of samples which may cause erroneous results with substantial consequences, South Africa's extant law appears to place the duties surrounding samples on those with some sort of professional qualification. For example, the NHA requires a registered medical practitioner or dentist to remove, use, or transplant tissue from a living person.[2] The General Control Regulations and the Human Biological Material Regulations only permit 'competent persons' to undertake activities relating to blood or blood products,[3] or

[1] This is so, unless the direct-to-consumer genetic test or company requires the involvement of a healthcare professional to gather the saliva sample on the consumer's behalf.
[2] Section 59(1) of the NHA.
[3] Regulation 1 of the General Control Regulations.

biological material respectively.[4] But what is the position in the context of direct-to-consumer genetic testing, where it is generally not professionals, but rather 'unqualified' consumers without specialist training, that are responsible for the collection of saliva samples? Are these consumers allowed, by law, and deemed 'competent' to collect their own saliva samples for the purpose of a direct-to-consumer genetic test?[5]

As a starting point, it is necessary to determine what is meant by 'competent person'. Section 56 of the NHA states that a *person* may use tissue removed from a living person.[6] The NHA does not define the term 'person', but it does provide that only registered medical practitioners or dentists are permitted to use or 'remove any tissue from a living person' (thus excluding laypersons)[7] – but only for the purposes in section 56.[8] This limits the scope of this provision to certain purposes relating to the use of inter alia tissue,[9] without which *any* removal or use of tissue would be illegal. Section 56(1) of the NHA allows tissue removed from a living person to be used 'only for such medical or dental purposes as may be prescribed'.[10] The NHA does not define 'medical or dental purposes', but the term 'as may be prescribed' leads to the Human Biological Material Regulations.[11] According to regulation 5 of the Human Biological Material Regulations, biological material may be removed from living persons for inter alia 'DNA, RNA and chromosome-based genetic testing'.[12] This suggests that 'medical or dental

[4] Regulation 1 of the Human Biological Material Regulations.
[5] It is only the *removal* of biological material that is relevant to consumers; the *use* of such material is dealt with by testing companies or their associated laboratories – either to obtain test results or to conduct research.
[6] Section 56(1) of the NHA.
[7] Ibid. In terms of direct-to-consumer genetic testing, it would seem that this provision requires the involvement of a healthcare professional.
[8] Ibid.
[9] Ibid.
[10] Ibid.
[11] This is because section 1 of the NHA defines 'prescribed' to mean 'prescribed by regulation', and section 68(1) of the NHA permits the Minister to make various regulations.
[12] Regulation 5(a) of the Human Biological Material Regulations. It is interesting to note that the Draft Testing and Research Regulations go into greater detail and specify the types of genetic testing that may be conducted. Regulation 4 of the Draft Testing and Research Regulations specify the medical and dental purposes for which biological material may be removed or withdrawn and used. These include –

'(a) DNA, RNA and chromosome-based genetic testing including:
(i) diagnostic tests;
(ii) testing for genetic carrier status;
(iii) antenatal diagnosis;
(iv) voluntary presymptomatic, predictive or susceptibility testing, screening tests, drug response or toxicity tests, identity or paternity testing'.

It is apparent that reference to 'DNA, RNA and chromosome-based genetic testing' in the Human Biological Material Regulations relates to such testing in the clinical context. This is because firstly there is no reference to direct-to-consumer genetic testing or any indications of self-testing, and secondly the Human Biological Material Regulations were promulgated shortly after the advent of direct-to-consumer genetic testing abroad (let alone in South Africa), making it unlikely that such situations were even contemplated when the Human Biological Material Regulations were drafted. While this may be the case, and although the analytical validity, clinical validity, and clinical utility of direct-to-consumer genetic tests have been questioned, the manner in which the

purposes as may be prescribed' in section 56(1) of the NHA includes genetic testing,[13] thus permitting saliva to be removed for this purpose.

The NHA's related regulations also do not contain a universal definition of 'competent person', and this differs based on the purpose for which the biological material is used.[14] According to the Human Biological Material Regulations, only a 'competent person' may remove biological material for 'genetic testing, genetic health research or therapeutic purposes',[15] and this must be undertaken in an authorised and prescribed institution.[16] Furthermore, the definition of 'competent person' in the Human Biological Material Regulations provides various categories under which a competent person may fall depending on the circumstance, activity, and the purpose.[17] The definition of 'competent person' in the Human Biological Material Regulations is broader than a registered medical practitioner or dentist (as per the NHA)[18] as it refers to an array of groups.[19] The Human Biological Material Regulations define certain activities for which biological material may be removed and used – such as the withdrawal of blood, gametes, ovum, or sperm[20] – and then stipulates the requisite 'competent persons' to undertake the task – such as medical practitioners, nurses, gynaecologists, and medical technologists or scientists, depending on the activity.[21] However,

saliva sample is used and the basic, process (of extraction, sequencing, and analysis) look to be the same as clinical genetic testing. Therefore, although the things used (saliva versus blood) and the manner in which biological material is removed and by whom may differ between genetic tests offered directly to consumers and those conducted in the clinical context, I suggest that the use of saliva and the purposes for which it is used do not vary greatly between the direct-to-consumer genetic testing context and the clinical setting.

[13] This is further enhanced by section 68(1)(*p*) of the NHA, which provides that the Minister may make regulations regarding the 'acquisition, storage, harvesting, utilisation or manipulation of tissue…for any purpose' – suggesting that genetic testing may be one such purpose. See also, M Nöthling Slabbert & MS Pepper '"A room of our own?" Legal *lacunae* regarding genomic sovereignty in South Africa' (2010) 73 *THRHR* 448.

[14] A definition of 'competent person' is provided for in regulation 1 of the Human Biological Material Regulations, regulation 1 of the General Control Regulations, regulation 1 of the Regulations Relating to Tissue Banks GN R182 GG 35099 of 2 March 2012 (Tissue Bank Regulations), and regulation 1 of the Draft Testing and Research Regulations. The Human Biological Material Regulations provide the most comprehensive definition of 'competent person' covering a wide range of activities, including research.

[15] Regulation 2(a) of the Human Biological Material Regulations.

[16] Regulation 2(b) of the Human Biological Material Regulations. Section 1 of the NHA defines an 'authorised institution' as 'any institution designated as an authorised institution in terms of section 54' of the NHA. section 54(1) of the NHA allows the Minister, through a notice in the Government Gazette, to appoint any institution besides those in section 63 of the NHA as an authorised institution.

[17] Regulations 1 and 2(a) of the Human Biological Material Regulations.

[18] Section 59(1) of the NHA.

[19] Regulation 1 of the Human Biological Material Regulations.

[20] Ibid.

[21] Ibid. For example, according to regulation 1 of the Human Biological Material Regulations, a person registered as a medical practitioner or a health professional trained as a phlebotomist and registered in terms of the Health Professions Act or as a nurse in terms of the Nursing Act 33 of 2005 (Nursing Act) is required for intravenous blood withdrawal; a medical practitioner registered, under the Health Professions Act, as a specialist in the procedure is required for the intra-arterial withdrawal of blood; and a urologist registered in terms of the Health Professions Act or a male reproductive health expert is required for the withdrawal of sperm.

prior to listing these categories, the definition states that '"competent person" means trained…'.[22] I suggest that this implies that a 'competent person' requires some type of professional training in a particular area and thus appears to exclude laypersons who generally lack any medical or scientific expertise.

Furthermore, the Buccal Sample and Blood Withdrawal Regulations provide that a healthcare provider or a person considered in section 56 of the NHA, who is not a healthcare provider, may take a buccal sample or remove blood from a living person in line with sections 55 and 56(1) of the NHA.[23] The NHA defines a 'health care provider' as 'a person providing health services in terms of any law' including inter alia the Health Professions Act.[24] As such, unless an individual is acting as both a patient and a provider of medical services, the Buccal Sample and Blood Withdrawal Regulations are not applicable to collecting a buccal sample from oneself. This brings into question saliva that is collected by an individual at home, as is generally the case with direct-to-consumer genetic testing – and not by a 'competent person' in an 'authorised institution'. The inescapable conclusion seems to be that the NHA and the Human Biological Material Regulations effectively ban the removal of saliva for the purpose of a direct-to-consumer genetic test by individuals themselves who are not 'competent persons' and are therefore not legally authorised to do so.

While the focus of this Chapter is on individual laypersons without any specialist training, I recognise that there are circumstances in which professionals who are 'competent persons' in terms of the Human Biological Material Regulations – such as medical practitioners – may collect their own saliva sample for a direct-to-consumer genetic test. In such a case, the medical practitioner is considered a 'competent person' and therefore authorised to remove saliva (even though it is their own) as they are legally registered and trained to perform such a task. Although medical practitioners may be competent persons, they are still acting unlawfully as they are performing an act for a purpose that brings it within the aegis of the NHA and the Human Biological Material Regulations, but they fail to adhere to the requirements thereof. Despite being a 'competent person', a medical practitioner still fails to adhere to various

[22] Regulation 1 of the Human Biological Material Regulations.
[23] Regulation 2 of the Buccal Sample and Blood Withdrawal Regulations refers to a 'buccal sample' as 'cellular material inside the mouth'. Regulation 4 of the Buccal Sample and Blood Withdrawal Regulations provides that when persons who are not healthcare professionals take a buccal swab, this may only be done by swabbing the inside of the cheek using specified equipment to gather 'a small quantity of cellular material sufficient for testing'.
[24] Section 1 of the NHA.

restrictions in these laws, including the requirement that the removal of saliva be done at an 'authorised institution'.[25]

Can it be that South Africa's extant law bans individuals from collecting their own saliva samples? Is this the writing on the wall for direct-to-consumer genetic testing? Anticipating potential pushback against the conclusion that the collection of saliva samples by individuals for the purpose of a direct-to-consumer genetic test is legally banned, in the following sections, I analyse six possible counter-arguments.

(a) The ban can be ignored based on the de minimis principle

The first, and perhaps most obvious, argument is that the ban on individuals collecting their own saliva samples can be excluded based on the maxim 'de minimis non curat lex' (the law does not concern itself with trifles), also known as the de minimis principle.[26] This principle allows one to escape punishment for an unlawful act if it is deemed to be trivial.[27] The de minimis principle has real world consequences and should not be ignored, but is the seemingly harmless collection of saliva samples by individuals something that the law should concern itself with? The issue with relying on the de minimis principle is that the decision of whether an issue is trifle or not is subjective. Those in opposition to direct-to-consumer genetic testing due to ethical concerns, such as the uncertain usefulness of test results and the ability of consumers to understand the information provided to them without professional assistance, would argue that the ban on individuals collecting their own saliva sample protects them from unscrupulous testing companies that function outside of the traditional healthcare system – an argument which may have force in a court of law. Therefore, it would be cavalier of testing companies that consider entering the market and offering their products and services in South Africa to assume that this is a trifle issue. Yet, I suggest that, for present purposes, this argument does not assist as the de minimis principle is not a principle of legal interpretation and does not aid with legal certainty.

[25] Regulation 2(b) of the Human Biological Material Regulations.

[26] The de minimis principle permits insignificant issues to not be subject to a legal requirement. LexisNexis 'De minimis definition' available at *https://www.lexisnexis.co.uk/legal/glossary/de-minimis*, accessed on 10 May 2021.

[27] See, *S v Kgogong* 1980 (3) SA 600 (AD) where the Appellate Division declined to convict the accused of theft because he stole a trivial piece of paper that was of no value. See also, *S v Dane* 1957 (2) SA 472 (N) where the appellant was acquitted of the charge of malicious damage to property for cutting part of the neighbour's hedge.

(b) The ban is superseded by the consumer's consent

The second argument against the ban on individuals collecting their own saliva samples is that of consent. Testing companies require individuals to provide consent prior to testing.[28] Generally, there are different stages of the testing process that consumers are required to consent to – namely, consent to testing and collection of the saliva sample,[29] consent to sequencing and analysis of the saliva sample to obtain DNA,[30] consent to storage (of either the saliva sample or extracted DNA),[31] and consent to research.[32]

Questions have been raised about whether the consent provided by consumers in the direct-to-consumer genetic testing context is informed. The Human Biological Material Regulations require written *informed* consent for the removal of biological material from living persons for 'genetic testing, genetic training, genetic health research or therapeutics'.[33] However, it has been found that informed consent is not always possible or adequately realised in direct-to-consumer genetic testing.[34] In the medical context, informed consent is paramount

[28] Eline M Bunnik, A Cecile, JW Janssens et al 'Informed consent in direct-to-consumer personal genome testing: The outline of a model between specific and generic consent' (2012) 28(7) *Bioethics* 343 & 346.

[29] See, Helix 'Helix privacy policy' 30 November 2020 available at *https://www.helix.com/pages/privacy-policy*, accessed on 9 January 2021; Myriad 'Myriad informed consent policies' available at *https://myriadwomenshealth.com/consent-policies/*, accessed on 9 January 2021; MyHeritage 'Privacy policy' 17 December 2020 available at *https://www.myheritage.com/privacy-policy*, accessed on 9 January 2021; AncestryDNA 'Ancestry terms and conditions' 23 September 2020 available at *https://www.ancestry.com/cs/legal/termsandconditions*, accessed on 9 January 2021; GenebyGene 'Gene by Gene privacy policy' available at *https://genebygene.com/wp-content/uploads/2019/08/GenebyGene_PrivacyPolicy.pdf*, accessed on 9 January 2021; Color 'Color informed consent' available at *https://www.color.com/informed-consent*, accessed on 9 January 2021.

[30] See, Helix 'Helix terms of service' 30 November 2020 available at *https://www.helix.com/pages/terms-of-service*, accessed on 9 January 2021; Helix op cit note 29; EasyDNA 'Privacy policy' 20 May 2018 available at *https://www.easydna.co.za/privacy-policy/*, accessed on 9 January 2021.

[31] See, 23andMe 'Biobanking consent document' available at *https://www.23andme.com/about/biobanking/*, accessed on 9 January 2021; Helix op cit note 29; Color op cit note 29.

[32] Those testing companies that conduct research often require separate, specific consent in order to utilise the genetic data or information (and sometimes DNA) of consumers for the purpose of research. See, 23andMe 'Individual data sharing consent' available at *https://www.23andme.com/about/individual-data-consent/*, accessed on 9 January 2021; MapmyGenome 'Terms and conditions' available at *https://mapmygenome.in/terms-and-conditions/*, accessed on 9 January 2021; 23andMe 'Research consent document' available at *https://www.23andme.com/about/consent/*, accessed on 9 January 2021; Helix op cit note 30; Helix op cit note 29; Myriad op cit note 29; MyHeritage op cit note 29; AncestryDNA 'AncestryDNA informed consent' 24 July 2018 available at *https://www.ancestry.com/dna/lp/informedconsent-v4-en*, accessed on 9 January 2021; AncestryDNA op cit note 29; GenebyGene op cit note 29; Color op cit note 29.

[33] Regulation 3(1)(*a*) of the Human Biological Material Regulations. Informed consent refers to the process of understanding the advantages, risks, aims, and possible consequences of a procedure or project. Thus, when an individual agrees to such, they do so with full knowledge of all aspects. Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018) 4.

[34] Testing companies, instead of using informed consent, often rely on Terms of Service (TOS) agreements which, according to Bunnik et al, is insufficient when such tests are offered commercially. Bunnik et al op cit note 28 at 344; Valerie Gutmann Koch 'Recreational genetics or research enterprise? Cloudy consent issues arising from direct-to-consumer genetic testing' *Bill of Health* 2 August 2018, available at *https://blog.petrieflom.law.harvard.edu/2018/08/02/recreational-genetics-or-research-enterprise-cloudy-consent-issues-arising-from-direct-to-consumer-genetic-testing/*, accessed on 26 June 2021; Sara A Mahmoud-

in any decision-making process as it aims to protect against harm and promote autonomous choice.[35] But a lack of healthcare professional involvement, questionable clinical validity and utility, and the amount of information which consumers may not understand present difficulties for informed consent and pre-test information in direct-to-consumer genetic testing[36] – thus eroding an individual's autonomy.[37] Autonomy is also impeded when testing companies mislead consumers. Although consumers decide to undergo direct-to-consumer genetic tests and determine their utility, this may be inaccurate if testing companies offer erroneous or misleading information.[38] The absence of professional oversight in many direct-to-consumer genetic testing business models necessitates the need for the provision of comprehensive and sound information.[39]

One of the common requirements for consent in direct-to-consumer genetic testing is that consumers understand the process and purpose for the collection of the saliva sample. But what if a consumer understands the process, consents to undergo direct-to-consumer genetic testing, and collects the saliva themselves? Section 55 of the NHA deals with the removal of inter alia tissue from living persons, which may not be done without the written consent of the individual and in terms of prescribed conditions.[40] In line with this, the General Control Regulations state that tissue may only be removed from a living person and with written consent.[41] As mentioned

Davis 'Direct-to-consumer genetic testing: Empowering EU consumers and giving meaning to the informed consent process within the IVDR and GDPR frameworks' (2020) 19(1) *Wash U Global Stud L Rev* 2 & 39.

[35] Bunnik et al op cit note 28 at 345.

[36] The suitability of informed consent in direct-to-consumer genetic testing has been questioned for two reasons. Firstly, it has been contended that these commercial companies are not subject to the same moral standards as the medical profession. Although the TOS agreements, commonly used on direct-to-consumer genetic testing websites, provide legal protection to companies instead of consumers, they do offer information on risks and possible negative consequences. Secondly, testing companies purport to offer informational or educational services rather than medical tests. The TOS often advise consumers to consult with a medical professional, thus making direct-to-consumer genetic tests appear as a contract for consumer goods – meaning that companies may claim that TOS, rather than informed consent, is sufficient. Bunnik et al op cit note 28 at 343, 344 & 346.

[37] Individual autonomy may be diminished without a comprehensive and impartial understanding of the various genetic testing options available as well as the information provided. Mahmoud-Davis op cit note 34 at 39.

[38] Given that testing companies operate for-profit, they may exaggerate facts in advertisements and distort information on their websites in order to promote their tests and increase sales. If true, this would be contra sections 4(5)(*b*), 24, 29, 41, and 51(1)(*a*)(*ii*) of the Consumer Protection Act 68 of 2008 (CPA), and thus unlawful. In addition, in extreme cases of fraud there would also be common law remedies. Bunnik et al op cit note 28 at 345; Mahmoud-Davis op cit note 34 at 39; Paula Saukko 'State of play in direct-to-consumer genetic testing for lifestyle-related diseases: Market, marketing content, user experiences and regulation' (2013) 72 *Proc Nutr Soc* 58-59; Mark Popovsky 'Exaggerated benefits and underestimated harms: The direct-to-market consumer genetic test market and how to manage it going forward' (2010) 8(2) *Dartmouth LJ* 78; Stuart Hogarth, Gail Javitt & David Melzer 'The current landscape for direct-to-consumer genetic testing: Legal, ethical, and policy issues' (2008) 9 *Annu Rev Genom Hum Genet* 169.

[39] Amanda Singleton, Lori Hamby Erby, Kathryn V Foisie et al 'Informed choice in direct-to-consumer genetic testing (DTCGT) websites: A content analysis of benefits, risks, and limitations' (2012) 21(3) *J Genet Counsel* 6.

[40] Section 55 of the NHA. Section 55 of the NHA deals with removal, while section 56 of the NHA covers use.

[41] Regulation 2 of the General Control Regulations.

above, the Human Biological Material Regulations require written informed consent.[42] Therefore, in terms of South African law, if an individual provides written consent either for the removal of tissue (in terms of the NHA and the General Control Regulations),[43] or written informed consent for the removal of biological material for genetic testing or genetic health research (as per the Human Biological Material Regulations), this permits the removal of saliva – a form of 'tissue' in the NHA and the General Control Regulations,[44] and a type of 'biological material' in the Human Biological Material Regulations.[45]

The question is then: By who? Under the NHA, written consent permits a 'person' to remove inter alia tissue.[46] Although the NHA does not define the term 'person', because only registered medical practitioners or dentists may remove and use tissue,[47] it appears that these professionals are considered to be such 'persons'. The Human Biological Material Regulations allow a 'competent person' to remove biological material with written informed consent.[48] The provision of consent allows another person to remove saliva; it is not consent for the individual to do so themselves. Where a person is not a healthcare provider, the Buccal Sample and Blood Withdrawal Regulations still permit such a person to take a buccal sample from a living person, subject to certain conditions.[49] While this may extend to laypersons (though a very tenuous connection), the Buccal Sample and Blood Withdrawal Regulations refer to buccal samples taken from other living persons, thereby excluding a situation in which a consumer takes such a sample themselves.

In terms of testing company policies, the provision of consent allows an individual to remove their own saliva sample. However, the NHA and the Human Biological Material

[42] Regulation 3(1) of the Human Biological Material Regulations.
[43] Section 55(a) of the NHA and regulation 2 of the General Control Regulations respectively.
[44] Ibid.
[45] This is because regulation 3(1)(*a*) of the Human Biological Material Regulations prohibits the removal of human biological material by a competent person for genetic testing, genetic training, genetic health research, or therapeutics without the 'written informed consent' of the individual.
[46] Section 55(a) of the NHA.
[47] Section 59(1) of the NHA.
[48] Regulation 3(1)(*a*) of the Human Biological Material Regulations.
[49] These conditions, contained in regulation 3 of the Buccal Sample and Blood Withdrawal Regulations, are that: (1) they have undergone the necessary training at a health establishment; and (2) their name is recorded in a register. Regulation 5(a) of the Buccal Sample and Blood Withdrawal Regulations states that training will only be offered to those able to understand the subject matter, and regulation 5(c) of the Buccal Sample and Blood Withdrawal Regulations provides that training will include (i) information regarding the obtaining of informed consent; (ii) preparing for and taking the buccal; (iii) establishing process quality; (iv) using the equipment; (v) disposing of material; (vi) information regarding the tests to be undertaken; (vii) interpreting the test results; and (viii) submitting the buccal for further administration.

Regulations have the force of law and override the rules established by testing companies.[50] South African legislation permits only certain persons with the relevant expertise to remove tissue or biological material from others (provided that consent has been obtained), and does not allow individuals to do so, or to do so themselves. Therefore, even if an individual consents, this does not invalidate the legal requirement that only certain defined 'competent persons' may remove saliva for the purpose of direct-to-consumer genetic testing and does not rescind the ban against the self-collection of saliva samples by consumers.

(c) Is there no intention to regulate the collection of saliva, or is there an intent to ban it?

The third possible argument against the ban on individuals collecting their own saliva samples for the purpose of a direct-to-consumer genetic test revolves around intention – does the non-inclusion of saliva under the definition of 'competent person' in the Human Biological Material Regulations[51] mean that there is no intention to regulate it (allowing individuals to do as they wish), or does this signal an intent to ban it?

The removal of saliva is not included in the definition of 'competent person' in the Human Biological Material Regulations and it rather refers to inter alia the removal of blood, gametes, ova, or sperm.[52] Does this mean that anyone can remove saliva for the purpose of a genetic test, and it does not require the involvement of a 'competent person'? While this may be thought to be the case, the Human Biological Material Regulations outlaw this in regulation 2(a) by specifying that '*[n]o person*, except a competent person, may remove biological material' (own emphasis).[53] This requirement pertains to the removal of biological material for certain purposes, one of which is genetic testing. As direct-to-consumer genetic testing falls under this category, only a 'competent person' is permitted to remove saliva for this purpose.

[50] It is interesting to question whether the situation would be different in terms of the hybrid direct-to-consumer genetic testing model, where healthcare professionals are involved in the testing process. If the healthcare professional merely orders the test and consumers still collect their own saliva sample, the situation would remain unchanged. But if the healthcare professional ordered the test and oversaw the collection of the saliva sample, the healthcare professional would be deemed the 'competent person' in such a situation. This is because they would generally have the requisite training for such purposes. Although the definition of 'competent person' in regulation 1 of the Human Biological Material Regulations denotes various categories under which a competent person may fall, a common thread amongst all categories, regardless of the purpose, is the requirement that the competent person be registered in terms of the Health Professions Act. I suggest that a healthcare professional would be registered as such in terms of the Health Professions Act and would therefore be regarded as a 'competent person', authorised to remove biological material from an individual with their written consent.

[51] Regulation 1 of the Human Biological Material Regulations.

[52] Ibid.

[53] Regulation 2(a) of the Human Biological Material Regulations. Although saliva is not explicitly mentioned under the definition of 'competent person' in regulation 1 of the Human Biological Material Regulations, as saliva is a type of biological material, in terms of regulation 2(a) of the Human Biological Material Regulations, saliva may be removed for certain purposes.

Although the Human Biological Material Regulations do not specifically define a 'competent person' in relation to the removal of saliva, interestingly the Draft Testing and Research Regulations – although never made into law – explicitly provide for this. The Draft Testing and Research Regulations which, in terms of collecting cells from the inside of the cheek (buccal swab), provide that a 'competent person' in such a case is 'any person who has been trained to perform such a procedure or the person himself/herself who provides the sample for genetic testing'.[54] Thus, the Draft Testing and Research Regulations recognise, and include, situations wherein individuals collect their own saliva samples for the purpose of a genetic test and deem them competent persons. While saliva and its collection by individuals themselves was included in the Draft Testing and Research Regulations, this was not carried through into the Human Biological Material Regulations. The logical conclusion for this is that the inclusion of the self-collection of saliva was contemplated, but excluded with the intention that it be disallowed.

This apparent ban on the collection of saliva by individuals, appears to carry through to the Buccal Sample and Blood Withdrawal Regulations which, in regulation 4, refer to taking buccal samples or removing blood by individuals who are not healthcare professionals, and provide that, in the case of saliva, it shall only be by means of a cheek swab to collect an adequate amount of cellular material for testing.[55] Although this appears to allow laypersons[56] to remove small quantities of saliva or blood themselves for testing purposes, other provisions in the Buccal Sample and Blood Withdrawal Regulations refer to the taking of a buccal sample or the removal of blood '*from another living person*'[57] (own emphasis), thereby excluding situations wherein individuals remove saliva or blood themselves.[58]

The Buccal Sample and Blood Withdrawal Regulations form part of the NHA's subsidiary legislation, and must thus be read with the NHA and its other regulations. This is based on the rule of statutory interpretation that –

---

[54] Regulation 1 of the Draft Testing and Research Regulations. However, the provisions of the Draft Testing and Research Regulations do not make specific mention of a competent person in such a situation. Besides the definition, the only other mention of 'competent person' in the Draft Testing and Research Regulations is in reference to utilising stem cells for therapeutic cloning. Regulation 11 of the Draft Testing and Research Regulations.

[55] Regulation 4 of the Buccal Sample and Blood Withdrawal Regulations.

[56] As persons who are not healthcare professionals, as per regulation 4 of the Buccal Sample and Blood Withdrawal Regulations.

[57] Regulations 2 and 3 of the Buccal Sample and Blood Withdrawal Regulations.

[58] This is contrary to the position taken by other academics, who view this provision as allowing diabetics to withdraw blood themselves. Marlise Richter, WD Francois Venter & Andy Gray 'Home self-testing for HIV: AIDS exceptionalism gone wrong' (2010) 100(10) *SAMJ* 638.

> '[E]very part of a statute should be construed so as to be consistent, so far as possible, with every other part of that statute, and with every other unrepealed statute enacted by the Legislature'.[59]

In line with this, statutes dealing with identical subject matter should be read jointly.[60] Therefore, although the collection of saliva is not specifically mentioned in the NHA or the Human Biological Material Regulations, its inclusion in the Buccal Sample and Blood Withdrawal Regulations signals an intention to regulate it, with the assistance of certain registered professionals – thereby regulating it with the intention that the self-collection of saliva by individuals be banned.[61]

(d) There is no intention to ban self-withdrawal of human biological material that entails no or minimal risk

The fourth argument against the ban on individuals collecting their own saliva samples is that there is no intention to ban the self-withdrawal of human biological material where there is no, or minimal, risk. Genetic testing using blood commonly requires a full sample.[62] However, the Human Biological Material Regulations cater for situations where a finger prick to obtain a drop of blood is sufficient for *testing* and, in such a case, define a 'competent person' as someone registered in terms of the Health Professions Act.[63] But is the involvement of a 'competent person' necessary in this instance? A finger prick is a simple and non-invasive procedure which can, and is, commonly done at home without professional guidance – for example, in the case of diabetics who prick their fingers to obtain a drop of blood that allows them to check their blood sugar levels.[64] In the same light, should 'competent persons' be

---

[59] *Chotabhai v Union Government (Minister of Justice)* 1911 AD 13 at 24, cited in *Independent Institute of Education (Pty) Limited v Kwazulu-Natal Law Society* 2020 (2) SA 325 (CC) para 38.

[60] *Independent Institute of Education (Pty) Limited v Kwazulu-Natal Law Society* supra note 59 para 38. In *Ruta v Minister of Home Affairs* 2019 (2) SA 329 (CC) para 42, the court held that '[w]ell-established interpretive doctrine enjoins us to read the statutes alongside each other, so as to make sense of their provisions together'.

[61] Where both the removal and use of biological material is done by individuals, this is beyond the scope of the Human Biological Material Regulations. But where the removal and/or use of biological material is done for purposes as prescribed in the Human Biological Material Regulations (such as genetic testing), healthcare professionals are required to be involved and these acts fall within the ambit of the Human Biological Material Regulations. But even where collection is undertaken by individuals, as is the case in direct-to-consumer genetic testing, the testing is not done by individuals (the purpose of removal or the use of biological material), so it falls within the ambit of the Human Biological Material Regulations.

[62] Mayo Clinic 'Genetic testing' 14 April 2020 available at *https://www.mayoclinic.org/tests-procedures/genetic-testing/about/pac-20384827*, accessed on 5 January 2021.

[63] Regulation 1 of the Human Biological Material Regulations. The Human Biological Material Regulations explicitly mention a finger prick to obtain blood for the purpose of testing – testing is not specifically stated in terms of intravenous or intra-arterial blood withdrawal.

[64] If the *testing* referred to in terms of a finger prick under the definition of 'competent person' in regulation 1 of the Human Biological Material Regulations, is the same purpose as that referred to in regulation 2(a) of the Human Biological Material Regulations, then such testing would be *genetic testing*. But it is unlikely for a finger prick to be used for genetic testing as this usually requires a full blood sample. Therefore, if it refers to testing in general,

required to undertake non-invasive saliva sample collection for the purpose of a direct-to-consumer genetic test?

The Human Biological Material Regulations mention 'genetic testing' as a purpose for which biological material may be removed. Given that this can only be undertaken by a 'competent person' which, besides research and reproductive matters, refers to the withdrawal of blood,[65] it can be implied that the genetic testing referred to in the context of the Human Biological Material Regulations is the type which utilises a blood sample to extract, sequence, and analyse an individual's DNA. This is also strengthened by the fact that no reference is made in the Human Biological Material Regulations to a buccal sample – which is specifically referred to, alongside blood, in the Buccal Sample and Blood Withdrawal Regulations. While utilising blood samples is common for genetic testing in the clinical context, direct-to-consumer genetic testing utilises saliva samples, which consumers can easily collect at home. This merely entails spitting into a tube or swabbing the inside of one's cheek to obtain the necessary cells from which DNA can be extracted, sequenced, and analysed.[66] Although there are risks involved in saliva collection, such as sample contamination[67] or compromised DNA quality and yield[68] leading to inaccurate results, this does not require the same level of skill necessary for blood withdrawal – which involves inserting a needle into a vein, thus requiring precision and some medical knowledge. It is therefore evident that professional training and skill are necessary for genetic testing using blood, while collecting a saliva sample is a relatively straightforward process. In addition, testing companies typically provide step-by-

this may include diabetics who prick their fingers to *test* their blood sugar levels, in which case the involvement of a 'competent person' is necessary. If not, regulation 2(a) of the Human Biological Material Regulations allows a competent person to remove biological material for 'therapeutic purposes' – is checking one's blood sugar levels a therapeutic purpose? The meaning of 'therapeutic purpose' is broad and ranges from monitoring disease to the treatment thereof. While checking blood sugar levels may be deemed a therapeutic purpose, it is often not undertaken by a 'competent person'. Even if checking blood sugar is not deemed to be a therapeutic purpose, when reading the Human Biological Material Regulations with the Buccal Sample and Blood Withdrawal Regulations, withdrawing blood for testing requires a healthcare professional. Therefore, does this mean that all diabetics are contravening the Human Biological Material Regulations? On a narrow reading, the answer is yes – but it would be against public policy to enforce this. One cannot fine or imprison all diabetics. Regulation 14 of the Human Biological Material Regulations, dealing with offences, states that '[a]ny person who contravenes these regulations or fails to comply with any provision of these regulations, is guilty of an offence, and liable upon conviction to a fine or imprisonment of not more than 10 years, or both such fine and such imprisonment'. Robert Edwin Rakel 'Therapeutics' *Britannica* 2021, available at *https://www.britannica.com/science/therapeutics*, accessed on 6 May 2021; Collins Dictionary 'Therapeutic' available at *https://www.collinsdictionary.com/dictionary/english/therapeutic*, accessed on 6 May 2021.

[65] Regulation 1 of the Human Biological Material Regulations.

[66] 23andMe 'How it works' available at *https://www.23andme.com/howitworks/*, accessed on 20 November 2020; Bermseok Oh 'Direct-to-consumer genetic testing: Advantages and pitfalls' (2019) 17(3) *Genomics Inform* 1.

[67] Sharon A Thrush & Ruth McCaffrey 'Direct-to-consumer genetic testing: What the nurse practitioner should know' (2010) 6(4) *J Nurse Pract* 272.

[68] 23andMe 'Providing saliva sample for DNA test kit' available at *https://customercare.23andme.com/hc/en-us/articles/202904530-Providing-Saliva-Sample-for-DNA-Test-Kit*, accessed on 9 January 2021.

step instructions on how to collect a saliva sample which, if followed, should present no complications.

Therefore, I suggest that there is no intention to ban the self-withdrawal of biological material where there are no, or minimal, risks involved.[69] In comparison to other procedures such as blood withdrawal, collecting saliva samples can be done without professional oversight – making a strong case for direct-to-consumer genetic testing.

(e) The ban violates personal autonomy

The fifth argument against the ban on individuals collecting their own saliva samples relates to personal autonomy. Autonomy refers to individual freedom or self-determination.[70] The Stanford Encyclopedia of Philosophy defines autonomy as:

> '[A]n idea that is generally understood to refer to the capacity to be one's own person, to live one's life according to reasons and motives that are taken as one's own and not the product of manipulative or distorting external forces'.[71]

In South African case law, autonomy is recognised as a constitutional value that underlies various rights such as human dignity, freedom, and privacy. In the case of *NM v Smith* (*NM*)[72] O'Regan J, in her dissenting judgment, expanded on autonomy and held as follows:

> Recognising the role of freedom of expression in asserting the moral autonomy of individuals demonstrates the close links between freedom of expression and other constitutional rights such as human dignity, privacy and freedom. Underlying all these constitutional rights is the constitutional celebration of the possibility of morally autonomous human beings independently able to form opinions and act on them…Our Constitution seeks to assert and promote the autonomy of individuals…'[73]

[69] In the case of diabetics, a finger prick can be painful and may cause bleeding or bruising, yet individuals nevertheless do this themselves at home. However, collecting a saliva sample is painless and, if done correctly, should not have any side effects.

[70] It is also defined as 'the quality or state of being self-governing'. Donrich W Jordaan 'Autonomy as an element of human dignity in South African case law' (2009) 9(3) *Journal of Philosophy, Science & Law* 4; Merriam-Webster 'Autonomy' available at *https://www.merriam-webster.com/dictionary/autonomy*, accessed on 3 March 2021.

[71] John Christman 'Autonomy in moral and political philosophy' *Stanford Encyclopaedia of Philosophy* 29 June 2020, available at *https://plato.stanford.edu/entries/autonomy-moral/*, accessed on 12 July 2020.

[72] 2007 (5) SA 250 (CC).

[73] *NM* supra note 72 paras 145–6. This was also cited with approval in the High Court case of *AB v Minister of Social Development* 2016 (2) SA 27 (GP) paras 65–6 (*AB*), as well as being quoted by the Supreme Court of Appeal (SCA) in *British American Tobacco South Africa (Pty) Ltd v Minister of Health* [2012] 3 All SA 593 (SCA) para 13.

The Constitutional Court in *Barkhuizen v Napier* (*Barkhuizen*)[74] explicitly recognised 'self-autonomy'[75] as 'the ability to regulate one's own affairs, even to one's own detriment'.[76] This asserts autonomy as a value underlying the Constitution. Individuals *choose* to participate in direct-to-consumer genetic testing for a variety of reasons; the basic reason being to obtain results that hold insight and information regarding their genes. Part of this process requires the collection of a saliva sample by an individual, which is clearly an act of autonomy and self-determination. The value of autonomy, as well as the constitutional rights linked thereto, are relevant to direct-to-consumer genetic testing in several ways. Below, I discuss the constitutional rights of human dignity, privacy, and bodily integrity and show how, because each of these rights are infused by the value of autonomy, prohibiting individuals from collecting their own saliva samples for direct-to-consumer genetic testing infringes autonomy and is thereby a violation of the associated constitutional rights.

Firstly, human dignity is a right explicitly recognised in the Constitution.[77] In *National Coalition for Gay and Lesbian Equality v Minister of Justice*,[78] it was held that, at a minimum, dignity necessitates acknowledging 'the value and worth of all individuals as members of our society'.[79] Furthermore, *Teddy Bear Clinic for Abused Children v Minister of Justice and Constitutional Development* (*Teddy Bear Clinic*),[80] held that 'dignity recognises the inherent worth of all individuals…as members of our society, as well as *the value of the choices that they make*' (own emphasis).[81] In *Barkhuizen*, it was held that autonomy is 'the very essence of freedom and a vital part of dignity'.[82] This position was elaborated on by the majority of the Constitutional Court in *Member of the Executive Council for Education: Kwazulu-Natal v Pillay* (*Pillay*)[83] as follows:

> 'A necessary element of freedom and of dignity of any individual is an "entitlement to respect for the unique set of ends that the individual pursues"…That we choose voluntarily rather than

[74] 2007 (5) SA 323 (CC).
[75] I agree with the views of Thaldar and Steytler who state that the term 'self-autonomy', as used by the Constitutional Court in *Barkhuizen* supra note 74 para 57, is an 'unnecessary tautology' and does not differ from the term 'autonomy'. Donrich W Thaldar & Michaela Steytler 'Time for Cinderella to go to the ball: Reflections on the right to freedom of scientific research' (2020) 138(2) *SALJ* 273.
[76] *Barkhuizen* supra note 74 para 57.
[77] Section 10 of the Constitution states that '[e]veryone has inherent dignity and the right to have their dignity respected and protected'.
[78] 1999 (1) SA 6 (CC).
[79] *Barkhuizen* supra note 74 para 57.
[80] 2014 (2) SA 168 (CC).
[81] *Teddy Bear Clinic* supra note 80 para 52 cited in *AB* supra note 73 para 89.
[82] *Barkhuizen* supra note 74 para 57.
[83] 2008 (1) SA 474 (CC).

through a feeling of obligation only enhances the significance of a practice to our autonomy, our identity and our dignity'.[84]

Accordingly, human dignity demands that an individuals' autonomy be respected. In terms of direct-to-consumer genetic testing, individuals deciding to undergo such testing for their own reasons, and thereby collect a salvia sample, exercise their autonomy in doing so. Therefore, it follows that collecting a saliva sample for direct-to-consumer genetic testing is protected by the right to dignity and the apparent ban on this self-collection is an infringement of the right to human dignity.

Secondly, privacy is another right recognised in the Constitution,[85] and shares links with both autonomy and dignity. The right to privacy acknowledges that individuals 'have a right to a sphere of intimacy and autonomy that should be protected from invasion'.[86] In *Bernstein v Bester* (*Bernstein*),[87] Ackermann J referred to privacy as the 'inner sanctum of a person'.[88] The right to privacy includes the right to live, within a personal realm, as one pleases,[89] free from interference.[90] An individual's personal choice to undergo direct-to-consumer genetic testing falls within this 'inner sanctum' which should not be interfered with – thus being protected by the constitutional right to privacy. Therefore, the collection of a saliva sample for direct-to-consumer genetic testing, as an individual's own choice, should not be interfered with and doing so violates the right to privacy.

Thirdly, from a human rights perspective, individuals are entitled to make decisions regarding their own bodies, free from unwarranted involvement by others.[91] Bodily integrity refers to each individual's right to self-determination and autonomy regarding their body.[92]

---

[84] *Pillay* supra note 83 para 64. This was also repeated in *AB* supra note 73 para 89, where Basson J expressly stated that autonomy is 'a vital part of human dignity'.

[85] Section 14 of the Constitution states that '[e]veryone has the right to privacy, which includes the right not to have –
(a) their person or home searched;
(b) their property searched;
(c) their possessions seized; or
(d) the privacy of their communications infringed'.

[86] *Khumalo v Holomisa* 2002 (5) SA 401 (CC) para 27. In *NM* supra note 72 para 131, O'Regan J also held that privacy 'presupposes personal space within which to live this life'.

[87] 1996 (2) SA 751 (CC).

[88] *Bernstein* supra note 87 para 67.

[89] *NM* supra note 72 para 33.

[90] Ibid para 45.

[91] Nienaber and Bailey refer to this as the right to 'physical integrity'. A Nienaber & KN Bailey 'The right to physical integrity and informed refusal: Just how far does a patient's right to refuse medical treatment go?' (2016) 9(2) *SAJBL* 74.

[92] Ibid; CRIN 'Bodily integrity' 2018 available at *https://archive.crin.org/en/home/what-we-do/policy/bodily-integrity.html*, accessed on 14 April 2021.

Section 12(2)(*b*) of the Constitution, relating to the right to bodily and psychological integrity, consists of two elements: 'security in', and 'control over', one's body.[93] The former represents protecting bodily integrity against external intrusions, whilst the latter indicates the protection of 'bodily autonomy or self-determination against interference'.[94] It forms part of the right to be left alone in having the freedom to live as one chooses.[95] In line with this, it is unconstitutional to prevent individuals from doing as they wish with their own bodies, such as collecting saliva samples themselves for a direct-to-consumer genetic test, provided that it does not cause harm to others.

In order to promote autonomy, individuals should have the freedom and privacy to do as they wish with their own bodies, free from governmental control, where the risk of harm to themselves and others is minimal. In line with this, I suggest that the Human Biological Material Regulations, insofar as they make it illegal for consumers to collect their own saliva sample as part of a direct-to-consumer genetic test, are clearly overbroad and violate autonomy. While there are several concerns pertaining to direct-to-consumer genetic testing in the absence of a healthcare professional, there is no conceivable legitimate government purpose in preventing individuals from *collecting* their own saliva sample, and interference by the state or others offends personal autonomy.[96] Therefore, the relevant provisions in South Africa's extant law that purport to ban individuals from collecting their own saliva samples violate the rights to human dignity, privacy, and bodily integrity, and are unconstitutional and thus invalid.

---

[93] Section 12(2)(*b*) of the Constitution provides that '[e]veryone has the right to bodily and psychological integrity', including the right to 'security in and control over their body'. The right to security in one's body prohibits undesirable interference of bodily integrity (such as forced medical treatment), while the right to control over one's body refers to the ability to make independent and autonomous decisions concerning one's body. Nienaber & Bailey op cit note 91 at 74.

[94] Iain Currie & Johan De Waal *The Bill of Rights Handbook* 6 ed (2013) 287.

[95] Ibid.

[96] Rights contained in the Bill of Rights may be limited in specific instances, where there is justifiable limitation in terms of section 36 of the Constitution. Section 36(1) of the Constitution states that '[t]he rights in the Bill of Rights may be limited only in terms of law of general application to the extent that the limitation is reasonable and justifiable in an open and democratic society based on human dignity, equality and freedom, taking into account all relevant factors, including –

(a) the nature of the right;
(b) the importance of the purpose of the limitation;
(c) the nature and extent of the limitation;
(d) the relation between the limitation and its purpose; and
(e) less restrictive means to achieve the purpose'.

This section protects individual rights by ensuring that government is held accountable for any actions which may limit rights. Nienaber & Bailey op cit note 91 at 74.

(f) The ban conflicts with a purposive interpretation

The sixth argument against the ban on individuals collecting their own saliva samples for the purpose of a direct-to-consumer genetic test is that a plain reading of the relevant provisions in the Human Biological Material Regulations are both incompatible with a purposive approach to statutory interpretation, and are also unconstitutional.[97] As mentioned above, this approach seeks to ascertain the legislation's intent,[98] in line with constitutional values.[99] This involves reading a statute in its entirety and placing the objective of a statute and the relationship between its provisions in context.[100]

The Human Biological Material Regulations were promulgated in terms of section 68 of the NHA. Section 68(1)(*p*) of the NHA permits the Minister to make regulations regarding 'the acquisition, storage, harvesting, utilisation or manipulation of tissue…for any purpose'.[101] In line with this, I suggest that the likely purpose of the Human Biological Material Regulations is to govern the removal and use of biological material for various medical and dental purposes, which is to be done under proper supervision *in order to mitigate risks* – thus the requirement for the involvement of 'competent persons'. Although there are various other risks associated with direct-to-consumer genetic testing, such as the undetermined accuracy and usefulness of test results, and the ability of consumers to understand them without professional assistance,[102] the *risks involved in the self-collection of saliva are minimal*.

A purposive approach to statutory interpretation would allow individuals to collect their own saliva samples, in line with the constitutional arguments advanced above. Banning

[97] For an explanation of a purposive interpretation, see *Bato Star* paras 90 and 91; *Bertie Van Zyl* para 21.
[98] Annette Singh *The Impact of the Constitution on Transforming the Process of Statutory Interpretation in South Africa* (unpublished PhD thesis, University of KwaZulu-Natal, 2014) 47.
[99] GK Goldswain 'The purposive approach to the interpretation of fiscal legislation – The winds of change' (2008) 16(2) *Meditari Accountancy Research* 114.
[100] A statute's purpose assists in determining 'a context that clarifies the scope and intended effect of a law'. In *S v Mhlungu* 1995 (3) SA 867 (CC) at 916 it was held that 'Judges do not go by the literal meaning of the words or by the grammatical structure of the sentence. They go by the design or purpose which lies behind it. When they come upon a situation which is to their minds within the spirit – but not the letter – of the legislation, they solve the problem by looking at the design and purpose of the legislature – at the effect it was sought to achieve. They then interpret the legislation so as to produce the desired effect…They ask simply: what is the sensible way of dealing with this situation so as to give effect to the presumed purpose of the legislation'. Ibid at 111. See also, *Bertie Van Zyl* supra note 97 para 21; Singh op cit note 98 at 47.
[101] Section 68(1)(*p*) of the NHA.
[102] There are also several sources of risk in direct-to-consumer genetic testing – whether in the procurement process, the handling process, the testing process, the interpretation process, or the process of delivering test results to consumers and giving them advice.

individuals from doing so does not advance the 'the spirit, purport and objects of the Bill of Rights',[103] and violates the 'the democratic values of human dignity, equality and freedom'.[104]

III CONCLUSION

This Chapter examined one of the primary steps in the direct-to-consumer genetic testing process: The collection of a saliva sample; as well as one of the first major legal hurdles: Whether consumers are deemed to be 'competent persons' in terms of legislation and thus legally permitted to collect their own saliva samples. Although the first three arguments, dealing with the de minimis principle, consent, and whether there is an intention to ban or not to regulate, do not assist greatly in opposing the legal ban on the self-collection of saliva for a direct-to-consumer genetic test,[105] the subsequent three arguments, which considered the regulation of saliva, personal autonomy, and a purposive approach to statutory interpretation, provide some impetus.[106] Based on a plain reading of the NHA and the Human Biological Material Regulations,[107] the first step in the direct-to-consumer genetic testing process – namely, the collection of saliva samples by consumers – is illegal. This is because individuals are not 'competent persons' and are therefore banned from collecting their own saliva for the purpose of a direct-to-consumer genetic test. Given the arguments advanced above, I suggest that this provisional conclusion is unsustainable, and the self-collection of saliva should not be unlawful.

Although the involvement of healthcare professionals in direct-to-consumer genetic testing appears to be a growing trend amongst testing companies, there are still those that bypass such professionals and involve only the individual. As such, I suggest that the Human Biological Material Regulations be amended to accommodate situations wherein individuals

---

[103] *Bato Star* supra note 97 para 72.
[104] Ibid.
[105] I established that, in terms of argument (1), the de minimis principle is speculative and does not assist with legal certainty and that, in terms of argument (2), consent by consumers cannot override the fact that legislation (specifically the NHA, the General Control Regulations, and the Human Biological Material Regulations) requires a 'competent person' to remove biological material and consent does not allow an individual to collect their own saliva sample. In terms of argument (3), while saliva and its collection were not included in the Human Biological Material Regulations, their inclusion in the Buccal Sample and Blood Withdrawal Regulations signifies that there is not an intention to ban such activities, but rather an intention to regulate them with professional assistance.
[106] Argument (4) showed that it is overbroad and nonsensical to prevent individuals from collecting their own saliva sample where it poses only a minimal risk of harm. Argument (5) showed that legislating on the removal of saliva by individuals infringes the constitutional rights to dignity, privacy, and bodily integrity, violates an individual's autonomy, and is thus unconstitutional. Finally, argument (6) considers how the ban on the self-collection of saliva samples by individuals conflicts with a purposive approach to statutory interpretation.
[107] A plain reading requires words to be given their literal or grammatical meaning. CJ Botha *Statutory Interpretation* 5 ed (2012) 193 cited in Singh op cit note 98 at 30.

collect their own biological material[108] – aligned with the Draft Testing and Research Regulations[109] – specifically in circumstances where the chance of inflicting physical self-harm is negligible or, at the very least, saliva is recognised as a type of biological material that can be removed by a 'competent person'. This includes broadening the scope of those deemed to be 'competent persons' in the Human Biological Material Regulations and allowing for the removal of biological material by individuals and outside of prescribed or authorised institutions.[110] Provided that consumers have consented to testing and understand that test results are not definitive for determining their predisposition to certain diseases and conditions, I see no reason why consumers cannot collect their own saliva samples for the purpose of a direct-to-consumer genetic test, and the law should allow for this.

[108] In the form of saliva for the purpose of a direct-to-consumer genetic test, or blood in terms of a finger prick for testing blood sugar levels.

[109] While regulation 1 of the Draft Testing and Research Regulations provide a definition of 'competent person' which includes individuals and allows them to collect their own saliva samples, this definition in the Draft Testing and Research Regulations conflicts with a plain reading of the NHA and the Human Biological Material Regulations.

[110] As per regulation 2(b) of the Human Biological Material Regulations. Although one way of making the wording of the Human Biological Material Regulations constitutional is to read in competent persons or persons themselves, this may not solve the problem as persons themselves cannot take their own blood. This may therefore necessitate the need for another clause to cater for certain categories of things that pose a minimal risk – such as buccal swabs or finger pricks.

CHAPTER 4

DIRECT-TO-CONSUMER GENETIC TESTS AS MEDICAL DEVICES

I INTRODUCTION AND CHAPTER OVERVIEW

In the previous chapters, I have outlined the law relating to the process of direct-to-consumer genetic testing. Now, I focus on the legal status of the tests themselves. In the US, direct-to-consumer genetic tests are regulated by the Food and Drug Administration (FDA) as medical devices. Conversely, the UK DoH and Health Canada do not view direct-to-consumer genetic tests, specifically those developed by 23andMe, as high-risk medical devices.[1]

In this Chapter, I examine South Africa's medical device legislation to determine whether direct-to-consumer genetic tests are medical devices, IVDs, or both – and if so, how they should be classified. This provides clarity regarding whether, and how, direct-to-consumer genetic tests align with extant laws, which holds implications for the licensing, registering, importing, exporting, and advertising of such products and services. This Chapter primarily examines the Medicines Act and the Medical Device Regulations.

II ARE DIRECT-TO-CONSUMER GENETIC TESTS MEDICAL DEVICES, IN VITRO DIAGNOSTIC DEVICES, OR BOTH?

Direct-to-consumer genetic tests are offered to individuals who may not be ill or at greater risk of disease, but are simply interested or worried about their predisposition.[2] However, direct-to-consumer genetic tests are generally stand-alone without confirmatory testing; they test for diseases and conditions which may have a severe impact on individuals, but lack confirmed analytical and clinical validity; and they entail dangers inherent to new devices – undetermined efficiency and deficient knowledge.[3] If direct-to-consumer genetic tests are treated as non-

[1] Jessica Cussins 'Direct-to-consumer genetic tests should come with a health warning' *The Pharmaceutical Journal* 15 January 2015, available at *https://pharmaceutical-journal.com/article/opinion/direct-to-consumer-genetic-tests-should-come-with-a-health-warning*, accessed on 23 February 2021; André Picard 'Controversial genetic self-testing kits coming to Canada' *The Globe and Mail* 2 October 2014, available at *https://www.theglobeandmail.com/life/health-and-fitness/health/genetic-self-testing-kits-to-come-to-canada/article20885678/?click=sf_globefb*, accessed on 7 December 2020.

[2] Harvard Women's Health Watch 'Direct-to-consumer genetic testing kits' *Harvard Health Publishing* September 2010, available at *https://www.health.harvard.edu/newsletter_article/direct-to-consumer-genetic-testing-kits*, accessed on 5 February 2021.

[3] Stuart Hogarth & David Melzer *The IVD Directive and Genetic Testing Problems and Proposals: A Briefing Presented to the 20th Meeting of Competent Authorities* (2007) 10.

medical devices, this may hold risks for consumer safety, but viewing such tests as medical devices may lead to untenable costs and restrictions for this expanding industry.[4]

(a) Medical device

While the supply and distribution of medicines was controlled by the Medicines Control Council (MCC), medical devices were unregulated in South Africa until the implementation of the Medical Device Regulations in 2016. Given that the FDA in the US informed various testing companies that their tests qualified as medical devices and required pre-market approval,[5] examining the regulation of medical devices becomes relevant. This is also due to concerns that testing companies make claims, unsupported by scientific evidence, regarding a test's value in health decision-making.[6] Thus, it is necessary to determine the meaning of

[4] Federica Lucivero & Barbara Prainsack 'The lifestylisation of healthcare? "Consumer genomics" and mobile health as technologies for healthy lifestyle' (2015) 4 *Appl Transl Genom* 47.

[5] In November 2013, the FDA barred direct-to-consumer genetic testing for health-related conditions in the US through the issuance of a warning letter to 23andMe, compelling the company to cease offering such tests until it received FDA authorisation. The FDA's letter stated that 23andMe's Saliva Collection Kit and Personal Genome Service (PGS) – which offered health-related information on numerous diseases and conditions as well as information on non-disease traits and genealogy – was a medical device, as per section 201(h) of the of the Federal Food, Drug, and Cosmetic Act, 21 USC § 321(h) (FDCA), and therefore required pre-market approval as there was also no evidence regarding the success of the tests and accuracy of the results. The FDA regulates devices that are intended to diagnose disease or those aimed at curing, treating, or preventing disease. As 23andMe's health-related direct-to-consumer genetic tests offered information on various diseases and conditions, and based on the manufacturers claims regarding test results, the FDA deemed them to be medical devices. For example, 23andMe marketed its PGS as offering health reports on numerous diseases and conditions, including 'carrier status' and 'health risks'. 23andMe also stated that its PGS was 'the first step in prevention' and allowed consumers to 'take steps toward mitigating serious diseases', suggestive of something 'intended for use in the diagnosis…or prevention of disease' in terms of the statutory definition of a device in section 201(h) of the FDCA. Following this, 23andMe declared its intention to halt offering health-related information to new consumers, but would continue marketing its PGS, limited only to 'ancestry-related genetic information and…raw data without 23andMe's interpretation'. Patricia J Zettler, Jacob S Sherkow & Henry T Greely '23andMe, the Food and Drug Administration, and the future of genetic testing' (2014) 174(4) *JAMA Internal Medicine* 493; Arthur A Daemmrich '23andMe: The business and ethics of personal genetics testing' (2015) *University of Kansas School of Medicine* 7–8. See also, Kelly Servick 'Frustrated U.S. FDA issues warning to 23andMe' *Science* 25 November 2013, available at *https://www.sciencemag.org/news/2013/11/frustrated-us-fda-issues-warning-23andMe*, accessed on 15 October 2019; Andrew Pollack 'FDA orders genetic testing firm to stop selling DNA analysis service' *The New York Times* 25 November 2013, available at *https://www.nytimes.com/2013/11/26/business/fda-demands-a-halt-to-a-dna-test-kits-marketing.html*, accessed on 15 October 2019; Seon-Hee Yim & Yeun-Jun Chung 'Reflections on the US FDA's warning on direct-to-consumer genetic testing' (2014) 12(4) *Genomics Inform* 151; Donna Dickenson 'Testing times for the consumer genetics revolution' (2014) 221(2951) *New Scientist* 26; Megan A Allyse, David H Robinson, Matthew J Ferber et al 'Direct-to-consumer testing 2.0: Emerging models of direct-to-consumer genetic testing' (2018) 93(1) *Symposium on Precision Medicine* 118; Robert C Green & Nita A Farahany 'The FDA is overcautious on consumer genomics' (2014) 505 *Nature* 286; Justin P Annes, Monica A Giovanni & Michael F Murray 'Risks of presymptomatic direct-to-consumer genetic testing' (2010) 363(12) *N Engl J Med* 1100; Amanda Holpuch 'FDA orders genetics company 23andMe to cease marketing of screening service' *The Guardian* 25 November 2013, available at *https://www.theguardian.com/science/2013/nov/25/genetics-23andme-fda-marketing-pgs-screening*, accessed on 15 October 2019.

[6] Harvard Women's Health Watch op cit note 2.

'medical device' in South African law and whether health-related direct-to-consumer genetic tests meet this definition. The Medicines Act defines a 'medical device' as –

> 'any instrument, apparatus, implement, machine, appliance, implant, reagent for in vitro use, software, material or other similar or related article… –
>
> (a) intended by the manufacturer to be used, alone or in combination, for humans or animals, for one or more of the following:
>
> (i) diagnosis, prevention, monitoring, treatment or alleviation of disease;
>
> …
>
> (vii) providing information for medical or diagnostic purposes by means of in vitro examination of specimens derived from the human body; and
>
> (b) which does not achieve its primary intended action by pharmacological, immunological or metabolic means, in or on the human or animal body, but which may be assisted in its intended function by such means'.[7]

The use of the word 'and' between subsections 1(h)(*a*) and (*b*) of the definition of 'medical device'[8] means that both criteria must be met for a direct-to-consumer genetic test to be deemed a 'medical device'.[9] This is bolstered by the fact that in the case of *Berman v Teiman*,[10] it was held that the words 'and/or' must be 'read disjunctively as well as *conjunctively*' (own emphasis).[11]

In South Africa, there is little guidance on whether direct-to-consumer genetic tests constitute medical devices.[12] According to the ASSAf Report, medical devices 'include diagnostic tests and would therefore also cover genetic tests'.[13] This leads the ASSAf Report to recommend that the South African Health Products Regulatory Authority (SAHPRA) should regulate genetic tests under the Medicines Act. However, no mention is made of direct-to-consumer genetic tests specifically. The ASSAf Report's reference to genetic tests likely refers to genetic tests in the clinical setting, and not direct-to-consumer genetic tests. This is because,

[7] Subsection (h) under the definition of 'medical device' in section 1 of the Medicines Act.
[8] Section 1 of the Medicines Act.
[9] The definition of 'medical device' has undergone alterations over the years, initially mentioned in the Medicines and Related Substances Act 101 of 1965, followed by the Medicines and Related Substances Amendment Act 72 of 2008, and finally the definition in the current Medicines Act.
[10] 1975 (1) SA 756 (W).
[11] *Berman v Teiman* supra note 10 at 757. See also, *Barnett v Estate Gumpert* 1926 CPD 363 at 364–5; *Aird v Hockly's Estate* 1937 EDL 34 at 42. There is no South African case law dealing with the meaning of the word 'and', only the words 'and/or'.
[12] Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018) 75.
[13] Ibid at 75–6.

while direct-to-consumer genetic tests sometimes purport to diagnose disease, they mostly do not due to their lack of analytical and clinical validity, making diagnosis problematic.

A pivotal aspect of the definition of 'medical device' in the Medicines Act is part (a), which refers to the intention of the manufacturer.[14] This indicates that the focus is on the manufacturer and what they deem the purpose of the device to be – including whether it is medical or diagnostic, or not. If the manufacturer, which in most cases is the testing company, does not intend the device to be used in inter alia the diagnosis, prevention, or monitoring of disease, or to provide information that is medical or diagnostic, then it does not qualify as a medical device as per section 1 of the Medicines Act.[15]

Testing companies, often on their websites, may make certain medical claims about their tests. If that is their intention, then the definition of 'medical device' in the Medicines Act applies.[16] I acknowledge that there are cases where these claims are ambivalent and those will therefore need to be dealt with on a case-by-case basis to determine whether a manufacturer has a medical or diagnostic intention.[17] However, for the purposes of this dissertation, my focus is not on particular testing companies, but rather on the abstract. The definition of 'medical device'[18] makes a binary distinction – either there is an intention or there is not. Therefore, I distinguish between those manufacturers with an intention – thus potentially falling within the definition of 'medical device'[19] – and those without.

(i) Non-medical devices

Although prima facie health-related direct-to-consumer genetic tests appear to be medical in nature, this is not necessarily what the manufacturer intends. This is evidenced by the fact that most testing companies (including those based in South Africa), in their terms and conditions, include statements that their tests are informational and do not constitute medical diagnoses.[20]

---

[14] As per subsection (a) under the definition of 'medical device' in section 1 of the Medicines Act.
[15] Despite the fact that testing companies do not intend a direct-to-consumer genetic test to be used for a certain purpose, they still see a possibility of it being used for that purpose which is why testing companies have disclaimers, and this would suffice for intention (proof of foreseeability is in the fact they have a label saying this is not meant to be used).
[16] Section 1 of the Medicines Act.
[17] As per the definition of 'medical device' in section 1 of the Medicines Act.
[18] Section 1 of the Medicines Act.
[19] Ibid.
[20] For example, see 23andMe 'Health + ancestry service' available at *https://www.23andme.com/dna-health-ancestry?mkpc=true*, accessed on 5 October 2019; 23andMe '23andMe for healthcare professionals' available at *https://medical.23andme.com/*, accessed on 5 October 2019; Ancestry 'Now your DNA reveals so much more with AncestryHealth' available at *https://www.ancestry.com/health*, accessed on 5 October 2019; MyHeritage 'Terms and conditions' available at *https://www.myheritage.com/terms-and-conditions*, accessed on 5 October 2019. For examples of South African testing companies, see DNAlysis 'Privacy policy' 2020 available at

When I speak of direct-to-consumer genetic tests as medical devices, I am not referring to all direct-to-consumer genetic tests – rather only those that intend to make medical or diagnostic claims or those that are regulated as medical devices.[21]

(ii) Medical devices

This section pertains to direct-to-consumer genetic tests that are medical devices as they are intended by the manufacturer to diagnose, prevent, monitor, treat, or alleviate disease, or provide 'information for medical or diagnostic purposes by means of in vitro examination of specimens derived from the human body'.[22]

The first part of the definition of 'medical device' in the Medicines Act mentions a variety of articles that are considered to be medical devices.[23] It is also inclusive of other 'similar or related' articles, meaning that it is not limited to what is expressly mentioned under the definition.[24] The various aspects of direct-to-consumer genetic tests can be seen to fall under the articles mentioned in the definition of 'medical device'.[25] For example, the tube which collects saliva samples can be deemed an 'instrument',[26] 'apparatus',[27] or 'implement'[28] (so too can the machines used for DNA extraction, sequencing, and analysis be considered instruments, apparatus, or implements as they are tools or devices used to perform a specific task, namely obtaining and examining DNA); the DNA extractor and sequencer can be seen as

https://dnalysis.co.za/privacy-policy/, accessed on 22 June 2020; GeneWay 'FAQ – Frequently asked questions' 2020 available at *https://www.geneway.co.za/faq-frequently-asked-questions*, accessed on 22 June 2020.

[21] 23andMe is one of the few testing companies with a medical intention, thus going the regulation route and having their tests governed as medical devices in the US. Helix is another company that is regulated as a medical device in the US, and has received approval to market inter alia its Genetic Health Risk App for late-onset Alzheimer's disease for over-the-counter use. Zettler et al op cit note 5 at 493; Daemmrich op cit note 5 at 7–8. Green & Farahany op cit note 5 at 286; Helix 'Helix Laboratory Platform granted the first and only FDA authorization for a whole exome sequencing platform' 11 January 2021 available at *https://www.helix.com/pages/helix-laboratory-platform-granted-fda-authorization*, accessed on 7 April 2021.

[22] As per the definition of 'medical device' in section 1 of the Medicines Act.

[23] Section 1 of the Medicines Act, subsection (a) under the definition of 'medical device'.

[24] Subsection (a) under the definition of 'medical device' in section 1 of the Medicines Act.

[25] Section 1 of the Medicines Act.

[26] 'Instrument' is defined as 'a tool or device that is used to do a particular task, especially a scientific task'. Collins Dictionary 'Instrument' available at *https://www.collinsdictionary.com/dictionary/english/instrument*, accessed on 6 January 2021.

[27] 'Apparatus' means 'a set of equipment or tools or a machine that is used for a particular purpose'. Cambridge Dictionary 'Apparatus' available at *https://dictionary.cambridge.org/dictionary/english/apparatus*, accessed on 6 January 2021.

[28] 'Implement' has been defined as 'a device used in the performance of a task'. Merriam-Webster 'Implement' available at *https://www.merriam-webster.com/dictionary/implement*, accessed on 6 January 2021.

a 'machine'[29] or 'appliance';[30] and it is 'software'[31] on these machines or appliances that assists in the extraction, sequencing, and analysis of DNA. Therefore, I suggest that the articles used in the various stages of the direct-to-consumer genetic testing process are included in this part of the definition of 'medical device' in the Medicines Act.[32]

But these individual aspects, as well as the direct-to-consumer genetic test in general, cannot be a medical device without being able to perform certain functions. The definition of 'medical device' stipulates certain purposes for which medical devices are intended to be used, either alone or jointly, the most relevant of which is subsection (a)(*vii*).[33] It refers to the provision of information 'for medical or diagnostic purposes by means of in vitro examination of specimens derived from the human body'.[34] This, specifically the meaning of 'medical or diagnostic purposes',[35] requires further examination.

The information aspect of subsection (a)(*vii*) under the definition of 'medical device' mentions that a medical device inter alia provides information for 'medical or diagnostic purposes'.[36] Firstly, in terms of the provision of information for 'diagnostic purposes',[37] direct-to-consumer genetic tests, due to their unclear accuracy are primarily informational tools that cannot be seen to offer definite diagnoses for disease – rather they provide risk estimates and identify whether an individual possesses a particular genetic mutation that *may* lead to the development of certain diseases or conditions. The uncertain analytical and clinical validity of direct-to-consumer genetic tests, the variations in results among testing companies, as well as environmental influences means that a definitive diagnosis, in any event, may be inaccurate. The terms and conditions available on some testing company websites typically state that they are not medical professionals, and the tests are merely for informational or educational

[29] 'Machine' means 'a piece of equipment with several moving parts that uses power to do a particular type of work'. Cambridge Dictionary 'Machine' available at *https://dictionary.cambridge.org/dictionary/english/machine*, accessed on 6 January 2021.
[30] 'Appliance' refers to 'a device, machine, or piece of equipment, especially an electrical one that is used in the house, such as a cooker or washing machine'. Cambridge Dictionary 'Appliance' available at *https://dictionary.cambridge.org/dictionary/english/appliance*, accessed on 6 January 2021.
[31] The meaning of 'software' is 'instructions that tell a computer what to do'. Software consists of all programs and procedures linked to the working of a computer system, as opposed to 'hardware', which is a computer's physical parts. Britannica 'Software' available at *https://www.britannica.com/technology/software*, accessed on 6 January 2021.
[32] Section 1 of the Medicines Act.
[33] Under the definition of 'medical device' in section 1 of the Medicines Act.
[34] Subsection (a)(*vii*) under the definition of 'medical device' in section 1 of the Medicines Act.
[35] Ibid.
[36] Ibid.
[37] Ibid.

purposes and do not diagnose disease.[38] But when a consumer discovers, through a direct-to-consumer genetic test, that they are at risk of developing Huntington's disease or that they possess the breast cancer (BRCA) gene, it is doubtful whether they will view this information as non-diagnostic.

Secondly, in terms of supplying information for medical purposes,[39] although direct-to-consumer genetic tests cannot be concretely used for diagnostic purposes, health-related direct-to-consumer genetic tests are relevant for medical purposes as they provide information on health and predisposition to disease. As many testing companies advise consumers to consult with healthcare professionals regarding their results, it can be inferred that these tests provide information that is medical in nature and, as a result, assistance is recommended to ensure proper understanding and avoid harm and potentially unnecessary health decisions. Additionally, as DNA derived from saliva samples of consumers is sequenced and analysed in vitro,[40] meaning outside of the body, I suggest that such tests meet the first part of the definition of 'medical device' in the Medicines Act.[41]

In terms of the second part of the definition of 'medical device',[42] I suggest that direct-to-consumer genetic testing cannot provide health-related information about an individual, its 'primary intended action',[43] without a saliva sample – from which DNA is extracted, sequenced, and analysed – and in vitro examination. But pharmacological, immunological, or metabolic means may assist in the testing process by providing genetic information that influences test results. Processes occurring within the body, as well as genetics, determine the

[38] For example, 23andMe states that their PGS test does not aim to diagnose disease, inform consumers of their current state of health, or make medical decisions without confirmation in the clinical context. It does not replace visits to healthcare practitioners for screenings or follow-ups. Ancestry provides that their test results are not diagnostic and do not indicate the likelihood of developing a disease or condition. They advise consulting with a healthcare practitioner prior to taking any action. MyHeritage says that their DNA Health Services are not meant to diagnose, prevent, or treat a disease or condition or be used in medical decision-making. Consumers are advised to consult with physicians, genetic counsellors, or other healthcare providers to acquire accurate results. The DNA Health Services does not constitute medical advice, form the practice of medicine, or establish a doctor-patient relationship. There are also several South African companies offering direct-to-consumer genetic testing, which have similar information in their terms and conditions. DNAlysis states that they do not intend to provide consumers with medical advice or diagnose disease, but offer information that will allow a greater understanding of health risks and benefits based on genotype. They appeal to consumers to visit a healthcare practitioner as the test results do not verify or alter diagnoses from healthcare practitioners. GeneWay informs consumers that their results are informational and should not be utilised in making medical or health decisions without consulting a healthcare professional as their genetic tests are not diagnostic or deterministic of potential risk outcomes. 23andMe op cit note 20; 23andMe op cit note 20; Ancestry op cit note 20; MyHeritage op cit note 20; DNAlysis op cit note 20; GeneWay op cit note 20.

[39] Subsection (a)(*vii*) under the definition of 'medical device' in section 1 of the Medicines Act.

[40] Ibid.

[41] This refers to subsection (a) under the definition of 'medical device' in section 1 of the Medicines Act.

[42] This refers to subsection (b) under the definition of 'medical device' in section 1 of the Medicines Act.

[43] Subsection (b) under the definition of 'medical device' in section 1 of the Medicines Act.

outcome of the test. Therefore, given the above, I suggest that direct-to-consumer genetic tests intended by the manufacturer to offer information for medical or diagnostic purposes meet the definition of 'medical device' in the Medicines Act.[44]

Therefore, while some direct-to-consumer genetic tests meet the definition of 'medical device',[45] others do not due to the manufacturer's intention. However, can it be that the manufacturer's intention, in addition to its profit-making goals, is the determining factor in the circumvention of regulation? The South African Medical Association (SAMA) cautioned the public against utilising HIV self-testing kits, noting that unmanaged self-testing is unsafe and may have devastating effects on individuals.[46] This is due to inter alia test accuracy, a dearth of counselling, and result misinterpretation.[47] Likewise, I am of the opinion that given the possible mental anguish that some direct-to-consumer genetic tests may cause because of the sensitive and largely unknown nature and implications of the information that it provides and due to the fact that proper genetic counselling is not necessarily offered, these tests require better regulation on the basis that they may cause psychological harm, which could outweigh the informational benefit. Thus, even if a direct-to-consumer genetic test proclaims to merely offer information, it may still have a psychological effect on consumers to the extent that it is a separate consideration for the manufacturer's intention. Based on South Africa's extant law, in the sections below, as well as the chapters that follow, I distinguish between the two scenarios established above: Direct-to-consumer genetic tests that are deemed to be medical devices, and those that are not.

(b) In vitro diagnostic device (IVD)

Direct-to-consumer genetic tests that are not medical devices, are automatically disqualified from being IVDs because the definition of 'IVD' makes mention of a 'medical device', meaning that to qualify as an IVD, the article in question must be a medical device. Like medical devices, IVDs provide information for various purposes – including that they offer consumers information about certain diseases and conditions as well as health status.[48]

[44] Section 1 of the Medicines Act.
[45] Ibid.
[46] These concerns were repeated by the DoH and the Treatment Action Campaign (TAC). Marlise Richter, WD Francois Venter & Andy Gray 'Home self-testing for HIV: AIDS exceptionalism gone wrong' (2010) 100(10) *SAMJ* 636.
[47] Ibid.
[48] Robyn Howes 'SALDA In Vitro Diagnostics in South Africa' (2014) *SALDA* 2.

Therefore, the next logical step is to determine whether direct-to-consumer genetic tests that are medical devices meet the definition of 'IVD'. The Medicines Act defines an 'IVD' as –

> '[A] medical device, whether used alone or in combination, intended by the manufacturer for the *in vitro* examination of specimens derived from the human body solely or principally to provide information for diagnostic, monitoring or compatibility purposes'.[49]

An identical definition of 'IVD' also appears in the Medical Device Regulations.[50] It includes specimen receptacles, control materials, reagents, calibrators, and software.[51] IVDs have no direct interaction with the human body and examine specimens in vitro. Because testing companies analyse the DNA of consumers extracted from saliva samples, the examination of 'specimens derived from the human body' (saliva containing DNA) occurs 'in vitro' – thus in line with the definition of 'IVD'.[52] By doing this, the main purpose of health-related direct-to-consumer genetic testing is to provide consumers with information regarding their susceptibility to various genetic diseases and conditions – this appears to be consistent with the definition of 'IVD'.[53] However, can this information provided by testing companies be used for 'diagnostic, monitoring or compatibility purposes'?[54]

'Diagnostic' is a relevant term to consider as this has generated debate in the direct-to-consumer genetic testing industry. In the US, the FDA prohibited 23andMe from offering *diagnostic* direct-to-consumer genetic tests for health-related conditions,[55] as it requires 'stricter regulatory oversight'.[56] This was partly due to their questionable success and result accuracy. There is still uncertainty regarding the analytical and clinical validity of direct-to-

[49] Section 1 of the Medicines Act.
[50] Regulation 1 of the Medical Device Regulations.
[51] But it precludes non-IVDs for general laboratory use unless, given their characteristics, they are utilised for the specific purposes mentioned in the definition of 'IVD'. Section 1 of the Medicines Act; regulation 1 of the Medical Device Regulations. See also, Howes op cit note 48 at 2.
[52] Subsection (f) under the definition of 'medical device' in section 1 of the Medicines Act; Regulation 1 of the Medical Device Regulations.
[53] Section 1 of the Medicines Act; regulation 1 of the Medical Device Regulations.
[54] Regulation 1 of the Medical Device Regulations.
[55] The FDA defined diagnostic tests as 'tests [that] are often used as the sole basis for major treatment decisions'. US Food and Drug Administration 'Review memorandum: 23andMe Personal Genome Service (PGS) test' 6 April 2017 available at *https://www.accessdata.fda.gov/cdrh_docs/pdf16/den160026.pdf,* accessed on 3 October 2019 cited in Allyse et al op cit note 5 at 118.
[56] The FDA regulates genetic tests as devices when they are 'intended for use in the diagnosis…or prevention of disease' (as per section 201(h) of the FDCA, but has refrained from regulating many health-related genetic tests. Allyse et al op cit note 5 at 118; Zettler et al op cit note 5 at 493. See also, Green & Farahany op cit note 5 at 286; Pollack op cit note 5; Annes et al op cit note 5 at 1100; Holpuch op cit note 5; Daemmrich op cit note 5 at 7–8.

consumer genetic tests, bringing into question their ability to offer definitive and accurate diagnostic information.[57]

Direct-to-consumer genetic tests may provide information for monitoring purposes.[58] Although 'monitoring' is not defined in the Medical Device Regulations, it can be seen to mean regular observation and checking.[59] As health-related direct-to-consumer genetic tests provide consumers with information regarding genetic predisposition, such tests may alert consumers to conditions that they may not have otherwise been aware of, allowing them to *monitor* and seek treatment, if necessary. In this regard, and given that such direct-to-consumer genetic tests have a health-related function, I suggest that they offer information for 'monitoring' purposes in line with the definition of 'IVD'.[60]

Despite this, the wording of the definition of 'IVD' states that IVDs are intended to provide information 'solely or *principally*'[61] (own emphasis) for such purposes. In the case of *Selection Park Investments (Pty) Ltd v Friedman* (*Friedman*),[62] it was held that the ordinary dictionary meaning of the word 'mainly', also meant inter alia 'principally'.[63] Based on this,

[57] Although the FDA has previously only approved tests for carrier status, in 2017 the agency, following a successful pre-market approval process, authorised the marketing of the first direct-to-consumer genetic test that offers information about predisposition to disease. This test, 23andMe's PGS Genetic Health Risk (GHR), allowed 23andMe to expand its health risk offerings and provides genetic health information for ten multifactorial conditions, including Parkinson's disease, celiac disease, and late-onset Alzheimer's disease. Subsequently, the FDA has approved further tests from 23andMe that determine the risk of developing a condition or disease. The agency also stated that direct-to-consumer genetic tests for further diseases or conditions would be exempt from pre-market review, as long as they satisfy the GHR category's requirements. Stephany Tandy-Connor, Jenna Guiltinan, Kate Krempely et al 'False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care' (2018) *Genet Med* 1; Abigail Hoglund-Shen 'Direct-to-consumer genetic testing, gamete donation, and the law' (2017) 55(3) *Family Court Review* 475; Amanda K Sarata 'FDA regulation of Laboratory-Developed Tests (LDTs)' (2019) *Congressional Research Service* 2; Allyse et al op cit note 5 at 118.

[58] Regulation 1 of the Medical Device Regulations.

[59] Merriam-Webster 'Monitor' available at *https://www.merriam-webster.com/dictionary/monitor*, accessed on 6 January 2021.

[60] Another purpose for which IVDs may be used is 'compatibility', as per regulation 1 of the Medical Device Regulations. Like 'monitoring', the Medical Devices Regulations do not contain a definition of 'compatibility'. However, 'compatibility' appears to relate to determining safety and congruency, especially in terms of a potential recipient (possibly for the purposes of donation or transplantation of biological material), thus making it inapplicable to direct-to-consumer genetic testing, which determines individual health status and not compatibility with others. 'Compatibility' could refer to those direct-to-consumer genetic tests that determine how an individual's body responds to, and metabolises, certain drugs and foods as the test results show what products and substances are 'compatible' with an individual based on their genes. However, this is peripheral to health-related direct-to-consumer genetic tests, and rather falls within the realm of nutrigenic or pharmacogenetic direct-to-consumer genetic tests. Australian Government Department of Health 'IVD medical devices: Definitions & links' 3 December 2010 available at *https://www.tga.gov.au/ivd-medical-devices-definitions-links*, accessed on 11 January 2021.

[61] Section 1 of the Medicines Act; regulation 1 of the Medical Device Regulations.

[62] 1941 (2) PH M41 (WLD).

[63] *Friedman* supra note 62 at 77.

the purposes for which IVDs provide information is not limited to 'diagnostic, monitoring or compatibility'[64] and may extend to other purposes, such as genetic propensity and disease risk.

To conclude, some direct-to-consumer genetic tests do not qualify as IVDs by virtue of the fact that they do not meet the definition of 'medical device' in the Medicines Act,[65] given that the testing company does not intend for these tests to be diagnostic or medical in nature. But direct-to-consumer genetic tests that are medical devices, I suggest, are also IVDs as they provide information, primarily for monitoring purposes, but also regarding disease risk, and involve the examination of samples in vitro.

(c) In vitro diagnostic device (IVD) medical device

Devices can either be medical devices (non-IVDs), IVDs, or IVD medical devices. While some direct-to-consumer genetic tests meet the definition of a 'medical device', they are also considered 'IVDs'. Thus, direct-to-consumer genetic tests that are medical devices, are generally IVD medical devices. This is also in line with SAHPRA's *Classification of Medical Devices and IVDs* (Classification Guidelines),[66] which distinguishes between medical devices (non-IVDs) and IVDs (IVD medical devices).

(d) Self-testing

Interestingly, the Medical Device Regulations contain a definition of 'self-testing' which is defined as 'testing performed by a lay person'.[67] But the definition does not specify the type of testing to which the definition is applicable. 'Self-testing' is only mentioned under labelling (for medical devices or IVDs) and instruction requirements (for IVDs) in the Medical Device Regulations.[68] The fact that self-testing devices are included under labelling and instruction requirements for medical devices or IVDs suggests that they are classified as such – but only where they have a medical or diagnostic intention.[69] Although prima facie direct-to-consumer

[64] Section 1 of the Medicines Act; regulation 1 of the Medical Device Regulations.
[65] Section 1 of the Medicines Act.
[66] South African Health Products Regulatory Authority (SAHPRA) *Classification of Medical Devices and IVDs* (2019).
[67] Regulation 1 of the Medical Device Regulations.
[68] Regulations 22(1)(*p*)(*vi*) and 24(1)(*d*) of the Medical Device Regulations. Regulation 22(1)(*p*) of the Medical Device Regulations explicitly refers to medical devices, although the title of the regulation mentions both medical devices and IVDs – indicating that such labelling for self-testing only applies to medical devices and not IVDs. Regulation 24 of the Medical Device Regulations specifically refers to IVDs, and regulation 24(1)(*d*) of the Medical Device Regulations mentions 'in vitro diagnostic use' showing that it is IVD specific. Therefore, there are differences in terms of the requirements that mention self-testing – labelling applies to self-testing medical devices, while instructions apply to self-testing IVDs.
[69] As per the definition of 'medical device' in section 1 of the Medicines Act.

genetic tests may be thought of as devices used for 'self-testing', I am of the view that this is not necessarily the case. The operative word is 'testing', and this is not something that the consumer does. Self-testing rather seems to refer to home pregnancy tests or blood sugar testing kits, where individuals both take the sample, administer the test, and receive the results themselves. Unlike pregnancy tests or blood sugar tests where the results are determinable (either one is pregnant or one is not, and one's blood sugar level either falls within normal parameters or it does not), direct-to-consumer genetic tests merely provide information regarding propensity to certain diseases and conditions without a definitive diagnosis, thus making 'self-testing' in this context seem futile.[70]

SAHPRA has published various guidelines relating to the classification, licensing, and essential principles of medical devices and IVDs, amongst others. While these documents are not binding, they serve to further enhance the Medicines Act and the Medical Device Regulations and may clarify the current legal status of direct-to-consumer genetic testing in South Africa. SAHPRA's Classification Guidelines define an 'IVD medical device for self-testing' as including –

> 'IVDs intended for use in the collection of a sample by a lay person and, if the sample is tested by another person (e.g. a laboratory) the results are returned directly to the person from whom the sample was taken without the direct supervision of a health professional who has formal training in a medical field or discipline to which the test relates'.[71]

This echoes the meaning of direct-to-consumer genetic testing – namely, genetic tests where saliva samples are collected by laypersons, where the testing process bypasses healthcare professionals, and where test results are sent directly to consumers. However, unlike the Medical Device Regulations, where 'self-testing' is seen to exclude situations wherein an individual does not perform the test themselves (as is the case with direct-to-consumer genetic testing), SAHPRA's Classification Guidelines specifically recognise certain IVD medical devices as being for self-testing even where the testing aspect is done elsewhere, such as in a laboratory – which is applicable to direct-to-consumer genetic testing. This creates confusion regarding the meaning of 'self-testing'. But South African legislation lacks clarity regarding the status of genetic testing in general, and direct-to-consumer genetic testing in particular. The inclusion of the definition of 'self-testing' in the Medical Device Regulations is a start; yet

[70] This is strengthened by the fact that healthcare professionals, such as doctors, often do not possess the necessary knowledge to interpret test results.
[71] SAHPRA op cit note 66 at 38.

greater certainty regarding its meaning and application throughout the Medical Device Regulations would be of assistance.

Given the above analysis, and to conclude this section, direct-to-consumer genetic tests can either be classified as medical devices or non-medical devices. Despite the provision of diagnostic information being questionable, since the various articles involved in the direct-to-consumer genetic testing process fall under the definition of 'medical device' in the Medicines Act,[72] and given that by being a medical device direct-to-consumer genetic tests may also be 'IVDs' and additionally meet its definition as DNA is examined in vitro for monitoring purposes as well as the provision of disease risk,[73] I suggest that direct-to-consumer genetic tests intended by the manufacturer to be medical devices additionally meet the definition of 'IVD medical device for self-testing', as per SAHPRA's Classification Guidelines.[74]

III CLASSIFYING DIRECT-TO-CONSUMER GENETIC TESTS

SAHPRA, established by the Medicines Act, aims to regulate (as well as monitor, evaluate, investigate, control, license, and register) health products, including medical devices and IVDs.[75] SAHPRA classifies medical devices and IVDs according to their quality, safety, and performance.[76] Medical devices and IVDs may fall into Class A (low risk), Class B (low-moderate risk), Class C (moderate-high risk), or Class D (high risk) depending on their risk to patients, users, or public health.[77] It is important to ascertain the classification of direct-to-consumer genetic tests as this impacts on inter alia registration, licensing, advertising, labelling, importing, and exporting. In what follows, I analyse the Medicines Act, the Medical Device Regulations, and SAHPRA's guidelines to determine which class direct-to-consumer genetic tests that are IVD medical devices should fall into.

[72] Section 1 of the Medicines Act.
[73] Regulation 1 of the Medical Device Regulations; section 1 of the Medicines Act.
[74] SAHPRA op cit note 66 at 38.
[75] Sections 2A & 2B(1)(*a*) of the Medicines Act; SAHPRA 'Medical devices' available at *https://www.sahpra.org.za/medical-devices/*, accessed on 5 May 2020; Julie Oppenheim 'Medicines and Related Substances Amendment Acts come into force' *Bowmans* 9 June 2017, available at *https://www.bowmanslaw.com/insights/pharmaceuticals-healthcare/medicines-related-substances-amendment-acts-come-force/*, accessed on 22 July 2020; SAHPRA 'About us' available at *https://www.sahpra.org.za/who-we-are/*, accessed on 5 May 2020.
[76] SAHPRA op cit note 66 at 1.
[77] Regulation 11(1) of the Medical Device Regulations.

(a) What is being classified?

What requires clarification is what aspect(s) of direct-to-consumer genetic testing is considered an IVD medical device, and how it should be classified.[78] Is it the saliva collection buccal? Is it the machines that extract, sequence, and analyse the DNA of consumers? Is it the software used to interpret and analyse the results? Or is it the entire service in combination? It may assist in distinguishing between the product, kit, or device used to measure a specific biomarker and the wider service in which tests are offered. While medical devices (the product) are legally governed through legislation, the service (the interpretation of test results) is subject to consumer protection and advertising laws.[79]

SAHPRA classifies various aspects, that are of relevance to the direct-to-consumer genetic testing process, differently. While IVD medical devices for self-testing are generally classified as Class C (subject to certain exceptions), an instrument for use in in vitro diagnostic procedures as well as specimen receptacles are Class A IVD medical devices.[80] However, specimen containers for use in self-testing and general laboratory tubes for containing and storing processed specimens are not specimen receptacles,[81] and are thus not IVD medical devices.[82]

---

[78] In 2017, the FDA approved 23andMe's PGS test as a Class II medical device for ten diseases and conditions (hereditary thrombophilia, alpha-1 antitrypsin deficiency, late onset Alzheimer's disease, Parkinson's disease, Gaucher disease, factor XI deficiency, celiac disease, Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency, early onset primary dystonia, and hereditary hemochromatosis), as well as specifying measures to lessen risks, such as erroneous test results, misunderstanding the test, and incorrect result interpretation. The FDA noted that the PGS test, rather than describing an individual's risk, shows that an individual has certain DNA variants linked to a condition. The PGS test has not been approved for: (1) prenatal testing; (2) cancer predisposition where test results may lead to screening, treatments, and procedures with morbidity or mortality (such as the BRCA1 and BRCA2 genes as individuals may undergo mastectomies or chemotherapy); (3) identifying genetic variants affecting metabolism, chance of negative effects, exposure response, or dosing for over-the-counter medications; and (4) determining the existence of deterministic autosomal dominant variants (for example, Huntington's disease is based on the inheritance of a single gene). Nicole Angelica 'FDA 23andMe Personal Genome Service (PGS) test – Evaluation of automatic class III designation – De novo request (memorandum)' *Duke SciPol* 24 October 2017, available at *https://scipol.duke.edu/track/fda-23andme-personal-genome-service-pgs-test-%E2%80%93-evaluation-automatic-class-iii-designation-%E2%80%93-de*, accessed on 27 November 2020.

[79] Caroline F Wright, Alison Hall & Ron L Zimmern 'Regulating direct-to-consumer genetic tests: What is all the fuss about?' (2011) 13(4) *Genetics in Medicine* 296.

[80] A 'specimen receptacle' is a device intended by the manufacturer 'for the primary containment and preservation of a specimen derived from the human body for the purpose of in vitro diagnostic examination'. SAHPRA considers a specimen receptacle to be an IVD medical device. SAHPRA op cit note 66 at 32, 33 & 41.

[81] Ibid at 41.

[82] Although prima facie the tube into which saliva is collected or the buccal swab is stored by consumers for the purposes of direct-to-consumer genetic testing does not appear to be a specimen receptacle and therefore is not an IVD medical device, some points can be argued. While a product used in general laboratory procedures is not an IVD medical device, this does not apply where the manufacturer intends the product, given its features, to be used for in vitro diagnostic examination. The intended use is gathered from: (a) instructions for use; (b) labelling; (c) advertising materials; and (d) technical documentation. The relevant product labelling related to the direct-to-consumer genetic test would appear on the box (or in the instructions) sent to the consumer containing the saliva collection tube and other objects (such as a swab) necessary for saliva collection. It can be implied that whatever

There are three distinct 'devices' used at various stages in the direct-to-consumer genetic testing process, which may be classified differently: (1) the tube into which saliva is collected, and which is used to transport the sample to the laboratory; (2) the machines that extract, sequence, and analyse DNA to produce genetic data and test results as well as the chemicals or reagents allowing it to perform;[83] and (3) the software that converts raw data into useable information. So, what happens when multiple IVD medical devices that form part of the same process are classified differently? And what happens when certain articles involved in the process are not IVD medical devices at all?

In terms of the classification of direct-to-consumer genetic tests, two situations may arise: (1) classifying direct-to-consumer genetic tests where the saliva sample is collected in South Africa, but sent to another jurisdiction for DNA extraction, sequencing, and analysis; and (2) classifying direct-to-consumer genetic tests where the saliva sample is collected and analysed in South Africa. In terms of (1), where saliva samples are extracted, sequenced, and analysed in laboratories in other jurisdictions, the saliva collection kit may be considered a low-risk medical device as it does not perform a diagnostic function and is only used to transport saliva samples.[84] However, in terms of (2), as saliva sample collection and analysis is conducted in South Africa, such direct-to-consumer genetic tests may be deemed to be moderate to high risk IVD medical devices.[85]

Some IVDs are used together with other IVDs, non-IVD medical devices, or accessories.[86] The rules for classification are independently applied to each device. Where several IVDs form part of a group, or where groups contain both IVDs and non-IVD medical devices, the highest class for any individual IVD or component determines the class of the group.[87] Adding to this, the Medical Device Regulations provide that where a medical device

labels appear on the packaging, instructions, or product apply to its contents. Thus, the labelling required by the Medical Device Regulations would make it apparent that the manufacturer intends the product to be used for in vitro diagnostic examination and thus, although for use in self-testing and general laboratory use, would qualify as an IVD medical device. Furthermore, although the tube is not directly used for in vitro diagnostic examination, it is necessary in order for the test to be performed; without the tube containing the saliva sample, the DNA cannot be extracted, sequenced, and analysed, and direct-to-consumer genetic test results cannot be ascertained. Therefore, I suggest that the tube into which saliva is collected or the buccal swab is stored for the purposes of a direct-to-consumer genetic test can be deemed to be an IVD medical device. Ibid at 5 & 41.

[83] Catherine M Sharkey 'Direct-to-consumer genetic testing: The FDA's dual role as safety and health information regulator' (2019) 68(2) *Depaul L Rev* 361.

[84] This was the position taken by Health Canada when 23andMe began marketing its tests in Canada. Joanne Kim 'Health technology update' (2017) 18 *CADTH* 11–2; Picard op cit note 1.

[85] See, Kim op cit note 84 at 11–2.

[86] An accessory is an item intended by the manufacturer to be used with an IVD, allowing the IVD to function as intended. Accessories are classified separately to the IVD. SAHPRA op cit note 66 at 30.

[87] Ibid at 30.

or IVD is classified into more than one class, it must be placed 'in the higher of the risk classes'.[88] Although there are certain indications that point towards different features of the direct-to-consumer genetic testing process and their classification, because direct-to-consumer genetic testing is provided as a group, the highest class applies. Even though the DNA extraction or sequencing machines on their own may be classified as a certain class of medical device, for example, they do not fall under self-testing as these machines (and their related software) are not used by consumers – it forms part of the testing process. Therefore, the most practical way in which to approach this is to view the IVD medical device as the entire direct-to-consumer genetic testing service as a whole, rather than examining each aspect individually – this is also in line with regulation 11(4) of the Medical Device Regulations.[89] Although the classification of direct-to-consumer genetic tests is discussed below, based on the above considerations, I preliminarily suggest that direct-to-consumer genetic tests are Class C IVD medical devices.

(b) What class do direct-to-consumer genetic tests fall into?

What makes the determination of a class challenging is that different jurisdictions may vary in their classification of medical devices.[90] In South Africa, SAHPRA classifies both medical devices and IVDs according to: (1) the device's intended use;[91] and (2) the level of risk to individuals and others (considering the possibility and severity of harm).[92] However, medical devices contain additional categories relevant to their classification, namely: (1) the extent of invasiveness; and (2) the period of use.[93] While the Medical Device Regulations provide that SAHPRA is responsible for establishing the classification of medical devices and IVDs,[94] SAHPRA's Classification Guidelines stipulate that manufacturers or distributors must

[88] Regulation 11(4) of the Medical Device Regulations. SAHPRA provides an example of a self-monitoring blood glucose system which clarifies this point. As each component of the system would be classified individually, the highest class in general must apply. A glucose metre, used for IVD procedures, is a Class A IVD; the glucose reagent test strips used in self-testing are Class C IVDs as incorrect results may be life-threatening; and the lancet for collecting blood samples are Class B medical devices. Thus, a self-testing blood glucose system would be classified as a Class C IVD. Ibid at 39.

[89] This is supported by Allyse et al, who state that from an examination of 23andMe's pre-market application approved by the FDA the whole direct-to-consumer genetic testing process – from the time of purchase to the receiving of test results – is one device. Allyse et al op cit note 5 at 119.

[90] While one country may view a direct-to-consumer genetic test as a medical device, another country may not. Brian Goemans & Robert McLaughlin *Medical Devices: An Innovation Guide from Lab to Commercialisation* (2018) 5.

[91] The intended use may be gathered from: (1) information accompanying the IVD, such as the instructions for use and labelling; (2) advertising information; and (3) if applicable, the design dossier. SAHPRA op cit note 66 at 29.

[92] Ibid at 5 & 29.

[93] Ibid at 5.

[94] Regulation 11(3) of the Medical Device Regulations.

ascertain a medical device or IVD's class based on the classification rules,[95] and SAHPRA only intervenes and determines the classification of a medical device or IVD where there is a dispute.[96] Greater harmony between the Medical Device Regulations and SAHPRA's Classification Guidelines would be of assistance to amend this disparity.

SAHPRA must 'consider the classification of a medical device or IVD individually, taking into account its design and intended use'.[97] While the intended use of direct-to-consumer genetic tests, from the perspective of testing companies, is generally to be informational or educational rather than medical or diagnostic, this may not necessarily be the intended use for consumers. This is also because the information regarding the intended use of the medical device is not always visible. In terms of SAHPRA's Classification Guidelines, which are based on the intention of the manufacturer, most direct-to-consumer genetic tests would not be intended to be used for diagnosis or health purposes and would rather provide information regarding *risk* of the development of certain diseases or conditions.

According to rule three in SAHPRA's Classification Guidelines,[98] IVDs intended for inter alia 'human genetic testing' are deemed to be Class C IVD medical devices.[99] This is because such IVDs pose a fair risk to public health or a great individual risk as incorrect results may cause individuals to make significant decisions regarding their health.[100] However, based on the other categories under this rule, it appears that the genetic testing referred to is that conducted in the clinical setting, and not that which is undertaken by consumers.[101] I suggest that this is also the case as SAHPRA distinguishes between IVD medical devices for genetic testing and those for self-testing (although not confined to genetic testing), this shows that there are two distinct categories. These IVDs also typically offer the crucial or only grounds for correct diagnosis.[102] This is not so with direct-to-consumer genetic testing in that, although

[95] SAHPRA op cit note 66 at 5 & 29.
[96] Ibid at 30.
[97] Regulation 11(5) of the Medical Device Regulations.
[98] Based on these Classification Guidelines, direct-to-consumer genetic tests do not appear to fall into the rules for the classification of medical devices (non-IVDs) and are therefore guided by the classification rules for IVD medical devices. Classification rule three deals with the 'detection of transmissible agents or biological characteristics posing a moderate public health risk or a high personal risk'. SAHPRA op cit note 66 at 31.
[99] SAHPRA provides examples of tests that detect Philadelphia chromosome, Huntington's disease, or cystic fibrosis. Ibid at 31–2 & 36.
[100] Ibid at 36.
[101] This is because no reference is made to self-testing. Other IVDs falling under this classification rule refer to 'patients' – a term used in the clinical setting. Furthermore, reference is made to inter alia 'screening for congenital disorders in the foetus' and 'to monitor levels of medicines, substances or biological components, when there is a risk that an erroneous result will lead to a patient management decision resulting in an immediate life-threatening situation for the patient' – things that would not be done by individuals themselves and would require the involvement of healthcare professionals. Ibid at 31–2.
[102] Ibid at 36.

erroneous test results may have a negative effect on consumers, such tests are not used in isolation and cannot be seen to provide an accurate diagnosis without further testing in the clinical setting, which involves healthcare professionals.

As discussed above, classification rule four in SAHPRA's Classification Guidelines specifically mentions 'IVD medical devices for self-testing', classifying them as Class C if the condition being tested for: (1) generally requires healthcare professionals to be involved in diagnosis or treatment; or (2) cannot be exactly understood by ordinary individuals, or needs supervision for safe treatment.[103] This is because such tests are used by laypersons, who usually lack medical training, with results bypassing healthcare professionals.[104] However, where a self-test does not detect a 'serious condition, ailment, or defect',[105] or the results are prefatory and require additional testing, SAHPRA provides alternative rules for classification – generally placing such tests into Class B.[106] Various scenarios may arise here. Certain direct-to-consumer genetic tests are health-related, while others are not; some consumers may approach healthcare professionals with their test results and request further testing (or healthcare professionals may advise this), and others may take the test results at face value and either use them to improve their health or continue as they had been.

Firstly, in terms of (1) and (2) above,[107] certain diseases and conditions that direct-to-consumer genetic tests identify typically necessitate oversight from healthcare professionals if undertaken in the clinical setting, and therefore such tests require professional assistance to interpret and understand the implications of the results. This is because of the complexity of the human genome, the lack of understanding on the part of individuals regarding their test results, the potential repercussions that may arise if a disease or condition manifests itself, and the current lack of treatments or cures for some diseases. These are health-related tests for conditions such as Parkinson's disease, Alzheimer's disease, Huntington's disease, and cardiovascular disease, and such tests are therefore classified as Class C IVD medical devices. Something to consider is: Should all health-related direct-to-consumer genetic tests not require the involvement of a healthcare professional? While consumers may think that they understand their test results, healthcare professionals such as genetic counsellors have received training specific to the field. This issue has been extensively debated in the literature, and tends to lean

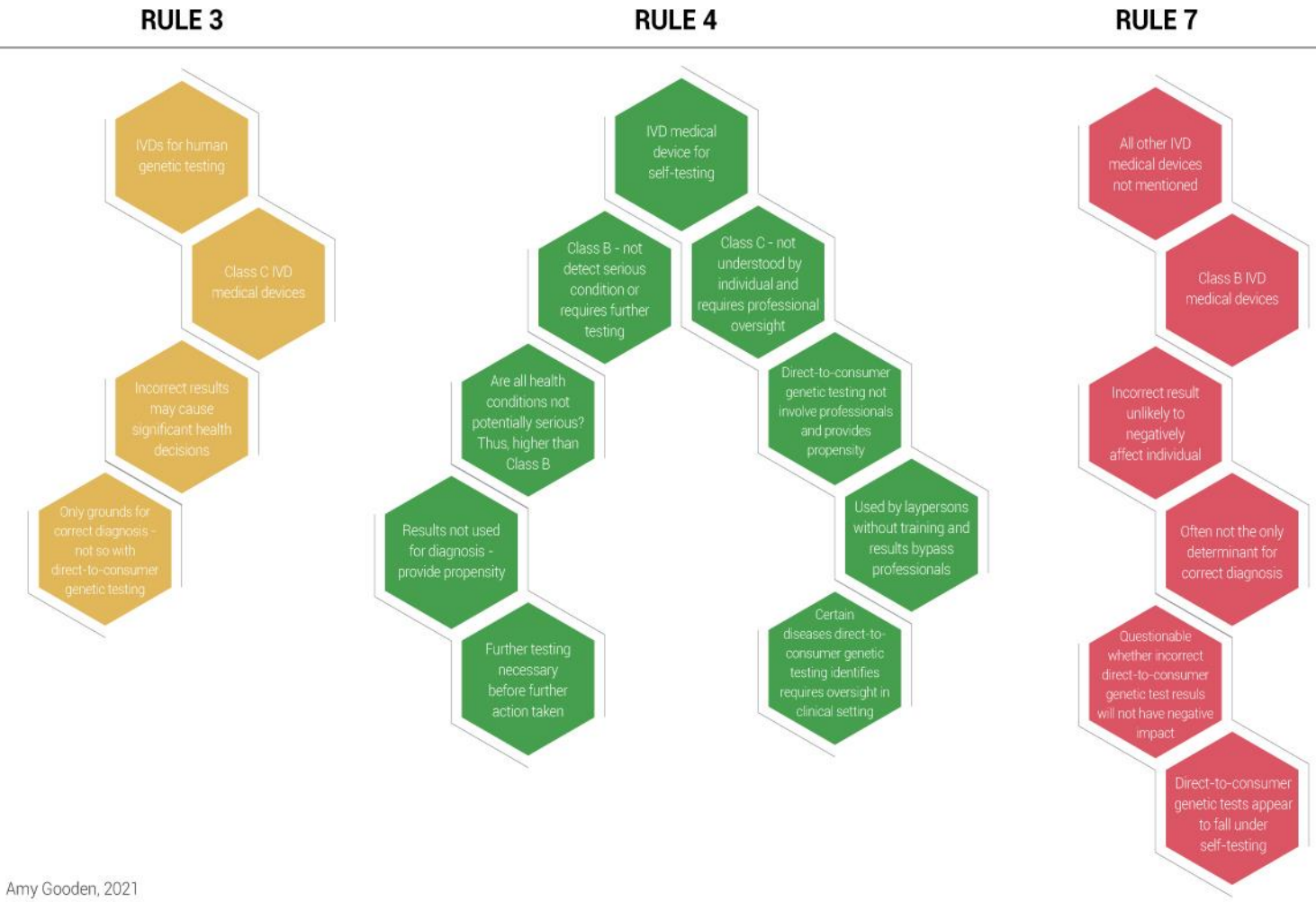
[103] Ibid at 38.
[104] Ibid at 38.
[105] Ibid at 32.
[106] For example, a positive pregnancy self-test will usually involve a follow-up medical consultation, causing them to be classified as Class B IVDs. Ibid at 32 & 39.
[107] Ibid at 38.

towards healthcare professionals, particularly genetic counsellors, being mandatory in the direct-to-consumer genetic testing process.[108]

Secondly, what qualifies as a 'serious condition, ailment, or defect'?[109] Are all health-related conditions not potentially serious? SAHPRA provides examples of pregnancy and fertility self-testing kits, self-monitoring tests for blood glucose, and urine self-test strips, but these relate to the preliminary nature of results – not to the determination of a serious condition. I suggest that greater clarification regarding what qualifies as a 'serious condition, ailment, or defect'[110] will assist in determining whether 'IVD medical devices for self-testing', including direct-to-consumer genetic tests, fall into Class B or Class C.[111]

In terms of classification rule seven in SAHPRA's Classification Guidelines,[112] all other IVD medical devices omitted in the other rules, are Class B IVD medical devices.[113] These devices pose a moderate risk to individuals. It is improbable for incorrect results to negatively affect individuals. These devices are often not the only source used for accurate diagnosis.[114] But it is questionable whether incorrect direct-to-consumer genetic test results will not have an adverse impact on consumers. As direct-to-consumer genetic testing already appears to fall under rule four of SAHPRA's Classification Guidelines, it is this rule that applies. The diagram below serves to illustrate these rules and the processes (Diagram 2).

[108] See, Cheryl Berg & Kelly Fryer-Edwards 'The ethical challenges of direct-to-consumer genetic testing' (2008) 77(1) *J Bus Ethics* 28–9; Ruslan Dorfman, Rabia Khan & Gouri Mukerjee 'Proposed regulatory framework for direct-to-consumer genetic testing: Diagnostics vs genetic screening' (2014) 60(11) *Clin Chem* 1456; Heidi Carmen Howard & Pascal Borry 'Survey of European clinical geneticists on awareness, experiences and attitudes towards direct-to-consumer genetic testing' (2013) 5(45) *Genome Med* 6; Anna Middleton, Álvaro Mendes, Caroline M Benjamin et al 'Direct-to-consumer genetic testing: Where and how does genetic counseling fit?' (2017) 14(3) *Per Med* 253; SK Delaney & MF Christman 'Direct-to-consumer genetic testing: Perspectives on its value in healthcare' (2016) 99(2) *Clin Pharmacol Ther* 148; Stuart Hogarth & Paula Saukko 'A market in the making: The past, present and future of direct-to-consumer genomics' (2017) 36(3) *New Genet Soc* 200.
[109] SAHPRA op cit note 66 at 32.
[110] Ibid at 32.
[111] This classification should also depend on what is to be tested for. The more serious the disease or condition, the higher the classification must be. Thus, the greater the uncertainty or chances for misinterpretation with serious consequences, the higher the classification of IVD medical device.
[112] SAHPRA op cit note 66 at 33.
[113] Ibid at 33.
[114] Ibid at 42.



Because direct-to-consumer genetic tests are not definitively diagnostic and because most testing companies recommend that consumers visit a healthcare professional with their results before making medical decisions (which may lead to additional testing), it must be questioned whether these less 'serious' tests are classified as Class B IVD medical devices. Given that evidence regarding the analytical validity, clinical validity, and clinical utility of many direct-to-consumer genetic tests is lacking, although additional testing may be required to confirm or rule out a condition, it is unlikely that healthcare professionals would immediately jump to this conclusion should a consumer approach them with their test results, preferring to rather conduct other preliminary examinations first. Therefore, I suggest that despite the above, health-related direct-to-consumer genetic tests should generally be classified as Class C IVD medical devices.

The classification rules as they relate to direct-to-consumer genetic tests are ambiguous. Despite the classification rules and the potential for direct-to-consumer genetic tests to be classified as Class B IVD medical devices, I suggest that direct-to-consumer genetic tests pose

a moderate to high risk to individuals given the psychological effects, the use of test results to make medical decisions, the lack of involvement of healthcare professionals, and the determination of risk for certain serious diseases or conditions – and classifying such tests as Class C IVD medical devices in most instances therefore seems appropriate. Based on international standards (such as the approach followed by the FDA in the US) and given the recognition of consumer protection in South Africa (in terms of section 3 of the CPA), I suggest that SAHPRA issue a directive clarifying this position.

IV CONCLUSION

This Chapter examined the legal status of medical devices and IVDs in South Africa and, through the Medicines Act and the Medical Device Regulations, determined that while many direct-to-consumer genetic tests are not considered IVD medical devices due to the manufacturer's intention,[115] there are tests that are indeed IVD medical devices and additionally meet SAHPRA's definition of an 'IVD medical device for self-testing'.[116] Furthermore, in this Chapter I assessed what is being classified and which class of IVD medical device direct-to-consumer genetic tests should fall into, ultimately concluding that although the stages of the direct-to-consumer genetic testing process involve various articles that may be classified differently, in line with the Medical Device Regulations,[117] the highest class (being Class C) applies.

While SAHPRA's Classification Guidelines place IVD medical devices for human genetic testing into Class C and all other IVD medical devices not covered by the Classification Guidelines into Class B, I suggest that for various reasons mentioned above, both of these categories are unsuited to direct-to-consumer genetic testing. Direct-to-consumer genetic tests rather fall into the classification rule for 'IVD medical devices for self-testing', which can either be Class B or Class C depending on the involvement of healthcare professionals, individual understanding, the severity of the condition, and the finality of the results.

While the intended use of a direct-to-consumer genetic test may be as an informational tool that shows genetic propensity to various diseases and conditions, the level of risk to individuals is paramount in this case. This is because the psychological harm that such tests may have on consumers is largely undetermined and it is uncertain how consumers will

[115] As per the definition of 'medical device' in section 1 of the Medicines Act.
[116] SAHPRA op cit note 66 at 38.
[117] Regulation 11(4) of the Medical Device Regulations.

interpret their test results.[118] Without the involvement of healthcare professionals or genetic counsellors in the direct-to-consumer genetic testing process, the *possibility* and *severity* of harm is high.[119] This is due to potential result misinterpretation and a lack of understanding of the testing process and what the results may mean.

Despite SAHPRA's Classification Guidelines pointing towards direct-to-consumer genetic tests being classified as Class B IVD medical devices, I suggest that given the psychological impact, the promotion of genetic determinism, utilising test results in making medical decisions,[120] the absence of healthcare professionals, and risk determination for particular serious diseases or conditions.[121] direct-to-consumer genetic tests that are IVD medical devices generally be classified as Class C.

Overall, there are inconsistences and uncertainties between the various laws, regulations, and guidelines regarding medical devices and IVDs. To amend this, I suggest that SAHPRA issue a directive clarifying these discrepancies, as well as elucidating the position and classification of direct-to-consumer genetic tests that qualify as IVD medical devices.

[118] While some consumers may not take their test results seriously, others may use them to make medical decisions which could have potentially adverse consequences.
[119] See, SAHPRA op cit note 66 at 29.
[120] Although it can be said that the more serious steps that consumers may take (such as surgery) will inevitably involve healthcare professionals, there are measures that consumers could adopt without healthcare professionals, and which could negatively impact their health, such as changing medication dosages on their own due to the results of a direct-to-consumer genetic test. It may cause consumers to stop taking important medication due to test results showing a decreased risk of a certain disease or condition which they have been diagnosed with by a healthcare professional. Thus, genetic counselling or healthcare professional involvement may ensure that consumers do not cease prescribed treatment or medication, or pursue needless treatment founded on their own result interpretation. See, Jane Kaye 'The regulation of direct-to-consumer genetic tests' (2008) 17 *Hum Mol Genet* 182.
[121] See, SAHPRA op cit note 66 at 29.

CHAPTER 5

OFFERING DIRECT-TO-CONSUMER GENETIC TESTS TO THE PUBLIC IN SOUTH AFRICA

I INTRODUCTION AND CHAPTER OVERVIEW

A unique feature of the direct-to-consumer genetic testing model is the fact that genetic tests, with potential health-related implications, are offered directly to the public often without professional oversight. Clearly, this can have far-reaching negative consequences. While the offering of direct-to-consumer genetic tests to the public may seem simple, South Africa's extant laws contain various requirements that must be adhered to prior to being able to sell products to the public. Furthermore, once products are available on the market, South African legislation regulates their advertising. Both aspects may hold implications for testing companies offering their products and services to the public in South Africa and raise questions regarding whether these protections are adequate to suit the (sometimes) international nature of the contractual relations between testing companies and consumers.

In this Chapter, I explore the various issues and legislative provisions linked to the offering of direct-to-consumer genetic tests to the South African public – and its implications for testing companies operating both in South Africa and abroad. This includes the licensing, registering, and selling of direct-to-consumer genetic tests; as well as their advertising, marketing, disclosure, and labelling. This Chapter examines the legal processes and requirements involved in offering direct-to-consumer genetic tests in South Africa through analysing inter alia the Medicines Act, the CPA, ECTA, POPIA, and the Medical Device Regulations.

The issues associated with offering direct-to-consumer genetic tests to the public in South Africa is broad. As such, this Chapter does not deal with warranties, defects, faults, the return of goods, potential harm and the liability of suppliers, or terms and conditions and privacy policies and whether they are fair, just, and reasonable and comply with the CPA and ECTA. This Chapter does not consider electronic contracts and their formation, requirements, and existence in detail – ECTA is only examined insofar as it relates to advertising, marketing, and disclosure.

II WHICH STATUTES APPLY TO DIRECT-TO-CONSUMER GENETIC TESTING?

Before proceeding with an analysis of the relevant statutes and regulations that relate to the offering of direct-to-consumer genetic tests in South Africa, it is prudent to first examine whether any of the identified statutes – namely the CPA, ECTA, and POPIA – apply to testing companies – particularly those based in other jurisdictions that offer their tests to consumers in South Africa.[1]

(a) The Consumer Protection Act 68 of 2008 (CPA)

The CPA, as South Africa's seminal consumer protection legislation, holds implications for testing companies based within and outside the country – should it apply. Direct-to-consumer genetic testing is a complex industry that falls at the intersection of health products and consumer goods – to which the CPA pertains. The CPA applies to all transactions that occur *within* South Africa, unless exempted by section 5(2),[2] or in terms of subsections 5(3) and (4) of the CPA.[3] The CPA also governs the promotion of goods or services *within* South Africa, unless exempted.[4] The following questions are then important to consider: (1) what is the meaning of 'within'; (2) what is the meaning of 'transaction'; and (3) what is the meaning of 'promotion'? Once these questions are answered, it will be possible to determine whether the CPA (or which provisions thereof) apply to various testing companies, both local and foreign.

---

[1] While 23andMe and AncestryDNA are two of the biggest testing companies globally, they do not offer their services or ship their products to South Africa. Other such companies that do not offer their products and services in South Africa include Color Genomics, Counsyl, GeneByGene, Helix, Veritas Genetics, Kailos, and FamilyTreeDNA. This is largely due to cost and logistical issues as well as various legal requirements that create impediments. However, MyHeritage as well as MapMyGenome are two testing companies based abroad that do ship their products to South Africa. 23andMe 'What countries do you ship to?' available at *https://int.customercare.23andme.com/hc/en-us/articles/214806628-What-countries-do-you-ship-to-*, accessed on 2 January 2020; AncestryDNA 'Need to ship to another country?' available at *https://www.ancestry.com/checkout/MLI?rtype=85&flow=3*, accessed on 2 January 2020.

[2] Section 5(2) of the CPA states that the CPA 'does not apply to any transaction –

(a) in terms of which goods or services are promoted or supplied to the State;
(b) in terms of which the consumer is a juristic person whose asset value or annual turnover, at the time of the transaction, equals or exceeds the threshold value determined by the Minister in terms of section 6;
(c) if the transaction falls within an exemption granted by the Minister in terms of subsections (3) and (4);
(d) that constitutes a credit agreement under the National Credit Act, but the goods or services that are the subject of the credit agreement are not excluded from the ambit of this Act;
(e) pertaining to services to be supplied under an employment contract;
(f) giving effect to a collective bargaining agreement within the meaning of section 23 of the Constitution and the Labour Relations Act, 1995 (Act No. 66 of 1995); or
(g) giving effect to a collective agreement as defined in section 213 of the Labour Relations Act, 1995 (Act No. 66 of 1995)'.

[3] In terms of subsections 5(3) and (4) of the CPA, upon application the Minister may grant an industry-wide exemption from certain provisions of the CPA based on the fact that they overlap: (1) other national legislation; or (2) a treaty, international law, convention, or protocol.

[4] Section 5(1)(*b*) of the CPA.

(i) What is the meaning of 'within'?

As mentioned above, the applicability of the CPA depends on whether the transaction occurs 'within in the Republic'.[5] As per section 5(1)(*a*) and (*b*) of the CPA, any transactions or promotions of goods and services within South Africa fall under the CPA. On a literal interpretation, the CPA does not apply to transactions occurring outside South Africa – this is one way for foreign testing companies to avoid being bound by the CPA.[6] But the meaning of the phrase 'within in the Republic'[7] is unclear, specifically where transactions take place online and only delivery of goods occurs within South Africa. As neither the courts nor the National Consumer Tribunal (NCT) have provided a position on this matter, it seems that determining whether the CPA applies in its entirety will be done on an ad hoc basis.[8]

There are two situations to consider in this regard: (1) local testing companies, namely those based in South Africa; and (2) foreign testing companies based in another jurisdiction. In terms of (1), as South African testing companies supply their products and offer their services to consumers within the country, the CPA applies to them.[9] In terms of (2), the situation is more complex. Section 5(8)(*a*) of the CPA notes that subsections 5(1) to 5(7) apply to matters regardless of whether the supplier[10] is located, or has its main office, within or outside South Africa[11] – thus prima facie appearing to bring foreign testing companies within the ambit of the CPA. For example, it is possible for a testing company based in another jurisdiction to have

[5] As per section 5(1)(*a*) of the CPA. The CPA's reference to 'every transaction occurring within the Republic' brings these types of transactions within its scope. Although contracts often have a governing law clause, specifying that the transaction is subject to the law of a certain country, parts of the transaction may nevertheless by subject to the CPA. Ismail Laher 'The Consumer Protection Act and cross-border transactions' *Polity* 9 November 2010, available at *https://www.polity.org.za/article/the-consumer-protection-act-and-cross-border-transactions-2010-11-09*, accessed on 8 March 2021.

[6] There are however certain provisions in the CPA which apply regardless of which legal system is used. Foreign companies that sell goods and services to South Africa must comply with the CPA and, if they contravene its provisions relating to product liability or labelling, may be sued for damages. If a consumer in South Africa suffers injury or loss due to the incorrect or insufficient labelling of a foreign product, liability may fall onto both the local retailer and the foreign supplier. Products containing risks which 'an ordinarily alert consumer' could not be expected to be aware of must be brought to their attention by the supplier in plain language. Laher op cit note 5; Werksmans Attorneys 'Local consumer law has international ramifications' 7 July 2010 available at *https://www.werksmans.com/legal-updates-and-opinions/local-consumer-law-has-international-ramifications/*, accessed on 8 March 2021; SAPA 'Consumer Protection Act has international implications' *Sunday Times* 26 July 2010, available at *https://www.timeslive.co.za/sunday-times/business/2010-07-26-consumer-protection-act-has-international-implications/*, accessed on 8 March 2021.

[7] As per section 5(1)(*a*) of the CPA.

[8] Thomas Reisenberger 'Does the Consumer Protection Act apply to foreign retailers selling goods online to South Africa?' *Legalese* 20 November 2018, available at *https://legalese.co.za/does-the-consumer-protection-act-apply-to-foreign-retailers-selling-goods-online-to-south-africa/*, accessed on 8 March 2021.

[9] This also applies where South African testing companies offer their services to consumers abroad.

[10] 'Supplier' is defined in section 1 of the CPA as 'a person who markets any goods or services'. As 'person' is inclusive of juristic persons (companies), a testing company that markets its products is considered a supplier under the CPA.

[11] Section 5(8)(*a*) of the CPA.

a corresponding South African domain.[12] In such a case, the provisions of the CPA apply in totality because of the South African domain – and thus provisions, such as those relating to advertising, would impose obligations as to what is required on testing company websites. I suggest that this would also be the case for foreign testing companies who advertise (through agreement with the host) on a South African domain owned by another party (such as pop-up advertisements). But what about foreign testing companies that do not use a South African domain? I answer this below.

(ii) What is the meaning of 'transaction'?

In addition to determining whether transactions occur *within* South Africa, thus bringing them within the CPA's ambit, another consideration is the meaning of 'transaction' mentioned in section 5(1)(*a*) of the CPA. 'Transaction' is a technical term defined as including not only the agreement between parties for the supply of goods or services, but also performance.[13] There is uncertainty regarding whether the CPA is applicable to foreign testing companies that supply their goods and services to South African consumers, and where transactions occur online.[14] Although in the case of foreign testing companies, the service, namely the extraction, sequencing, and analysis of saliva samples occurs overseas and is not subject to the CPA (given that the transaction occurs in a foreign jurisdiction), delivery of the testing kits and test results (the goods) – as part of the contract with foreign testing companies – occurs in South Africa. This constitutes performance under the definition of 'transaction',[15] and as per section 5(1)(*c*) of the CPA which stipulates that the CPA applies (in its entirety) to any supply and performance in terms of the transaction as per section 5(1). The consequence of this is that the CPA applies to testing companies which tender performance in South Africa. Thus, where testing companies, wherever they may be based, offer their services to consumers in South Africa and tender performance themselves, the CPA applies.

[12] For example, a '.co.za' domain.
[13] Section 1 of the CPA.
[14] Reisenberger op cit note 8.
[15] Section 1 of the CPA.

(iii) What is the meaning of 'promotion'?

'Promotion' is defined in section 1 of the CPA as inter alia advertising,[16] displaying, or offering to supply goods or services to the public.[17] Given this, any testing company that wishes to advertise its products and services in South Africa needs to comply with the CPA provisions relating to advertising and marketing – including those of disclosure.

South African testing companies are currently not prominent advertisers. They may advertise on Facebook and other social media platforms, or on their own websites – which then spreads by referral or word of mouth – but there is no direct advertising of direct-to-consumer genetic tests to the public in South Africa – in the form of billboard, flyers, and magazine or television advertisements. The most likely way for consumers to find out about direct-to-consumer genetic testing themselves is through testing company websites. These websites often contain what would be classified as 'advertisements',[18] aimed at attracting consumers to their tests. The CPA defines the term 'advertisement' to include both direct and indirect forms of 'visual or oral communication transmitted by any medium',[19] with the aim of alerting the public to the existence of a supplier or goods and services for the purposes of promoting the supply of such goods and services.[20] For example, any testing company's website, including a South African one, must comply with the CPA's relevant provisions relating to advertisements.[21]

---

[16] Section 1 of the CPA defines 'advertisement' as –
'any direct or indirect visual or oral communication transmitted by any medium, or any representation or reference written, inscribed, recorded, encoded upon or embedded within any medium, by means of which a person seeks to –
(a) bring to the attention of all or part of the public –
(i) the existence or identity of a supplier; or
(ii) the existence, nature, availability, properties, advantages or uses of any goods or services that are available for supply, or the conditions on, or prices at, which any goods or services are available for supply;
(b) promote the supply of any goods or services; or
(c) promote any cause'.

[17] Section 1 of the CPA.

[18] As per the definition of 'advertisement' in section 1 of the CPA. However, such advertisements would be more indirect, rather than direct.

[19] Section 1 of the CPA.

[20] Ibid.

[21] There are no laws that relate specifically to online advertising. The Advertising Regulatory Board's (ARB) *Code of Advertising Practice* (ARB Code) mentions social media marketing. But the ARB Code is only binding on members of the ARB, making the ARB Code voluntary. Advertising Regulatory Board *Code of Advertising Practice, Appendix K* (2021); Livia Dyer & Craig Kennedy 'Digital business in South Africa: Overview' *Thomson Reuters* 1 March 2021, available at *https://uk.practicallaw.thomsonreuters.com/w-007-8319?transitionType=Default&contextData=(sc.Default)&firstPage=true*, accessed on 5 April 2021.

Testing companies based in foreign jurisdictions do not advertise *within* South Africa for the purposes of this provision, unless they have a South African domain or otherwise, as described above. However, as already mentioned, the CPA applies in its entirety when performance takes place in South Africa. Whilst it may seem untenable to stipulate that the provisions relating to advertising apply to foreign testing companies and their websites for example, it is important to recognise that other South African statutes, such as ECTA (discussed below), also have requirements relating to disclosure,[22] which apply as long as there is a South African consumer involved.

I suggest that from this reading, the CPA in its entirety applies to testing companies where a transaction involves a South African consumer – even those that do not have offices in South Africa and operate wholly outside of the country. It seems that the legislature intended, by the inclusion of this catch-all clause,[23] to protect consumers from risks posed by foreign companies.

(b) The Electronic Communications and Transactions Act 25 of 2002 (ECTA)

Testing companies, due to the nature of their business model, tend to function largely online. Information is sent to consumers electronically, terms and conditions are accepted online, and contracts are signed digitally – these electronic transactions bring about the provisions of ECTA. ECTA was enacted to both facilitate and regulate electronic transactions in the age of internet communication technologies. All electronic contracts are facilitated via 'data messages', which are defined in section 1 of ECTA as 'data generated, sent, received or stored by electronic means'.[24]

As testing companies based in foreign jurisdictions tend to transact with consumers online, ECTA regulates the conclusion of the contract through section 22(2), which provides that an agreement 'concluded between parties by means of data messages is concluded at the time when and place where the acceptance of the offer was received by the offeror'.[25] This means that consensus is reached, and thus the contract is formed, at the time and place where the consumer's acceptance is received by the testing company. Typically, this is where the testing company's main offices are located. Therefore, where the testing company is based

[22] Section 43 of ECTA.
[23] Section 5(8)(*a*) of the CPA.
[24] Section 1 of ECTA.
[25] Section 22(2) of ECTA. While some testing companies rely on the postal service, others (while utilising the post to send and receive testing kits) require consumers to complete their consent form (and agreement to terms and conditions) online.

abroad, the laws of the country where the testing company is based apply (and this is also where the contract is concluded, as per section 22(2) of ECTA) – rather than the South African legal system.

There are however certain provisions of ECTA that always apply when South African consumers are involved. Section 47 of ECTA states that the entirety of Chapter VII (sections 42 to 49) and the protections that it affords consumers apply to all electronic contracts 'irrespective of the legal system applicable to the agreement in question'.[26] Importantly, this means that all testing companies that supply their products and services to consumers in South Africa are bound by requirements such as information disclosure on their websites in section 43 of ECTA and performance obligations in section 46 of ECTA.[27]

(c) The Protection of Personal Information Act 4 of 2013 (POPIA)

As South Africa's central data protection legislation, if POPIA does not apply to foreign testing companies, this could have serious consequences for consumers and the privacy of their personal information. If POPIA does apply, this imposes obligations on testing companies looking to expand their consumer market to South Africa. Although POPIA is discussed in subsequent chapters, it is also relevant to the broader consumer protection framework.

In the consumer setting, personal information is commonly processed for the purpose of selling products or services, and for direct marketing.[28] POPIA relates to 'the processing of personal information entered in a record by or for a responsible party'.[29] Firstly, Chapter 2 established that the definition of 'personal information' under POPIA[30] encompasses genetic data. But in the present consumer protection context, the personal information to which POPIA relates is the names, addresses, and contact details of consumers that are collected when they

[26] Section 47 of ECTA. This means that even for transactions or agreements that occur with testing companies overseas, the provisions of ECTA that relate to consumer protection apply regardless.

[27] Section 46 of ECTA states that –

'(1) The supplier must execute the order within 30 days after the day on which the supplier received the order, unless the parties have agreed otherwise.

(2) Where a supplier has failed to execute the order within 30 days or within the agreed period, the consumer may cancel the agreement with seven days' written notice.

(3) If a supplier is unable to perform in terms of the agreement on the grounds that the goods or services ordered are unavailable, the supplier must immediately notify the consumer of this fact and refund any payments within 30 days after the date of such notification'.

[28] Adéle da Veiga, Ruthea Vorster, Colin Pilkington et al 'Compliance with the Protection of Personal Information Act and consumer privacy expectations: A comparison between the retail and medical aid industry' *16th International Information Security South Africa Conference* (August 2017 Johannesburg, South Africa) 17.

[29] Section 3(1)(*a*) of POPIA. The use of the word 'and' in section 3(1) of POPIA means that both conditions (a) and (b) must be complied with in order for POPIA to be applicable.

[30] Section 1 of POPIA.

sign up for a direct-to-consumer genetic test. POPIA only applies to genetic data once it has been sequenced and 'entered into a record'.[31] This is because 'personal information' in POPIA only includes genetic data, and not the containers of such, like saliva and DNA (discussed in Chapter 2). Genetic data in relation to research and POPIA is dealt with in Chapter 7. Secondly, 'record', which is broadly defined in POPIA,[32] refers to any recorded information (regardless of form or medium) including written, recorded, digital, or drawn[33] – and despite who possesses, controls, or creates it, or when it came into existence.[34]

According to section 3(1)(*b*), POPIA only applies where the responsible party is: (1) domiciled in South Africa; or (2) not domiciled in South Africa, but uses automated or non-automated processing means in the country, unless such means only forward personal information through South Africa.[35] Unlike local testing companies, many international testing companies are neither domiciled in South Africa, nor do they offer their products and services in the country – causing POPIA to be inapplicable to them. But do those testing companies not domiciled in South Africa make use of 'automated or non-automated means' in the country?[36] These means are used to process personal information, and as processing activities, such as receipt, storage, use, and destruction,[37] occur in the jurisdiction where the testing company is located, the laws of that country apply. However, the definition of 'processing' in POPIA[38]

---

[31] Section 3(1)(*a*) of POPIA. See also, Donrich Thaldar 'Why POPIA does not apply to DNA' (2021) 117(7/8) *S Afr J Sci* 2.

[32] Section 1 of POPIA.

[33] Section 1 of POPIA states that –

'"record" means any recorded information –

(a) regardless of form or medium, including any of the following:
  (i) Writing on any material;
  (ii) Information produced, recorded or stored by means of any tape-recorder, computer equipment, whether hardware or software or both, or other device, and any material subsequently derived from information so produced, recorded or stored;
  (iii) label, marking or other writing that identifies or describes any thing of which it forms part, or to which it is attached by any means;
  (iv) book, map, plan, graph or drawing;
  (v) photograph, film, negative, tape or other device in which one or more visual images are embodied so as to be capable, with or without the aid of some other equipment, of being reproduced'.

[34] Section 1 of POPIA.

[35] Section 3(1)(*b*) of POPIA. Thus, POPIA's scope is more limited than that of the EU's GDPR which applies to anyone processing personal data from the EU, regardless of their location. Hunton Andrews Kurth 'South Africa's Protection of Personal Information Act, 2013, goes into effect July 1' *The National Law Review* 29 June 2020, available at *https://www.natlawreview.com/article/south-africa-s-protection-personal-information-act-2013-goes-effect-july-1*, accessed on 9 October 2020; Alt Advisory 'POPIA compliance' available at *https://altadvisory.africa/popia/#applicationofpopia*, accessed on 9 October 2020; Preeta Bhagattjee 'South Africa – Data protection overview' *Data Guidance* July 2020, available at *https://www.dataguidance.com/notes/south-africa-data-protection-overview*, accessed on 9 October 2020.

[36] Section 3(1)(*b*)(*ii*) of POPIA. According to section 3(4) of POPIA, 'automated means' refers to 'any equipment capable of operating automatically in response to instructions given for the purpose of processing information'.

[37] As per section 1 of POPIA.

[38] Ibid.

includes the 'collection' of personal information which, in this context, entails obtaining names, addresses, and contact details. This information is provided by consumers in South Africa via the testing company's website (automated means). Accordingly, foreign testing companies not domiciled in South Africa do make use of automated means in the country, hence such testing companies will always be subject to POPIA in this context.[39]

Although direct-to-consumer genetic testing dominates the US and European markets, it has not yet gained such traction in South Africa. But as awareness and interest increase, it is possible that testing companies from foreign jurisdictions may begin to offer their products and services in South Africa – making the potential harms to the privacy of personal information a primary concern. As POPIA will always apply to testing companies with regards to the names, addresses, contact details, and other personal information of consumers, the implications of this on consumers must briefly be assessed. As POPIA aims to safeguard the privacy of consumers' personal information, testing companies may only lawfully process personal information in line with the eight conditions in POPIA.[40] These are: (1) accountability;[41] (2) processing limitation;[42] (3) purpose specification;[43] (4) further processing limitation;[44] (5) information quality;[45] (6) openness;[46] (7) security safeguards;[47] and (8) data subject participation.[48] Testing companies must ensure that personal information is complete, precise, and updated where required.[49] Personal information must be processed lawfully,[50] in a manner that is reasonable, not excessive, and in accordance with the given purpose.[51] The implications of POPIA on testing companies mean that personal information must be stored and discarded securely, and consumers must be informed of how their information is used and processed.

[39] Personal information in this context is collected in South Africa and is not simply forwarded through the country. Therefore, the exception contained in section 3(1)(*b*)(*ii*) of POPIA – that POPIA is irrelevant if 'automated or non-automated means' are only utilised to forward personal information through South Africa – does not apply because South Africa is not merely a conduit.
[40] Section 4(1) of POPIA requires the processing of personal information to comply with eight conditions to be lawful.
[41] Section 8 of POPIA.
[42] Sections 9 to 12 of POPIA.
[43] Sections 13 and 14 of POPIA.
[44] Section 15 of POPIA.
[45] Section 16 of POPIA.
[46] Sections 17 and 18 of POPIA.
[47] Sections 19 to 22 of POPIA.
[48] Sections 23 to 25 of POPIA.
[49] Section 16 of POPIA.
[50] Section 8 of POPIA.
[51] Sections 9 to 12 of POPIA. Testing companies must also be transparent in their processing, informing consumers and allowing them to partake in the processing, and control (in terms of access, correction, and deletion), of their personal information. Section 23 of POPIA.

Personal information, once collected, cannot be shared (unless authorised by the consumer).[52] Testing companies are only permitted to collect that which is essential to their object – in this instance, gathering personal details for the purpose of a direct-to-consumer genetic test.

III SOUTH AFRICA'S REGULATORY FRAMEWORK: CONSIDERATIONS FOR DIRECT-TO-CONSUMER GENETIC TESTING COMPANIES

Since the applicability of South Africa's legal regime relating to the offering of direct-to-consumer genetic tests to the public has been discussed, I now consider the substantive elements of the relevant legislation and clarify the legal position regarding the licensing, registering, and selling of direct-to-consumer genetic tests in South Africa.

(a) Licensing, registering, and selling direct-to-consumer genetic tests in South Africa

Chapter 4 established that while some direct-to-consumer genetic tests meet the definitions of both 'medical device'[53] and 'IVD',[54] many tests are not medical devices (and thus not IVDs), given the manufacturer's intention.[55] Devices that are not intended to be medical in nature do not need to comply with the laws relating to the licensing and registration of medical devices. Therefore, this section is only relevant to direct-to-consumer genetic tests that are IVD medical devices. This elucidates what is required of local, as well as foreign, testing companies wanting to offer their products in South Africa.

(i) Licensing direct-to-consumer genetic tests

Medical device establishment licenses are used to alert SAHPRA to manufacturers, importers, and distributors of medical devices in South Africa and their risk classification.[56] Medical devices and IVDs cannot be manufactured, imported, exported, sold, or distributed in South Africa without a valid medical device establishment license[57] – the requirements for which are expounded in the Medicines Act and the Medical Device Regulations.[58] One of three types of

[52] If this does occur, consumers can complain to the Information Regulator. Edward-John Bottomley 'Less than 100 days before SA's strict new privacy rules. Here's how it will affect you' *Business Insider South Africa* 25 March 2021, available at *https://www.businessinsider.co.za/popia-what-your-business-needs-to-do-2021-3*, accessed on 7 April 2021.
[53] As per section 1 of the Medicines Act.
[54] As per section 1 of the Medicines Act and regulation 1 of the Medical Device Regulations.
[55] As per the definition of 'medical device' in section 1 of the Medicines Act.
[56] South African Health Products Regulatory Authority (SAHPRA) *Guideline for a Licence to Manufacture, Import, Export or Distribute Medical Devices & IVDs* (2019) 3.
[57] Section 22C(6) of the Medicines Act; SAHPRA 'Medical devices' available at *https://www.sahpra.org.za/medical-devices/*, accessed on 5 May 2020.
[58] Section 22C(1)(*b*) of the Medicines Act; Regulation 5 of the Medical Device Regulations.

licenses must be applied for, and testing companies may need to apply for one or more such licenses, depending on the circumstances: (1) a manufacturer license;[59] (2) a distributor license;[60] or (3) a wholesaler license.[61]

(1) Manufacturers

Testing companies that design, manufacture, package, and label their tests prior to marketing are deemed to be manufacturers.[62] Most testing companies manufacture their own products and market them under their name.[63]

(2) Distributors

Testing companies that import or export completed and registered medical devices or IVDs with the intention of marketing and selling them under their name without changes to the form or packaging are distributors.[64] There are certain direct-to-consumer genetic tests that are

[59] In order to manufacture, label, pack, service, import, or export medical devices or IVDs. Regulation 5(1)(*a*)(*i*)(*aa*) of the Medical Device Regulations

[60] In order to import, export, and distribute medical devices or IVDs. Regulation 5(1)(*a*)(*i*)(*bb*) of the Medical Device Regulations.

[61] In order to store, transport, and deliver medical devices or IVDs. Companies that manufacture and wholesale medical devices are required to apply for both types of licenses. Regulation 5(1)(*a*)(*i*)(*cc*) of the Medical Device Regulations; See also, SAHPRA op cit note 57; Catherine Tomlinson 'IN-DEPTH: The tangled web of medical device regulation in SA' *Spotlight* 3 September 2020, available at *https://www.spotlightnsp.co.za/2020/09/03/in-depth-the-tangled-web-of-medical-device-regulation-in-sa/*, accessed on 5 December 2020.

[62] According to regulation 1 of the Medical Device Regulations, manufacturer 'means –

(a) a natural or legal person with the responsibility for the design, manufacture, packaging and labelling of a medical device or IVD before it is placed on the market under the natural or legal person's own name, or in the name of a firm or company, regardless of whether these operations are carried out by that person by himself or on his or her behalf by a third party; or

(b) any other person who assembles, packages, reprocesses, refurbishes or labels one or more ready-made products or assigns to them their intended purpose as a medical device or IVD, with a view to their being placed on the market under the natural or legal person's own name, except a person who assembles or adapts medical devices or IVDs already on the market to their intended purpose for patients'.

This may also be undertaken by a third party on the company's behalf. Manufacturers are also those testing companies that assemble, package, or label existing products, and place them on the market under their name. Regulation 1 of the Medical Device Regulations.

[63] Regulation 1 of the Medical Device Regulations. DNAlysis is a South African company, but states that their products and services are 'distributed worldwide by Nordic Laboratories and are available globally'. EasyDNA appears to be an international company that operates in different countries worldwide (including South Africa) under the same name. DNAlysis 'About' available at *https://dnalysis.co.za/about/*, accessed on 22 June 2020; EasyDNA 'Home' available at *https://www.easydna.co.za/*, accessed on 22 June 2020.

[64] Regulation 1 of the Medical Device Regulations defines a distributor as 'a natural or legal person who –

(a) imports or exports a medical device or IVD, which is on the register for medical devices or on the register for IVDs in its final form, wrapping and packaging, with a view to the medical device or IVD being placed on the market under the natural or legal person's own name; and

(b) sells the medical device or IVD to a healthcare professional, healthcare institution, wholesaler or the user'.

developed in a single laboratory and distributed globally, sometimes under a single name or by several different testing companies.[65]

(3) Wholesalers

Testing companies that buy medical devices or IVDs from a manufacturer or distributor and sell them to retailers are wholesalers.[66] Wholesalers are only permitted to purchase medical devices or IVDs from the original manufacturer or primary importer,[67] and can only sell them into the retail sector.[68]

Prior to registering a medical device or IVD, a medical device establishment license must be obtained.[69] As per section 22C(1)(*b*) of the Medicines Act, SAHPRA may grant a license to manufacture, distribute, or wholesale medical devices.[70] The licensing requirements for medical devices, as per the Medical Device Regulations, pertain to companies based in South Africa that import, export, manufacture, distribute, and wholesale medical devices in the country.[71] Although these licensing requirements apply to local testing companies, they hold implications for foreign manufacturers that export their products to South Africa.[72] They must provide importers and/or distributors in South Africa with device information including Global Medical Device Nomenclature (GMDN) codes,[73] and Certificates of Free Sale (CFS) for Class C and Class D IVD medical devices.[74] The South African importer or distributor must also

[65] Two examples of such companies are DNAlysis and EasyDNA. DNAlysis op cit note 63; EasyDNA op cit note 63.
[66] Regulation 1 of the Medical Device Regulations defines a wholesaler as 'a dealer who purchases medical devices or IVDs from a manufacturer or distributor and sells them to a retailer'.
[67] Section 22H(1)(*a*) of the Medicines Act.
[68] Section 22H(1)(*b*)(*i*) of the Medicines Act. It is unclear how many testing companies are wholesalers as it seems that most testing companies develop their own tests in their laboratories.
[69] T Saidi & TS Douglas 'Medical device regulation in South Africa: The Medicines and Related Substances Amendment Act 14 of 2015' (2018) 108(3) *S Afr Med J* 169.
[70] Without this, medical devices and IVDs cannot be imported or exported. SAHPRA op cit note 56 at 5.
[71] This does not apply to foreign manufacturers, although they must supply device information to importers and local distributors. This includes Global Medical Device Nomenclature codes and Certificates of Free Sale. A Certificate of Free Sale is proof that the medical device has received regulatory approval and is sold or distributed legally and freely. Saidi & Douglas op cit note 69 at 168–9.
[72] Stewart Eisenhart 'South African medical device regulatory system set for implementation' *Emergo* 22 August 2016, available at *https://www.emergobyul.com/blog/2016/08/south-african-medical-device-regulatory-system-set-implementation*, accessed on 18 June 2020.
[73] The GMDN is a list of generic names to identify, and internationally standardise, all medical devices – with the aim of allowing authorities worldwide to regulate medical devices. Through a naming system, the primary purpose of the GMDN is to supply health authorities, regulatory, healthcare providers, and manufacturers with a means of sharing medical device information. GMDN *GMDN User Guide: A Comprehensive Guide to the Global Medical Device Nomenclature* (2018) 9; GMDN Agency 'What is GMDN' available at *https://www.gmdnagency.org/services/gmdn*, accessed on 7 December 2020.
[74] A CFS is proof that the medical device has received regulatory approval and is sold or distributed legally and freely. Saidi & Douglas op cit note 69 at 168–9.

appoint an Authorised Representative, a natural person residing in South Africa, to oversee legal compliance.[75]

The level of risk and intended use of an IVD medical device determines the regulations applicable to its manufacture, import, export, distribution, and wholesale.[76] SAHPRA requires manufacturers, importers, exporters, or distributors of Class B, Class C, or Class D IVD medical devices to be licensed with them.[77] The requirements for manufacturer (including import or export) and distributor licenses for Class B, Class C, or Class D IVD medical devices are the same.[78] Class A IVD medical devices do not have to be licensed.[79]

When applying for a license, the particulars of the applicant must be provided,[80] and the type of business activities (whether it is manufacture, import, distribution, wholesale, or export) must be specified. Furthermore, information regarding the medical device or IVD must be furnished, including: (1) its name and/or group;[81] (2) GMDN code; and (3) risk classification.[82] As part of the licensing process, an Authorised Representative must also be appointed. If SAHPRA is satisfied that the applicant as well as the license application adheres to the prescribed conditions, a license is issued.[83]

---

[75] The Authorised Representative is a natural person responsible for activities such as importing, transporting, storage, distribution, marketing, and sales. Regulation 1 of the Medical Device Regulations define an 'Authorised Representative' as a 'natural person, resident in the Republic of South Africa, who –

(a) has the written mandate to represent a manufacturer, importer, distributor, wholesaler, retailer or service provider in the Republic;
(b) acts on behalf of a manufacturer, importer, distributor, wholesaler, retailer or service provider for specified tasks with regard to the latter's obligations and in whose name the manufacturer licence, distributor licence, wholesaler licence or certificate of registration is issued; and
(c) is responsible for all aspects of the medical device or IVD, including performance, quality, safety and compliance with conditions of registration, clinical trials or clinical investigations'.

The Authorised Representative will also manage the manufacturing, distribution, and wholesaling of medical devices or IVDs. Regulation 5(1)(*a*)(*ii*) of the Medical Device Regulations; SAHPRA op cit note 57; Eisenhart op cit note 72.

[76] Saidi & Douglas op cit note 69 at 169.

[77] SAHPRA op cit note 56 at 4.

[78] The following information must be provided: (1) a list of all medical devices or IVDs imported into South Africa with the Global Medical Device Nomenclature Code; (2) for Class C and Class D medical devices or IVDs, proof of pre-market approval or registration from certain overseas regulatory authorities; (3) for Class B, Class C, and Class D medical devices or IVDs, Certificate of Free Sale from country of manufacture; (4) license holders for Class C and Class D medical devices or IVDs must produce technical documentation if requested by SAHPRA; and (5) a certificate of conformance or analysis, where relevant. Ibid at 6 & 7.

[79] Ibid at 1; Tomlinson op cit note 61.

[80] This includes the full name of the legal person, individual identity number or company registration number, physical and postal address, and telephone number. Regulation 5(1)(*c*) of the Medical Device Regulations; SAHPRA op cit note 56 at 4.

[81] Regulation 5(1)(*d*) of the Medical Device Regulations; SAHPRA op cit note 56 at 5.

[82] For Class C and Class D medical devices and IVDs, the name and address of the manufacturer must be supplied. Moreover, the applicant must submit an application form and pay an application fee. Regulations 5(1)(*b*) and (*e*) of the Medical Device Regulations; SAHPRA op cit note 56 at 5.

[83] Regulation 5(4) of the Medical Device Regulations.

(ii) Registering and selling direct-to-consumer genetic tests

Unlike medicines, medical devices currently do not appear to have a registration process in place prior to being sold in South Africa.[84] There are overlapping requirements by SAHPRA, and in terms of the Medicines Act and the Medical Device Regulations relating to the registration of medical devices and IVDs. A medical device or IVD, which is subject to registration, cannot be sold in South Africa unless it is registered.[85]

Both the Medicines Act and the Medical Device Regulations contain requirements relating to applications for the registration of medical devices or IVDs.[86] Section 15(3)(*a*) of the Medicines Act states that a certificate of registration shall be granted if a medical device or IVD: (1) is fit for its intended purpose; (2) meets the stipulated requirements; and (3) is safe, effective, of proper quality, and functions as intended.[87] If successful, each medical device or IVD is provided with a name[88] and registration number which is recorded in the register and on the certificate of registration.[89]

Despite the above, SAHPRA is still developing mechanisms for establishing a medical device's quality, efficacy, and safety.[90] As the registration process for medical devices remains

[84] SAHPRA's registration of medical devices will entail a 'call-up' of particular products or classes to be registered by a notice in the Government Gazette. SAHPRA has published a draft call-up plan which details the risk-based approach to be used by SAHPRA in calling-up medical devices, prioritising those that are higher risk and vital for public health. Tomlinson op cit note 61.

[85] Section 14(1) of the Medicines Act. Regulation 8(1) of the Medical Device Regulations provides that any natural or legal person residing and conducting business in South Africa may apply to register a medical device or IVD.

[86] According to section 15(1) of the Medicines Act, applications for the registration of medical devices or IVDs must include: (1) the prescribed particulars; (2) where possible, samples of the medical device or IVD; and (3) the registration fee. The Medical Device Regulations expand on these requirements and provide that an application for registration must include: (1) the application form; (2) a proposed label; (3) the instructions for use; and (4) the application fee. The application form must contain: (1) the details of the prospective holder of the certificate of registration; and (2) the particulars of the medical device or IVD, which includes inter alia the name and group; proposed use or function; classification and registration status in identified authorities outside of South Africa as well as intended classification in the country. Where the medical device or IVD has not been manufactured in South Africa, the original manufacturer's name and address is required and, where applicable, clinical investigation sites must be provided. Moreover, an application for the registration of a medical device or IVD must adhere to SAHPRA's Essential Principles for Safety and Performance of Medical Devices, containing requirements for safety, performance, and quality. Where a medical device or IVD is registered outside of South Africa, the application must include: (1) a certified copy of the registration certificate or pre-market approval, where applicable; (2) instructions for use, where applicable; and (3) conditions of registration. SAHPRA may also request any other information that it deems applicable. Regulations 8(5), (6), and (9) of the Medical Device Regulations. Where applicable, additional documentation may be required, including a copy of the manufacturer or distributor licence as well as a conformity assessment certificate of a Quality Management System for the medical device establishment or for the medical device or IVD to be registered. Regulation 8(3) of the Medical Device Regulations.

[87] Section 15(3)(*a*) of the Medicines Act.

[88] Section 15(4) of the Medicines Act.

[89] Section 15(5) of the Medicines Act.

[90] Tomlinson op cit note 61.

in development,[91] SAHPRA's licensing process contains certain 'quasi-registration' requirements, which involve the provision of information on both the company and the medical device.[92] Companies must demonstrate the existence of suitable quality management systems that enable the safe manufacture or handling of medical devices that are of sound quality.[93] Furthermore, the application for an establishment license (which forms part of the 'quasi-registration' requirements), requires manufacturers and distributors of Class C and Class D IVD medical devices to show pre-market approval or registration in Australia, Brazil, Canada, the EU, Japan, the US, or pre-qualification by the World Health Organisation (WHO).[94]

As it is questionable whether SAHPRA will cope with the monumental task of testing the numerous medical devices on the market when they are called-up for registration by notice in the Government Gazette,[95] relying on the authorisation or registration of medical devices in other countries may offer an approach for local registration. But the absence of testing and certification of medical devices in South Africa means that local testing companies must obtain foreign pre-authorisation or registration in a recognised country to be granted an establishment licence, and thus permitted to market medical devices or IVDs in South Africa, especially where they are higher risk.[96] This means that as direct-to-consumer genetic tests that are Class C IVD medical devices must be registered in another jurisdiction to enable them to be sold in South Africa.

To conclude: Manufacturing, distributing, or selling direct-to-consumer genetic tests without a license in South Africa is illegal. Based on my suggestion in Chapter 4 that direct-to-consumer genetic tests that are IVD medical devices should be classified as Class C, testing companies operating in South Africa are required to apply for a manufacture, distribution, or

[91] SAHPRA op cit note 57.
[92] This includes a list of all medical devices to be manufactured, distributed, or wholesaled in South Africa. Ibid; Tomlinson op cit note 61; SAHPRA op cit note 56 at 5 & 8.
[93] A quality management system ensures the implementation of all elements of quality assurance, including: agreements and contracts; documents and records; audits, both internal and external; facility installation, maintenance, and cleanliness; manufacturing, final product handling, and storage; purchasing; training; handling of complaints; quality assurance; plans for emergencies and recalls; international regulatory controls; distribution (such as transportation, delivery, and temperature control); and documentation showing proof of export. SAHPRA op cit note 57; SAHPRA op cit note 56 at 5 & 8; Tomlinson op cit note 61.
[94] These regulatory authorities are: Australia's Therapeutic Goods Administration (TGA); Brazil's National Health Surveillance Agency (ANVISA); Canada's Medical Device Licence to market; the EU's CE marking certificate in line with the Medical Devices Directive (93/42/EEC); Japan's Marketing Authorization Holder (MAH); and the US FDA's Center for Devices and Radiological Health (CDRH) pre-market approval or pre-market notification 510(k) clearance. IVDs accepted in terms of the WHO's Prequalification of In Vitro Diagnostics Programme will also be recognised. Tomlinson op cit note 61; Saidi & Douglas op cit note 69 at 169; SAHPRA op cit note 56 at 6 & 7.
[95] Tomlinson op cit note 61.
[96] Ibid.

wholesale license depending on their activities – without which these tests cannot be sold to the public.[97] Additionally, as Class C IVD medical devices pose a greater risk, testing companies offering these direct-to-consumer genetic tests must also show proof of pre-market authorisation or registration from overseas. While regulating establishments is a partial solution, it is imperative that SAHPRA further develops and refines these registration processes for IVD medical devices and the requirements in order to guarantee the quality, efficacy, and safety of the relevant medical devices.[98]

IV CONSUMER REGULATION IN SOUTH AFRICA

The regulatory environment relating to the advertising, marketing, disclosure, and labelling of direct-to-consumer genetic tests is important to assess as it dictates what testing companies can and cannot do when promoting their products and services in South Africa. Below I analyse the poignant provisions of the CPA, ECTA, the Medicines Act, the Medical Devices Regulations, the *Medical Device Code of Ethical Marketing and Business Practice* (SAMED Code),[99] and the *South African Code of Marketing Practice for Health Products* (MCA Code)[100] in turn.

(a) Advertising and marketing direct-to-consumer genetic tests

(i) The Consumer Protection Act 68 of 2008 (CPA)

The CPA is South Africa's primary marketplace regulation for goods and services,[101] which seeks to make transactions and dealings fair, safe, and transparent and to create mechanisms for consumer redress.[102]

---

[97] Direct-to-consumer genetic tests that do not detect a serious condition or consist of preliminary results that require additional testing are likely to be classified as Class B IVD medical devices and will also have to be licensed. For example, a positive pregnancy self-test will usually involve a follow-up medical consultation, causing them to be classified as Class B IVDs. South African Health Products Regulatory Authority (SAHPRA) *Classification of Medical Devices and IVDs* (2019) 32 & 39.

[98] Tomlinson op cit note 61.

[99] South African Medical Device Industry Association (SAMED) *Medical Device Code of Ethical Marketing and Business Practice* (2017).

[100] Marketing Code Authority (MCA) *The South African Code of Marketing Practice for Health Products Code & Guideline, Version 14* (2021).

[101] Of particular importance to this dissertation are parts D (the right to disclosure and information), E (the right to fair and responsible marketing), F (the right to fair and honest dealing), G (the right to fair, just, and reasonable terms and conditions), and H (the right to fair value, good quality, and safety) of the CPA.

[102] According to section 55(2) of the CPA, consumers have the right to receive goods that: (i) are appropriate for their intended purpose; (ii) are in working order, of good quality, and free from defects; and (iii) will last for a reasonable time. The CPA provides its own enforcement mechanisms for consumers such as ombudsmen, consumer courts, the National Consumer Commission (NCC), and the National Consumer Tribunal (NCT). This is in addition to the existing common law remedies available to consumers. These bodies can impose fines on

Chapter 2 of the CPA covers fundamental consumer rights, including equality in the market,[103] privacy,[104] the right to choose,[105] and the right to disclosure and information.[106] Importantly, parts E and F in Chapter 2 of the CPA deal with marketing standards and deception. Section 29(a) provides that goods or services must not be marketed in such a way that is reasonably likely to mislead or give a false representation, concerning a material fact, to consumers[107] – including the nature and use of goods or services.[108] Suppliers are prohibited from using inter alia unfair tactics in the marketing of goods or services.[109] These safeguards prevent consumers from being erroneously led into purchasing direct-to-consumer genetic tests.[110] Many testing company websites try to persuade consumers to buy a test, especially given their profiteering interests. In this light, for there to be informed choice, it is imperative to present consumers with a balanced view of the limits, risks, and benefits of the direct-to-consumer genetic tests.[111]

The CPA also gives consumers the right to good quality and safe goods.[112] In determining this, regard must be had to inter alia: (1) the way that, and the reasons for which, goods are packaged, displayed, and marketed, the use of trade descriptions or marks, and any instructions or warnings;[113] and (2) the various uses of the goods.[114] If testing companies convey that their products consist of qualities, ingredients, performance characteristics, accessories, uses, or

suppliers who contravene provisions of the CPA and may set in motion criminal prosecutions. The CPA encourages consumers to resolve issues between parties or via the ombudsman. If there is no resolution through this route, consumers may approach provincial consumer courts. Section 69 of the CPA.

[103] Part A of Chapter 2 in the CPA (sections 8, 9, and 10 of the CPA).

[104] Part B of Chapter 2 in the CPA (sections 11 and 12 of the CPA).

[105] Part C of Chapter 2 in the CPA (sections 13 to 21 of the CPA).

[106] Part D of Chapter 2 in the CPA (sections 22 to 28 of the CPA).

[107] Sections 29(a) and 41(1)(*a*) of the CPA. In terms of section 41(1)(*b*) of the CPA, suppliers must also not exaggerate or use innuendo or ambiguity regarding a material fact, or knowingly allow a consumer to believe, and fail to disclose, a false, misleading, or deceptive fact.

[108] This includes the nature, properties, advantages, or uses, as well as price. Subsections 29(b)(*ii*) and (*iii*) of the CPA; Nicky Campbell & Stephen Logan *Consumer Protection Guide for Lawyers* (2011) 54.

[109] Section 40(1) of the CPA.

[110] Marietjie Botes 'Direct-to-consumer genetic tests: A treacherous road' *Caveat Legal* available at *http://www.caveatlegal.com/direct-to-consumer-genetic-tests-a-treacherous-road/*, accessed on 2 June 2020.

[111] Amanda Singleton, Lori Hamby Erby, Kathryn V Foisie et al 'Informed choice in direct-to-consumer genetic testing (DTCGT) websites: A content analysis of benefits, risks, and limitations' (2012) 21(3) *J Genet Counsel* 6.

[112] Section 55(2) of the CPA states that 'every consumer has a right to receive goods that—

(a) are reasonably suitable for the purposes for which they are generally intended;
(b) are of good quality, in good working order and free of any defects;
(c) will be useable and durable for a reasonable period of time, having regard to the use to which they would normally be put and to all the surrounding circumstances of their supply; and
(d) comply with any applicable standards set under the Standards Act, 1993 (Act No. 29 of 1993), or any other public regulation'.

[113] Section 55(4)(*a*) of the CPA. See also, Campbell & Logan op cit note 108 at 86–7.

[114] Section 55(4)(*b*) of the CPA. See also, Campbell & Logan op cit note 108 at 86–7.

benefits;[115] or are of a certain quality or standard when this is untrue,[116] it amounts to a false, misleading, or deceptive representation.[117] Because direct-to-consumer genetic tests have been found to have questionable analytical and clinical validity and clinical utility, these aspects must be clearly explained to consumers. To comply with the CPA, South African testing companies marketing direct-to-consumer genetic tests in South Africa or via a website with a South African domain must avoid marketing such tests as a diagnostic tool with definitive test results. Consumers must be informed of things such as the fact that test results merely show genetic predisposition – which is affected by other factors, such as environment and lifestyle, and may never express itself phenotypically, that results do not constitute medical diagnoses and are generally for informational purposes.[118]

I suggest that it would be best practice for testing companies to highlight this fact to consumers on their websites, on the testing kits, in the contracts (and terms and conditions), and in the test results. While this is already done in some instances, more visible emphasis should be placed on the limitations of direct-to-consumer tests.[119]

---

[115] In terms of section 41(3)(*b*)(*i*) of the CPA.

[116] In terms of section 41(3)(*b*)(*ii*) of the CPA.

[117] Simplex commendatio refers to mere commendation or puffery. Puffery is a statement that is neither intended to be taken seriously, nor could any reasonable person attach any serious consideration to it. However, this does not mean that individuals can make any statement that they like. For example, if a testing company states that their tests are completely accurate and will diagnose disease, but it turns out that there were false positive results and the test only showed predisposition to disease (rather than concrete diagnosis), this is not a mere puff and the testing company may be liable for misrepresentation. In *Carlill v Carbolic Smoke Ball Co* [1892] 2 QB 484, the defendant (Carbolic Smoke Ball Company) advertised their products (which they claimed would prevent colds and influenza) in a newspaper, stating that any individual who purchased and used their product as instructed, but still contracted influenza would be entitled to a £100 reward. The defendant guaranteed this in their advertisement by stating that they had deposited £1000 in the bank as a show of their sincerity. Believing the defendant's advertisement, the plaintiff (Lilli Carlill) bought a Carbolic Smoke Ball and used it as directed, but nevertheless contracted influenza. She approached the Court to recover the £100 from the defendants. The primary issue in this case was whether the language used in the advertisement, regarding the £100 reward, was intended to be an express promise or a mere puff. The Court found in favour of the plaintiff and held that the advertisement was not a puff because the statement by the defendant in their advertisement regarding the deposit of £1000 was proof of their sincerity to pay the reward.

[118] This must be clearly indicated in marketing materials to bring it to the attention of consumers. Due to exaggerated claims about benefits and clinical utility and false information regarding risk, the advertising of health-related direct-to-consumer genetic tests may create unfounded beliefs or anxiety and confusion. Martina C Cornel, Carla G van El & Pascal Borry 'The challenge of implementing genetic tests with clinical utility while avoiding unsound applications' (2012) *J Community Genet* 5; Alice K Hawkins & Anita Ho 'Genetic counseling and the ethical issues around direct to consumer genetic testing' (2012) 21 *J Genet Counsel* 368.

[119] Advertising of direct-to-consumer genetic testing has been found to overemphasise the advantages while overlooking possible risks, and information on the disadvantages of testing and its clinical value are lacking. Where risks are discussed, they tend to be over-simplified and deficient. AD Schickedanz & RC Herdman 'Direct-to-consumer genetic testing: The need to get retail genomics right' (2009) 86(1) *Clinical pharmacology & Therapeutics* 18–9.

(ii) The Medicines and Related Substances Act 101 of 1965 (Medicines Act) and the Regulations Relating to Medical Devices and In Vitro Diagnostic Medical Devices (Medical Device Regulations)

The Medicines Act was enacted to control matters related to medicines and to establish the MCC, which has been replaced by SAHPRA.[120] The Medicines Act refers to advertising, specifically focusing on medical devices, and thus only relevant to direct-to-consumer genetic tests that are regulated as such.[121] 'Advertising' in the Medicines Act, is described as any visual or verbal work that is shared or brought to the attention of the public.[122]

The Medicines Act states that medical devices or IVDs cannot be advertised for sale unless they adhere to certain conditions,[123] and prohibits the publication and distribution of false advertisements regarding medical devices or IVDs.[124] Statements in advertisements must not conflict with evidence in the application for registration of a medical device or IVD in terms of its quality, safety, or performance where it has been accepted by SAHPRA and forms part of the instructions for use.[125] This means that direct-to-consumer genetic testing advertisements can neither make false claims regarding the efficacy and result, nor can they recommend that a medical device or IVD should be used for a purpose contrary to SAHPRA's mandate.[126]

---

[120] As discussed above, the Medicines Act also oversees licenses to manufacture, wholesale, or distribute medical devices. Preamble to the Medicines Act.

[121] As per the definition of 'medical device' in section 1 of the Medicines Act.

[122] Section 1(a) of the Medicines Act defines 'advertisement' as 'any written, pictorial, visual or other descriptive matter or verbal statement or reference –

(a) appearing in any newspaper, magazine, pamphlet, electronic media (including radio and television) or other publication;
(b) distributed to members of the public; or
(c) brought to the notice of members of the public in any manner whatsoever,

which is intended to promote the sale of that medicine, Scheduled substance, medical device or IVD'.
The definition of 'sell' in section 1 of the Medicines Act includes advertising, and is defined broadly to include inter alia importing, offering, advertising, conveying, or delivering. According to section 19(1) of the Medicines Act, in order to sell a medical device or IVD, it must comply with prescribed requirements.

[123] Section 18(2) of the Medicines Act.

[124] Section 20(1)(*a*) of the Medicines Act.

[125] Regulation 21(1)(*c*) of the Medical Device Regulations. The Medical Device Regulations refer to the 'Council' – which refers to the MCC, as evidenced by the Preamble to the Medical Device Regulations, as well as the definition of 'as determined by Council', which means 'as determined by the Medicines Control Council'. But this has since been replaced by SAHPRA. Regulation 21(1)(*d*) of the Medical Device Regulations provide that written advertisements for medical devices or IVDs must include their name and, where applicable, the registration number.

[126] Section 20(1) of the Medicines Act.

Unlike in the US, where direct-to-consumer genetic testing is widely advertised on television and in magazines,[127] South Africa currently has a smaller market – and seemingly stricter standards. The Medical Device Regulations permit the advertising of only Class A and Class B IVD medical devices to the public or laypersons.[128] However, the Medical Device Regulations refer to the initial advertising of Class C or Class D medical devices or IVDs to a 'prospective user',[129] who must be provided with written information, including at least the instructions for use.[130] It has been suggested that Class C and Class D IVD medical devices may only be advertised to inter alia medical practitioners and pharmacists.[131] But the wording 'prospective user' in regulation 21(1)(*e*) of the Medical Device Regulations does not take into account direct-to-consumer genetic testing. In such instances, the prospective user of the test is the consumer – who is generally a member of the public or a layperson, as per regulation 21(1)(*a*) of the Medical Device Regulations.[132] This implies that direct-to-consumer genetic tests which are classified as Class C IVD medical devices cannot be advertised to consumers. However, direct-to-consumer genetic tests that are not medical devices can be advertised freely to the public like other consumer goods.

(iii) Medical Device Code of Ethical Marketing and Business Practice (SAMED Code)

The South African Medical Device Industry Association (SAMED) established the SAMED Code,[133] to regulate the ethical marketing of medical devices. Although there is a chapter on

[127] Furthermore, direct-to-consumer genetic tests are handed out at National Football League (NFL) games in the US, they are advertised as ideal holiday gifts, and feature on common reality television and talk shows. This shows that there is a greater demand and uptake of direct-to-consumer genetic tests in other countries, which has resulted in commercial advertising and marketing of the tests. Seon-Hee Yim & Yeun-Jun Chung 'Reflections on the US FDA's warning on direct-to-consumer genetic testing' (2014) 12(4) *Genomics Inform* 152; Valerie Gutmann Koch & Kelly Todd 'Research revolution or status quo: The new common rule and research arising from direct-to-consumer genetic testing' (2018) 56(1) *Hous L Rev* 83.

[128] Regulation 21(1)(*a*) of the Medical Device Regulations.

[129] Regulation 21(1)(*e*)(*i*) of the Medical Device Regulations.

[130] Referred to in regulations 23 and 24 of the Medical Device Regulations. When there is subsequent advertising of the medical device or IVD, the information must be available if requested. Regulation 21(1)(*e*) of the Medical Device Regulations.

[131] Martha Smith 'Marketing, manufacturing, packaging & labelling, advertising' *Pharma Boardroom* 12 October 2018, available at *https://pharmaboardroom.com/legal-articles/marketing-manufacturing-packaging-labelling-advertising-south-africa/*, accessed on 16 January 2021.

[132] If Class C and Class D medical devices and IVDs can only be advertised to persons such as medical practitioners, then these are the people 'to whom the advertisement is directed' (as per regulation 21(1)(*e*) of the Medical Device Regulations). But a medical practitioner is not the prospective user of such a device in the case of direct-to-consumer genetic testing. Even if a healthcare professional is involved in the direct-to-consumer genetic testing process, they do not use the device, they assist consumers in using it.

[133] SAMED op cit note 99. It appears that SAMED changed its name from the South African Medical Technology Industry Association, as referred to in previous codes. A previous version (version 7) of the South African Medical Technology Industry Association *Medical Device Code of Ethical Marketing and Business Practice* made extensive reference to the advertising and promotion of medical devices and IVDs. However, the latest ratified

the advertising of medical devices and IVDs, the SAMED Code does not deal with this, making it the responsibility of members to comply with 'relevant legislation and/or regulations' relating to the advertising of medical devices or IVDs.[134] Although the SAMED Code constitutes a mechanism of self-regulation and is only binding on its members,[135] those advertising direct-to-consumer genetic tests that are medical devices must adhere to the provisions of the CPA and the Medical Device Regulations relating to advertising.

(iv) South African Code of Marketing Practice for Health Products (MCA Code)

The MCA Code was issued in terms of section 18C of the Medicines Act and contains provisions relating to the advertising of medical devices and IVDs.[136] The MCA Code extensively covers the marketing and advertising of health products in South Africa, which must be in line with the Medicines Act.[137] 'Health products' are defined in the MCA Code as inter alia 'Medical Devices, and IVDs as regulated by the Medicines Act'[138] – thereby encompassing direct-to-consumer genetic tests that are IVD medical devices.

While the MCA Code is comprehensive, there are relevant aspects which must be highlighted. The MCA Code confirms that the advertising of medical devices and IVDs must be in accordance with the Medicines Act, meaning that only Class A and Class B IVD medical devices may be advertised to the public.[139] Class C IVD medical devices, which I suggest include some direct-to-consumer genetic tests, cannot be advertised to the public.[140]

The MCA Code further states that companies must not provide consumers with information or guidance regarding their personal medical issues where requested, instead

version of the SAMED Code has removed this information. However, some of the omitted information does appear in the MCA op cit note 100.

[134] These include the Advertising Standards Authority of South Africa's (ASA) *Advertising Code of Practice*, the CPA, and the Medical Device Regulations. Infringements or complaints regarding the advertising of medical devices or IVDs are also not covered in the SAMED Code. SAMED op cit note 99 at 37; South African Medical Device Industry Association (SAMED) *An Overview of the Medical Device Code of Ethical Marketing and Business Practice* 12.

[135] SAMED op cit note 99 at 7 & 10.

[136] MCA op cit note 100 at 1.

[137] Ibid at 14.

[138] Ibid at xii.

[139] Ibid at 66.

[140] However, it seems that advertisements for such products can still target healthcare practitioners. Furthermore, the MCA Code provides that non-promotional information regarding Class C and Class D medical devices and IVDs may be provided to consumers, in cases of direct queries from individuals or by distribution through channels such as press conferences and announcements, and television and radio reports. Such information must be fair, credible, and must not advocate for consumers to request healthcare practitioners to prescribe a particular product. Ibid at 67.

advising them to consult with their healthcare practitioner.[141] The promotion of health products, including those using electronic or digital media, must follow the MCA Code.[142] Advertisements must neither compromise advice pertaining to healthy lifestyles, nor must they discourage the obtaining of medical advice,[143] or include inappropriate, ambiguous, or distressing claims regarding recovery.[144] Advertising is prohibited from offering virtual diagnoses, advice, or treatment.[145] Material must be sufficiently comprehensive, thereby allowing consumers to develop their own assessment of a product's therapeutic value.[146] In line with the MCA Code, advertisements by testing companies are required to observe the 'Minimum Requirements',[147] and must be inter alia: (1) accurate and balanced;[148] (2) coherent;[149] (3) unambiguous, reflective of current evidence,[150] and able to be verified;[151] and (4) presented objectively without exaggeration.[152] Such advertisements must not: (1) be

[141] Further, 'help-seeking advertisements' aimed at consumers are permitted, as long as material for Class C and Class D medical devices and IVDs does not: (1) include, or imply, the product's name; (2) make, or refer to, medicinal or therapeutic claims; and (3) utilise safety or risk information. Advertisements must alert consumers to the fact that treatment exists for a medical condition, and contain the phrase, 'for more information, refer to your HCP' (healthcare practitioner). Ibid at 66 & 68.

[142] According to the MCA Code, electronic or digital media encompasses 'the Internet, websites, applications, social media, electronic signage and e-commerce'. In terms of social media, companies are responsible for their activities, which must adhere to the MCA Code. Companies that display public testimonials or comments on their Facebook and Twitter pages will be liable for their falsity or deception. Social media content must be controlled by companies, and unsuitable content must be removed within 24 hours. Ibid at xi, 33 & 36–7.

[143] Ibid at 15.

[144] Ibid.

[145] Ibid.

[146] Ibid at 21.

[147] 'Minimum requirements' denotes the legislative requirements for written advertisements provided for in the General Regulations made in terms of the Medicines and Related Substances Act, 1965, as Amended GN R510 GG 24727 of 10 April 2003 (General Regulations for Medicines) and the Medical Device Regulations. Ibid at xii.

[148] Ibid at 66.

[149] Ibid at 18.

[150] Claims must be able to exist independently without the use of footnotes or links. Ibid at 21.

[151] However, substantiation is not required for claims in the Instructions for Use approved by SAHPRA. Evidence in support of a claim must not be ambiguous, emotive, unlimited, or incorrect, and able to be substantiated, scrutinised, and independently reviewed. Advertisers must show that a systematic approach to inspecting evidence has been adopted. Acceptable evidence includes: (1) data published in peer-reviewed journals; (2) textbooks; and (3) unpublished company data accepted by its medical or regulatory departments, while improper evidence consists of: (1) that which is outdated due to recent studies and scientific progress; and (2) newspaper reports and other editorial material that are anecdotal and unsupported by clinical evidence. References, of a scientific journal standard, must appear in the advertisement. Ibid at 26–7.

[152] Exaggerated or absolute claims cannot be made, and superlatives must not be utilised, unless they link to a distinct and substantiated fact regarding the product. Claiming that a product is 'the best' treatment of a certain condition contains too many variables to confirm the claim. Moreover, use of the words 'unique' and 'ultimate' must be approached with caution. Ibid at 21–2.

frivolous;[153] (2) create false hope for successful treatment;[154] or (3) guarantee the safety, quality, or efficacy of a product.[155]

Some testing companies offer the provision of genetic counselling as part of their service and advertise this – predominantly on their websites. But such counselling is often not offered face-to-face, with testing companies instead relying on email communication and telephone or video calls.[156] Offering genetic counselling in direct-to-consumer genetic testing advertisements contravenes the MCA Code, which prohibits offering virtual diagnoses, advice, or treatment.[157]

Advertising and marketing by testing companies has been recognised as potentially ambiguous.[158] Based on the MCA Code, scientific claims made by companies must be supported with evidence and device safety, quality, and efficacy must not be guaranteed.[159] Information must not be scant or misleading, and should be sufficient enough to allow consumers to decide to undergo direct-to-consumer genetic testing or rather avoid it. Testing companies may make claims, or use words, regarding their products and services that exaggerate the safety, usefulness, or non-diagnostic nature of their tests.[160] In order to comply with the MCA Code, testing companies must refrain from doing so.

---

[153] Ibid at 18.
[154] Ibid at 67.
[155] Additionally, the wording and images used in advertisements is important and is included in the MCA Code. For example, the word 'safe', its alternatives, or words referring to safety must not indicate that a product has no dangers or side effects. The word 'serious' must only be utilised in describing conditions, defects, or ailments generally acknowledged as being: (1) inappropriate to be diagnosed and/or treated without a healthcare practitioner; and/or (2) unable to be correctly assessed and safely treated without professional oversight. Artwork used in advertisements – which includes illustrations, logos, graphs, tables, and trade dress – must not: (1) be unsuitable; (2) misrepresent the nature of a product or claim; (3) downplay its safety; or (4) disclose any additional information prohibited by the Medicines Act. Ibid at 19 & 25.
[156] Some testing companies, as a substitute for face-to-face genetic counselling, include informational materials and videos on their websites, telephonic support, and online counselling by professionals contracted by the company. Teresa Pàmpols Ros, José Miguel García Sagredo, Antonio Pérez Aytése et al 'Directed to consumer genetic testing: Perspective from the Ethics Commission of the Spanish Society for Human Genetics' (2019) 153(1) *Med Clin (Barc)* 37.
[157] MCA op cit note 100 at 15.
[158] Ruth Saunders 'Legal implications of direct-to-consumer genetic testing for common diseases' (2010) 1 *QMLJ* 77.
[159] Overseas, Covolo et al note that direct-to-consumer genetic testing is advertised despite the lack of supporting evidence. Loredana Covolo, Sara Rubinelli, Elisabetta Ceretti et al 'Internet-based direct-to-consumer genetic testing: A systematic review' (2015) 17(12) *J Med Internet Res* 11–2.
[160] For example, 23andMe's website marketed its products and service as 'the first step in prevention', allowing consumers to 'take steps toward mitigating serious diseases'. Other companies use claims like 'let your DNA help you plan for the important things in life. Take charge of your health and wellness today', which exaggerate direct-to-consumer genetic tests' value in improving health. Other direct-to-consumer genetic testing advertisements claim that 'knowledge is power', but for information to be empowering, it must be correct and relevant. Ryan Jaslow 'FDA warns 23andMe, tells genetic testing firm to halt sales' *CBS News* 25 November 2013, available at *https://www.cbsnews.com/news/fda-warns-23andme-tells-genetic-testing-firm-to-halt-sales/*, accessed on 25 October 2020; Megan A Allyse, David H Robinson, Matthew J Ferber et al 'Direct-to-consumer testing 2.0:

(b) Disclosure and direct-to-consumer genetic tests

(i) The Consumer Protection Act 68 of 2008 (CPA)

Part D of the CPA deals with disclosure – specifically sections 22 to 24. These sections give effect to the consumer's right to information, which is important insofar as any marketing materials, contracts, and notices are concerned, requiring companies to disclose information in 'plain language'.[161] In terms of the common law, key information or contractual terms should also be clearly disclosed and brought to the attention of consumers.[162] This requires testing companies, when making use of more complex terminology dealing with things such as genetics or when explaining how the testing process works and how results are determined, to ensure that this is explained clearly and reduced to plain language when advertising to, or contracting with, consumers.

(ii) The Electronic Communications and Transactions Act 25 of 2002 (ECTA)

ECTA requires that suppliers provide consumers with certain basic information in electronic transactions and, because some testing companies market their products electronically and require consumers to enter into electronic transactions, this is relevant. The notable provisions are those contained in section 43 of ECTA,[163] which relate to disclosure requirements and

---

Emerging models of direct-to-consumer genetic testing' (2018) 93(1) *Symposium on Precision Medicine* 117; Sara Chandros Hull & Kiran Prasad 'Reading between the lines: Direct-to-consumer advertising of genetic testing' (2001) 31(3) *Hastings Center Rep* 33.

[161] Section 22(1)(*b*) of the CPA. Section 22(2) of the CPA attempts to define plain language and states that –

'(2) For the purposes of this Act, a notice, document or visual representation is in plain language if it is reasonable to conclude that an ordinary consumer of the class of persons for whom the notice, document or visual representation is intended, with average literacy skills and minimal experience as a consumer of the relevant goods or services, could be expected to understand the content, significance and import of the notice, document or visual representation without undue effort, having regard to –

(a) the context, comprehensiveness and consistency of the notice, document or visual representation;
(b) the organisation, form and style of the notice, document or visual representation;
(c) the vocabulary, usage and sentence structure of the notice, document or visual representation; and
(d) the use of any illustrations, examples, headings or other aids to reading and understanding'.

Plain language is key to determining whether the supplier engaged in unconscionable conduct (section 41 of the CPA) or made false, misleading, or deceptive representations (sections 23 and 24 of the CPA) in the process of dealing with the consumer.

[162] See, *Durban's Water Wonderland (Pty) Ltd v Botha* 1999 (1) SA 982 (SCA).

[163] The relevant parts of section 43(1) of ECTA read as follows: '(1) A supplier offering goods or services for sale, for hire or for exchange by way of an electronic transaction must make the following information available to consumers on the web site where such goods or services are offered:

(a) Its full name and legal status;
(b) its physical address and telephone number;
(c) its web site address and e-mail address;
(d) membership of any self-regulatory or accreditation bodies to which that supplier belongs or subscribes and the contact details of that body;
…

information to be provided to consumers. This section compels testing companies to make information available to consumers on their websites. This includes:

(a) Details of the testing company (such as their name, address, registration number, and contact details);[164]

(b) Details regarding the products and services offered (such as adequate descriptions of their primary features to enable consumers to make informed decisions, the price as well as tax and shipping fees, the payment method, and return policy);[165]

(c) Details regarding the privacy policy and security measures that deal with the processing of payment and personal information.[166]

I suggest that section 47 of ECTA makes it clear that any testing company – which is either based in South Africa, has a website with a South African registered domain, or offers its products and services to the South African public – is bound by this section of ECTA. Inter alia, testing companies must comply with the requirements listed above on their advertising platforms including their websites, promotional materials, and emails.

(c) Labelling and direct-to-consumer genetic tests

(i) The Consumer Protection Act 68 of 2008 (CPA)

Labelling forms an important part of direct-to-consumer genetic testing and is covered by the CPA. This includes the tags or stickers, information, and images contained on the testing kit. The requirements for labelling in the CPA determine what testing companies are required to include on their products. The CPA uses the term 'trade description' to refer to statements made

(f) in the case of a legal person, its registration number, the names of its office bearers and place of registration;
…
(h) a sufficient description of the main characteristics of the goods or services offered by that supplier to enable a consumer to make an informed decision on the proposed electronic transaction;
(i) the full price of the goods or services, including transport costs, taxes and any other fees or costs;
(j) the manner of payment;
(k) any terms of agreement, including any guarantees, that will apply to the transaction and how those terms may be accessed, stored and reproduced electronically by consumers;
…
(n) the return, exchange and refund policy of that supplier;
…
(p) the security procedures and privacy policy of that supplier in respect of payment, payment information and personal information'.

[164] Sections 43(1)(*a*), (*b*), (*c*), (*d*), (*e*), (*f*), and (*g*) of ECTA.
[165] Sections 43(1)(*h*), (*i*), (*j*), (*k*), (*l*), (*m*), (*n*), and (*o*) of ECTA.
[166] Section 43(1)(*p*) of ECTA. In terms of section 43(2) of ECTA, consumers must also be given the opportunity to review the electronic transaction, correct any errors, and withdraw before an order is placed.

in advertisements, labels, or packaging that describe inter alia the 'number, quantity, measure, weight or gauge'[167] of goods; the name of the producer; the ingredients or materials; and the place where the goods are made.[168] Trade descriptions must appear on the goods in a visible and readable manner in plain language.[169] The CPA requires that labelling and product packaging must not deceive consumers.[170] This links to the common theme that is present throughout the CPA that consumers must not be misled and should have goods and services offered in a transparent and understandable manner.

(ii) The Medicines and Related Substances Act 101 of 1965 (Medicines Act) and the Regulations Relating to Medical Devices and In Vitro Diagnostic Medical Devices (Medical Device Regulations)

The Medicines Act prohibits any person from selling a medical device or IVD without a label,[171] which is defined as any brand, mark, or description visible on, affixed to, or packed with an article, and that refers to an article. This must appear on the device or packaging (where practical) and include stipulated details as determined by SAHPRA.[172] The Medical Device Regulations expand on the particulars that a medical device or IVD label must contain, which include: Its name,[173] description,[174] intended use,[175] the name and business address of the manufacturer,[176] the lot and serial numbers (where applicable),[177] and any warnings or precautions.[178] Additionally, the intended use of the medical device must be indicated as: (1)

[167] Section 1 of the CPA.
[168] Ibid; Simone Monty 'South Africa: Things to consider when packaging your products' *Mondaq* 7 November 2011, available at *https://www.mondaq.com/southafrica/consumer-law/149662/things-to-consider-when-packaging-your-products*, accessed on 15 March 2021.
[169] It is adequate for trade descriptions to be visible near the goods, rather than attached to them. Section 24(2)(*b*) of the CPA provides that one must not 'alter, deface, cover, remove or obscure a trade description or trade mark applied to any goods in a manner calculated to mislead consumers'. Sections 22 and 24 of the CPA; Campbell & Logan op cit note 108 at 47–8.
[170] Section 24 of the CPA.
[171] Section 1 of the Medicines Act defines a 'label' as 'when used as a noun, means any brand or mark or any written, pictorial or other descriptive matter appearing on or attached to or packed with and referring to any article or the package containing any article'.
[172] Section 18(1)(*b*) of the Medicines Act. SAHPRA is responsible for approving labels on medical devices and IVDs, and may prescribe its own conditions regarding a label's contents and format. Subsections 18(3) and (4) of the Medicines Act.
[173] Regulation 22(1)(*a*) of the Medical Device Regulations.
[174] Regulation 22(1)(*b*) of the Medical Device Regulations.
[175] Ibid.
[176] Regulation 22(1)(*d*) of the Medical Device Regulations.
[177] Regulations 22(1)(*g*) and (*h*) of the Medical Device Regulations.
[178] Regulation 22(1)(*o*) of the Medical Device Regulations. In terms of regulation 22(1) of the Medical Device Regulations, other information that a medical device or IVD label must contain are inter alia: (1) a product catalogue, where applicable; (2) the name and address of the holder of the registration certificate; (3) an indication that the medical device includes a biological substance, where relevant; (4) the expiry or manufacturing date; (5)

single use;[179] (2) clinical investigation or pre-market clinical performance study;[180] (3) non-clinical research, teaching, or testing;[181] (4) presentation or demonstration;[182] (5) IVD use or Laboratory Developed Tests (LDTs);[183] and (6) where appropriate, 'for professional use only', 'near patient testing', 'point of care', or 'self- testing'.[184]

Medical device or IVD labels are required to be in at least English and must appear on: (1) the actual medical device or IVD;[185] or (2) each item's packaging;[186] and (3) the packaging of numerous medical devices or IVDs.[187] Labelling links to marketing and, as per regulation 21 of the Medical Device Regulations, advertisements for medical devices or IVDs may not consist of statements that differ from, or contravene, evidence in the registration application.[188] To sell their products, testing companies must ensure that their tests are suitably labelled and do not contain any information that deviates from that which has been approved by SAHPRA.

To conclude, the CPA, the Medicines Act, and the Medical Device Regulations all contain provisions relating to labelling. While the Medicines Act and the Medical Device Regulations only apply to medical devices, the CPA governs the labelling of all goods offered within South Africa. Therefore, even direct-to-consumer genetic tests that are not medical devices must still comply with the CPA's labelling requirements.

V CONCLUSION

(a) Licensing, registering, and selling considerations

In order to manufacture, import, export, sell, or distribute medical devices and IVDs in South Africa, a license – obtained prior to registering such medical device or IVD – is required. While neither Class A medical devices, nor direct-to-consumer genetic tests that do not qualify as medical devices, do not need to be licensed, it is necessary to license those posing a higher risk. As such, Class C IVD medical devices – which I suggest certain direct-to-consumer genetic tests should be classified as – require a license. These licensing requirements only apply to

---

the relevant storage or handling requirements; and (6) the net quantity of the contents, where applicable (this can be expressed in weight, volume, numerical count, or any other suitable measure).

[179] Regulation 22(1)(*p*)(*i*) of the Medical Device Regulations.
[180] Regulation 22(1)(*p*)(*ii*) of the Medical Device Regulations.
[181] Regulation 22(1)(*p*)(*iii*) of the Medical Device Regulations.
[182] Regulation 22(1)(*p*)(*iv*) of the Medical Device Regulations.
[183] Regulation 22(1)(*p*)(*v*) of the Medical Device Regulations.
[184] Regulation 22(1)(*p*)(*vi*) of the Medical Device Regulations.
[185] Regulation 22(2)(*a*) of the Medical Device Regulations.
[186] Regulation 22(2)(*b*) of the Medical Device Regulations.
[187] Regulation 22(2)(*c*) of the Medical Device Regulations.
[188] Smith op cit note 131.

South African testing companies, but foreign testing companies that export their tests to South Africa must also provide importers and/or distributors with certain medical device information.[189]

As medical devices currently seem to lack an established registration pathway in South Africa, this aspect is more challenging. Without being registered, direct-to-consumer genetic tests cannot be sold in the country.[190] Therefore, testing companies offering to direct-to-consumer genetic tests that are medical devices must show proof of pre-market authorisation or registration from overseas to sell their tests in South Africa. For South African testing companies, this is an arduous and costly task, but it is a necessary part of the licensing process and one which allows testing companies to manufacture, distribute, or wholesale their tests. It may assist for SAHPRA to introduce guidance that updates licensing and registration processes, addresses the overlapping legislation, provisions, and codes, and covers testing companies more fully.

(b) Relevant consumer protection requirements

Offering direct-to-consumer genetic tests to the public in South Africa requires numerous considerations to be taken into account, primarily in terms of advertising, marketing, disclosure, and labelling. The CPA requires testing companies to act in such a way that does not mislead or deceive consumers, providing them with sufficient, accurate, clear, and understandable information regarding their advertisements, terminology in their contracts, and other information – including on their websites. ECTA contains requirements pertaining to electronic transactions and consumer protection – relevant to testing companies that operate and transact online.[191]

In this Chapter, POPIA is relevant to personal information in the form of names, addresses, and contact details of consumers that are entered into testing company websites prior to undergoing a test. In terms of POPIA, entering these details into a website in South Africa – even where it is to be received overseas – is considered 'automated means'[192] and thus, POPIA applies to South African testing companies as well as those companies not domiciled in South Africa, but which make use of 'automated means' in the country.[193] For testing companies to

[189] This includes GMDN codes and CFSs, as well as appointing an Authorised Representative in South Africa.
[190] Section 14(1) of the Medicines Act.
[191] Section 43 of ECTA stipulates the information that must be disclosed by testing companies such as their particulars, the products and services offered, as well as their privacy policies and security measures.
[192] As per section 3(1)(*b*) of POPIA.
[193] Section 3(1)(*b*)(*ii*) of POPIA.

ensure consumer trust and adhere to POPIA – which has various provisions in place that aim to protect the privacy of consumers' personal information – testing companies must ensure that personal information is processed lawfully.

Labelling is covered by the CPA, the Medicines Act, and the Medical Device Regulations. Labels should be visible and clear, using understandable language with no deceit.[194] The abovementioned detailed requirements relating to labels for medical devices and IVDs indicates the specificity of labelling in South African law.[195] To legally offer direct-to-consumer genetic tests in South Africa, labels must have the appropriate information contained therein.

The Medicines Act outlaws the false advertising of medical devices and IVDs,[196] thus bringing it in line with the CPA's requirements relating to deception. However, an important consideration in the Medical Device Regulations is the fact that Class C IVD medical devices – under which I suggest that certain direct-to-consumer genetic tests fall – cannot be advertised to the public or laypersons.[197] This means that consumers technically have access to direct-to-consumer genetic tests, but such tests cannot be advertised to them – similar to the advertising of tobacco products in South Africa.[198] Therefore, both South African and foreign testing companies offering direct-to-consumer genetic tests that are medical devices to South African consumers are acting illegally in terms of the Medical Device Regulations. However, the Medical Device Regulations refer to the advertising of Class C medical devices or IVDs to a 'prospective user'.[199] It seems that this was drafted with healthcare professionals in mind, where certain Class C medical devices or IVDs are to be used by them in the course of their work, and not situations wherein individuals have access to medical devices or IVDs.

The provisions in the CPA[200] and ECTA[201] are also applicable to testing companies in foreign jurisdictions, and such companies must ensure that their products and services comply with this legislation in order to function legally in South Africa.

---

[194] Sections 22 and 24 of the CPA.
[195] Smith op cit note 131.
[196] Section 20(1)(*a*) of the Medicines Act.
[197] Regulation 21(1)(*a*) of the Medical Device Regulations.
[198] Section 3(1)(*a*) of the Tobacco Products Control Act 83 of 1993 (as amended) states that '[n]o person shall advertise or promote, or cause any other person to advertise or promote, a tobacco product through any direct or indirect means, including through sponsorship of any organisation, event, service, physical establishment, programme, project, bursary, scholarship or any other method'.
[199] Regulation 21(1)(*e*)(*i*) of the Medical Device Regulations.
[200] In terms of section 5 of the CPA.
[201] In terms of section 47 of ECTA.

Offering direct-to-consumer genetic tests to the public in South Africa is a complex issue that involves several, overlapping statutes and legal instruments and contains many requirements with which testing companies must comply. Prior to being able to advertise and market, and thereby sell, direct-to-consumer genetic tests in South Africa, testing companies must follow the licensing and registration requirements set out in the Medicines Act, the Medical Device Regulations, and SAHPRA's guidance documents. Thereafter, the CPA and ECTA predominantly govern requirements pertaining to the advertising, marketing, and labelling of direct-to-consumer genetic tests as well as requirements for disclosure, understandable information, and fair terms and conditions. This aims to protect consumers from harm and prevent testing companies from misleading consumers in an attempt to induce sales.

CHAPTER 6

THE IMPORTING AND EXPORTING OF DIRECT-TO-CONSUMER GENETIC TESTS

I INTRODUCTION AND CHAPTER OVERVIEW

The importing and exporting associated with direct-to-consumer genetic testing is not straightforward as different stages of the process may occur in different jurisdictions, and it does not follow the typical methods used for importing and exporting samples and data into and out of South Africa as consumers usually purchase tests privately and export the saliva sample themselves through the post.[1] I have identified five primary issues associated with the importation and exportation of direct-to-consumer genetic tests: (1) importing the testing kit itself, where the testing company is based in another jurisdiction; (2) exporting saliva samples by consumers out of South Africa to the testing company in another jurisdiction (where relevant), exporting saliva samples by testing companies in South Africa for processing in overseas laboratories; (3) exporting extracted DNA by testing companies in South Africa for analysis in overseas laboratories; (4) importing genetic data and test results from overseas, where the testing company is based in South Africa, but samples are sent elsewhere for analysis; and (5) exporting genetic data out of South Africa, to be used for research.

To address these issues, I establish the legal landscape related to the importing and exporting of direct-to-consumer genetic tests as well as its associated samples and data, through an examination of the five abovementioned issues, to determine the legal status of such activities in South Africa. This is done by analysing the NHA, POPIA, the Medicines Act, the Import and Export Regulations, and the Medical Device Regulations.

It should be mentioned at the outset that the topic of exportation (of samples and data) is broad. Thus, the relevant legal instruments are discussed briefly, but are not analysed in depth due to the focus of this dissertation. They are touched on insofar as they may be relevant to direct-to-consumer genetic testing, but require further, comprehensive analysis to do them justice. My focus in this Chapter is on the non-research aspects of direct-to-consumer genetic

[1] However, this may change with the involvement of healthcare practitioners in the process. In such instances, healthcare practitioners will order a test on the consumer's behalf and facilitate the collection of the saliva sample. Thereafter, the healthcare practitioner sends the sample to the testing company who will either analyse the sample themselves or send it to an overseas laboratory for analysis.

testing (although research is briefly mentioned where relevant). Cases where there is a possibility of research are dealt with in Chapter 7.[2]

II IMPORTING THE TESTING KIT INTO SOUTH AFRICA

The rules regarding importation only apply where the consumer is in South Africa, but the testing company is based in another jurisdiction – it neither applies to situations where both the consumer and the testing company are in South Africa, nor where a South African testing company creates and develops its own tests locally.

While the importing of the testing kit is relevant, the exporting of such kits does not require in depth examination. This is because, firstly, South African testing companies are not as established as those operating overseas, and it is therefore unlikely that they will export testing kits out of South Africa to consumers who have ordered them in other jurisdictions.[3] Secondly, when consumers who import direct-to-consumer genetic testing kits send them back to the testing company, they are not exporting the kit,[4] but rather the saliva sample, which I deal with below.

(a) Non-medical devices

Direct-to-consumer genetic tests that do not meet the definition of 'medical device' in the Medicines Act[5] – and are thus not IVDs – follow a different importation path to direct-to-consumer genetic tests that are medical devices. Such testing kits are treated the same way as most other consumer goods purchased from abroad – they can simply be bought by consumers

[2] Therefore, mechanisms such as the SA MTA are not relevant to importing and exporting where no research is involved.

[3] There are international testing companies that have offices in South Africa and distribute their products globally, but it is the testing kits that is imported into South Africa and then offered to consumers locally. Examples of such testing companies are HomeDNADirect, EasyDNA, and DNAlysis. HomeDNADirect 'About us' available at *https://www.homednadirect.co.za/about-us/*, accessed on 22 June 2020; EasyDNA 'About us' available at *https://www.easydna.co.za/about-us/*, accessed on 22 June 2020; DNAlysis 'About' available at *https://dnalysis.co.za/about/*, accessed on 22 June 2020.

[4] Although the same box that the testing kit came in is generally used when returning the saliva sample to the testing company (as is the case with 23andMe in the US and GeneWay in South Africa), other testing companies (such as MyHeritage) provide consumers with a separate padded envelope into which they place their vials containing the saliva samples. However, these boxes are merely used for the purposes of protecting the samples during transport, and the important focus is rather on the exportation of the saliva sample. For more information, see 23andMe 'Returning DNA sample to the lab' available at *https://customercare.23andme.com/hc/en-us/articles/202904570-Returning-DNA-Sample-to-the-Lab*, accessed on 25 June 2020; GeneWay 'I want to get tested, how does it work?' available at *https://geneway.co.za/i-want-get-tested-how-does-it-work/*, accessed on 25 June 2020; MyHeritage 'How do I collect and send back my DNA sample to the lab?' available at *https://faq.myheritage.com/en/article/how-do-i-collect-and-send-back-my-dna-sample-to-the-lab*, accessed on 25 June 2020.

[5] Section 1 of the Medicines Act.

and sent by testing companies into South Africa without the need for licenses or other documentation. The South African Revenue Service (SARS) confirms that Customs can clear the importing of personal goods and, in most instances, individuals are not required to register as importers and obtain importer codes.[6]

(b) Medical devices

Although the importing of direct-to-consumer genetic testing kits that are not medical devices by consumers is simple, direct-to-consumer genetic tests that are classified as Class C IVD medical devices due to the potential harm that they pose, come with certain importation requirements – which I analyse in the paragraphs that follow.

Importing or exporting medical devices or IVDs into or out of South Africa requires an import or export license respectively. As mentioned in Chapter 5, only those who are licensed in terms of section 22C(1)(*b*) of the Medicines Act may import medical devices or IVDs into South Africa.[7] While the Medical Device Regulations contain a provision on the importation of medical devices and IVDs into the country, there is no similar provision regarding exportation. The Medicines Act refers to 'a medical device or IVD establishment, manufacturer, wholesaler or distributor'[8] and, as consumers are generally individual laypersons who rather use direct-to-consumer genetic tests for their own purposes, it is unlikely that they will be holders of licenses and will thus not be authorised to import direct-to-consumer genetic tests themselves. Where direct-to-consumer genetic tests are not developed in South Africa by a testing company's own laboratory, the solution is for testing companies to apply for licenses to import direct-to-consumer genetic tests and allow consumers to purchase the tests locally from them.

[6] However, this only applies where: (1) only three transactions (which are less than R50 000 each) occur in a calendar year; (2) goods are imported for personal use, not for resale or business purposes; (3) the individual is in South Africa; and (4) the individual's identity number, passport number, or taxpayer reference number is entered in the declaration form. If purchases do not meet these requirements, individuals must register and obtain an importer code. South African Revenue Service 'FAQ: Do I need to register as an importer if I buy personal goods from abroad e.g. from Amazon?' 9 March 2021 available at *https://www.sars.gov.za/faq/faq-do-i-need-to-register-as-an-importer-if-i-buy-personal-goods-from-abroad-e-g-from-amazon/*, accessed on 7 May 2021.

[7] Regulation 3(3)(*a*) of the Medical Device Regulations. Regulation 5 of the Medical Device Regulations deals with licenses to import, export, manufacture, distribute, or wholesale medical devices or IVDs. Regulation 3(1) of the Medical Device Regulations stipulates that importing medical devices into South Africa must be done via certain ports of entry. These ports of entry are Cape Town International Airport or harbour, Port Elizabeth Airport or harbour, King Shaka International Airport or Durban harbour, or OR Tambo International Airport.

[8] Section 22C(1)(*b*) of the Medicines Act.

SAHPRA's *Guideline for a Licence to Manufacture, Import, Export or Distribute Medical Devices & IVDs* (Licensing Guidelines),[9] prohibit any person from ordering or importing Class B, Class C, or Class D medical devices or IVDs that are unregistered in South Africa for personal use, unless SAHPRA has granted authorisation.[10] As I suggest that direct-to-consumer genetic tests are generally classified as Class C IVD medical devices, this provision affects them. The meaning of 'personal use' is unclear, but it appears that if a medical device or IVD is registered in South Africa, then it can be ordered by any person for personal use. However, SAHPRA's registration process is currently in development, meaning that medical devices or IVDs are not registered in South Africa, but rather registered elsewhere and approved for use in South Africa.[11] Given this, the licensing requirements must be relied on – meaning that an import license for medical devices or IVDs, as per the Medicines Act, must be obtained. As such a license is not issued to consumers,[12] it must instead be acquired by a testing company in South Africa (if they are acquiring testing kits from manufacturers overseas).

On the other hand, consumers who order testing kits from South African testing companies that ship their products within the country do not need an import permit or license. This is because the transaction occurs within South Africa and does not transcend its borders.[13] However, direct-to-consumer genetic tests that are classified as Class C IVD medical devices must nevertheless be licensed with SAHPRA.

To conclude: Individual consumers wanting to import direct-to-consumer genetic tests from a testing company in another jurisdiction are prohibited from doing so in terms of South Africa's legislation governing the importation of medical devices and IVDs. As consumers do not qualify to apply for a license under the Medicines Act,[14] they are prohibited from importing direct-to-consumer genetic tests into South Africa themselves. This seems to conflict with SAHPRA's Licensing Guidelines, which allow Class C IVD medical devices to be imported into South Africa for *personal use* if registered. However: (1) SAHPRA's registration process

[9] South African Health Products Regulatory Authority (SAHPRA) *Guideline for a Licence to Manufacture, Import, Export or Distribute Medical Devices & IVDs* (2019).
[10] Ibid at 5; T Saidi & TS Douglas 'Medical device regulation in South Africa: The Medicines and Related Substances Amendment Act 14 of 2015' (2018) 108(3) *S Afr Med J* 169.
[11] Catherine Tomlinson 'IN-DEPTH: The tangled web of medical device regulation in SA' *Spotlight* 3 September 2020, available at *https://www.spotlightnsp.co.za/2020/09/03/in-depth-the-tangled-web-of-medical-device-regulation-in-sa/*, accessed on 5 December 2020.
[12] Such licenses are only issued to a 'medical device or IVD establishment, manufacturer, wholesaler or distributor'. Section 22C(1)(*b*) of the Medicines Act.
[13] Two South African testing companies that operate in this manner are GeneWay and KnowU. GeneWay 'How it works' available at *https://geneway.co.za/i-want-get-tested-how-does-it-work*, accessed on 21 November 2020; KnowU 'About us' available at *https://knowudna.com/about-us/#labs*, accessed on 21 November 2020.
[14] Section 22C(1)(*b*) of the Medicines Act.

remains in development; and (2) SAHPRA's guidelines are only binding on those with obligations to SAHPRA.[15] Therefore, the Medicines Act is authoritative and thereby prevents consumers from importing direct-to-consumer genetic tests (as Class C IVD medical devices) into South Africa.

III EXPORTING SALIVA SAMPLES OUT OF SOUTH AFRICA

Once saliva samples are collected by consumers, these consumers send the sample back to the testing company.[16] It must be mentioned that while some testing companies process and analyse the saliva samples of consumers in their own laboratories, others outsource their testing services to an external laboratory[17] which, for the purposes of this dissertation, may either be based in South Africa or abroad,[18] making the exportation of these samples relevant. If the testing company is based in South Africa, the sample remains within the country; if they are based overseas, the saliva sample is exported by consumers. But if the testing company is based in South Africa and makes use of overseas laboratories for DNA extraction, sequencing, and analysis, the saliva sample is sent, by consumers, to the testing company in South Africa, who is then responsible for exporting the saliva sample overseas.

This section applies to the exporting of saliva samples: (1) by testing companies based in South Africa that send saliva samples to an overseas laboratory for DNA extraction, sequencing, and analysis; or (2) by consumers out of South Africa to a testing company in another jurisdiction. This appears to be something that has not yet been considered in depth. Most of the existing research and legal instruments apply to the exportation of tissue and other biological samples (usually in bulk) for research purposes by institutions or groups. It is unclear

[15] Section 35(1) of the Medicines Act permits the Minister of Health, together with SAHPRA, to issue regulations. In the cases of both *Allergan Pharmaceuticals (Pty) Ltd v Medicines Control Council* [2015] 3 All SA 173 (GP) (*Allergan*) and *Gelderma Laboratories South Africa (Pty) Ltd v Medicines Control Council* [2014] ZAGPPHC 360 (*Gelderma*), prior to the existence of the Medical Device Regulations, the courts held that without regulations being promulgated, the then MCC were not authorised to manage inter alia the registering, manufacturing, importing, exporting, selling, or using medical devices in terms of their safety, quality, and efficacy (*Allergan* para 54; *Gelderma* para 32). Conversely, in *Omegalabs (Pty) Ltd v Medicines Control Council* [2016] ZAGPPHC 1157 para 31 (*Omegalabs*), the court held that as the Medicines Act grants the MCC the power to license the manufacture, import, export, distribution, and wholesale of medical devices, it also has the power and ability to classify and register medical devices without first requiring the Minister of Health to promulgate regulations. However, these cases occurred in the context of no regulations. Now that the Medical Device Regulations have been promulgated, it seems that guidance documents by SAHPRA are not binding. See also, Beverley A Townsend 'Software as a medical device: Critical rights issues regarding artificial intelligence software-based health technologies in South Africa' (2020) 4 *TSAR* 751–2.

[16] This may also be done by healthcare professionals, where relevant.

[17] G Lippi, EJ Favaloro & M Plebani 'Direct-to-consumer testing: More risks than opportunities' (2011) 65(12) *Int J Clin Pract* 1222.

[18] This outsourcing could either be for both the processing and analysis of the saliva samples and extracted DNA, or it could just be for the analysis of DNA taken from already processed (extracted and sequenced) saliva samples.

what the legal position is where individuals personally export their saliva samples for the purpose of a direct-to-consumer genetic test.

To clarify this, individuals are not excluded from obtaining export permits as the Import and Export Regulations refer to both a 'person'[19] and an 'applicant'[20] as those who may apply for such permits. Moreover, an 'applicant' is defined in the Import and Export Regulations as including a '*person* applying for an export or import permit' (own emphasis).[21] This means that, provided an individual has obtained an export permit, they are legally allowed to export their saliva sample to a testing company in another jurisdiction.

Neither the Medicines Act, the Medical Device Regulations, nor SAHPRA's guidelines apply in this instance as they are relevant to the importing and exporting of medical devices and IVDs, and not biological samples. The Import and Export Regulations govern the importing and exporting of inter alia tissue into, and out of, South Africa.[22] Given that Chapter 2 established that saliva is a type of 'tissue' as per the NHA,[23] the Import and Export Regulations are applicable to the exporting of saliva samples as part of a direct-to-consumer genetic test. The importation or exportation of tissue requires a permit,[24] granted by the Director-General of the DoH following the submission of an application that satisfies certain requirements.[25] Moreover, the following criteria must be met when seeking a permit to export tissue:

(1) The application must contain written proof that the tissue sought to be exported was donated and, once exported, will be used for the purpose for which it was removed, in terms of the NHA;[26]

(2) The tissue exported from South Africa must accord with the laws and regulations of the country that it is being imported into;[27]

[19] Regulations 2(1), (2), and (3) of the Import and Export Regulations refer to a 'person'.
[20] Regulations 2(4) and 3(1) of the Import and Export Regulations refers to an 'applicant'.
[21] Regulation 1 of the Import and Export Regulations.
[22] See, C Staunton & K Moodley 'Data mining and biological sample exportation from South Africa: A new wave of bioexploitation under the guise of clinical care?' (2016) 106(2) *SAMJ* 136–7; Department of Health *South Africa Frequently Asked Questions (FAQs) Import/Export Permit Programme* (2015) 2.
[23] Section 1 of the NHA.
[24] Regulation 2(1) of the Import and Export Regulations. According to regulation 2(2) of the Import and Export Regulations, an import or export permit for inter alia tissue requires a written application to the Director-General of the DoH.
[25] Regulations 2 (3) and (4) of the Import and Export Regulations.
[26] Regulation 3(1) of the Import and Export Regulations.
[27] Regulation 4(10)(*c*) of the Import and Export Regulations.

(3) A permit for the export of tissue may only be to a member state of the Southern African Development Community (SADC);[28] and

(4) Where a permit is issued, transportation must comply with the relevant biological standards.[29]

To expand on (1) above, samples cannot be exported unless they have been acquired in line with the NHA (consent was granted prior to removal), and will be used according to the terms of the NHA.[30] To ensure some form of oversight, a register must be kept of all samples exported out of South Africa.[31] As consumers generally collect their own saliva samples and post them back to the testing company themselves (where the testing company is based overseas), it is unlikely that such oversight will be possible.[32]

All saliva samples exported out of South Africa require export permits. This includes testing companies based in South Africa that export saliva samples to overseas laboratories for DNA extraction, sequencing, and analysis,[33] as well as consumers. However, if each consumer is required to apply for, and obtain, a permit to export their saliva sample to overseas testing companies, this will increase the workload of the Director-General who assesses applications and grants permits and, with an application for each individual, is likely to create a backlog. Consumers may also be dissuaded by going through a laborious process just for a direct-to-consumer genetic test, rather opting not to undergo them. This, and the register requirement above, are impractical, constitute an infringement of freedom, and represent an area that is over-regulated, and unnecessarily so.

[28] Regulation 4(10)(*a*) of the Import and Export Regulations. The SADC is an inter-governmental organisation aimed at economic development. It consists of 16 Member States: Angola, Botswana, Comoros, Democratic Republic of Congo, Lesotho, Madagascar, Malawi, Mauritius, Mozambique, Namibia, Seychelles, South Africa, eSwatini, Tanzania, Zambia, and Zimbabwe. Southern African Development Community 'Member States' available at *https://www.sadc.int/member-states/*, accessed on 18 January 2021.

[29] Regulation 4(7) of the Import and Export Regulations. In regulation 1 of the Import and Export Regulations, 'biological standards' are defined as 'norms or guidelines used to ensure the preservation of biological substances of human origin for the purpose which these substances are intended to be used'.

[30] Staunton & Moodley op cit note 22 at 137.

[31] Ibid.

[32] It is difficult to know how this would be monitored, and keeping track of it would be almost impossible.

[33] In this case, consumers post their saliva samples to the testing company within the country and do not require export permits. However, the company does. It is unclear if the testing company would wait for several saliva samples before sending them off in bulk or whether they would send one at a time as they come. It is important to mention that the provisions relating to exporting do not only apply to research and therefore even those South African testing companies that gather and export saliva samples for the purpose of DNA extraction, sequencing, and analysis in order to provide test results to consumers and do not undertake research, are still required to obtain export permits and the same provisions apply.

IV EXPORTING EXTRACTED DNA OUT OF SOUTH AFRICA

A tissue permit is needed to export saliva samples to another jurisdiction, but what about DNA, which is not 'tissue' as per the NHA?[34] Sometimes, South African testing companies may extract DNA from saliva samples and then send the extracted, but unsequenced, DNA to an overseas laboratory for analysis. This is because whilst the DNA extraction stage (illustrated in Diagram 1 in Chapter 2) is simple, DNA sequencing is an automated and more costly process that uses a different machine. Once DNA has been extracted from saliva, it is no longer tissue and is instead a type of biological material.

The title of the Import and Export Regulations mention 'human tissue'; no reference is made to biological material.[35] The Import and Export Regulations do not contain a definition of 'tissue' and therefore, as it forms part of the NHA's subsidiary legislation, the definition utilised in the NHA must be employed. As established in Chapter 2, the definition of 'tissue' in the NHA excludes sub-cellular components, such as extracted DNA. On a plain reading, regulation 2(1) of the Import and Export Regulations limit the scope of application to include tissue, but not biological material such as DNA – as a permit is required for importing and exporting 'any tissue or any blood, blood product, cultured cells, gametes, stem cells or embryos'[36] – DNA is not included in this list.[37] Given this, and the fact that the definition of 'tissue' in the NHA excludes extracted DNA, it is not covered by the Import and Export Regulations and extracted DNA need not comply with its provisions relating to permits.[38]

There are also testing companies based in South Africa that operate entirely within the country.[39] In such instances, no tissue, biological material, or genetic data leaves or enters South Africa. Therefore, the laws relating to importing or exporting do not apply, but such testing companies must ensure compliance with other legislation, such as the NHA, in terms of removal and use of tissue, and POPIA, in terms of processing personal information.

---

[34] Section 1 of the NHA.
[35] The Import and Export Regulations are entitled 'Regulations relating to the Import and Export of *Human Tissue*, Blood, Blood Products, Cultured Cells, Stem Cells, Embryos, Foetal Tissue, Zygotes and Gametes' (own emphasis).
[36] Regulation 2(1) of the Import and Export Regulations.
[37] Regulation 1 of the Import and Export Regulations contain a definition of 'DNA' (which is identical to the definition in the Human Biological Material Regulations), but the term is not used in the Import and Export Regulations.
[38] Thus, only saliva (as a type of tissue) is subject to the Import Export Regulations; DNA and other biological material is excluded.
[39] Two examples of such testing companies are GeneWay and KnowU. GeneWay 'Home' available at *https://geneway.co.za/*, accessed on 26 July 2020; KnowU 'Home' available at *https://knowudna.com/*, accessed on 26 July 2020.

V IMPORTING GENETIC DATA INTO, AND EXPORTING GENETIC DATA OUT OF, SOUTH AFRICA

In this section, importation entails the importing of genetic data from overseas laboratories, where the testing company is based in South Africa, but the sample is sent elsewhere for DNA extraction, sequencing, and analysis – with the genetic data thereafter being sent back to the testing company that relays test results to consumers and conducts research in South Africa. Exportation consists of the exporting of genetic data gathered from direct-to-consumer genetic testing out of South Africa, to be used for research.[40] Therefore, it is important to briefly touch on the legal rules relevant to the importation and exportation of data, specifically genetic data.[41]

Once an overseas laboratory has undertaken DNA extraction, sequencing, and analysis (or just DNA sequencing and analysis in some instances), it is likely to send the genetic data back to the testing company in South Africa to enable them to generate test results,[42] and to undertake research, if they do so. This means that genetic data is transported between countries, coming from overseas into South Africa.[43] The Import and Export Regulations do not refer to the importation or exportation of data. Therefore, answers must be sought from other South African legislation. Given that POPIA covers transfers of personal information from South Africa to another country and, as genetic data falls within the ambit of POPIA, it becomes relevant. It must be noted that POPIA is not applicable to genetic data coming into South Africa. Thus, the ensuing discussion is only relevant to testing companies that do the extraction and sequencing in South Africa and then send the genetic data abroad for analysis, or South African testing companies that also conduct the analysis, but send the results abroad for research.

[40] Testing companies, like EasyDNA and the Wellness Revolution, appear to operate and distribute their products internationally, albeit under different names. In such a case, if these companies conduct research, it is likely that the genetic data from consumers worldwide would be sent to one laboratory, or the saliva samples or DNA would be sent to the laboratory where they would be analysed. EasyDNA op cit note 3; The Wellness Revolution 'How it works' available at *https://thewellnessrevolution.co.za/how-it-works/*, accessed on 22 June 2020.

[41] According to Staunton and Moodley, data (unlike samples) are not physically transported and are thus excluded from the export permit system. But as a sample may remain in South Africa and its related data may be exported, oversight on access to data and its transfer is necessary. Staunton & Moodley op cit note 22 at 138.

[42] It is also possible that some testing companies, especially those that distribute their tests internationally would have the saliva samples extracted, sequenced, and analysed in a laboratory overseas, that would then send the test results, based on the genetic data, back to the testing company in South Africa.

[43] It is unclear what would happen to the saliva samples – whether it would be the responsibility of the testing company to destroy them (in which case the samples would need to be sent back to South Africa), or whether this would be done by the laboratory overseas.

Prior to POPIA, the sharing of genetic data was unregulated.[44] POPIA limits data sharing within, as well as beyond, South Africa. Section 72 of POPIA controls the transfer of data,[45] by providing that, in order for personal information to be transferred to another country, a legal ground must exist,[46] thereby prohibiting the transfer of personal information outside of South Africa, unless: (1) the recipient in another jurisdiction is subject to a law, binding corporate rules, or a contract (between the recipient and the data subject) that provides an 'adequate level of protection' for, and contains provisions 'substantially similar' to, POPIA;[47] (2) consent is obtained from the data subject; (3) the transfer is needed for contractual performance between the responsible party and the data subject; (4) the transfer is required for the conclusion or performance of a contract between the responsible party and a third party in the interests of the data subject; or (5) the transfer benefits the data subject and, although not 'reasonably practicable' to obtain their consent, the data subject would likely have consented.[48] As consumers consent to undergo a direct-to-consumer genetic test – which often includes consenting to the processing and analysis of their saliva samples to obtain test results and for possible research, which may be done in other jurisdictions – this seems to satisfy the provision in POPIA relating to consent.[49] This is also the case in terms of contractual performance between testing companies and consumers because, without the analysis of DNA obtained from saliva samples in a laboratory (which may be located in another jurisdiction), consumers cannot obtain their test results and testing companies cannot fulfil their obligations in terms of the agreement between the parties.[50]

---

[44] Donrich W Thaldar, Marietjie Botes & Annelize Nienaber 'South Africa's new standard material transfer agreement: Proposals for improvement and pointers for implementation' (2020) 21(85) *BMC Med Ethics* 6.

[45] POPIA does this and thus protects personal information, by specifying conditions that a responsible party must comply with to transfer personal information overseas. Section 72 of POPIA.

[46] Section 72 of POPIA. See also, Beverley Townsend 'The lawful sharing of health research data in South Africa and beyond' (2021) *Information & Communications Technology Law* 8.

[47] Section 72(1)(*a*) of POPIA. This may be done through a data transfer agreement (DTA). Data protection frameworks often restrict cross-border data transfers. But POPIA's protection stays with the data, requiring the recipient country to have data laws and protections, often in a binding agreement, which are, at a minimum, 'substantially similar' to POPIA's protections. Townsend op cit note 46 at 8 & 17.

[48] Section 72(1) of POPIA.

[49] Section 72(1)(*b*) of POPIA. Although saliva and DNA do not fall within POPIA's ambit, by agreeing to undergo direct-to-consumer genetic testing, depending on the company's location and processes, consumers consent to their saliva sample (or extracted DNA) being exported out of South Africa to the company's laboratory for DNA extraction (where relevant), sequencing, and analysis. Staunton and Moodley recognise that if unaware of their sample being exported, individuals may assume that it will be controlled in accordance with South African law. Thus, individuals should be informed if their data will be exported and also possibly used for research purposes. DNAlysis and Muhdo (The Wellness Revolution) are two South Africa based testing companies that specifically mention that their laboratories are based overseas. DNAlysis states that their products and services are distributed by Nordic Laboratories in Denmark and Muhdo analyses the DNA sample at a laboratory in Europe. Staunton & Moodley op cit note 22 at 137; DNAlysis op cit note 3; The Wellness Revolution op cit note 40.

[50] Section 72(1)(*c*) of POPIA.

Where 'special personal information' is transferred to another country that does not offer adequate protection, POPIA contains a further requirement.[51] To transfer 'special personal information', namely genetic data in this instance,[52] from South Africa to another country that does not offer a suitable level of protection, prior authorisation must be obtained from the Information Regulator.[53] But if a code of conduct for the relevant sector has been accepted by the Information Regulator, no prior authorisation is necessary.[54] A code of conduct seeks to apply the provisions of POPIA to a specific sector, promoting adherence thereto. According to POPIA, a code of conduct must –

> '(a) incorporate all the conditions for the lawful processing of personal information or set out obligations that provide a functional equivalent of all the obligations set out in those conditions; and
>
> (b) prescribe how the conditions for the lawful processing of personal information are to be applied, or are to be complied with, given the particular features of the sector or sectors of society in which the relevant responsible parties are operating'.[55]

Currently, there is no code of conduct that has been approved for the genetic testing industry or for genetic research, although a Code of Conduct for research is currently being drafted by ASSAf.[56] This may provide a potential pathway for the cross-border transfer of genetic data. But this applies to personal information leaving South Africa and not data coming into the country from elsewhere. POPIA neither defines which countries are considered to have 'an adequate level of protection',[57] nor does it specify how these countries will be determined.[58] Similar to the 'White List' published by Malaysia's Personal Data Protection Commissioner

---

[51] Townsend op cit note 46 at 9.

[52] Special personal information is dealt with in section 26(a) of POPIA, and encompasses personal information regarding a data subject's health and biometric information. As genetic data is a form of biometric information, genetic data falls under special personal information in POPIA.

[53] Section 57(1)(*d*) of POPIA. Section 57 of POPIA deals with situations wherein prior authorisation is required for the processing of personal information. In terms of section 57(4) of POPIA, this prior authorisation only needs to be obtained once, rather than every time personal information is processed, unless the processing differs from what has been permitted.

[54] Section 57(3) of POPIA. Thaldar et al op cit note 44 at 5. See also, Townsend op cit note 46 at 9.

[55] Section 60(2) of POPIA.

[56] Academy of Science of South Africa 'POPIA: A code of conduct for research' available at *https://www.assaf.org.za/index.php/2-uncategorised/798-popia-a-code-of-conduct-for-research*, accessed on 6 May 2021. See also, Rachel Adams, Fola Adeleke, Dominique Anderson et al 'POPIA Code of Conduct for research' (2021) 117(5/6) *S Afr J Sci* 1–12; Rachel Adams, Susan Veldsman, Michèle Ramsay et al 'Drafting a Code of Conduct for research under the Protection of Personal Information Act No. 4 of 2013' (2021) 117(5/6) *S Afr J Sci* 1–3.

[57] Section 72(1)(*a*) of POPIA.

[58] Noor Kapdi, Shahid Sulaiman & Daniél Hofmeyr 'South Africa: Transferring personal information across borders' *Mondaq* 2 February 2016, available at *https://www.mondaq.com/southafrica/data-protection/462800/transferring-personal-information-across-borders*, accessed on 4 July 2020.

(PDPC) of jurisdictions deemed adequate for personal information to be transferred to[59] – and as recommended by Botes et al[60] – I suggest that it may assist, and provide clarity, for the Information Regulator to create a similar list for South Africa. This would allow for efficient and lawful cross-border data sharing.[61]

While POPIA covers the sending of genetic data out of South Africa, the position where genetic data comes back into the country depends on the laws of the country from where it is sent[62] – making the legal position where genetic data is sent from a foreign country to South Africa, as in the present case with direct-to-consumer genetic testing, less clear.

---

[59] The PDPC published *Personal Data Protection (Transfer of Personal Data to Places Outside Malaysia) Order* Public Consultation Paper No. 1/2017 (2017). The White List will allow controllers to transfer personal information to approved countries without reliance on exemptions. Mark Parsons & Louise Crawford 'Malaysia publishes draft "White List" for personal data exports' *Hogan Lovells* 27 April 2017, available at *https://www.hoganlovells.com/en/publications/malaysia-publishes-draft-white-list-for-personal-data-exports*, accessed on 6 May 2021; Graham Greenleaf '2014-2017 update to Graham Greenleaf's *Asia Data Privacy Laws – Trade and Human Rights Perspectives*' (2017) 47 *UNSWLRS* 27. See also, M Botes, A Olckers & M Labuschaigne 'Data commercialisation in the South African health care context' (2021) 24 *PER/PELJ* 25.

[60] Botes et al op cit note 59 at 25.

[61] Ibid.

[62] For example, the EU – and specifically the GDPR – requires that when transferring personal information from the EU to other countries, the level of protection that the EU provides should not be diminished. From the perspective of the EU, South Africa is not seen to provide such an adequate level of protection – the country is neither on the European Commission's adequacy list, nor does there exist a Safe Harbour Agreement or a Privacy Shield Framework between the EU and South Africa. The Safe Harbour Agreement consisted of principles governing data transfers between the US and the EU. It applied to various organisations and businesses that collected personal data. In 2015, the Court of Justice of the European Union (CJEU) declared the Safe Harbour Agreement invalid due to its determination of the inadequacy of US data protection laws in *Schrems v Data Protection Commissioner* (C-362/14) (*Schrems I*). In this case, Schrems challenged the transfer of data by Facebook from the EU to the US, and whether the Safe Harbour Agreement and Standard Contractual Clauses (SCCs) are adequate to protect personal data when it enters and/or leaves the EU. The invalidation of the Safe Harbour Agreement meant that US companies needed 'model contract clauses' for each data transfer. However, the invalidation of the Safe Harbour Agreement resulted in the EU-US Privacy Shield. This permitted the transfer of data from the EU to the US while adhering to the CJEU's requirements. But in 2020, the Privacy Shield was also declared invalid by the CJEU in *Data Protection Commissioner v Facebook Ireland and Maximillian Schrems* (C-311/18) (*Schrems II*). The CJEU found that US data laws failed to protect personal data of EU citizens, were inadequate in terms of the standards required in EU law, and contravened the GDPR. *Schrems II* confirmed that SCCs are valid, but that those importing or exporting data must confirm that the laws of the recipient country offer adequate protection. Where this is not the case, further safeguards must be specified and included in the transfer agreement to ensure that individuals are protected. Therefore, in order to transfer personal information from the EU into South Africa, the EU processor must offer 'appropriate safeguards' confirming that rights and legal remedies exist for EU data subjects to ensure that 'adequate' protection is offered in South Africa. These 'safeguards' may be done through standard data protection clauses accepted by the European Commission. *Schrems II* makes it necessary to consider two additional aspects when deciding on the existence of suitable safeguards and legal remedies as well as enforceable rights: (1) the SCCs agreed between the EU processor and the recipient of the transfer in South Africa; and (2) whether South African public authorities have access to the transferred personal data, as well as further features of South Africa's legal system which could have an impact on these rights. Therefore, in addition to the data protection offered by POPIA, there must also exist enforcement measures and judicial solutions. On the other hand, to enable data to be transferred into the country, South Africa must ensure that it meets an adequate level of protection. This requires the existence of an applicable legal framework, as well as its successful implementation. As US laws do not protect EU privacy rights, and given that POPIA bears similarities to the GDPR, Botes et al state that it is unlikely for US laws to offer the adequate level of protection required by POPIA. Townsend op cit note 46 at 9 & 17–8; Experian 'What is the Safe Harbour Agreement?' available at *https://www.experian.co.uk/business/glossary/safe-harbour-agreement/*, accessed on 13

In summary, POPIA allows genetic data to be sent from South Africa to another country subject to various safeguards. These include consent from the data subject, or an adequate level of protection offered by the laws in the country where the genetic data is being sent. Genetic data may also be transferred from another country to South Africa, but POPIA does not apply, and this depends on the data protection laws of that country and its provisions relating to transborder flows of information.

VI DISCUSSION

The above analysis shows that direct-to-consumer genetic testing may follow different routes in terms of importation and exportation of either testing kits or samples and data. Below, through various scenarios, I illustrate who is authorised to import or export, the things that may be imported or exported, and the applicable South African legislation to be complied with.

(a) Scenario (1)

The testing company is based in South Africa and manufactures its own tests in South Africa, and the sending of saliva samples by consumers, as well as DNA extraction, sequencing, and analysis occur in South Africa. In this scenario, all the things involved in the stages of the direct-to-consumer genetic testing process – namely, saliva, DNA, and genetic data – remain within the country. Therefore, as the testing company manufactures and provides its own tests locally, it does not need to obtain a license, in terms of the Medicines Act and the Medical Device Regulations, to import direct-to-consumer genetic testing kits.[63] Furthermore, the testing company and consumers do not need to apply for a permit, as per the Import and Export Regulations, to export saliva samples abroad.

(b) Scenario (2)

The testing company is based abroad, where saliva samples are sent by consumers and where the processing of samples occurs overseas. The testing kit can only be imported into South

May 2021; Martin A Weiss & Kristin Archick 'U.S.-EU data privacy: From Safe Harbor to Privacy Shield' (2016) *Congressional Research Service* 1; Sameul Gibbs 'What is "safe harbour" and why did the EUCJ just declare it invalid?' *The Guardian* 6 October 2015, available at *https://www.theguardian.com/technology/2015/oct/06/safe-harbour-european-court-declare-invalid-data-protection*, accessed 13 May 2021; Noah Ramirez 'The EU-US Privacy Shield invalidated: What this means for you' *Osano* 1 June 2021, available at *https://www.osano.com/articles/privacy-shield-invalidated*, accessed on 11 May 2021; Adams et al op cit note 56 at 10; Avani Singh 'Data without borders: How to manage cross-border data transfers in South Africa' *Alt Advisory* 7 November 2017, available at *https://altadvisory.africa/2017/11/07/data-without-borders-manage-cross-border-data-transfers-south-africa/*, accessed on 4 July 2020; Botes et al op cit note 59 at 18–20.

[63] But they would need a license to manufacture, distribute, and sell such tests. This is dealt with in Chapter 5.

Africa by those who hold a license in terms of the Medicines Act, which excludes consumers. Testing companies based in other jurisdictions can import their testing kits into South Africa provided that they have a license from SAHPRA, they comply with the prescribed requirements for the license application, and there is an Authorised Representative in South Africa to oversee the process and conformity to legislation.[64] Consumers are permitted to export their saliva samples, in line with the Import and Export Regulations, provided that they have an export permit. In terms of South African law, the overseas testing company does not need any import or export permits as consumers export their saliva samples to them. The testing company then extracts, sequences, and analyses the DNA in their laboratories, stores the genetic data and other information in databases overseas, and sends consumers their test results online (usually via email or on a mobile app). Although the direct-to-consumer genetic test results contain information about a consumer's genes based on an analysis of their genetic data, which falls under the definition of 'personal information' in POPIA (established in Chapter 2),[65] POPIA is not relevant as the foreign testing company is not domiciled in South Africa. As the DNA is extracted, sequenced, and analysed outside of South Africa, local laws do not apply, and it is instead the laws of that particular jurisdiction which govern these things that is applicable.

(c) Scenario (3)

The testing company is based in South Africa, but saliva samples are sent overseas, and DNA extraction, sequencing, and analysis occur in a laboratory in another jurisdiction. Obtaining the testing kit is not an issue as the testing company is based in South Africa and can send its kits locally without the need for an import license. Moreover, the sending of saliva samples by consumers to the testing company is straightforward as it occurs within South Africa, thus consumers are not subject to any import or export laws – however the testing company is. Given that once the testing company receives saliva samples from consumers, they are sent to a laboratory in another jurisdiction for DNA extraction, sequencing, and analysis, the testing company must comply with certain legal requirements. Firstly, the testing company must obtain an export permit to send saliva samples overseas, in line with the Import and Export Regulations.[66] As 'tissue' is not covered by POPIA and given that saliva samples are exported before genetic data is obtained, POPIA does not apply. Secondly, once saliva has been

[64] Regulation 1 of the Medical Device Regulations.
[65] Section 1 of POPIA.
[66] This is also because saliva falls under the definition of 'tissue' in section 1 of the NHA, and tissue is referred to in the Import and Export Regulations as a thing that requires an export permit.

extracted, sequenced, and analysed by the overseas laboratory, the resulting genetic data is sent back to the testing company in South Africa, allowing them to generate test results for consumers, and for potential research. Therefore, genetic data comes from overseas into South Africa. Its importation depends on the laws of the country from which it is sent. As the overseas testing company is domiciled, and processes information, outside of South Africa, it does not need to comply with POPIA – even though it uses the personal information of South Africans. However, the testing company must comply with the data protection laws of the country in which it is domiciled.[67]

(d) Scenario (4)

The testing company is based in South Africa and saliva samples are sent locally and DNA extraction happens in South Africa, but the sequencing and analysis of DNA occurs in a laboratory abroad. The first part of this scenario is similar to scenario (3) above in that the testing kits and saliva samples (sent by consumers to the testing company) are sent within South Africa and are thus not subject to import and export provisions. However, as saliva samples are not sent overseas by the testing company, this scenario may have different implications. As extracted and unsequenced DNA is not subject to the Import and Export Regulations, an export permit is not required to send DNA to a laboratory in another jurisdiction. Furthermore, POPIA does not apply as DNA is not 'personal information'.[68] The extracted DNA is analysed, and genetic data obtained. This genetic data is sent back to the testing company in South Africa and used to create test results, and possibly for research. Like scenario (3) above, genetic data is sent to South Africa from overseas. Because the overseas testing company is domiciled, and processes information, beyond South Africa, it is exempt from complying with POPIA. But the testing company must adhere to the data protection laws of its own country.[69] POPIA's export requirements only apply where a South African testing company extracts and sequences DNA from saliva samples locally, resulting in genetic data, which is subsequently sent overseas.

(e) Conclusion on the scenarios

The applicability of South African legislative provisions, as well as the need for licenses and permits, relating to importing and exporting of both direct-to-consumer genetic testing kits, either by consumers or local testing companies, as well as samples (either saliva, DNA, or

[67] John Giles 'Must I comply with the POPI Act?' *Michalsons* 12 February 2020, available at *https://www.michalsons.com/blog/must-i-comply-with -the-popi-act/41827,* accessed on 3 March 2021.
[68] Section 1 of POPIA.
[69] Giles op cit note 67.

genetic data) depends on the location of the testing company, their associated laboratory, and where the DNA extraction, sequencing, and analysis occur.

VII CONCLUSION

From a legal perspective, POPIA, the NHA, the Medicines Act, and the associated regulations cover aspects of the direct-to-consumer genetic testing process. But DNA reveals areas of freedom from over-regulation in South Africa's extant law. The legitimate government purpose served by the Import and Export Regulations may be to control the export of biological samples from South Africa to ensure that there is no exploitation of South Africans, especially given past instances of overseas researchers using the samples of Africans, but withholding research benefits from participants and scientists.[70] Therefore, I suggest that the Import and Export Regulations, while potentially useful for bulk exports of biological samples for commercial or research purposes by groups or institutions, curtail the freedom of individuals and should be revised to exclude individuals from being subject to the export permit system, specifically for the purpose of exporting saliva samples for a direct-to-consumer genetic test. This entails amending the definition of 'applicant' in the Import and Export Regulations to exclude individuals. Such a permit system infringes on the freedom of individuals and wastes valuable state resources to detect saliva samples that are being sent overseas for a consumer's own reasons, and for which regulation is nonsensical.

Based on the above analysis, there are several best business models for South African testing companies to follow in terms of the importing and exporting of saliva samples, extracted DNA, and genetic data. Firstly, a local laboratory can extract DNA from saliva samples, which would then neither be covered by the Import and Export Regulations, nor the NHA, and this extracted DNA can then be sent abroad for sequencing and analysis – this is the easiest way to avoid the Import and Export Regulations. Secondly, if a South African testing company extracts and sequences DNA locally, the resulting genetic data would bring about the provisions of POPIA. As the second option involves having to follow certain regulatory requirements, I suggest that the first option of extracting DNA locally and then sending it overseas for sequencing and analysis is the simplest path for South African testing companies to follow.

[70] Annelize Nienaber 'Consent to and authorisation of the export and use of human biological specimens for future research – Perspectives from three African countries' (2011) 44(2) *CILSA* 226.

CHAPTER 7

CAN DIRECT-TO-CONSUMER GENETIC TESTING COMPANIES CONDUCT RESEARCH USING GENETIC DATA GATHERED FROM CONSUMERS?

I INTRODUCTION AND CHAPTER OVERVIEW

Given the potential of genetic data in the diagnosis, prevention, and treatment of disease,[1] research conducted by testing companies often forms an important part of their business model.[2] Repeat consumers are unlikely, which has caused many testing companies to turn their focus towards big data.[3] Through consumers, testing companies can gather, store, and share large amounts of genetic data which could serve to benefit research and advance genetic discoveries.[4]

In this Chapter, I consider the research that certain testing companies conduct using the genetic data gathered from consumers who undergo direct-to-consumer genetic testing. This Chapter examines the NHA, POPIA, the Human Biological Material Regulations, the SA

[1] See, Donrich W Thaldar, Marietjie Botes & Annelize Nienaber 'South Africa's new standard material transfer agreement: Proposals for improvement and pointers for implementation' (2020) 21(85) *BMC Med Ethics* 2.

[2] Stuart Hogarth & Paula Saukko 'A market in the making: The past, present and future of direct-to-consumer genomics' (2017) *New Genet Soc* 204.

[3] 'Big data' refers to substantial data sets which, when analysed, uncover links, patterns, and trends, and are used to for predictions and discoveries. Dusty-Lee Donnelly *Privacy by (re)Design: A Comparative Study of the Protection of Personal Information in the Mobile Applications Ecosystem under United States, European Union and South African Law* (Doctor of Laws thesis, University of KwaZulu-Natal, 2020) 76.

[4] The collection of genetic data by testing companies allows them to establish an important resource for drug development, discover rare genetic markers, and identify populations for clinical trials. The business models of testing companies 'do not focus on profits from the sale of genetic tests, but from gathering the genetic and personal data that can be licensed and sold to institutions, academic researchers, or drug companies' – thus forming a pillar of a 'two-sided data-banking market model'. Elizabeth A Varga 'You want to do what? My mother's choice to have direct-to-consumer genetic testing' (2012) *J Genet Counsel* 385; Andrew S Robertson 'Taking responsibility: Regulations and protections in direct-to-consumer genetic testing' (2009) 24(1) *Berkeley Tech LJ* 218; Hogarth & Saukko op cit note 1 at 204; Valerie Gutmann Koch 'PGTandMe: Social networking-based genetic testing and the evolving research model' (2012) 22(1) *Health Matrix* 61–2; Elizabeth R Pike 'Securing sequences: Ensuring adequate protections for genetic samples in the age of big data' (2015) 37 *Cardozo L Rev* 1995–6; Henri-Corto Stoeklé, Marie-France Mamzer-Bruneel, Guillaume Vogt et al '23andMe: A new two-sided data-banking market model' (2016) 17(19) *BMC Med Ethics* 3; Michael Grothaus 'How 23andMe is monetizing your DNA' *Fast Company* 5 January 2015, available at *https://www.fastcompany.com/3040356/what-23andme-is-doing-with-all-that-dna*, accessed on 21 March 2020 cited in Valerie Gutmann Koch & Kelly Todd 'Research revolution or status quo: The new common rule and research arising from direct-to-consumer genetic testing' (2018) 56(1) *Houston L Rev* 91; Jennifer Cacchio 'What you don't know can hurt you: The legal risk of peering into the gene pool with direct-to-consumer genetic testing' (2018) 87 *UMKC L Rev* 229–30; Mauro Turrini 'Online Genomes: Problematizing the disruptiveness of direct-to-consumer genetic tests' (2018) *Sociology Compass* 8.

MTA, and the Human Research Participant Regulations – all with the intention of ultimately determining whether testing companies are legally permitted to conduct such research.

While research is a vast topic that contains many aspects requiring investigation, in this Chapter I only examine the main issues that relate to direct-to-consumer genetic testing. Although there are various privacy issues associated with direct-to-consumer genetic testing relating to the safeguarding of confidential data; the conservation and control of samples and data; the sale of data to third parties; and the fate of samples and data in the case of a testing company closing or going bankrupt, as has occurred previously,[5] this Chapter purely focuses on privacy in terms of research and these other issues, though relevant, fall outside of the scope of this Chapter. This Chapter also does not determine ownership, control, and commercialisation issues in relation to biological material and its associated data.

As a point of clarity at the outset, research is not conducted on saliva, but on the DNA sequence (the A, G, C, and T nucleotide bases) found in the saliva – also referred to as genetic data.[6] Genetic data is typically the vital source of information and the object of research,[7] rather than the biological material.[8]

[5] In 2009, deCODEme filed for bankruptcy and the sales of its genetic tests were discontinued. Patricia J Zettler, Jacob S Sherkow & Henry T Greely '23andMe, the Food and Drug Administration, and the future of genetic testing' (2014) 174(4) *JAMA Inter Med* 493; Teresa Pàmpols Ros, José Miguel García Sagredo, Antonio Pérez Aytése et al 'Directed to consumer genetic testing: Perspective from the Ethics Commission of the Spanish Society for Human Genetics' (2019) 153(1) *Med Clin (Barc)* 36; Bartha Maria Knoppers, Denise Avard & Heidi Carmen Howard 'Direct-to-consumer genetic testing: Driving choice?' (2010) 10(8) *Expert Rev Mol Diagn* 965–6.

[6] While some testing companies state that they use DNA for research, others conduct research using genetic data. But a common mistake is to refer to the sequence or genetic data as DNA. It is inevitable that, even where testing companies claim to conduct research on DNA, they use the sequence derived therefrom, rather than the DNA.

[7] Though testing companies may vary in specifying the things used in research, genetic data holds the most value. Although it is unlikely that researchers would find value in saliva samples, some testing companies indicate that research may include the use of samples. For example, Myriad and Color Genomics use samples and any information derived therefrom. Other testing companies use DNA and other related information. GeneWay and MyHeritage use DNA, DNA results, and other DNA information – although GeneWay only uses this for internal research. There are testing companies that mention that genetic data (or information) is used in research. For example, MapMyGenome, Ancestry, Helix, GenebyGene, Myriad, 23andMe and Color Genomics all make use of genetic information or data for research. Myriad 'Myriad informed consent policies' available at *https://myriadwomenshealth.com/consent-policies/*, accessed on 9 January 2021; Color 'Color informed consent' available at *https://www.color.com/informed-consent*, accessed on 9 January 2021; MyHeritage 'Privacy policy' 17 December 2020 available at *https://www.myheritage.com/privacy-policy*, accessed on 9 January 2021; Helix 'Helix privacy policy' 30 November 2020 available at *https://www.helix.com/pages/privacy-policy*, accessed on 9 January 2021; MapmyGenome 'Terms and conditions' available at *https://mapmygenome.in/terms-and-conditions/*, accessed on 9 January 2021; AncestryDNA 'AncestryDNA informed consent' 24 July 2018 available at *https://www.ancestry.com/dna/lp/informedconsent-v4-en*, accessed on 9 January 2021; GenebyGene 'Gene by Gene privacy policy' available at *https://genebygene.com/wp-content/uploads/2019/08/GenebyGene_PrivacyPolicy.pdf*, accessed on 9 January 2021; 23andMe 'Research consent document' available at *https://www.23andme.com/about/consent/*, accessed on 9 January 2021; Joel C Eissenberg 'Direct-to-consumer genomics: Harmful or empowering?' (2017) 114(1) *Mo Med* 29.

[8] Biological material is a container for genetic data and is disposed of once the genetic data has been extracted.

II DONATION AND RESEARCH

Donation is relevant to direct-to-consumer genetic testing as consumers provide their saliva sample, which is extracted, sequenced, and analysed to obtain genetic data that is used by testing companies for research without remunerating consumers. The Human Biological Material Regulations state that donation refers to 'donation of human biological material for genetic testing, genetic training, and genetic health research for therapeutic purposes'.[9] Given that saliva is a type of biological material (established in Chapter 2), the donation thereof is relevant to consider. Section 63 of the NHA states that –

> 'A human body, tissue, blood, blood products or gametes may be donated by any person contemplated in section 55(*a*) or 62 to any prescribed institution or person for any purpose contemplated in section 56 or 64(1)'.[10]

Firstly, section 63 of the NHA, when describing the categories eligible for donation, uses the word 'or',[11] meaning that any one of the mentioned categories may be donated – one of which is tissue. Secondly, any person 'in section 55(*a*) or 62' of the NHA may donate inter alia tissue,[12] but such tissue cannot be removed from a living person without their written consent.[13] Section 63 of the NHA also allows for tissue to be donated to a 'prescribed institution or person for any purpose contemplated in section 56 or 64(1)' of the NHA.[14] Section 54(1) of the NHA states that the Minister may 'designate any institution other than an institution contemplated in section 63 as an authorised institution'.[15] A testing company must apply to the Minister to be designated as an authorised institution which, if successful, would mean that consumers may provide such testing companies with their saliva samples, but only for certain purposes.[16] This is because section 56 of the NHA allows tissue removed from living persons to be used 'only

[9] Regulation 1 of the Human Biological Material Regulations. It is interesting to note that regulation 1 of the Draft Testing and Research Regulations define 'donation' to mean 'donation of human DNA, RNA, cultured cells, stem cells; blastomeres, polar bodies, embryos, embryonic tissue and small tissue biopsies for genetic testing, health research or therapeutic purposes'.
[10] Section 63 of the NHA.
[11] *In Re Estate Cullingworth* 1936 NPD 251 (NPD) at 265 referred to the 'ordinary strictly disjunctive meaning' of the word 'or'.
[12] Section 63 of the NHA. However, section 62 of the NHA deals with the donation of inter alia tissue of deceased persons and is therefore not relevant to consider in the context of this Chapter. But section 55(*a*) of the NHA is applicable.
[13] Section 55(*a*) of the NHA.
[14] Section 63 of the NHA.
[15] Section 54(1) of the NHA. Section 54(2) of the NHA provides that an authorised institution may inter alia: (i) obtain or use any tissue lawfully imported or removed for any purpose referred to in section 56 or 64 of the NHA; and (ii) supply any that it preserves to a person or institution in section 63 of the NHA for any purpose referred to in section 58 or 64 of the NHA. Sections 54(2)(*b*) and (*c*) of the NHA.
[16] Section 56(1) of the NHA.

for such medical or dental purposes as may be prescribed'.[17] Therefore, regard must be had to regulation 5 of the Human Biological Material Regulations, which provides the medical and dental purposes for which biological material may be removed.[18] The fact that the Human Biological Material Regulations refer to removing and using biological material for the 'medical and dental' purpose of inter alia health research implies that this forms part of the purposes prescribed in section 56(1) of the NHA – and thus the NHA applies.

While section 64(1) of the NHA explicitly mentions that tissue may be donated for inter alia health research,[19] this only applies to deceased persons. Without a similar provision in the NHA relating to living persons, tissue can only be donated for 'any purpose contemplated in section 56' of the NHA – unless such a purpose includes health research.[20] As established above, as per the Human Biological Material Regulations, biological material may be removed for several medical and dental purposes, including health research.[21] However, I suggest that 'health research' (for which tissue may be donated) also be explicitly included under *living* persons in the NHA – which will be a step towards the NHA including provisions relating to the field of genetics.

The implications of the above mean that, in terms of the NHA, consumers are permitted to donate saliva samples to testing companies (if they are designated as authorised institutions) for health research. Although saliva samples are sent by consumers for the purpose of undergoing a direct-to-consumer genetic test, if research is part of the business model of the testing company, it forms part of the transaction from the onset (which consumers consent to) – even if genetic data, rather than saliva, is used in such research – saliva is still donated for the purpose of research as genetic data is derived from processing the saliva sample.[22] Hence,

---

[17] Ibid.

[18] Regulation 5 of the Human Biological Material Regulations. Referred to in section 69(3) of the NHA. See also, Donrich W Jordaan 'The boy and his microscope: Interpreting section 56(1) of the National Health Act' (2009) 2(1) *SAJBL* 13.

[19] Section 64(1)(*b*) of the NHA. 'Health research' is defined in section 1 of the NHA as –

'any research which contributes to the knowledge of –

(a) The biological, clinical, psychological or social processes in human beings;
(b) improved methods for the provision of health services;
(c) human pathology;
(d) the causes of disease;
(e) the effects of the environment on the human body;
(f) the development or new application of pharmaceuticals, medicines and related substances; and
(g) the development of new applications of health technology'.

[20] Section 63 of the NHA. Section 56(1) of the NHA states that a person may use inter alia tissue removed 'from a living person only for such medical or dental purposes as may be prescribed'.

[21] Regulation 5(b) of the Human Biological Material Regulations.

[22] This is so despite the fact that research generally uses genetic data (and not the actual saliva sample), which is not mentioned under the list of things that can be donated in section 63 of the NHA, as the genetic data is obtained

the transaction has a dual nature: (1) a consumer wanting genetic information about themselves; and (2) a testing company wanting to build its genetic database for commercial research purposes. As such, the research-related provisions in the law are applicable from the outset.

III THE REMOVAL OF BIOLOGICAL MATERIAL FOR RESEARCH

As established above, the Human Biological Material Regulations permit biological material to be removed for various medical and dental purposes, one of which is health research.[23] As mentioned in Chapter 3, only a 'competent person', as per the Human Biological Material Regulations, may remove and use biological material for inter alia 'genetic health research'.[24] With regards to research, a 'competent person' is a 'medical technologist or scientist registered as such in terms of the Health Professions Act'.[25]

In terms of direct-to-consumer genetic testing, saliva (a form of biological material) is *removed* by consumers as part of the testing process, but genetic data is *used* by medical technologists and scientists to provide test results to consumers and/or for research.[26] In determining the relevance of the research considerations in the Human Biological Material Regulations to direct-to-consumer genetic testing, the question to ask is: Where does the research start? Does it begin when one analyses the data, or when one obtains the source material? Broadly construed, I suggest that research commences with the collection of the saliva sample. Given the formulation of regulation 2(a) of the Human Biological Material Regulations, the focus is on the *purposes* for which biological material may be removed; it is not required that biological material be removed directly for the purposes for which it will be used. Therefore, if biological material is used for genetic testing or genetic health research,[27]

---

through extraction of DNA from the saliva sample and the subsequent sequencing and analysis. Therefore, consumers are permitted to donate their saliva samples for research, even though the saliva is not actually used in such research.

[23] Regulation 5(b) of the Human Biological Material Regulations state that biological material may be removed for 'health research referred to in section 69(3) of the Act'. Section 69(3) of the NHA lays out the duties of the National Health Research Committee (NHRC), which are restricted to public health research activities. These include determining and organising the health research to be undertaken by public health authorities, and which health research agendas and resources concentrate on urgent health issues. See, M Nöthling Slabbert & MS Pepper '"A room of our own?" Legal *lacunae* regarding genomic sovereignty in South Africa' (2010) 73 *THRHR* 448.

[24] Regulation 2(a) of the Human Biological Material Regulations.

[25] Regulation 1 of the Human Biological Material Regulations. Moreover, removing biological material for genetic health research may only be done at an authorised institution, a prescribed institution, or a research institution. Regulation 2(b) of the Human Biological Material Regulations; sections 54(1) and 63 of the NHA.

[26] The removal and use of biological material are dealt with separately in the Human Biological Material Regulations. Regulation 2 of the Human Biological Material Regulations deals with the removal of human biological material and regulation 3 of the Human Biological Material Regulations covers the removal or withdrawal of biological samples from living persons, while regulation 5 of the Human Biological Material Regulations provides for the use of human biological material.

[27] As per regulation 2(a) of the Human Biological Material Regulations.

as is the case here, then the Human Biological Material Regulations apply, and the removal and use must be undertaken by a 'competent person'.

While 'health research' is defined and referred to in the NHA, the Human Biological Material Regulations refer to 'genetic health research',[28] and stipulate several legal requirements relating to such, including that genetic research be undertaken at an authorised or prescribed institution,[29] and that such an institution have registers for recording genetic research.[30] 'Genetic health research' is not defined in the NHA or the Human Biological Material Regulations and, as the meaning of 'health research' is only elucidated in the NHA – as the enabling legislation upon which the Human Biological Material Regulations are based – regard must be had to this definition.[31] The term 'health research' in the NHA[32] includes '*any* research which contributes to knowledge of…' (own emphasis).[33] Prima facie, the word 'any' is 'a word of wide and unqualified generality'.[34] In *Hayne & Co v Kaffrarian Steam Mill Co Ltd*,[35] it was held that '[i]n its natural and ordinary sense "any" – unless restricted by the context – is an indefinite term which includes all the things to which it relates'.[36] Genetic research commonly concerns the investigation of the role that genes play in disease development.[37] Such

[28] Regulation 2(a) of the Human Biological Material Regulations. The Human Biological Material Regulations also mention 'health research' in regulation 5(b). 'Genetic health research' is referred to in regulations 2 and 3 of the Human Biological Material Regulations regarding the removal of biological material (from living persons). But regulation 5 of the Human Biological Material Regulations, in terms of the use of biological material, only refers to 'health research'.

[29] Regulation 2(b) of the Human Biological Material Regulations.

[30] Regulation 12(1) of the Human Biological Material Regulations.

[31] Regulation 1 of the Human Biological Material Regulations states that '[i]n these Regulations any word or expression to which a meaning has been assigned in the Act shall have such meaning and, unless the context otherwise indicates'. The South African Law Reform Commission, in considering revisions to the Interpretation Act 33 of 1957 (Interpretation Act) proposed that reference to 'the Act' in subordinate legislation 'must be read as a reference to the enabling legislation in terms of which that subordinate legislation was enacted'. Furthermore, words or expressions used in subordinate legislation, and which are defined in the enabling legislation, have the meaning given in the enabling legislation. South African Law Reform Commission *Discussion Paper 112 Statutory Revision: Review of the Interpretation Act 33 of 1957 (Project 25)* (2006) 408.

[32] Section 1 of the NHA.

[33] Ibid.

[34] *R v Hugo* 1926 AD 268 at 271. *Dutch Reformed Church, De Aar v Joubert* 1921 CPD 9 at 13 cited two cases in which the meaning of the word 'any' was discussed. *Beckford v Wade* 17 Ves at 91 held that general words 'must receive a general construction, unless you find in the statute itself some ground for limiting and restraining their meaning by reasonable construction, and not by arbitrary addition or retrenchment'. And in *Duck v Bates* 12 QBD 79 it was held that 'any' is a word 'which excludes limitation or qualification'. See also, *S v Wood* 1976 (1) SA 703 (A) at 706 where it was held that '[j]udicially the word "any" has been defined as a word of very wide import, "and *prima facie* the use of it excludes limitation" (*Clarke-Jervoise v Scutt* (1920) 1 Ch 382 at 388)'.

[35] 1914 AD 363.

[36] *Hayne & Co v Kaffrarian Steam Mill Co Ltd* supra note 35 at 371.

[37] Genetic research typically includes genetic testing and gene therapy. Nature Research 'Genetics research' available at *https://www.nature.com/subjects/genetics-research*, accessed on 7 January 2021; World Health Organization 'Genetic research' available at *https://www.who.int/genomics/research/en/*, accessed on 7 January 2021.

research examines, amongst other things, biological processes, human pathology,[38] the causes of disease, and the application or development of medicines and pharmaceuticals – all of which fall within the definition of 'health research' in the NHA.[39] Therefore, I suggest that as the definition of 'health research' in the NHA is broad, and given that the research undertaken by testing companies relates to health and involves the use of genetic data, it is considered 'genetic health research'[40] – bringing both the NHA and the Human Biological Material Regulations into application.

Also relevant is the NHA's review procedure by a Health Research Ethics Committee (HREC)[41] of all proposed health research in South Africa.[42] According to the NHA, all institutions in South Africa conducting health research must either have access to, or establish their own, HREC,[43] registered with the National Health Research Ethics Council (NHREC),[44] that must approve all proposed health research.[45] The Human Biological Material Regulations prohibit genetic health research unless it has been approved by a registered HREC.[46] The Human Research Participant Regulations also require research involving human participants[47] to be subject to independent review by a registered HREC.[48] HRECs, with a focus on the protection of research participants' interests,[49] have a statutory duty to review health research proposals, and must confirm that proposed research studies have a health purpose and satisfy the HREC's ethical standards.[50] The NHA also requires the NHREC to establish national norms

[38] 'Pathology' refers to the study of diseases in humans. Merriam-Webster 'Pathology' available at *https://www.merriam-webster.com/dictionary/pathology*, accessed on 20 August 2020; Cambridge Dictionary 'Pathology' available at *https://dictionary.cambridge.org/dictionary/english/pathology*, accessed on 20 August 2020.
[39] Section 1 of the NHA.
[40] The same argument was advanced by Thaldar et al in relation to research on human germline editing. Donrich Thaldar, Marietjie Botes, Bonginkosi Shozi et al 'Human germline editing: Legal-ethical guidelines for South Africa' (2020) 116(9/10) *S Afr J Sci* 2.
[41] This is a statutory body selected by the Minister of Health. Section 72(2) of the NHA.
[42] DW Thaldar & BA Townsend 'Exempting health research from the consent provisions of POPIA' (2021) 24 *PER/PELJ* 4.
[43] Section 73(1) of the NHA.
[44] Section 73 of the NHA.
[45] Section 73(2) of the NHA.
[46] Regulation 3(2) of the Human Biological Material Regulations. This is referred to in section 73(1) of the NHA.
[47] Regulation 1 of the Human Research Participant Regulations define a 'human participant' as 'a living person about whom a researcher obtains data or specimens or identifiable private information through intervention or interaction with that person'.
[48] Regulation 2(g) of the Human Research Participant Regulations.
[49] Donrich Thaldar 'One material transfer agreement to rule them all? A call for revising South Africa's new standard material transfer agreement' (2020) 7(105) *Humanities & Social Sciences Communications* 3; Department of Health (DoH) *Ethics in Health Research: Principles, Processes and Structures* 2 ed (2015) 40.
[50] Section 73(2) of the NHA.

and standards.[51] The DoH Ethics Guidelines is one such source.[52] As the Human Research Participant Regulations, in regulation 2(a), state that health research involving human participants 'must comply with the Department of Health national ethical guidelines for research with human participants',[53] the DoH Ethics Guidelines are legally binding on HRECs and pertain to all research involving biological material and related data.[54]

To conclude: The removal of biological material is for 'genetic health research' the moment removal is done with that purpose in mind. I established that 'health research' in the NHA is broad enough to encompass 'genetic health research' and thus includes research undertaken by testing companies. This means that both the NHA and the Human Biological Material Regulations are relevant, and all health research undertaken by testing companies in South Africa require approval from a HREC.[55]

IV CONSENT TO RESEARCH

One of the most important South African statutes in relation to research conducted by testing companies is POPIA. Given that genetic data, used in research, falls under the definition of 'personal information'[56] (established in Chapter 2), POPIA's research provisions may apply. It must be mentioned that in terms of personal information and special personal information, the personal details of individuals – such as their names, contact numbers, addresses, and payment information – are also relevant. However, what is at stake in research is genetic data and this is therefore my focus in this Chapter.

Consent is vital for genetic research and has become a highly debated area. South Africa's legal landscape defines the type of consent that must be obtained from participants for data collection, research, storage, and sharing.[57] Consent is covered by numerous South African statutes and guidelines, albeit in varying degrees and in differing ways. Consent is defined as an 'indication of agreement to participate in research, based on adequate knowledge and

[51] Section 72(6)(*c*) of the NHA.
[52] DoH op cit note 49.
[53] Regulation 2(a) of the Human Research Participant Regulations.
[54] Regulations 2(a) and 6(b) of the Human Research Participant Regulations; Thaldar et al op cit note 1 at 3.
[55] As testing companies must have their research protocol approved by a HREC (in terms of section 73 of the NHA), they will need to determine what research they plan to undertake. While testing companies may propose a research project that entails blanket consent, such as building a genetic database for research, it is unlikely that the HREC will approve a project involving blanket consent due to it not being recommended by the DoH Ethics Guidelines. As such, it is advisable that testing companies have a research protocol that is likely to be approved by the HREC. See, DoH op cit note 49 at 31.
[56] Section 1 of POPIA.
[57] Thaldar & Townsend op cit note 42 at 4.

understanding of relevant information, and freely given'.[58] Notably, section 12(2)(*c*) of the Constitution safeguards against exploitative research, providing that –

> 'Everyone has the right to bodily and psychological integrity, which includes the right…not to be subjected to medical or scientific experiments without their informed consent'.[59]

The DoH *Guidelines for Good Practice in the Conduct of Clinical Trials with Human Participants in South Africa* state that informed consent is 'an essential component of ethical research'.[60] Similarly, the NHA requires informed consent for both participation in research and for the removal of tissue.[61] Written informed consent is required to have biological material removed for inter alia genetic health research, in terms of the Human Biological Material Regulations.[62] The Human Research Participant Regulations provide that research involving human participants must 'be undertaken with appropriate consent processes'.[63]

There are different types of consent used in genetic research, the most common of which are broad consent[64] and specific consent.[65] While broad consent allows individuals to consent to their samples and/or data being used widely for present as well as undetermined future studies,[66] specific consent defines a particular study, limiting sample and/or data use to that purpose.[67] Blanket consent, which neither defines a study nor restricts the type of research that may be undertaken, is not recommended by the DoH Ethics Guidelines.[68]

---

[58] DoH op cit note 49 at 53.
[59] Section 12(2)(*c*) of the Constitution.
[60] The DoH *Guidelines for Good Practice in the Conduct of Clinical Trials with Human Participants in South Africa* also state that informed consent suggests that information has been provided to participants concerning the nature and purpose of the research as well as other options. Informed consent entails free choice by individuals regarding research participation based on information allowing an informed decision. Department of Health *Guidelines for Good Practice in the Conduct of Clinical Trials with Human Participants in South Africa* 2 ed (2006) 11; DoH op cit note 49 at 21.
[61] Once participants are aware of the purpose of the research as well as benefits and risks, they can consent based on this information. Section 55 of the NHA; C Staunton & K Moodley 'Data mining and biological sample exportation from South Africa: A new wave of bioexploitation under the guise of clinical care?' (2016) 106(2) *SAMJ* 137.
[62] Regulation 3(1)(*a*) of the Human Biological Material Regulations.
[63] Regulation 2(f) of the Human Research Participant Regulations.
[64] Broad consent defines a wide variety of studies and entails an individual permitting the use of their sample for current research, storage, and potential future research. Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018) 50–1; DoH op cit note 49 at 31; Thaldar et al op cit note 1 at 4.
[65] Consent is for a particular research purpose. Further use means new consent must be obtained. Although specific and informed consent aim to respect autonomy, re-contacting and re-consenting individuals can be costly and presents difficulties in situations where individuals relocate and fail to update contact details. ASSAf op cit note 64 at 50; DoH op cit note 49 at 31; Thaldar et al op cit note 1 at 4.
[66] ASSAf op cit note 64 at 51.
[67] Ibid at 50.
[68] Blanket consent is open-ended and does not limit or restrict the use and sharing of samples or data. DoH op cit note 49 at 31; Thaldar et al op cit note 1 at 4.

The DoH Ethics Guidelines, published in 2015, are binding on researchers and institutions conducting health research with human participants. The DoH Ethics Guidelines provide three types of consent deemed acceptable for use in research, namely narrow (restrictive) consent, tiered consent,[69] and broad consent.[70] Although these forms of consent are satisfactory, the DoH Ethics Guidelines – in stark contrast with POPIA, which specifies that consent must demonstrate an 'expression of will',[71] and information must be collected for a 'specific' purpose[72] – state that consent should be 'broad enough to allow for future and secondary uses of data, in line with the opportunities to use such data in advancing knowledge to improve health'[73] – thereby purporting to allow, and prefer, broad consent for genetic research.[74] The Human Research Participant Regulations, as part of the NHA's subsidiary legislation, stipulate the information that research participants must be informed of for consent to be given[75] – one of which is 'the purpose of the research'.[76] As noted by Thaldar and Townsend, this requirement of a recognisable purpose seems to imply that blanket consent is prohibited. But as it is possible for multiple studies to have a single, over-arching purpose, it appears that broad consent is allowed under the Human Research Participant Regulations.[77]

POPIA defines 'consent' as 'any voluntary, *specific* and informed expression of will in terms of which permission is given for the processing of personal information' (own emphasis).[78] In contrast to the DoH Ethics Guidelines, the requirement in POPIA that consent be *specific*, has caused contention regarding the type of consent required for genetic research in South Africa.[79] POPIA's specific consent requirement differs from the norm in South Africa where genetic research is typically conducted using broad consent, allowing researchers to utilise, and participants to consent to the use of, genetic data for a wide range of current as well as prospective, unknown purposes.[80] Thus, there have been disputes surrounding POPIA and

[69] Tiered consent combines specific and broad consent and allows for consent individually to different aspects of a study. Tiered consent allows an individual to decide to partake in the main study and to authorise their sample to be stored and used in the future. Tiered consent also requires the provision of further information. For example, the storage tier must explain the location and duration of sample storage as well as their fate once the storage period comes to an end. ASSAf op cit note 64 at 50–1; DoH op cit note 49 at 31; Thaldar et al op cit note 1 at 4.
[70] DoH op cit note 49 at 31.
[71] Section 1 of POPIA.
[72] Section 13(1) of POPIA; ASSAf op cit note 64 at 52.
[73] DoH op cit note 49 at 31; Thaldar et al op cit note 1 at 4.
[74] DoH op cit note 49 at 31.
[75] Regulation 5 of the Human Research Participant Regulations.
[76] Regulation 5(a) of the Human Research Participant Regulations.
[77] Thaldar & Townsend op cit note 42 at 5.
[78] Section 1 of POPIA.
[79] Thaldar & Townsend op cit note 42 at 11.
[80] However, problems may arise in terms of the broad consent model as it may not always be feasible to inform consumers of every research study in which their data will be used. Staunton and Moodley argue that broad

utilising an individual's genetic data using broad consent. Staunton et al argue that a purposive interpretation of the provisions of POPIA pertaining to consent for research, allow broad consent.[81] However, Thaldar and Townsend reason that interpreting POPIA purposively supports its ordinary meaning – that genetic research requires specific consent.[82] I suggest that, given the reference to 'specific' in section 13(1) of POPIA,[83] as well as the definition of 'consent',[84] consent for the collection of personal information is to be specific. Moreover, reference to the singular 'purpose' in section 13(1) of POPIA implies that each specific purpose must be consented to, and not general or all-encompassing consent to a broad range of different purposes.[85]

As alluded to above, there is a conflict between POPIA and the DoH Ethics Guidelines, which appear to favour broad consent for research (although narrow and tiered consent are also permitted).[86] In terms of POPIA, personal information must be collected, and consent must be provided, for a specific purpose[87] which, in this case, is undergoing a direct-to-consumer genetic test (and preferably to research), and then relying on the research exceptions or the further processing provisions in POPIA.[88] However, consumers must nevertheless consent to research in terms of the DoH Ethics Guidelines.[89] Given that direct-to-consumer genetic testing often has a dual purpose, it can potentially be argued that consumers are required to give

consent may encompass sample exportation as participants consent to the storage and reuse of samples for future, unidentified research, which could encompass research abroad. DW Thaldar & B Townsend 'Genomic research and privacy: A response to Staunton *et al*' (2020) 110(3) *SAMJ* 172; Staunton & Moodley op cit note 61 at 137.

[81] Essentially, Staunton et al contend that genetic research and innovation would be hampered by specific consent as research participants that provided broad consent would have to be 're-contacted and re-consented'. Further, based on the premise that genetic research is in the public interest, Staunton et al argue that specific consent undermines this, and the public interest requires POPIA to be interpreted purposively to 'permit broad consent for the processing of personal information for research'. C Staunton, R Adams, M Botes et al 'Safeguarding the future of genomic research in South Africa: Broad consent and the Protection of Personal Information Act No. 4 of 2013' (2019) 109(7) *SAMJ* 468.

[82] In *Bato Star* para 91, the Constitutional Court adopted a purposive approach. Although a purposive approach to interpretation focuses on context, it does not overlook the ordinary meaning or significance of the words used. In *Bertie Van Zyl* para 22, the Constitutional Court held that a 'purposive reading of a statute must…remain faithful to the actual wording of the statute'. Therefore, according to Thaldar et al, attempting to alter the meaning of 'specific' to 'broad' in POPIA appears to overly rely on purposive interpretation. See also, *South African Airways (Pty) Ltd v Aviation Union of South Africa* 2011 (3) SA 148 (SCA) paras 25–30; *Natal Joint Municipal Pension Fund v Endumeni Municipality* 2012 (4) SA 593 (SCA) para 18; Thaldar & Townsend op cit note 80 at 172; Thaldar et al op cit note 1 at 4. See also, Beverley A Townsend & Donrich W Thaldar 'Navigating uncharted waters: Biobanks and informational privacy in South Africa' (2019) 35(4) *SAJHR* 329–50.

[83] Section 13(1) of POPIA states that personal information must be collected for a 'specific, explicitly defined and lawful' purpose, of which data subjects must be aware.

[84] Section 1 of POPIA.

[85] Donnelly op cit note 3 at 244.

[86] This includes consent to partake in research as well as gathering the biological material to be used in research. DoH op cit note 49 at 31.

[87] Section 13(1) of POPIA.

[88] Section 15(3)(*e*) of POPIA.

[89] DoH op cit note 49 at 30.

specific consent for the purpose of a direct-to-consumer genetic test and narrow, tiered, or broad consent for research in line with the DoH Ethics Guidelines, as well as broad consent under the Human Research Participant Regulations. But, in terms of POPIA, can research also be considered a specific purpose? I deal with this question below.

POPIA also requires consent to be 'informed'.[90] This entails that data subjects understand all the particularities associated with research, as well as potential risks or harms, and agree to such. In terms of direct-to-consumer genetic testing, consumers must be informed, in a comprehensible manner, of inter alia the personal information to be collected,[91] the purpose of its collection,[92] and the protection of personal information where the third party is based in another jurisdiction[93] – without which consent is invalid.[94] This is bolstered by section 13(2) of POPIA, which requires that steps are taken to notify data subjects of the purpose of collection of personal information.[95] However, notification is not necessary if individuals consent to non-compliance,[96] or if the personal information is used for research.[97]

Consent is inferred from the conduct of data subjects prior to the collection of personal information – as long as it meets the requirements of being specific, voluntary, and informed.[98] Because of the direct-to-consumer genetic testing model, consent is generally attained through online means. Consumers are required to tick a box to indicate agreement[99] – thus taking the form of click-wrap agreements.[100] However, as discussed in Chapter 3, it has been questioned whether the type of consent given in direct-to-consumer genetic testing is informed. Unlike traditional genetic testing, where informed consent is central,[101] direct-to-consumer genetic

[90] In terms of the definition of 'consent' in section 1 of POPIA.
[91] Section 18(1)(*a*) of POPIA.
[92] Section 18(1)(*c*) of POPIA.
[93] Section 18(1)(*g*) of POPIA.
[94] See, Donnelly op cit note 3 at 242–3.
[95] Section 13(2) of POPIA.
[96] Section 18(4)(*a*) of POPIA.
[97] Section 18(4)(*f*)(*ii*) of POPIA.
[98] Donnelly op cit note 3 at 245.
[99] Eline M Bunnik, A Cecile, JW Janssens et al 'Informed consent in direct-to-consumer personal genome testing: The outline of a model between specific and generic consent' (2012) 28(7) *Bioethics* 346.
[100] Andelka M Phillips 'Genomic privacy and direct-to-consumer genetics: Big consumer genetic data – What's in that contract? (2015) *IEEE CS Security and Privacy Workshops* 61. In South African law, click-wrap agreements are, as stated in *Stellenbosch University Law Clinic v Lifestyle Direct Group International (Pty) Ltd* [2021] 4 All SA 219 (WCC) para 48, 'agreements concluded electronically when a consumer ticks a box on a website prior to submitting an online application'. This positive act then implies an acceptance of the terms and a readiness to continue – even if the terms are not read, it indicates an awareness of the terms on the part of the consumer. However, the enforceability of such contracts is still to be determined by South African courts.
[101] Through genetic counsellors the test's purpose, its advantages and drawbacks, possible results, apprehensions regarding privacy, consequences for individuals and their families, and insurance concerns are discussed. This is done with the aim of providing patients with sufficient information to enable them to voluntarily consent to undergo testing. Jill Furnival 'The trouble with UNinformed consent in direct-to-consumer genetic testing'

testing merely requires consumers to review and agree to the testing company's terms of service, privacy policy, and research consent (if applicable) prior to purchase.[102] This is often done without consumers reading these documents and,[103] even where the terms and conditions are read, these may not be understood by consumers given the complicated subject matter and language,[104] thereby vitiating free and informed consent.[105]

In summary, consent in POPIA must be given: (1) before collection; (2) in the affirmative; (3) for specific purposes; and (4) willingly.[106] The specific consent requirement is complied with by consumers consenting to the direct-to-consumer genetic test. Although testing companies may not know what all of their potential future research may entail, POPIA provides an exception for research, which is discussed below: As long as personal information is collected legally – in other words, for a specific purpose for which there was consent and provided that the information is not published in an identifiable form[107] – there is no need to obtain further consent to conduct research using consumers' genetic data.

V COLLECTING AND PROCESSING PERSONAL INFORMATION FOR RESEARCH

(a) Collecting personal information

Because the definition of 'personal information' in POPIA[108] encompasses genetic data (established in Chapter 2), the collection of personal information is relevant to genetic research.[109] Although collection is a type of processing,[110] I first deal with collection and then move onto the broader concept of processing. To reiterate, section 13(1) of POPIA requires that personal information be collected 'for a specific, explicitly defined and lawful purpose'.[111] As demonstrated in Chapter 2, saliva and DNA do not fall within the ambit of POPIA, and

SoundRocket 19 May 2021, available at *https://soundrocket.com/trouble-informed-consent-dtc-genetic-testing/*, accessed on 8 June 2021.

[102] For example, 23andMe. 23andMe op cit note 7.

[103] Sara A Mahmoud-Davis 'Direct-to-consumer genetic testing: Empowering EU consumers and giving meaning to the informed consent process within the IVDR and GDPR frameworks' (2020) 19(1) *Wash U Global Stud L Rev* 45.

[104] Furnival op cit note 101.

[105] Mahmoud-Davis op cit note 103 at 45.

[106] Donnelly op cit note 3 at 246.

[107] Section 15(3) of POPIA.

[108] Section 1 of POPIA.

[109] As per section 13 of POPIA.

[110] As per the definition of 'processing' in section 1 of POPIA.

[111] Section 13 of POPIA further requires that the purpose of collection of personal information relate 'to a function or activity of the responsible party'.

therefore its collection is not relevant for present purposes.[112] Instead, it is only the collection of *identified* genetic data that is pertinent to POPIA[113] – which only applies once it has been sequenced, stored, or 'entered into a record'.[114]

As mentioned above, POPIA creates uncertainties surrounding whether research constitutes a specific purpose.[115] I suggest that either: (1) research is not a specific purpose in terms of POPIA; or (2) research constitutes a specific purpose under POPIA. In terms of (1), I suggest instead that the specific purpose for which consumers provide consent is the direct-to-consumer genetic test (this includes consent to gathering personal details as well as saliva samples – and may include consent to research using their genetic data). Thereafter, research falls under the exceptions and further processing provisions in POPIA. Testing companies tend not to specify a particular research study and rather list a broad range of projects for which consumers must provide consent[116] – this arguably does not constitute a specific purpose. However, to comply with POPIA's further processing provisions as well as the DoH Ethics Guidelines, it is sufficient for testing companies to state that the genetic data of consumers will be used in various types of future research that is generally described (broad consent) – such as drug discovery or studies on conditions like Parkinson's disease or Lupus.

In terms of (2), research is merely one potential use of genetic data. In the context of direct-to-consumer genetic testing, saliva samples are collected for two purposes: (1) to undergo a direct-to-consumer genetic test; and (2) for research. It cannot be said that the direct-to-consumer genetic test is the primary purpose and research is a secondary activity – one is not more important than the other. If testing companies explicitly state, or leave the possibility open, that genetic data may be used for research, then research is a purpose for which personal

[112] However, although not directly related to research, POPIA's provisions would generally apply from the time of saliva sample collection by consumers. This is because it is not only the saliva sample but also the consumer's personal information such as their name, contact details, and other health information that are collected by the testing company – these are types of 'personal information' protected by POPIA. In terms of research, this personal information is generally stored separately from the genetic data in order to ensure anonymity.
[113] The unknown genetic information contained within the DNA of a saliva sample before it is extracted and sequenced is also beyond POPIA's scope.
[114] Section 3(1)(*a*) of POPIA.
[115] As per section 13(1) of POPIA.
[116] For example, 23andMe's research page states that '[o]n average, a customer who chooses to opt into research contributes to over 230 studies on topics that range from Parkinson's disease to lupus to asthma and more'. Furthermore, 23andMe's Research Consent Document provides that research topics include 'simple traits such as hair color or freckles, serious diseases such as Parkinson's disease or diabetes, and less serious conditions such as migraine headaches'. While 23andMe does have a page that links to their published papers, participants considering partaking in research are only advised broadly of various studies that 23andMe may undertake. 23andMe 'Becoming part of something bigger' available at *https://www.23andme.com/research/*, accessed on 9 January 2021; 23andMe op cit note 7; 23andMe 'Publications' available at *https://www.23andme.com/publications/*, accessed on 9 January 2021.

information is collected. But as this may be deemed too broad, the question that arises is: What is a specific purpose in terms of research?[117] The specificity required for the purpose is undetermined.[118] If simply stating research as a purpose is too broad, does the discipline then need to be specified, for example genetic research? Or is one required to go into further detail and specify the type of genetic research or the gene to be studied, for example cancer research on the BRCA gene? Requiring this level of specificity becomes a slippery slope. Limiting research in this way would violate freedom, as well as the advancement of science and medicine. It can also be argued that such debates are inapplicable to researchers given the further processing provisions in POPIA.[119]

Additionally, POPIA requires that, when collecting personal information, data subjects are made aware of inter alia the information being collected,[120] the purpose of collection,[121] and whether there is an intention to transfer the personal information to a third party in another country and the level of protection afforded by such country,[122] unless section 18(4) of POPIA applies.[123] Section 18(4) of POPIA exempts a responsible party from complying with the provisions in section 18(1) of POPIA if inter alia the data subject consents to the non-compliance;[124] or the data subject's identity remains confidential, or the information is used for research.[125] Prima facie it appears that POPIA requires testing companies to inform consumers of their personal information to be collected, and whether it will be sent elsewhere. However, genetic data is collected for research, in which case section 18(4) of POPIA applies, and it seems that consumers do not need to be informed of the purpose for which their personal information is collected.

(b) Processing personal information

Both the collection and processing of personal information are important. In the present case, processing encompasses the collection of personal information for research and its ensuing

[117] As per section 13(1) of POPIA.
[118] POPIA does not define the term 'specific', but it does define 'consent' in section 1, and there is reference to a 'specific' purpose in section 13(1) of POPIA.
[119] Section 15 of POPIA.
[120] Section 18(1)(*a*) of POPIA.
[121] Section 18(1)(*c*) of POPIA. Similarly, regulation 5 of the Human Research Participant Regulations provide that participants must be informed of inter alia: (1) the research purpose; (2) the processes and methods; (3) the possible harms and risks of harm; (4) the anticipated advantages of the research; and (5) the choice to participate or withdraw from the research without reason or consequence.
[122] Section 18(1)(*g*) of POPIA.
[123] Section 13(2) of POPIA.
[124] Section 18(4)(*a*) of POPIA. Section 18(4) of POPIA only provides for an exemption from section 18(1) of POPIA and not the entire Act.
[125] Section 18(4)(*f*) of POPIA.

analysis, storage, use, modification, transmission, and deletion.[126] The processing of personal information is permitted only if its collection followed section 13(1) of POPIA.[127] Researchers and institutions must ensure adherence to POPIA's eight conditions for the processing of personal information, unless exempted.[128]

In addition, POPIA allows for the further processing of personal information in certain situations.[129] This would permit additional activities beyond those initially consented to, such as research, provided that such processing is 'in accordance or compatible with the purpose for which it was collected in terms of section 13' of POPIA.[130] The further processing of personal information may be consistent with the purpose of collection if, amongst other things: (1) the data subject consents to the further processing;[131] or (2) the information is used for research purposes only and will not be published in an identifiable manner.[132] Based on section 15(1) of POPIA, it appears that if personal information was not initially collected for a 'specific, explicitly defined and lawful purpose',[133] further processing is unlawful.[134] Testing companies must either rely on consumer's consenting to their genetic data being used for research to further process it for research purposes or, as genetic data is fundamentally identifiable, testing companies must ensure that the genetic data is not published – in which cases, research is permitted without requiring new consent.

The DoH Ethics Guidelines briefly discuss the secondary use of human biological material or data.[135] Often where biological material is collected for diagnostic or therapeutic

[126] Staunton et al op cit note 81 at 469.
[127] Thaldar & Townsend op cit note 80 at 174.
[128] As mentioned in Chapter 5, these eight conditions are: (1) accountability (section 8 of POPIA); (2) processing limitation (sections 9 to 12 of POPIA); (3) purpose specification (sections 13 and 14 of POPIA); (4) further processing limitation (section 15 of POPIA); (5) information quality (section 16 of POPIA); (6) openness (sections 17 and 18 of POPIA); (7) security safeguards (sections 19 to 22 of POPIA); and (8) data subject participation (sections 23 to 25 of POPIA). See also, Thaldar & Townsend op cit note 42 at 12.
[129] Section 15(1) of POPIA; Joanne van Harmelen, Ridwaan Boda & Rakhee Dullabh 'Genetic information: A new resource to be mined?' *International Law Office* 20 October 2020, available at *https://www.internationallawoffice.com/Newsletters/Healthcare-Life-Sciences/South-Africa/ENSafrica/Genetic-information-a-new-resource-to-be-mined*, accessed on 16 February 2021.
[130] Section 15(1) of POPIA. This is determined by examining: (1) the relationship between the purposes of collection and further processing; (2) the nature of the information; (3) the implications of further processing on the data subject; (4) how the information was collected; and (5) contractual rights and duties between the parties. Subsections 15(2)(*a*) to (*e*) of POPIA.
[131] Section 15(3)(*a*) of POPIA.
[132] Section 15(3)(*e*) of POPIA. The operative word used in this section is 'publish'. Thus, the genetic data can be entirely identifiable – as long as it is not published. See also, Ciara Staunton, Rachel Adams, Edward S Dove et al 'Ethical and practical issues to consider in the governance of genomic and human research data and data sharing in South Africa: A meeting report' (2019) *AAS Open Research* 5; Staunton et al op cit note 81 at 469.
[133] In terms of section 13(1) of POPIA.
[134] Thaldar & Townsend op cit note 42 at 13.
[135] DoH op cit note 49 at 32.

purposes, the informed consent does not cater for use in research, and thus the question is whether use of biological material or data for unforeseen research requires new informed consent. Where there is no broad consent encompassing research, the DoH Ethics Guidelines recommend that: (1) utilising collected material for diagnostic or clinical purposes for research entails a determination of whether the subsequent use of biological material or data was considered in the previous consent which, if found to fall within the ambit of the research proposal, does not need new consent; (2) if the research proposal's purview differs, new consent may be necessary; (3) where samples have been anonymised and the research results would not pose a risk of harm, there is no need to obtain new consent; (4) if there is a connection to identifiers, but it is not given to the researchers and the research results would not pose a risk of harm, new consent is unneeded; (5) the individual in possession of the link or code should sign a written agreement prohibiting the release of the identifiers to the researchers; and (6) where samples are identifiable, a case-by-case determination is required by Research Ethics Committees (RECs) to determine if a full or expedited review is necessary.[136]

While the DoH Ethics Guidelines only require a consideration of whether new informed consent is necessary when there is no broad consent for research, POPIA only permits specific consent. The DoH Ethics Guidelines refer to biological material that was initially collected for diagnostic or therapeutic purposes. In terms of POPIA, such a purpose for collection differs from the purpose of research and thus new consent would be required. Both the DoH Ethics Guidelines and POPIA are similar in terms of the anonymisation or de-identification of data, in which case there is no need for new consent. Where the DoH Ethics Guidelines, in terms of identifiable samples, require a case-by-case determination of whether new consent is necessary, POPIA does not require new consent if the genetic data is for research.

Additionally, section 32(5) of POPIA allows health information regarding inherited characteristics to be processed for, amongst others, 'research activity'.[137] Given that direct-to-consumer genetic testing aims to identify an individual's predisposition to various genetic conditions, and because research undertaken by testing companies studies genetic diseases and conditions in order to discover more effective treatments, I suggest that this information relates to inherited characteristics and can therefore be processed, in terms of POPIA, for research.[138]

---

[136] Ibid at 32.

[137] Rachel Adams, Fola Adeleke, Dominique Anderson et al 'POPIA Code of Conduct for research' (2021) 117(5/6) *S Afr J Sci* 6–7; van Harmelen et al op cit note 129.

[138] On the other hand, section 33 of POPIA, relating to authorisations regarding the processing of personal information for criminal behaviour or biometric information does not include research. As Chapter 2 established

Exemptions may also be granted by the Information Regulator,[139] allowing the processing of personal information that has been excluded under POPIA.[140] One such exemption is if processing is in the public interest and substantially exceeds intruding into a data subject's privacy.[141] Section 37(2)(*e*) of POPIA provides that the public interest includes 'research activity'.[142] As research is deemed by POPIA to be in the public interest,[143] this means that, should the Information Regulator grant an exemption, processing genetic data for this purpose will be permitted by POPIA – but this only applies in the case of personal information, not in the case of *special* personal information.

(c) Processing special personal information

Section 26 of POPIA refers to 'special personal information' and contains prohibitions on its processing. Of relevance is section 26(*a*) of POPIA, which states that '[a] responsible party may, subject to section 27, not process personal information concerning…health or sex life or biometric information of a data subject'.[144] Though section 26 of POPIA prohibits the processing of certain special personal information (inclusive of genetic data),[145] section 27 of POPIA contains exceptions to this. The processing of special personal information is not prohibited if inter alia: (1) the data subject consents to processing;[146] (2) the processing is for 'historical, statistical or research purposes';[147] or (3) processing has been authorised by the Information Regulator with suitable safeguards.[148] The most straightforward way to achieve this is to obtain consumers' consent. This is generally done when consumers consent to undergo a direct-to-consumer genetic test as there is often an option to consent to their genetic data being used in research.[149]

---

that genetic data is a type of biometric information that falls under the definition of 'personal information' in section 1 of POPIA, prima facie this means that biometric information cannot be processed for research.

[139] Section 39 of POPIA provides for the establishment of the Information Regulator, who is a juristic person that has jurisdiction throughout South Africa. Section 40 of POPIA defines the duties of the Information Regulator, which include educating, monitoring and enforcing compliance, consulting with parties, handling complaints, conducting research, and overseeing codes of conduct.

[140] Section 37(1) of POPIA. Such exemptions are granted by the Information Regulator through a notice in the Government Gazette.

[141] Section 37(1)(*a*) of POPIA.

[142] Section 37(2)(*e*) of POPIA.

[143] Ibid.

[144] Section 26(*a*) of POPIA.

[145] This is because genetic data is a type of personal information, special personal information, and biometric information.

[146] Section 27(1)(*a*) of POPIA.

[147] Section 27(1)(*d*) of POPIA.

[148] Section 27(2) of POPIA.

[149] Some companies also give consumers the option to decide to participate in research at a later stage too, for example 23andMe. See, 23andMe op cit note 7.

In terms of (2), that the processing of special personal information is permitted for inter alia 'research purposes', this requires two conditions to be met before relying on this legal ground: (a) the research is in the public interest and processing is fundamental to achieving its purpose; or (b) obtaining consent is impractical or would entail a 'disproportionate effort', provided that adequate assurances are implemented to protect the privacy of data subjects.[150] In terms of (a), distinct from the exemption for personal information, processing special personal information for research purposes does not automatically fall within the public interest and it must be shown that this is in fact the case.[151] In terms of (b), even if testing companies can show that the processing of genetic data is for research which will serve to assist in medical and therapeutic advances in the public interest, it is difficult to see how obtaining the consent of consumers for such research would be impossible as testing companies generally have terms and conditions to which consumers must agree before proceeding with testing, and consent to research is often included in such conditions. However, the above requirements only pertain to special personal information and there must still be compliance with the other conditions in POPIA for the processing of genetic data for research by testing companies to be permissible.[152]

For 'special personal information',[153] POPIA requires inter alia the data subject's consent.[154] But this requirement is voided where data has 'deliberately been made public by the data subject'[155] and, in such a case, it may be collected from another source,[156] and further processed.[157] POPIA does not contain guidance on when information is deemed to have been made 'public'. In terms of direct-to-consumer genetic testing, consumers may share their test results (and sometimes raw data) online – either on social media, forums, or testing company websites. If, by doing so, this information is deemed to have been made public, then consent is not needed from data subjects in order to process their genetic data.

[150] Section 27(1)(*d*) of POPIA. See also, Thaldar et al op cit note 1 at 4.
[151] Although other statutes deal with public interest, case law is yet to emerge on the meaning of public interest in terms of POPIA. Thaldar & Townsend op cit note 42 at 17.
[152] Ibid at 16.
[153] Section 26 of POPIA.
[154] Section 27(1)(*a*) of POPIA.
[155] Section 27(1)(*e*) of POPIA.
[156] Section 12(2)(*a*) of POPIA. However, in this instance the data subject nevertheless is entitled to be informed of the collection of the data. Donnelly op cit note 3 at 226–7.
[157] Section 15(3)(*b*) of POPIA.

VI PRIVACY ISSUES IN RESEARCH

Privacy issues in direct-to-consumer genetic testing relate to the safeguarding of confidential data through means of de-identification and the storage of data – which I deal with briefly below. The main source of law dealing with the privacy of genetic data is POPIA. All data, including genetic data, must remain confidential.[158] The Human Research Participant Regulations provide that health research with human participants must, amongst other things, respect the right to privacy.[159] The Health Professions Council of South Africa (HPCSA) has established various guidelines for research, including matters related to privacy and confidentiality, which they express must be guarded when personal information is collected, used, stored, or destroyed for research.[160] In terms of POPIA, it is the duty of the responsible party to guard the confidentiality and integrity of personal information through reasonable steps aimed at preventing: (1) loss, damage, or unintended destruction; and (2) illegal access or processing.[161]

[158] Confidentiality denotes the obligation to ensure that information remains classified as far as possible in order to identification of an individual – specifically in the context of research. Confidentiality concerns the agreement between the researcher and the participant regarding the handling, management, and dissemination of identifiable information. Information used for research must be protected against unlawful access, disclosure, use, modification, theft, or loss. Privacy relates to people, while confidentiality involves data. South African Medical Research Council *The South African Medical Research Council Guidelines on the Responsible Conduct of Research* (2017) 10. See also, Sharon Young 'Privacy versus confidentiality' *Office of the Saskatchewan Information and Privacy Commissioner* 8 December 2017, available at *https://oipc.sk.ca/privacy-versus-confidentiality/#:~:text=In%20terms%20of%20information%2C%20privacy,used%2C%20and%2For%20disclosed.&text=Confidentiality%20is%20the%20duty%20to,only%20to%20the%20extent%20possible.*, accessed on 20 April 2020; Association for the Accreditation of Human Research Protections 'Privacy vs. confidentiality – What's the difference?' (2009) *University of Kentucky* 1; DoH op cit note 49 at 15 & 53.

[159] In terms of the Human Research Participant Regulations, participants must also be informed of inter alia the degree of protection to their privacy and confidentiality. Privacy refers to an individual's right of control over the collection, use, and disclosure of their personal information. It relates to 'persons and to their interest in controlling the access of others to themselves'. Privacy is also involves who can access personal information, and covers possible harms to individuals from the collection, use, and disclosure of personal information for research. The right to privacy is protected in section 14 of the Constitution, as well as by common law. Regulations 2(h) and 5(g) of the Human Research Participant Regulations; Joan E Sieber 'Privacy and confidentiality: As related to human research in social and behavioral science' (2001) *Online Ethics Center at the National Academy of Engineering*; Department of Health op cit note 49 at 15 & 54; ASSAf op cit note 64 at 55.

[160] Similarly, the DoH Ethics Guidelines state that researchers must ensure privacy and confidentiality in the course of research, including the publication of results. Health Professions Council of South Africa (HPCSA) *Guidelines for Good Practice in the Health Care Professions: General Ethical Guidelines for Health Researchers (Booklet 13)* (2016) 3; DoH op cit note 49 at 15.

[161] Section 19(1) of POPIA. This requires the responsible party to: (1) determine all reasonably foreseeable risks to personal information, both internal and external; (2) create and manage suitable safeguards against these risks; (3) routinely check the effectiveness of the safeguards; and (4) assure that safeguards are regularly revised for new risks or inadequacies in previous safeguards. Section 19(2) of POPIA.

(a) De-identification and anonymisation

Genetic research entails balancing the sharing of data in order to improve scientific progress with protecting individual privacy.[162] De-identification is a method commonly employed when disclosing sensitive information.[163] It entails removing certain personal identifiers, making the discovery of the source more challenging and therefore less likely to cause harm.[164] But assuring the confidentiality of research participants, or those genetically related to them is problematic, given the uniqueness of genetic information combined with increased data sharing. Once an individual has been re-identified, it is possible that additional personal information may be disclosed. An issue for research involving the sharing of genetic data is whether it is possible to merely *distinguish* or actually *identify* an individual from sequence data.[165] Malin et al contend that although genomic sequence data is generally 'highly distinguishing',[166] this is 'insufficient to claim that the corresponding individual's privacy will actually be compromised'.[167] Because sequence data needs to be paired with the data of a named individual, for privacy to be infringed, there must be a means of linking the identified and de-identified data. It is then understood that an individual's privacy interest does not come into play where information is anonymous.[168]

Although 'de-identification' and 'anonymisation' are sometimes used interchangeably, the terms do differ. While both entail the removal of personal identifiers, where anonymisation makes re-identifying the source almost impossible,[169] de-identification is reversible and there is a possibility of re-identification.[170] 'De-identification', as used in POPIA,[171] involves

[162] National Human Genome Research Institute 'Privacy in genomics' *NIH* 24 February 2020, available at *https://www.genome.gov/about-genomics/policy-issues/Privacy*, accessed on 3 March 2020.
[163] Xinghua Shi & Xintao Wu 'An overview of human genetic privacy' (2017) 1387(1) *Ann N Y Acad Sci* 3.
[164] Pike op cit note 4 at 2016–7; Cacchio op cit note 4 at 231.
[165] Jane Kaye 'The tension between data sharing and the protection of privacy in genomics research' (2012) 13 *Annu Rev Genomics Hum Genet* 10–1.
[166] Bradley Malin, David Karp & Richard H Scheuermann 'Technical and policy approaches to balancing patient privacy and data sharing in clinical and translational research' (2010) 58(1) *J Investig Med* 4.
[167] Ibid.
[168] Kaye op cit note 165 at 10.
[169] Anonymisation commonly involves the permanent removal of personal identifiers from data, so that it cannot be re-linked to an individual. Recital 26 of the GDPR defines anonymous information as 'information which does not relate to an identified or identifiable natural person or to personal data rendered anonymous in such a manner that the data subject is not or no longer identifiable'. Kaye op cit note 165 at 10; Educause 'Guidelines for data de-identification or anonymization' July 2015 available at *https://www.educause.edu/focus-areas-and-initiatives/policy-and-security/cybersecurity-program/resources/information-security-guide/toolkits/guidelines-for-data-deidentification-or-anonymization*, accessed on 21 May 2021. See also, Lee Swales 'The Protection of Personal Information Act and data de-identification' (2021) 117(7/8) *S Afr J Sci* 2.
[170] Unlike anonymisation, de-identification entails removing personally identifying information, which has the potential to be re-linked to the data. Kaye op cit note 165 at 10; Swales op cit note 169 at 2; Pike op cit note 4 at 2018.
[171] Sections 1 and 6(1)(*b*) of POPIA.

removing all personal information identifying a data subject.[172] In terms of health research, data is de-identified when an individual in a research institution is unable to utilise or link data to identify a data subject through a 'reasonably foreseeable method'.[173] Although providing a definition, there is no specific provision in POPIA dealing with de-identification. As suggested by Swales, one way to solve this is for the Information Regulator to provide guidance, or for a code of conduct to communicate best practice.[174]

Testing companies, specifically when using genetic data for research, may emphasise the fact that such information has been de-identified to protect consumers' privacy. POPIA does not apply to de-identified[175] personal information that is incapable of being re-identified.[176] Given its de-identification,[177] genetic data may not meet the definition of 'personal information'[178] – namely, information that relates to 'an *identifiable*, living, natural person' (own emphasis), and thus POPIA may not apply.[179] However, the question is: Can genetic data

[172] See, Donnelly op cit note 3 at 228. The DoH Ethics Guidelines do refer to 'anonymous' data, specimens, or information and 'anonymised' information. An 'anonymous data or specimen' refers to 'data or material without any overt identifying information or link to a specific participant or donor' and 'anonymised information' means information that is 'irrevocably stripped of direct identifiers'. An 'identifier' refers to names, addresses, folder numbers, or biometric identifiers (such as fingerprints) that make it possible to identify individuals. While these terms refer to an end result, de-identification is seen to denote a process. The term 'pseudo-anonymisation' is mentioned by Adams et al in their article on a code of conduct for research in terms of POPIA. Adams et al make reference to article 4 of the GDPR, which uses 'pseudoanonymisation' to refer to information that is capable of re-identification, and defines it as 'the processing of personal data in such a manner that the personal data can no longer be attributed to a specific data subject without the use of additional information provided, that such additional information is kept separately and is subject to technical and organisational measures to ensure that the personal data are not attributed to an identified or identifiable natural person'. Adams et al op cit note 137 at 3; DoH op cit note 49 at 30 & 54.

[173] As per the definition of 'de-identify' in section 1 of POPIA. To avoid the application of POPIA to data, it is imperative that researchers remove all personal information identifying data subjects, and that re-identification is impossible. Even if a single individual in a research institute is able to identify a data subject (through accessing a master file or linking data) data is not de-identified and POPIA applies. Swales op cit note 169 at 2.

[174] Ibid.

[175] Section 6(1)(*b*) of POPIA. Section 1 of POPIA states that de-identify 'in relation to personal information of a data subject, means to delete any information that –

(a) identifies the data subject;
(b) can be used or manipulated by a reasonably foreseeable method to identify the data subject; or
(c) can be linked by a reasonably foreseeable method to other information that identifies the data subject'.

[176] Ibid. Section 1 of POPIA states that re-identify 'in relation to personal information of a data subject, means to resurrect any information that has been de-identified, that –

(a) identifies the data subject;
(b) can be used or manipulated by a reasonably foreseeable method to identify the data subject; or
(c) can be linked by a reasonably foreseeable method to other information that identifies the data subject'.

[177] Testing companies can, and do, de-identify genetic *information* and genetic *data*. For genetic information, see Consumer Reports *Direct-to-Consumer Genetic Testing: The Law Must Protect Consumers' Genetic Privacy* (2020) 6. For genetic data, see Grayson L Ruhl, James W Hazel, Ellen Wright Clayton et al 'Public attitudes toward direct to consumer genetic testing' (2019) *AMIA Annu Symp Proc* 781. For both genetic information and genetic data, see Yaniv Erlich, James B Williams, David Glazer et al 'Redefining genomic privacy: Trust and empowerment' (2014) 12(11) *PLoS Biol* 1.

[178] Section 1 of POPIA.

[179] Ibid. This also applies in terms of the definition of 'biometrics' in section 1 of POPIA, where the results of DNA analysis, the obtained genetic information and data, may identify an individual.

be truly de-identified? Although de-identification is utilised as a justification in both the consensual and non-consensual use and sharing of genetic data,[180] it has been found that complete de-identification of genetic data is impossible. Records, databases, and various other techniques are available, and may be used in triangulation to re-identify individuals.[181] Additionally, all DNA, except for that of identical twins, is unique and, because of this, can be traced back to a specific individual.[182] Therefore, as genetic data is inherently identifiable, I suggest that it always falls within the ambit of POPIA – unless an individual intentionally makes the information public.[183]

Regulation 13(h) of the Human Biological Material Regulations provides that information used for research should be treated anonymously.[184] The DoH Ethics Guidelines articulate that research utilising only secondary anonymous information or anonymous human biological material is generally not required to undergo formal ethics review, as long as no identifiable information is produced.[185] Some testing companies claim to use anonymised data in research.[186] But in de-identifying data to the point of anonymisation, identifiability must be balanced against its usefulness. The removal of too much data may decrease its scientific value and hamper research.[187] The DoH Ethics Guidelines also recognise that anonymisation may

[180] Pike op cit note 4 at 2016.

[181] The idea that genetic data can be completely anonymised is not absolute and it is possible to link samples to an individual. There has been an increasing number of examples of allegedly de-identified data being re-identified by combining multiple, disparate databases. This can be done through statistical methods with information about an individual's phenotypic characteristics, such as age, gender, and disease status. This not only holds implications for the individual concerned, but their close relatives too. The insufficiency of de-identification of genetic data was further demonstrated by Gymrek et al in 2013. Using only the date of birth and state of residence, the researchers identified the source simply by juxtaposing sections of an individual's genetic sequence with those shared in publicly accessible databases – allowing anyone possessing internet research skills to repeat this process. Pike op cit note 4 at 2016–7; Cacchio op cit note 4 at 231; ASSAf op cit note 64 at 57; RB Altman 'Direct-to-consumer genetic testing: Failure is not an option' (2009) 86(1) *Clin Pharmacol Ther* 16; David B Resnik 'Direct-to-consumer genomics, social networking, and confidentiality' (2009) 9(6-7) *Am J Bioeth* 46; Melissa Gymrek, Amy L McGuire, David Golan et al 'Identifying personal genomes by surname inference' (2013) 339(6117) *Science* 321–4; Erlich et al op cit note 177 at 1.

[182] S de Wet, H Oosthuizen & J Visser 'DNA profiling and the law in South Africa' (2011) 14(4) *PER/PELJ* 173–4. See also, Linda Nordling 'A new law was supposed to protect South Africans' privacy. It may block important research instead' *Science* 22 February 2019, available at *https://www.sciencemag.org/news/2019/02/new-law-was-supposed-protect-south-africans-privacy-it-may-block-important-research#:~:text=South%20Africa's%20current%2C%202015%20research,%E2%80%9Caware%20of%20the%20purpose.%E2%80%9D*, accessed on 17 February 2020; *S v Orrie* [2003] ZAWCHC 63 para 18; *S v Nyembe* 2014 (1) SACR 105 (GSJ) para 7; *S v Maqhina* 2001 (1) SACR 241 (T).

[183] Section 27(1)(*e*) of POPIA.

[184] Regulation 13(h) of the Human Biological Material Regulations.

[185] DoH op cit note 49 at 9.

[186] 90% of AncestryDNA consumers agreed to share 'their anonymized data for research purposes'. Heidi C Howard, Bartha Maria Knoppers & Pascal Borry 'Blurring lines: The research activities of testing companies raise questions about consumers as research subjects' (2010) 11(8) *EMBO Rep* 580; Pike op cit note 4 at 1996.

[187] The purpose of using genetic data in research is to examine the role of genes (rather than environment or lifestyle) in disease, but anonymising this data may conceal vital information. Pike op cit note 4 at 2018.

have implications on research participants by impeding the disclosure of significant findings or the ability to withdraw material from research.[188]

To conclude, although POPIA contains safeguards to protect privacy, once data has been de-identified, POPIA no longer applies. However, given that genetic data cannot be truly de-identified and remains identifiable to a certain extent, I suggest that POPIA continues to apply and will govern genetic research undertaken by testing companies in South Africa.

(b) The fate of an individual's genetic data

What happens to consumers' sample and genetic data following a direct-to-consumer genetic test is usually contained in the privacy policies of testing companies,[189] although the information contained therein tends to vary.[190]

POPIA requires that personal information not be kept for longer than needed to accomplish the purpose of collection or processing. But POPIA permits longer retention periods if records are for research, provided that the responsible party has suitable protections to ensure that the data is only used for such purposes.[191] In terms of direct-to-consumer genetic testing, this means that as soon as the testing process is complete, samples and data must be destroyed. However, if the testing company conducts research using genetic data and consumers have consented to this, data may be stored for a longer period.

---

[188] DoH op cit note 49 at 31.

[189] Currently, 23andMe and AncestryDNA require express consent from consumers prior to sharing their data with third parties. Rani Molla 'Why DNA tests are suddenly unpopular' *Vox* 13 February 2020, available at *https://www.vox.com/recode/2020/2/13/21129177/consumer-dna-tests-23andme-ancestry-sales-decline*, accessed on 15 April 2020.

[190] Companies such as 23andMe give consumers the freedom to decide on the extent of their participation, such as whether their saliva samples or DNA are stored. 23andMe also states that unless otherwise stated, samples are stored for at least one year, but not more than ten years, at their certified laboratory. South African testing company GeneWay retains samples for a certain time period stipulated by law, but are destroyed thereafter. Two other South African testing companies, HomeDNADirect and EasyDNA, state that leftover samples are destroyed in accordance with the laboratory's standard operating procedures and legal requirements. Cacchio op cit note 4 at 229; 23andMe 'Biobanking consent document' 2020, available at *https://www.23andme.com/about/biobanking/*, accessed on 22 June 2020; GeneWay 'FAQ – Frequently asked questions' 2020 available at *https://www.geneway.co.za/faq-frequently-asked-questions*, accessed on 22 June 2020; HomeDNADirect 'Terms and conditions – General' *HomeDNADirect* 24 May 2018, available at *https://www.homednadirect.co.za/terms-and-conditions/*, accessed on 23 June 2020; EasyDNA 'Terms and conditions' 24 May 2018 available at *https://www.easydna.co.za/terms-of-service/*, accessed on 22 June 2020.

[191] Section 14(2) of POPIA. It must be questioned what would be considered a 'record' in terms of genetic research. the most applicable category under section 1 of POPIA would be 'information produced, recorded or stored by means of any tape-recorder, computer equipment, whether hardware or software or both, or other device, and any material subsequently derived from information so produced, recorded or stored'. As DNA is extracted from the saliva sample and analysed (presumably using some sort of computer or machine) and the resulting genetic data is then stored in a database, I suggest that genetic data is part of a record.

The DoH Ethics Guidelines recommend that proposals for research studies must include an explanation of the manner in which data records will be protected, the duration of their retention, and the party accountable for storage and/or disposal.[192] The HPCSA promotes the secure storage of specimens and data used for research.[193] It is interesting to note that, although not binding, the HPCSA defines the minimum period for which data should be stored – two years after publication or six years where there was no publication of the findings.[194] Testing companies are not always explicit in addressing the period of time that data used in research will be stored. While there is the possibility of general data storage provisions also applying to data used in research, the lack of specific provisions may mean that such data will be stored indefinitely, save for the consumer requesting its deletion.[195] However, with the various laws and guidelines in South Africa, this may not be the case.

As per the Human Biological Material Regulations, 'genetic material records and other individually identifiable or related health information' must be destroyed as soon as their purpose is achieved.[196] In terms of research, material and data must be destroyed once the study is complete. But if material is stored for a longer period or relates to research findings, written informed consent is required. As the Human Biological Material Regulations specifically refer to individually *identifiable* information, it is assumed that its provisions do not apply where information has been de-identified.

Although policies appear to differ between testing companies, the common approach is that genetic data will be stored for certain time periods (sometimes stipulated by law), unless the consumer consents to their genetic data being used for research, in which case it may be stored indefinitely, unless the consumer requests that it be destroyed. Genetic data and reports may be stored indefinitely by some testing companies even if no research is being conducted.[197]

[192] DoH op cit note 49 at 20.
[193] HPCSA op cit note 160 at 11–2.
[194] Ibid at 11–2.
[195] Linnea I Laestadius, Jennifer R Rich & Paul L Auer 'All your data (effectively) belong to us: Data practices among direct-to-consumer genetic testing firms' (2017) 19(5) *Genet Med* 517.
[196] Regulation 13 of the Human Biological Material Regulations. The meaning of 'genetic material records' unclear, but it is assumed that this relates to anything dealing with genetic material – such as samples, DNA, and genetic data.
[197] Two South African testing companies that appear to do this are GeneWay and Muhdo (The Wellness Revolution). The Wellness Revolution 'FAQ' available at *https://thewellnessrevolution.co.za/faq/*, accessed on 22 June 2020.

VII TRANSFERRING GENETIC DATA FOR THE PURPOSES OF RESEARCH

It is not only possible for research to be conducted within South Africa, but genetic data may be sent elsewhere for such purposes – especially where testing companies are based in other jurisdictions (examined in Chapter 6). As mentioned above, individuals who consent to their personal information being used in research must be informed of whether there is an intention to transfer this information to a third party in another country and the standard of protection that the country offers,[198] unless inter alia their personal information will be utilised in research.[199]

In 2018, the SA MTA was published. A Material Transfer Agreement (MTA) is a written agreement between two parties; the provider of human biological material and the recipient that plans on conducting research using the material.[200] A MTA, based on the SA MTA, must be utilised when biological material is transferred, for research purposes, where at least one party is in South Africa[201] – thus obliging parties in other jurisdictions to use the SA MTA.[202]

The SA MTA contains provisions for transferring human biological material and associated data into, out of, and within South Africa[203] – although it is only a framework that serves to guide parties.[204] The SA MTA applies to the transfer of: (1) human biological material *only*; and (2) *both* human biological material and associated data. Where only data is transferred, there is no legal obligation to use the SA MTA.[205] However, data transfer may be controlled by a MTA or a data transfer agreement (DTA).[206] The SA MTA's definition of 'data' (provided in Chapter 2)[207] excludes transfers of biological material or data for purposes other

[198] Section 18(1)(*g*) of POPIA.
[199] Section 18(4)(*f*) of POPIA.
[200] A MTA also defines the rights and duties of parties. Thaldar et al op cit note 1 at 2; Thaldar op cit note 49 at 2.
[201] Thaldar et al op cit note 1 at 6; Thaldar op cit note 49 at 1.
[202] Thaldar et al op cit note 1 at 2.
[203] Thaldar & Townsend op cit note 42 at 7.
[204] Thaldar et al op cit note 1 at 6.
[205] This is because The SA MTA refers to human biological material and associated data collectively as 'Materials'. Thaldar et al note that capitalising the first letter indicates that the word is bound by a definition rather than its typical meaning. However, the notice in the Government Gazette mentions 'biological material' in lowercase, signalling that it carries its traditional meaning. This means that the SA MTA must only be used for transfers consisting of 'biological material' in its ordinary meaning. Thus, if the transfer only involves data, the parties are not obliged to use the SA MTA as a framework. Para 2.13 of the SA MTA; ibid at 6; Thaldar & Townsend op cit note 42 at 7.
[206] Thaldar et al op cit note 1 at 2.
[207] Para 2.7 of the SA MTA defines data as 'any information, including personal information in any form derived directly or indirectly during the conduct of research or clinical care'.

than research.[208] As testing companies transfer saliva, DNA, and/or genetic data (depending on the circumstances) for research, the SA MTA applies.

In contrast to other legal instruments, the SA MTA establishes a type of 'dynamic consent'.[209] Firstly, the SA MTA requires informed consent to be obtained from individuals for their biological material and associated data to be provided to recipients for a research study,[210] and thereafter it necessitates 'an on-going information sharing process'[211] permitting participants to consent to 'whether and how' their biological material and data are to be used.[212] Secondly, further use of an individual's materials for research beyond the original informed consent must have approval from a HREC that is a party to the MTA,[213] and additional informed consent must be obtained for each research project, but only where 'reasonably possible'.[214] Therefore, the broad secondary use of genetic data may be in contravention of the SA MTA unless the requirements in POPIA regarding the collection and further processing of personal information for research are adhered to.[215] The SA MTA also departs from the DoH Ethics Guideline's preference to use broad consent. In instances of conflict, it is a rule of interpretation that the later legislation supersedes the earlier legislation.[216] Thus, where human

[208] For example, where an overseas laboratory sends genetic data and test results back to a South African testing company to enable them to generate reports for consumers. Although relevant to biological material and not genetic data, which is the focus of the present analysis, it also appears that the SA MTA only applies to 'biological material for use in research or clinical trials', as per the preamble of the SA MTA, and would therefore not be required for the transfer of biological material for other purposes – for example, where a consumer in South Africa undergoes a direct-to-consumer genetic test and sends their saliva sample to a company overseas, or where a South African testing company sends saliva samples to an overseas laboratory for analysis, or where genetic data resulting from the DNA analysis is sent from an overseas laboratory back to a testing company in South Africa where it is not going to be used for research.

[209] 'Dynamic consent' is an approach to informed consent, allowing personalised communication and engagement with participants in which they can opt-in or opt-out of subsequent activities that utilise their biological material and data, and allows them to check consent options. Dynamic consent often utilises digital platforms, allowing the novel presentation of information (such as videos) and permitting participants to enter information, and fill out online questionnaires, themselves. This type of consent is 'dynamic' because it enables continuous interaction over a prolonged period and can adapted to suit the research and participant views, including when consent and communication are required at certain stages. Thaldar & Townsend op cit note 42 at 3; Harriet JA Teare, Megan Prictor & Jane Kaye 'Reflections on dynamic consent in biomedical research: the story so far' (2021) 29 *Eur J Hum Genet* 649.

[210] Para 10.1 of the SA MTA.

[211] Para 2.12 of the SA MTA.

[212] Thaldar & Townsend op cit note 42 at 7.

[213] Para 2.12 of the SA MTA.

[214] Paras 4.3 and 10.3 of the SA MTA. Para 2.12 of the SA MTA defines informed consent as 'a formal agreement that a Donor (with legal capacity to do so) signs to give permission for donation of Materials, after being informed about the project and includes an on-going information sharing process which allows a Donor to consent to participate and determine whether and how their Materials will be utilised in the Project, as approved by the HREC from time to time'.

[215] van Harmelen et al op cit note 129.

[216] If there are two different conflicting pieces of legislation, a solution must be sought by reading them together. If a solution cannot be found where both enactments deal with the same issue, the later one will take precedence over the earlier one. *New Modderfontein Gold Mining Company v Transvaal Provincial Administration* 1919 AD 367 at 400. See also, CJ Botha *Statutory Interpretation* 5 ed (2012) 136.

biological material (and data) is transferred, bringing the SA MTA into application, there must be adherence to its stricter consent provisions, as opposed to the comparably lenient provisions permitting broad and tiered consent in the DoH Ethics Guidelines.[217]

In cases where only genetic data is transferred for research, there is no obligation to follow the SA MTA.[218] However, POPIA applies to the transfer of genetic data. As mentioned in Chapter 6, where genetic data is to be transferred from South Africa to a foreign jurisdiction that lacks an adequate level of protection, prior authorisation must be obtained from the Information Regulator – unless a code of conduct exists.[219] Where a South African research institution plans to transfer personal information to a foreign research institution, without the specific consent of research participants, such a transfer may occur if the laws of the foreign country provide a suitable level of protection, or if there is an agreement between the parties.[220] As genetic data may be transferred by testing companies for research, a standard DTA may be necessary. A DTA would include provisions encompassing data ownership, accepted use of data, the sharing of data with third parties, ascribing credit, and intellectual property.[221] This can establish that an adequate level of protection exists.[222] However, data transfer is amiss in the current SA MTA. This is something that should be addressed.

VIII CAN DIRECT-TO-CONSUMER GENETIC TESTING COMPANIES CONDUCT RESEARCH USING GENETIC DATA COLLECTED FROM CONSUMERS?

In the above sections, I analysed the pertinent aspects of research conducted on the genetic data of consumers by testing companies, as well as the South African statutes that govern it. It is now necessary to consider various scenarios in which research may occur, and the implications of such for direct-to-consumer genetic testing, based on South Africa's extant law.

(a) Scenario (1)

The testing company is based in South Africa and research is conducted in the country. Given that the research and genetic data remain within South Africa, local laws apply – POPIA governs the collection and processing of this personal information. Research is allowed,

[217] Thaldar & Townsend op cit note 42 at 7.
[218] This is because the SA MTA applies where biological material, or both biological material and genetic data are transferred for research, and is therefore relevant to saliva samples and extracted DNA.
[219] Section 57(3) of POPIA. See also, Donrich Thaldar & Beverley Townsend 'Protecting personal information in research: Is a code of conduct the solution?' (2021) 117(3/4) *S Afr J Sci* 1.
[220] Ibid at 1–2.
[221] ASSAf op cit note 64 at 79.
[222] Thaldar & Townsend op cit note 219 at 1–2.

provided that consent – for the specific purpose of testing in terms of POPIA (and possibly for research if this is deemed to be a specific purpose), and broadly for research under the DoH Ethics Guidelines – is obtained from consumers. As the testing company is in South Africa, the SA MTA is not applicable – unless one testing company sequences the DNA and another conducts research. Further research is permitted if the genetic data is not published in an identifiable form and steps are taken to protect individual privacy. As it is questionable whether genetic data can truly be de-identified, POPIA will continue to govern this form of personal information and the research conducted using it.

(b) Scenario (2)

The testing company is based in another jurisdiction, where saliva samples are sent by South African consumers and where the processing of samples and research using genetic data occurs abroad. Although POPIA is committed to protecting the privacy of data subjects in South Africa,[223] because the testing company is based overseas, POPIA does not apply. However, the testing company must comply with the laws governing research in the country in which it is domiciled.[224]

(c) Scenario (3)

The testing company is based in South Africa, but saliva samples are sent overseas for DNA extraction, sequencing, and analysis. As genetic data is generated overseas, POPIA does not apply. However, the SA MTA governs the transfer of saliva samples from South Africa abroad. If the South African testing company, having obtained the genetic data from overseas, conducts research locally, POPIA and other research considerations are relevant. POPIA also applies if the testing company decides to send the genetic data overseas for research.

(d) Scenario (4)

The testing company is based in South Africa and DNA extraction happens locally, but DNA sequencing and analysis occur in another jurisdiction. Like scenario (3) above, genetic data is generated abroad through analysing the extracted DNA and, as such, POPIA does not apply. Rather, the laws of the jurisdiction in which the testing company is based govern the genetic data and research, if this is undertaken overseas. But the SA MTA is relevant to extracted DNA

[223] Thaldar & Townsend op cit note 80 at 172.
[224] John Giles 'Must I comply with the POPI Act?' *Michalsons* 12 February 2020, available at *https://www.michalsons.com/blog/must-i-comply-with -the-popi-act/41827*, accessed on 3 March 2021.

and, given that this is transferred from South Africa to another party abroad for research, the SA MTA applies. Like scenario (3) above, once the South African testing company receives the genetic data from overseas and undertakes its own research, POPIA and other research provisions are pertinent. If the testing company sends the genetic data to another jurisdiction for research, POPIA governs this transfer.

(e) Conclusion on the scenarios

The legal implications of testing companies conducting research using the genetic data gathered from consumers who undergo direct-to-consumer genetic testing depend on where the research is undertaken. If the research is conducted in South Africa, South African laws apply. But if the South African testing company sends the genetic data overseas for research, or if the testing company is based in another jurisdiction, the NHA applies to the collection of samples in South Africa and POPIA applies to the transfer of personal information overseas.

IX CONCLUSION

This Chapter explored the research undertaken by testing companies using the genetic data of consumers and the South African legislation relevant to such activities. Based on the above analysis, provided that certain conditions, primarily in POPIA and the DoH Ethics Guidelines, are complied with, such research is permitted. However, problematic aspects may arise if the data is merged with another dataset from abroad, if the testing company gives a third party access to its database or sells the data, or if the data is exported. Therefore, the answer to the question – can testing companies conduct further research utilising the genetic data collected from consumers – is a qualified yes. This area is regulated, primarily by POPIA, and thus there are certain safeguards to protect the rights of consumers.

An issue that arises in terms of POPIA is whether research constitutes a 'specific' purpose for which personal information may be collected in terms of section 13(1) of POPIA. While there are two opposing positions on the matter, it remains undetermined. I suggest that a Code of Conduct for research, as is currently being drafted by ASSAf, may assist in clarifying this issue as well as other questions in terms of the collection, use, and sharing of genetic data for the purposes of research.

CHAPTER 8

WHERE TO FROM HERE? CONCLUSION AND RECOMMENDATIONS

I INTRODUCTION

There is limited South African literature on direct-to-consumer genetic testing, and the little that exists asserts that the industry is *unregulated.*[1] Throughout this dissertation, I explored this assertion by conducting a legal analysis of the most pertinent aspects of direct-to-consumer genetic testing based on South Africa's extant law and determined whether they are indeed regulated by statute. Under the headings below, I answer each of my research questions (mentioned in Chapter 1) in turn, summarising the key findings of this dissertation.

(a) What statutory framework is relevant to direct-to-consumer genetic testing in South Africa?

Given that South Africa does not have *dedicated* legislation regulating direct-to-consumer genetic testing, the different aspects of the industry fall under a variety of statutes – the most prominent of which are briefly mentioned. The NHA and its various regulations, specifically the Human Biological Material Regulations, govern the human biological material involved in direct-to-consumer genetic testing – namely saliva and DNA. The NHA also covers the removal of human biological material,[2] who it may be done by,[3] and the purposes for which it may be removed,[4] the donation of inter alia tissue for research,[5] and the approval of research protocols by a HREC.[6]

The Medicines Act as well as the Medical Device Regulations, govern licenses to manufacture, import, export, distribute, or wholesale medical devices or IVDs as well as their registration.[7] The Medicines Act and Medical Device Regulations also include conditions for the advertising and labelling of medical devices and IVDs.[8] The Medical Device Regulations

[1] C Dandara, J Greenberg, L Lambie et al 'Direct-to-consumer genetic testing: To test or not to test, that is the question' (2013) 103(8) *SAMJ* 512; Academy of Science of South Africa *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018) 38.
[2] Section 55 of the NHA.
[3] Section 56 of the NHA.
[4] Ibid.
[5] Section 63 of the NHA.
[6] Section 72(2) of the NHA.
[7] Sections 15 and 22C of the Medicines Act; regulations 5 and 8 of the Medical Device Regulations.
[8] Sections 18 and 20 of the Medicines Act; regulations 21 and 22 of the Medical Device Regulations.

specifically regulate medical devices and IVDs and contain provisions relating to their classification.[9] In addition, the Medicines Act provides for the establishment of SAHPRA, which has published various guidelines regarding the licensing and classification of medical devices and IVDs to assist those seeking to license and register their products in South Africa.[10]

The CPA and ECTA govern the advertising, marketing, disclosure, and labelling related to the offering of direct-to-consumer genetic tests in South Africa. This is bolstered by the SAMED Code,[11] and the MCA Code,[12] which relate specifically to the advertising of medical devices.

POPIA regulates the names, contact details, and addresses of consumers as 'personal information', and genetic data as both 'personal information' and 'special personal information'. POPIA provides for the collection and processing of this information,[13] as well as the exporting of genetic data related to direct-to-consumer genetic tests.[14] POPIA is also relevant to research using genetic data.

The Import and Export Regulations also comprise provisions relating to importing and exporting, which applies to saliva.[15] Lastly, the SA MTA provides a framework for the transfer of biological material (saliva and extracted DNA) for research.[16]

(b) Are direct-to-consumer genetic tests considered to be medical devices?

Determining whether direct-to-consumer genetic tests are medical devices and/or IVDs is not clear-cut, because the intention of the manufacturer – which is not always explicit – is central.[17] As testing companies offering health-related direct-to-consumer genetic tests to the public tend to make various, and sometimes conflicting, claims regarding the medical or diagnostic nature of their tests, deciding whether a direct-to-consumer genetic test is a medical device must be

[9] Regulation 11 of the Medical Device Regulations.
[10] South African Health Products Regulatory (SAHPRA) Authority *Guideline for a Licence to Manufacture, Import, Export or Distribute Medical Devices & IVDs* (2019); South African Health Products Regulatory Authority (SAHPRA) *Classification of Medical Devices and IVDs* (2019).
[11] South African Medical Device Industry Association (SAMED) *Medical Device Code of Ethical Marketing and Business Practice* (2017).
[12] Marketing Code Authority (MCA) *The South African Code of Marketing Practice for Health Products Code & Guideline, Version 14* (2021).
[13] Section 13 of POPIA.
[14] Section 72 of POPIA.
[15] Regulation 2 of the Import and Export Regulations.
[16] Donrich W Thaldar, Marietjie Botes & Annelize Nienaber 'South Africa's new standard material transfer agreement: Proposals for improvement and pointers for implementation' (2020) 21(85) *BMC Med Ethics* 6; Donrich Thaldar 'One material transfer agreement to rule them all? A call for revising South Africa's new standard material transfer agreement' (2020) 7(105) *Humanities & Social Sciences Communications* 1.
[17] As per the definition of 'medical device' in section 1 of the Medicines Act.

done on a case-by-case basis. Direct-to-consumer genetic tests that are not medical devices are automatically not IVDs either, since to qualify as an IVD, the object must be a medical device.[18]

For direct-to-consumer genetic tests that are medical devices, the classification thereof becomes relevant. SAHPRA recognises IVD medical devices for self-testing,[19] which are similarly defined to direct-to-consumer genetic tests. The three primary devices used in direct-to-consumer genetic testing – namely the tube for saliva collection, the machines that extract, sequence, and analyse DNA, and the software that converts raw data into useable information – are classified differently. But where individual IVDs form part of a group, as is the case with direct-to-consumer genetic tests, SAHPRA provides that the class of the group is determined by the highest class of any individual IVD or component.[20] In line with SAHPRA's Classification Guidelines, as most testing companies advise consumers to consult with healthcare professionals regarding their results,[21] and given the lack of evidence regarding the accuracy and usefulness of tests and the potential psychological implications on consumers, I suggest that health-related direct-to-consumer genetic tests be classified as Class C IVD medical devices.

(c) Can direct-to-consumer genetic testing be legally offered and undertaken in South Africa?

In terms of the first step in the direct-to-consumer genetic testing process – namely the collection of saliva samples by consumers – I suggest that on a plain reading of the relevant provisions in the NHA and the Human Biological Material Regulations, the self-collection of saliva samples by consumers is unlawful because consumers are not 'competent persons' as per the Human Biological Material Regulations.[22] But such a plain reading is both irreconcilable with a purposive interpretation and unconstitutional as it infringes individual autonomy.

Offering direct-to-consumer genetic tests to the public in South Africa entails numerous considerations. Firstly, there are licensing and registration requirements for those tests that are IVD medical devices.[23] A medical device establishment license is required to manufacture,

[18] This is because the definition of 'IVD' in section 1 of the Medicines Act and Regulation 1 of the Medical Device Regulations includes the term 'medical device'.
[19] SAHPRA op cit note 10 at 38.
[20] Additionally, the regulation 11(4) of the Medical Device Regulations provide that where a medical device or IVD is classified into more than one class, it must be placed 'in the higher of the risk classes'. Ibid at 30.
[21] Ibid at 38.
[22] Regulation 1 of the Human Biological Material Regulations.
[23] As per the Medicines Act, the Medical Device Regulations, and SAHPRA's guidelines.

import, export, sell, or distribute inter alia Class C IVD medical devices in South Africa.[24] To sell IVD medical devices in South Africa, they must be registered,[25] but, at present, there appears to be no registration process for medical devices.[26] Thus, direct-to-consumer genetic tests that are medical devices must be registered in another jurisdiction to permit their sale in South Africa.

Secondly, there are conditions imposed by the CPA, ECTA, and other industry codes[27] in terms of the advertising, marketing, disclosure, and labelling of direct-to-consumer genetic tests – whether medical devices or not. The CPA, as well as the MCA Code,[28] prohibit testing companies from misleading or deceiving consumers, requiring them to provide adequate, balanced, correct, plain, and comprehensible information in terms of their advertisements, terminology, meaning and implications of test results, and websites. The Medicines Act forbids the false advertising of medical devices and IVDs,[29] in alignment with the CPA's deception requirements. ECTA stipulates the information that is to be disclosed by testing companies that transact online, including their particulars, products and services, privacy policies, and security measures.[30]

Thirdly, there is the question of whether this South African legislation applies to testing companies based in foreign jurisdictions. Although at first glance it may appear as though foreign testing companies that contract online cannot be subject to South African legislation. However, I suggest that this is not the case. Amongst other things, the CPA applies to all 'transactions' occurring within South Africa.[31] As the delivery of testing kits and results to consumers – as part of the contract with foreign testing companies – occurs in South Africa, this constitutes performance, thereby bringing it within the scope of 'transaction' in the CPA.[32] Although ECTA does not apply where online contracts between consumers and testing companies are concluded in foreign jurisdictions,[33] there are provisions of ECTA applicable to

[24] Section 22C(6) of the Medicines Act.
[25] Section 14(1) of the Medicines Act.
[26] Catherine Tomlinson 'IN-DEPTH: The tangled web of medical device regulation in SA' *Spotlight* 3 September 2020, available at *https://www.spotlightnsp.co.za/2020/09/03/in-depth-the-tangled-web-of-medical-device-regulation-in-sa/*, accessed on 5 December 2020.
[27] SAMED op cit note 11; MCA op cit note 12; Advertising Regulatory Board *Code of Advertising Practice, Appendix K* (2021); Advertising Standards Authority of South Africa *Advertising Code of Practice.*
[28] MCA op cit note 12 at 18, 21–2, 26–7 & 66.
[29] Section 20(1)(*a*) of the Medicines Act.
[30] Section 43 of ECTA.
[31] Section 5(1) of the CPA. According to section 1 of the CPA, 'transaction' includes the agreement between parties for the supply of goods or services as well as performance.
[32] Section 1 of the CPA.
[33] This is in line with section 22(2) of ECTA.

all electronic contracts notwithstanding the legal system,[34] binding testing companies that supply their products and services in South Africa to requirements such as website disclosure and performance obligations. Furthermore, POPIA applies to the information of consumers. Entering their details into a website in South Africa brings local testing companies as well as those testing companies not domiciled in South Africa but using 'automated means' in the country within POPIA's ambit[35] – thus mandating them to process personal information in line with the eight conditions in POPIA.[36]

(d) What is South Africa's legal position regarding the importing and exporting of direct-to-consumer genetic tests, as well as associated samples and data?

The importing and exporting of direct-to-consumer genetic testing kits, as well as samples and data, is not straightforward due to its inter-jurisdictional nature. Unlike direct-to-consumer genetic tests that are not IVD medical devices which can be imported into South Africa in the same manner as most other consumer goods, importing IVD medical devices into South Africa requires an import license. As these licenses are only issued to a 'medical device or IVD establishment, manufacturer, wholesaler or distributor',[37] South African consumers are excluded and therefore cannot import direct-to-consumer genetic tests from testing companies in other jurisdictions.

All saliva samples exported out of South Africa require export permits,[38] which must be obtained by testing companies based in South Africa that export saliva samples to overseas laboratories for DNA extraction, sequencing, and analysis, as well as consumers who send saliva samples to testing companies in other jurisdictions. Where extracted (but unsequenced) DNA is exported out of South Africa for analysis in a laboratory overseas, it is not subject to the Import and Export Regulations since it is not 'tissue' as defined in the NHA,[39] but rather a type of biological material which is not regulated by the Import and Export Regulations, and therefore does not require an export permit.

Where genetic data is generated overseas and transferred to South Africa, POPIA does not apply, and such a transfer must accord with the laws of that country. But POPIA permits the sending of genetic data from South Africa to other jurisdictions based on certain conditions,

[34] Section 47 of ECTA.
[35] Section 3(1)(*b*) of POPIA.
[36] Section 4(1) of POPIA.
[37] Section 22C(1)(*b*) of the Medicines Act.
[38] Regulation 2(1) of the Import and Export Regulations.
[39] Section 1 of the NHA.

including consent, contractual performance,[40] or suitable protection provided by the laws of the country to which the genetic data is sent.[41] Consumers consenting to undergo direct-to-consumer genetic tests – often inclusive of consent to processing and analysis of saliva samples to obtain test results and possible research (which may take place in other jurisdictions) – or contractual performance based on the testing company's duty to provide consumers with test results from DNA analysis (which may occur in laboratories in other jurisdictions), allow genetic data to be sent overseas, and POPIA governs this transfer. Transferring 'special personal information',[42] which encompasses genetic data, from South Africa to another country that does not offer adequate protection, requires prior authorisation from the Information Regulator,[43] unless a code of conduct exists.[44] Where biological material (saliva samples and extracted DNA) as well as genetic data are transferred to another jurisdiction for research, the SA MTA is applicable.[45]

(e) Can direct-to-consumer genetic testing companies conduct research using genetic data gathered from consumers?

Biological material (saliva and DNA) may be removed,[46] and saliva samples (as a type of 'tissue' in the NHA,[47] and 'biological material' in the Human Biological Material Regulations)[48] may be donated,[49] for health research which, according to the NHA and the Human Biological Material Regulations, must be approved by a HREC.[50] Unlike POPIA which requires specific and informed consent,[51] the DoH Ethics Guidelines allow broad consent for genetic research.[52] POPIA permits consent to be obtained for the specific purpose of undergoing a direct-to-consumer genetic test (and preferably to research) following which testing companies can rely on the research exceptions or section 15 of POPIA.[53] But consumers are also required to provide consent for research in terms of the DoH Ethics Guidelines.[54]

[40] Section 72(1) of POPIA.
[41] Section 72(1)(*a*) of POPIA.
[42] Section 26(a) of POPIA.
[43] Section 57(1)(*d*) of POPIA.
[44] Section 57(3) of POPIA.
[45] Thaldar et al op cit note 16 at 6.
[46] Regulation 5(b) of the Human Biological Material Regulations.
[47] Section 1 of the NHA.
[48] Regulation 1 of the Human Biological Material Regulations.
[49] Section 63 of the NHA.
[50] Section 72(2) of the NHA and regulation 3(2) of the Human Biological Material Regulations respectively.
[51] Sections 1 and 13(1) of POPIA.
[52] Department of Health (DoH) *Ethics in Health Research: Principles, Processes and Structures* 2 ed (2015) 31.
[53] Section 15(3)(*e*) of POPIA.
[54] DoH op cit note 52 at 30.

Provided that personal information is collected for a specific purpose for which there was consent,[55] and as long as the information is used for research and is not identifiable,[56] further consent for research using genetic data is not necessary.

As a Code of Conduct for research, drafted by ASSAf, is still being developed, the application of POPIA to research remains a greenfield with several uncertainties – one of which is whether research can be considered a specific purpose for which testing companies may collect personal information. Projecting the DoH Ethics Guidelines onto POPIA is one possibility, but the Code of Conduct for research may clarify this position.[57]

II RECOMMENDATIONS

South Africa's extant laws, although they do regulate direct-to-consumer genetic testing, do not always cater for its unique characteristics – and are therefore in need of reform in some respects. As such, I recommend the following:

1. Although the collection of saliva samples by consumers is regulated, the wording is problematic, and I suggest that the Human Biological Material Regulations be amended to accommodate situations where individuals are allowed to collect their own saliva samples for the purpose of a direct-to-consumer genetic test – aligned with the Draft Testing and Research Regulations – specifically in circumstances where the chance of inflicting physical self-harm is negligible or, at the very least, saliva is recognised as a type of biological material that can be removed by a 'competent person'. One way of accomplishing this is by broadening the scope of those deemed to be 'competent persons' in the Human Biological Material Regulations and allowing for the removal of biological material by individuals and outside of prescribed or authorised institutions.[58]

2. The prohibition in the Medical Device Regulations on advertising Class C medical devices or IVDs to the public or laypersons,[59] but only to a 'prospective user'[60] should be revised.[61] There are two possible ways to address this. Firstly, to amend

[55] Section 15(3)(*a*) of POPIA.
[56] Section 15(3)(*e*) of POPIA.
[57] The DoH Ethics Guidelines are broad, and the meanings and usage of terminology in the DoH Ethics Guidelines cannot simply be projected onto POPIA; it is a different statute, with a different context, and different interpretations.
[58] As per regulation 2(b) of the Human Biological Material Regulations.
[59] Regulation 21(1)(*a*) of the Medical Device Regulations.
[60] Regulation 21(1)(*e*)(*i*) of the Medical Device Regulations.
[61] Ibid.

the meaning of 'prospective user' to include consumers. The term 'prospective user'[62] is not defined in the Medical Device Regulations.[63] Therefore, such an amendment would involve adding the phrase 'which may include the public and laypersons where relevant' to regulation 21(1)(*e*) of the Medical Device Regulations where reference is made to a 'prospective user'.[64] Secondly, to create an exception in the Medical Device Regulations to allow for the advertising of direct-to-consumer genetic tests that are Class C IVD medical devices. Regulation 21(1)(*b*) of the Medical Device Regulations contains an exception, namely that condoms – despite generally being classified as Class C – may be advertised to the public.[65] I suggest that a similar exception may be made to the Medical Device Regulations, allowing direct-to-consumer genetic tests that are IVD medical devices (and thereby likely falling into Class C) to be advertised to the public. By prohibiting the advertising of direct-to-consumer genetic tests to consumers, this limits choice in terms of which tests consumers may want to undergo (without involving a healthcare professional), and thereby violates their freedom and autonomy to decide.

3. The Import and Export Regulations be amended to create an exemption for direct-to-consumer genetic testing, specifically in terms of the requirement to obtain export permits for saliva. This is because, given the Import and Export Regulations' broad scope of application, requiring individuals and testing companies to apply for export permits to send saliva samples, as part of a direct-to-consumer genetic test, to testing companies or laboratories in other jurisdictions is impractical, constitutes an infringement of freedom (of both individuals and testing companies), wastes valuable state resources, and represents an area that is over-regulated, and unnecessarily so.

4. In line with the recommendation by Botes et al,[66] if the Information Regulator were to institute a mechanism like the 'White List' published by Malaysia's PDPC of

[62] As per regulation 21(1)(*e*)(*i*) of the Medical Device Regulations.
[63] This is evidenced by the fact that these categories are mentioned separately in the Medical Device Regulations. Regulation 21(1)(*a*) of the Medical Device Regulations refers to advertising to 'the public or a lay person', while regulation 21(1)(*e*) of the Medical Device Regulations provides for advertising to a 'prospective user'.
[64] Regulation 21(1)(*e*) of the Medical Device Regulations.
[65] Regulation 21(1)(*b*) of the Medical Device Regulations.
[66] M Botes, A Olckers & M Labuschaigne 'Data commercialisation in the South African health care context' (2021) 24 *PER/PELJ* 25.

jurisdictions offering adequate levels of protection for the transfer of personal information,[67] this may aid the cross-border sharing of data.

5. Codes of conduct under POPIA aim to guide the interpretation of POPIA for a specific sector or industry as well as provide prior authorisation.[68] In line with chapter 7 of POPIA, a Code of Conduct for research is currently being developed by ASSAf which,[69] once approved by the Information Regulator,[70] will become legally binding.[71] The Code of Conduct for research was developed to further guide, and uniformly apply, POPIA in relation to research,[72] and to ensure that the research community in South Africa complies with the provisions of POPIA.[73]The approval of a Code of Conduct for research – which inter alia caters specifically for genetic research – will provide greater clarity and guidance to researchers in this area, and may assist in clarifying whether research constitutes a specific purpose in terms of section 13(1) of POPA as well as other questions in terms of the collection, use, processing and its research exceptions, and cross-border sharing of genetic data for research.

6. Although a DTA – which may elucidate situations regarding the transfer of data for the purposes of research – has not yet been published by the Minister of Health through a notice in the Government Gazette (as was the case with the SA MTA), the SA MTA has illustrated that such a template will not be of assistance in these instances.[74] Where a researcher wishes to transfer genetic data from South Africa to a foreign country lacking an 'adequate level of protection for the processing of personal information',[75] prior authorisation must be obtained from the Information Regulator.[76] Thaldar et al recommend, in the context of the SA MTA, summarising POPIA's relevant provisions into contractual terms that are binding on a foreign

[67] See, Mark Parsons & Louise Crawford 'Malaysia publishes draft "White List" for personal data exports' *Hogan Lovells* 27 April 2017, available at *https://www.hoganlovells.com/en/publications/malaysia-publishes-draft-white-list-for-personal-data-exports*, accessed on 6 May 2021; Graham Greenleaf '2014-2017 update to Graham Greenleaf's *Asia Data Privacy Laws – Trade and Human Rights Perspectives*' (2017) 47 *UNSWLRS* 27.
[68] In section 57 of POPIA. Rachel Adams, Susan Veldsman, Michèle Ramsay et al 'Drafting a Code of Conduct for research under the Protection of Personal Information Act No. 4 of 2013' (2021) 117(5/6) *S Afr J Sci* 1.
[69] Ibid at 1. See also, Rachel Adams, Fola Adeleke, Dominique Anderson et al 'POPIA Code of Conduct for research' (2021) 117(5/6) *S Afr J Sci* 1–12.
[70] Section 57(3) of POPIA.
[71] Adams et al op cit note 68 at 1.
[72] Ibid at 1–2.
[73] Ibid at 1–2.
[74] See, Thaldar et al op cit note 16 at 1–13.
[75] Section 57(1)(*d*) of POPIA.
[76] Ibid; Thaldar et al op cit note 16 at 12.

recipient.[77] Therefore, in line with this, I suggest the development by ASSAf of a set of standard clauses, approved by the Information Regulator in terms of POPIA, be included in any DTA to ensure suitable protection when data is exported to non-adequate jurisdictions.[78] This may be useful, especially in instances where genetic data is transferred by testing companies for research. Furthermore, it is beneficial to consider this now before a DTA is published.

Although the above recommendations represent primary areas of concern that I identified, these are by no means the only ones, and there are other more nuanced aspects of direct-to-consumer genetic testing in South Africa that require consideration. These include:

1. Whether informed consent is possible, or necessary, in the direct-to-consumer genetic testing context.
2. Ascertaining the intention of the manufacturer to determine whether a direct-to-consumer genetic test is a medical device in terms of the Medicines Act.[79]
3. The fact that the Medical Device Regulations already contain a definition of 'self-testing'[80] signals a recognition of such situations. But this contrasts the Human Biological Material Regulations which prohibit the self-collection of saliva, implying that the next step – namely self-testing – is not permitted.
4. Given the inconsistences and uncertainties between the various laws, regulations, and guidelines regarding medical devices and IVDs, I suggest that SAHPRA issue a directive clarifying these discrepancies, as well as elucidating which aspect(s) of the direct-to-consumer genetic testing service is considered an IVD medical device, and the classification thereof.
5. Interpreting what constitutes a 'serious condition, ailment, or defect'[81] to assist in determining whether 'IVD medical devices for self-testing' are classified as Class B or Class C.
6. Whether direct-to-consumer genetic testing should be regulated as either a hybrid or sui generis form of health product or service, much like gene therapies and other advanced biomedicine therapies, because of its incorporation of both medicines, devices, and/or vectors for different purposes.

[77] Thaldar et al op cit note 16 at 12.
[78] One such jurisdiction is the US.
[79] As per subsection (a) under the definition of 'medical device' in section 1 of the Medicines Act.
[80] Regulation 1 of the Medical Device Regulations define 'self-testing' as 'testing performed by a lay person'.
[81] SAHPRA op cit note 10 at 32.

7. Considering whether genetic counselling or some form of professional oversight is required for all health-related direct-to-consumer genetic tests. If this is the case, regard must be had to the MCA Code, which bans the offering virtual diagnoses, advice, or treatment,[82] thereby prohibiting testing companies from advertising the offering of genetic counselling via email, telephone, or video calls.
8. Developing SAHPRA's registration process for medical devices and IVDs, as well as a means for determining their safety, quality, and efficacy. Doing so will seemingly allow individuals to order or import Class B, Class C, or Class D IVD medical devices for 'personal use'.[83]
9. Assessing the advertisements and marketing claims made by testing companies – specifically whether their products and services are marketed as being medical or diagnostic in nature, the wording used in advertisements and on their websites, the terminology and information provided to consumers regarding inter alia analytical validity, clinical validity, clinical utility, and concreteness of test results.
10. That individuals are excluded from importing direct-to-consumer genetic tests, that are considered IVD medical devices, themselves given that they are not 'a medical device or IVD establishment, manufacturer, wholesaler or distributor'[84] and can therefore not obtain an import license as required by the Medical Device Regulations.[85]
11. That the NHA be amended to include 'health research' (for which tissue may be donated) under the provision relating to *living* persons.[86] This will be a step towards the NHA including provisions relating to the field of genetics.
12. Whether research constitutes a specific purpose in terms of the collection and processing of personal information under POPIA.[87]

[82] MCA op cit note 12 at 15.
[83] SAHPRA op cit note 10 at 5. See also, T Saidi & TS Douglas 'Medical device regulation in South Africa: The Medicines and Related Substances Amendment Act 14 of 2015' (2018) 108(3) *S Afr Med J* 169.
[84] Section 22C(1)(*b*) of the Medicines Act.
[85] Regulation 3(3)(*a*) of the Medical Device Regulations.
[86] Section 63 of the NHA. Section 64(1) of the NHA states that tissue may be donated for inter alia health research, but this applies to deceased persons. Section 56(1) of the NHA states that a person may use inter alia tissue removed 'from a living person only for such medical or dental purposes as may be prescribed'.
[87] Sections 1, 13, and 15 of POPIA.

13. Further guidance on when special personal information is deemed to have been made 'public', and can thus be processed without the data subject's consent, in terms of POPIA.[88]

14. Clarifying the situation related to the de-identification of genetic data, and that it remains inherently identifiable due to its uniqueness – thus posing challenges for the privacy of research participants, or their genetic relatives. Differentiating between de-identification and anonymisation, and recognising that anonymising genetic data too far may decrease its usefulness.[89]

III CONCLUSION

Through this dissertation, consisting of a legal analysis of South Africa's legislative framework relevant to direct-to-consumer genetic testing, it is clear that South Africa's current legal landscape does impact on direct-to-consumer genetic testing in a variety of ways. I have found that direct-to-consumer genetic testing is indeed regulated in South Africa – contrary to what has been suggested in the literature on the topic. Although there is no *dedicated* legislation for direct-to-consumer genetic testing in the country, the industry is governed by a variety of existing statutes that apply to different aspects of the testing process, and in some instances may even overlap or be inconsistent with one another – thereby creating a complex and multi-layered regulatory landscape. Therefore, it cannot be said that direct-to-consumer genetic testing is unregulated; it is regulated, but there are issues that should be addressed.

Word count: 46989 words (excluding footnotes)

[88] Section 27(1)(*e*) of POPIA.
[89] See, Elizabeth R Pike 'Securing sequences: Ensuring adequate protections for genetic samples in the age of big data' (2015) 37 *Cardozo L Rev* 2018.

BIBLIOGRAPHY

I PRIMARY SOURCES

(a) Constitution

Constitution of the Republic of South Africa Act 108 of 1996

(b) Statutes

(i) South Africa

Choice on Termination of Pregnancy Act 92 of 1996

Consumer Protection Act 68 of 2008

Draft Regulations Regarding the use of Human DNA, RNA Cultured Cells, Stem Cells, Blastomeres, Polar Bodies, Embryos, Embryonic Tissue and Small Tissue Biopsies for Diagnostic Testing, Health Research and Therapeutics GN R7 GG 29526 of 5 January 2007

Electronic Communications and Transactions Act 25 of 2002

General Regulations made in terms of the Medicines and Related Substances Act, 1965, as Amended GN R510 GG 24727 of 10 April 2003

Health Professions Act 56 of 1974

Insurance Act 18 of 2017

Interpretation Act 33 of 1957

Material Transfer Agreement for Human Biological Materials GN R719 GG 41781 of 20 July 2018

Medicines and Related Substances Act 101 of 1965

Medicines and Related Substances Amendment Act 14 of 2015

Medicines and Related Substances Amendment Act 59 of 2002

National Health Act 61 of 2003

National Health Laboratory Service Act 37 of 2000

National Health Laboratory Service Amendment Act 5 of 2019

National Heritage Resources Act 25 of 1999

Nursing Act 33 of 2005

Old Persons Act 13 of 2006

Promotion of Equality and Prevention of Unfair Discrimination Act 4 of 2000

Protection of Personal Information Act 4 of 2013

Regulations Regarding the General Control of Human Bodies, Tissue, Blood, Blood Products and Gametes GN R180 GG 35099 of 2 March 2012

Regulations Relating to the Import and Export of Human Tissue, Blood, Blood Products, Cultured Cells, Stem Cells, Embryos, Fetal Tissue, Zygotes and Gametes GN R181 GG 35099 of 2 March 2012

Regulations Relating to Medical Devices and In Vitro Diagnostic Medical Devices (IVDs) GN R1515 GG 40480 of 9 December 2016

Regulations Relating to Research with Human Participants GN R719 GG 38000 of 19 September 2014

Regulations Relating to Stem Cell Banks GN R183 GG 35099 of 2 March 2012

Regulations Relating to the Taking of Buccal Sample or Withdrawal of Blood from a Living Person for Testing (Amendment) GN R944 GG 34750 of 11 November 2011

Regulations Relating to the Use of Human Biological Material GN R177 GG 35099 of 2 March 2012

Regulations Relating to Tissue Banks GN R182 GG 35099 of 2 March 2012

South African Medical Research Council Act 58 of 1991

(ii) Foreign

1976 Medical Device Amendments to the Food, Drug, and Cosmetic Act 21 USC § 301–392

Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes CETS No. 203

Australian Privacy Act 1988

Austrian Gene Technology Act of 1995 (Gentechnikgesetz 1994)

Canadian Food and Drugs Act (RSC 1985 c. F–27)

Canadian Personal Information Protection and Electronic Documents Act SC 2000 c. 5

Clinical Laboratory Improvement Amendments of 1988 102 Stat. 2903

European Convention on Human Rights and Biomedicine ETS No.164

European Union General Data Protection Regulations 2016/679

French Civil Code 2016 created by Ordonnance n° 2016–131 of 10 February 2016

French Loi n°. 2004–800 rélative à la bioéthique

German Human Genetic Examination Act 2009 BGBL I at 2529

Hungarian Genetic Act 2008

In Vitro Diagnostic Medical Devices Directive 98/79/EC

In Vitro Diagnostic Medical Devices Regulation 2017/746

Italian General Authorization for the Processing of Genetic Data 2014

Lithuanian Order No. V–220 2003

Medical Devices Directive 93/42/EEC

Medicines and Medical Devices Bill (HC Bill 90)

Netherlands Decree on In Vitro Diagnostic Devices (2001)

Portuguese Law 12/2005 of 26 January 2005 (Law 12/2005, 26 Jan)

Spanish Boletín Oficial del Estado No159 2007

Swiss Federal Act on Human Genetic Testing 2007 (8 oct 2004 (1.4.2007))

United Kingdom Data Protection Act 1998

United Kingdom Human Tissue Act 2004 c 30

US Code: Federal Food, Drug, and Cosmetic Act 21 USC §§ 301–392

US Genetic Information Non-Discrimination Act of 2008 122 Stat. 881

US Health Insurance Portability and Accountability Act of 1996 110 Stat. 1936

Verifying Accurate, Leading-Edge IVCT Development Act S.3404 – 116th Congress (2020)

(c) Case law

(i) South Africa

A to Z Bazaars (Pty) Ltd v Minister of Agriculture 1975 (3) SA 468 (A)

AB v Minister of Social Development 2016 (2) SA 27 (GP)

AB v Minister of Social Development 2017 (3) SA 570 (CC)

Affordable Medicines Trust v Minister of Health 2006 (3) SA 247 (CC)

Aird v Hockly's Estate 1937 EDL 34

Allergan Pharmaceuticals (Pty) Ltd v Medicines Control Council [2015] 3 All SA 173 (GP)

Attorney-General, Transvaal v Additional Magistrate for Johannesburg 1924 AD 421 (A)

Barkhuizen v Napier 2007 (5) SA 323 (CC)

Barnett v Estate Gumpert 1926 CPD 363

Bato Star Fishing (Pty) Ltd v Minister of Environmental Affairs and Tourism 2004 (4) SA 490 (CC)

Beadica 231 CC v Trustees for the time being of the Oregon Trust 2020 (5) SA 247 (CC)

Berman v Teiman 1975 (1) SA 756 (W)

Bernstein v Bester NO 1996 (2) SA 751 (CC)

Bertie Van Zyl (Pty) Ltd v Minister for Safety and Security 2010 (2) SA 181 (CC)

Bhe v Magistrate, Khayelitsha (Commission for Gender Equality as Amicus Curiae); Shibi v Sithole; South African Human Rights Commission v President of the Republic of South Africa 2005 (1) SA 580 (CC)

Bhyat v Commissioner for Immigration 1932 AD 125

Botha v Smuts [2020] ZAECPEHC 19

British American Tobacco South Africa (Pty) Ltd v Minister of Health [2012] 3 All SA 593 (SCA)

Buthelezi v Ndaba 2013 (5) SA 437 (SCA)

Cape Explosives Works Ltd v South African Oil and Fat Industries Ltd (1) 1921 (CPD) 244

Case v Minister of Safety and Security; *Curtis v Minister of Safety & Security* 1996 (3) SA 617 (CC)

Castell v de Greef 1994 (4) SA 408 (C)

Chotabhai v Union Government (Minister of Justice) 1911 AD 13

Clarke v Member of the Executive Council for Health Western Cape [2018] ZAWCHC 64

Daniels v Minister of Defence 2016 (6) SA 561 (WCC)

De Beers Industrial Diamond Division (Pty) Ltd v Ishizuka 1980 (2) SA 191 (T)

Dibowitz v Commissioner for Inland Revenue 1952 (1) SA 55 (AD)

Dikoko v Mokhatla 2006 (6) SA 235 (CC)

Dilworth v Commissioner of Stamps 1899 AC

Durban's Water Wonderland (Pty) Ltd v Botha 1999 (1) SA 982 (SCA)

Dutch Reformed Church, De Aar v Joubert 1921 CPD 9

EMS Industries (Pty) Ltd v Inteletrack CC (Patent 2010/01326) [2015] ZAGPPHC 696

Everfresh Market Virginia (Pty) Ltd v Shoprite Checkers (Pty) Ltd 2012 (1) SA 256 (CC)

Ferreira v Levin NO; Vryenhoek v Powell NO 1996 (1) SA 984 (CC)

Gelderma Laboratories South Africa (Pty) Ltd v Medicines Control Council [2014] ZAGPPHC 360

Goliath v Member of the Executive Council for Health, Eastern Cape 2015 (2) SA 97 (SCA)

Hayne & Co v Kaffrarian Steam Mill Co Ltd 1914 AD 363

HMBMP Properties (*Pty*) *Ltd v King* 1981 (1) SA 906 (N)

In Re Estate Cullingworth 1936 NPD 251 (NPD)

Independent Institute of Education (Pty) Limited v Kwazulu-Natal Law Society 2020 (2) SA 325 (CC)

Isaacs v Pandie [2012] ZAWCHC 47

Jafta v Ezemvelo KZN Wildlife (2009) 30 ILJ 131 (LC)

Jaga v Dönges NO; Bhana v Dönges NO 1950 (4) SA 653 (A)

Jordan v State [2002] (11) BCLR 1117 (CC)

Katzeff v Canal Walk Limited t/a Canal Walk [2005] ZAWCHC 58

Kergeulen Sealing and Whaling Co Ltd v Commissioner for Inland Revenue 1939 AD 487

Khumalo v Holomisa 2002 (5) SA 401 (CC)

Loryan (Pty) Ltd v Solarsh Tea and Coffee (Pty) Ltd 1984 (3) SA (WLD)

M v Vallabh [2017] ZAGPJHC 397

Member of the Executive Council for Education: KwaZulu-Natal v Pillay 2008 (1) SA 474 (CC)

Meyers v Member of the Executive Council, Department of Health, Eastern Cape 2020 (3) SA 337 (SCA)

Minister of Health v New Clicks South Africa (Pty) Ltd 2006 (2) SA 311 (CC)

Minister of Justice and Correctional Services v Estate Late James Stransham-Ford 2017 (3) SA 152 (SCA)

Minister of Justice and Constitutional Development v Masingili 2014 (1) SACR 437 (CC)

Minister of Safety and Security v Xaba 2003 (2) SA 703 (D)

Minister of the Interior v Machadodorp Investments (*Pty*) *Ltd* 1957 (2) SA 395 (A)

Mpisi v Trebble 1994 (2) SA 136 (AD)

Napier v Barkhuizen 2006 (4) SA 1 (SCA)

Natal Joint Municipal Pension Fund v Endumeni Municipality 2012 (4) SA 593 (SCA)

National Coalition for Gay and Lesbian Equality v Minister of Justice 1999 (1) SA 6 (CC)

New Modderfontein Gold Mining Company v Transvaal Provincial Administration 1919 AD 367

NM v Smith 2007 (5) SA 250 (CC)

Norden v Bhanki 1974 (4) SA 647 (AD)

Nyembezi v Law Society, Natal 1981 (2) SA 752 (A)

Omegalabs (Pty) Ltd v Medicines Control Council [2016] ZAGPPHC 1157

Oppelt v Head: Health, Department of Health Provincial Administration: Western Cape 2016 (1) SA 325 (CC)

Port Elizabeth Municipality v Various Occupiers 2005 (1) SA 217 (CC)

Poswa v MEC for Economic Affairs, Environment and Tourism, Eastern Cape 2001 (3) SA 582 (SCA)

Potgieter v Member of the Executive Committee for Health and Social Development: Limpopo [2012] ZAGPPHC 342

R v Debele 1956 (4) SA 570 (AD)

R v Hugo 1926 AD 268

Rex v Ah Tong 1919 AD 186

Richter v Estate Hammann 1967 (3) SA 226 (C)

Robb NO v Standard Bank Ltd 1979 (2) SA 420 (R)

Roodepoort City Council v Shepherd 1981 (2) SA 720 (A)

Rosen v Rand Townships Registrar 1939 WLD 5 (WLD)

Ruta v Minister of Home Affairs 2019 (2) SA 329 (CC)

S v Crawford 1979 2 SA 48 (A)

S v Dane 1957 (2) SA 472 (N)

S v Jordan (Sex Workers Education and Advocacy Task Force as Amici Curiae) 2002 (6) SA 642 (CC)

S v Kgogong 1980 (3) SA 600 (AD)

S v Makwanyane 1995 (3) SA 391

S v Manamela (Director-General of Justice Intervening) 2000 (3) SA 1 (CC)

S v Maqhina 2001 (1) SACR 241 (T)

S v Mhlungu 1995 (3) SA 867 (CC)

S v Nyembe 2014 (1) SACR 105 (GSJ)

S v Orrie [2003] ZAWCHC 63

S v Weinberg 1979 (3) SA 89 (A)

S v Williams [2005] ZAWCHC 35

S v Wood 1976 (1) SA 703 (A)

Secretary for Inland Revenue v Charkay Properties (Pty) Ltd 1976 (4) SA 872 (A)

Shaik v Minister of Justice and Constitutional Development 2004 (3) SA 599 (CC)

Shoprite Checkers (Pty) Limited v Member of the Executive Council for Economic Development, Environmental Affairs and Tourism, Eastern Cape 2015 (6) SA 125 (CC)

Smeiman v Volkersz 1954 (4) SA 170 (C)

Sobekwa v Member of the Executive Council for the Department of Health [2014] ZAECMHC 25

Somke v Ostrofsky [2013] ZAWCHC 82

Soobramoney v Minister of Health (Kwazulu-Natal) 1998 (1) SA 765 (CC)

South African Airways (Pty) Ltd v Aviation Union of South Africa 2011 (3) SA 148 (SCA)

South African Police Service v Solidarity obo Barnard 2014 (6) SA 123 (CC)

Southern Life Association Ltd v Commissioner for Inland Revenue 1985 (2) SA 267 (C)

Stellenbosch University Law Clinic v Lifestyle Direct Group International (Pty) Ltd [2021] 4 All SA 219 (WCC)

Stoffberg v Elliot 1923 CPD 148

Stransham-Ford v Minister of Justice and Correctional Services 2015 (4) SA 50 (GP)

Teddy Bear Clinic for Abused Children v Minister of Justice and Constitutional Development 2014 (2) SA 168 (CC)

Thabela v Nedgroup Medical Aid Scheme/Medscheme 2020 (1) SA 318 (GJ)

Transvaal Consolidated Land and Exploration Co Ltd v Johannesburg City Council 1972 (1) SA 88 (W)

Tshabalala-Msimang v Makhanya 2008 (6) SA 102 (W); 2008 (3) BCLR 338 (W)

Union Government (*Minister of Finance*) *v Mack* 1917 (AD)

Venter v R 1907 TS 910 (TS)

Volks NO v Robinson 2005 (5) BCLR 446 (CC)

Volschenk v Volschenk 1946 TPD 486

Waylite Diary CC v First National Bank Ltd 1995 (1) SA 645 (A)

Zonele v Member of the Executive Council of the Department of Health of the Eastern Cape Government Bhisho [2014] ZAECBHC 17

(ii) Foreign

Beckford v Wade 17 Ves

Carlill v Carbolic Smoke Ball Co [1892] 2 QB 484

Data Protection Commissioner v Facebook Ireland and Maximillian Schrems (C–311/18)

Duck v Bates 12 QBD 79

Schrems v Data Protection Commissioner (C–362/14) (6 October 2015)

II SECONDARY SOURCES

(a) Books

Beauchamp, TL & JF Childress *Principles of Biomedical Ethics* 4 ed New York: Oxford Press, (1994).

Beauchamp, TL & JF Childress *Principles of Biomedical Ethics* 5 ed New York: Oxford Press, (2001).

Beauchamp, TL & JF Childress *Principles of Biomedical Ethics* 7 ed New York: Oxford Press, (2013).

Beyleveld, D & R Brownsend *Human Dignity in Bioethics and Biolaw* 1 ed Oxford University Press, (2001).

Botha, CJ *Statutory Interpretation* 5 ed Cape Town: Juta, (2012).

Braverman, I (ed) *Gene Editing, Law, and the Environment: Life Beyond the Human* 1 ed New York: Routledge, (2017).

Burley, J & J Harris (eds) *A Companion to Genethics* Massachusetts: Wiley-Blackwell, (2004).

Burley, J (eds) *The Genetic Revolution and Human Rights* 1 ed New York: Oxford University Press, (1999).

Carstens, P & D Pearmain *Foundational Principles of South African Medical Law* Durban: LexisNexis, (2007).

Currie, I & J De Waal *The Bill of Rights Handbook* 6 ed Cape Town: Juta, (2013).

Dada, MA & DJ McQuoid-Mason *Introduction to Medico-Legal Practice* Durban: LexisNexis, (2001).

Devenish, GE *Interpretation of Statutes* Cape Town: Juta, (1992).

Dhai, A & D McQuoid-Mason *Bioethics, Human Rights and Health Law* 1 ed Cape Town: Juta, (2011).

Donne, J *Devotions Upon Emergent Occasions* Toronto: Ambassador Books Ltd, (1959).

Dove, ES 'Collection and protection of genomic data' in *Handbook of Genomics, Health & Society, Research Paper Series* London: Routledge, (2018) 161–168.

Du Plessis, L 'Interpretation' in S Woolman, M Bishop & J Brickhill (eds) 2 ed *Constitutional Law of South Africa* Juta: Cape Town, (2008) 32–161.

Du Plessis, LM 'Statute law and interpretation' in LM Du Plessis (ed) 2 ed *The Law of South Africa* 25(1) Durban: Butterworths, (2011).

Dworkin, G *The Theory and Practice of Autonomy* Cambridge: Cambridge University Press, (1988).

Firth, JD, CP Conlon & TM Cox (eds) *Oxford Textbook of Medicine* 6 ed Oxford: Oxford University Press, (2020).

Fox, D *Birth Rights and Wrongs: How Medicine and Technology are Remaking Reproduction and the Law* New York: Oxford University Press, (2019).

Frankfurt, H *The Importance of What We Care About* New York: Cambridge University Press, (1988).

Friedman, M 'Autonomy and social relationships: Rethinking the feminist critique' in DT Meyers (ed) *Feminist theory and politics. Feminists rethink the self* Westview Press, (1997) 40–61.

Fukuyama, F *Our Posthuman Future: Consequences of the Biotechnology Revolution* New York: Picador, (2003).

Glannon, W *Genes and Future People: Philosophical Issues in Human Genetics* 1 ed Michigan: Avalon Publishing, (2001).

Gyekye, K ‘Person and community in African thought’ in Coetzee & Roux (eds) 3 ed *Philosophy from Africa: A Text with Readings* London: Oxford University Press, (2002) 297–312.

Habermas, J *The Future of Human Nature* 1 ed Cambridge: Polity Press, (2003).

Hanna, KE ‘The ethical, legal, and social implications program of the National Center for Human Genome Research: A missed opportunity?’ in *Society’s Choices: Social and Ethical Decision Making in Biomedicine* Washington: The National Academies Press, (1995) 432–457.

Harris, J *Enhancing Evolution: The Ethical Case for Making Better People* New Jersey: Princeton University Press, (2007).

Hernandez, LM (ed) *Implications of Genomics for Public Health: Workshop Summary* Washington, DC: The National Academies Press, (2005).

Higgs, R ‘On telling patients the truth’ in H Kuhse, U Schüklenk & P Singer (eds) 3 ed *Bioethics: An Anthology* Chichester: John Wiley & Sons, (2016) 621–627.

Howard, HC, S Sterckx, J Cockbain et al ‘The convergence of direct-to-consumer genetic testing companies and biobanking activities: The example of 23andMe’ in M Wienroth & E Rodrigues (eds) 1 ed *Knowing New Biotechnologies: Social Aspects of Technological Convergence* London: Routledge, (2015) 59–74.

Kaan, T & WC Ho (eds) *Genetic Privacy: An Evaluation of the Ethical and Legal Landscape* London: Imperial College Press, (2013).

Kalokairinou, L, HC Howard & P Borry ‘Direct-to-consumer genetic testing’ in John Wiley & Sons Ltd (ed) *eLS* Chichester: John Wiley & Sons Ltd, (2014) 1–5.

Kant, I *Groundwork of the Metaphysics of Morals: A German-English Edition* New York: Cambridge University Press, (2011).

Kaveny, C *Law’s Virtues: Fostering Autonomy and Solidarity in American Society* Washington, DC: Georgetown University Press, (2012).

Klug, WS, MR Cummings, CA Spencer et al *Concepts of Genetics* 12 ed New York: Pearson, (2018).

Laurie, GT *Genetic Privacy: A Challenge to Medico-Legal Norms* Cambridge: Cambridge University Press, (2002).

Mackenzie, C & N Stoljar (eds) *Relational Autonomy: Feminist Perspectives on Autonomy, Agency and the Social Self* Oxford University Press, (2000).

Mappes, TA & D Degrazia (eds) *Biomedical Ethics* 4 ed New York: McGraw Hill Inc, (1996).

Mathews, CK, KE van Holde, DR Appling et al *Biochemistry* 4 ed Toronto: Pearson, (2012).

Mill, JS *On Liberty* 4 ed London: Longman, Roberts & Green, (1999).

Mill, JS *Utilitarianism, Liberty, and Representative Government* London: JM Dent & Sons, (1910).

Moses, LB '*Sui Generis* Rules' in GE Marchant, BR Allenby & JR Herkert (eds) *The Growing Gap Between Emerging Technologies and Legal-Ethical Oversight: The Pacing Problem* London: Springer, (2011).

Mothibe, ME & M Sibanda 'African traditional medicine: South African perspective' in C Mordeniz (ed) *Traditional and Complementary Medicine* IntechOpen, (2019).

Munson, R 'Geneticize me! The case for direct-to-consumer genetic testing' in RW Kolb (ed) *The Ethics of Genetic Commerce* Oxford: Blackwell Publishing Ltd, (2007) 55–69.

National Academy of Sciences *Privacy Issues in Biomedical and Clinical Research* Washington, DC: National Academy Press, (1998).

O'Neill, O *Autonomy and Trust in Bioethics* Cambridge: Cambridge University Press, (2002).

Patrinos, GP (ed) *Applied Genomics and Public Health* London: Academic Press, (2020).

Phillips, AM *Buying Your Self on the Internet: Wrap Contracts and Personal Genomics* Edinburgh: Edinburgh University Press, (2019).

Prainsack, B 'DIY genetics: The right to know your own genome' in R Chadwick, M Levitt & D Shickle (eds) *The Right to Know and the Right Not to Know: Genetic Privacy and Responsibility* 2 ed Cambridge: Cambridge University Press, (2014) 100–115.

Prainsack, B 'Understanding participation: The "citizen science" of genetics' in B Prainsack, S Schicktanz & G Werner-Felmayer (eds) *Genetics as Social Practice: Transdisciplinary Views on Science and Culture* Farnham: Ashgate, (2014) 147–164.

Pugh, J *Autonomy, Rationality, and Contemporary Bioethics* Oxford: Oxford University Press, (2020).

Raz, J *The Morality of Freedom* (1986) New York: Oxford University Press.

Schafer, A 'Privacy: A philosophical overview' in D Gibson (ed) *Aspects of Privacy Law: Essays in Honor of John M Sharp* Toronto: Butterworths, (1980) 1–20.

Schermer, M 'Philosophical and ethical perspectives on autonomy' in M Schermer (ed) *The Different Faces of Autonomy: Patient Autonomy in Ethical Theory and Hospital Practice* Dordrecht: Kluwer Academic Publishers, (2002) 1–22.

Silver, LM *Challenging Nature: The Clash between Biotechnology and Spirituality* 1 ed New York: Ecco, (2006).

Silver, LM *Remaking Eden – Cloning, Genetic Engineering and the Future of Humankind?* London: Phoenix, (1999).

Smith, J *The Law of Contract* 3 ed Cape Town: Juta, (1996).

Stevenson, S (ed) *The National Health Act Guide* 3 ed Cape Town: Siber Ink CC, (2019).

Stoutjesdyk, A *The Bible and the Protestant Reformation* London: Trinitarian Bible Society, (2017).

Taelman, P & J Werbrouck 'Procedural autonomy and Belgian civil procedure law: A turbulent cohabitation' in B Krans & A Nylund (eds) *Procedural Autonomy Across Europe* Cambridge: Intersentia, (2020) 13–36.

Taylor, M *Genetic Data and the Law: A Critical Perspective on Privacy Protection* Cambridge: Cambridge University Press (2012).

Ten Have, H 'The right to know and the right not to know in the era of neoliberal biopolitics and bioeconomy' in R Chadwick, M Levitt & D Shickle (eds) *The Right to Know and the Right Not to Know: Genetic Privacy and Responsibility* 2 ed Cambridge: Cambridge University Press, (2014) 133–150.

The British Medical Association *Complete Family Health Encyclopaedia* London: Colour Library Books, (1997).

Ursin, LØ 'Duties and rights of biobank participants: Principled autonomy, consent, voluntariness and privacy' in JH Solbakk, S Holm & B Hofmann (eds) *The Ethics of Research Biobanking* Boston: Springer, (2009) 69–84.

Wachbroit, R ‘Genetic determinism, genetic reductionism, and genetic essentialism’ (2002) in TH Murray & MJ Mehlman (eds) *Encyclopedia of Ethical, Legal and Policy Issues in Biotechnology* New York: John Wiley & Sons, (2000) 352–356.

Weil, J *Psychosocial Genetic Counseling* 1 ed New York: Oxford University Press, (2000).

(b) Guidelines, reports, documents, and statements

Academy of Science of South Africa (ASSAf) *Human Genetics and Genomics in South Africa: Ethical, Legal and Social Implications* (2018).

Advertising Regulatory Board *Code of Advertising Practice* (2021).

Advertising Standards Authority of South Africa *Advertising Code of Practice*.

American College of Medical Genetics *ACMG Statement on Direct-to-consumer Genetic Testing* (2008).

American College of Obstetricians and Gynaecologists *Access to Genetic Testing: Position Statement* (2018).

Australian Government National Health and Medical Research Council *DNA Genetic Testing in the Australian Context: A Statement from the National Health and Medical Research Council (NHMRC)* (2010).

Campbell, N & S Logan *Consumer Protection Guide for Lawyers* (2011).

Canadian Association of Genetic Counsellors *CAGC Statement on Direct-to-consumer Genetic Testing* (2018).

Carpenter, CC *If the Genes Fit, Wear Them: A Proposal for Light-Handed Regulation of Direct-to-consumer Genetic Tests from the Federal Trade Commission as a Solution to a Regulatory Commons Problem* (2011).

Consumer Reports *Direct-to-consumer Genetic Testing: The Law Must Protect Consumers' Genetic Privacy* (2020).

Department of Health *Ethics in Health Research: Principles, Processes and Structures* 2 ed Pretoria: Department of Health (2015).

Department of Health *Guidelines for Good Practice in the Conduct of Clinical Trials with Human Participants in South Africa* 2 ed (2006).

Department of Health *South Africa Frequently Asked Questions (FAQs) Import/Export Permit Programme* (2015).

Department of Science and Technology *2010-2013 Corporate Strategy* (2010).

Department of Science and Technology *The Bio-Economy Strategy* (2013).

Department of Science and Technology *The Ten-Year Plan for Science and Technology* (2008).

Department of Science and Technology *White Paper on Science, Technology and Innovation* (2019).

European Academies of Science Advisory Council (EASAC) & Federation of European Academies of Medicine (FEAM) *Direct-to-consumer Genetic Testing for Health-Related Purposes in the European Union* (2012).

European Parliament – Science and Technology Options Assessment 'Direct to consumer genetic testing study' (2008) *Institute of Science and Technology*.

European Parliament *Science and Technology Options Assessment: Annual report 2015 (No. PE 563.507)* (2016).

European Technology Assessment Group *Direct-to-consumer Genetic Testing Final Report* (2008).

Future of Privacy Forum *Privacy Best Practices for Consumer Genetic Testing Services* (2018).

Lister Hill National Center for Biomedical Communications *Help Me Understand Genetics: Direct-to-Consumer Genetic Testing* (2019).

GMDN *GMDN User Guide: A Comprehensive Guide to the Global Medical Device Nomenclature* (2018).

Goemans, B & R McLaughlin *Medical Devices: An Innovation Guide from Lab to Commercialisation* (2018).

Government of the Republic of South Africa *South Africa's National Research and Development Strategy* (2002).

H3Africa Working Group on Ethics and Regulatory Issues for the Human Heredity and Health in Africa (H3Africa) Consortium *H3Africa Guideline for Informed Consent* 3 ed (2017).

Health Professions Council of South Africa *Guidelines for Good Practice in the Health Care Professions: Confidentiality: Protecting and Providing Information (Booklet 5)* (2016).

Health Professions Council of South Africa *Guidelines for Good Practice in the Health Care Professions: General Ethical Guidelines for Health Researchers (Booklet 13)* (2016).

Health Professions Council of South Africa *Standards of Practice for Genetic Counsellors* (2013).

HM Government & Association of British Insurers *Code on Genetic Testing and Insurance* (2018).

Hogarth, S, D Melzer & R Zimmern *The Regulation of Commercial Genetic Testing Services in the UK: A Briefing for the Human Genetics Commission* (2005).

House of Lords Science and Technology Committee *Genomic Medicine* (2009) 2(1) Report HL Paper 107–I.

Human Genetics Commission *A Common Framework of Principles for Direct-To-Consumer Genetic Testing Services* (2010).

Human Genetics Commission *Genes Direct: Ensuring the Effective Oversight of Genetic Tests Supplied Directly to the Public* (2003).

Human Genetics Commission *More Genes Direct: A Report on Developments in the Availability, Marketing and Regulation of Genetic Tests Supplied Direct to the Public* (2007).

Institute of Medicine and National Research Council *Direct-to-Consumer Genetic Testing: Summary of a Workshop* Washington, DC: The National Academies Press, (2011).

International Bioethics Committee *Report of the IBC on Updating Its Reflection on the Human Genome and Human Rights* (2015).

Marketing Code Authority *The South African Code of Marketing Practice for Health Products Code & Guideline, Version 14* (2021).

Medicines & Healthcare Products Regulatory Agency *Guidance on Legislation: Borderlines with Medical Devices* (2016).

National Department of Health *Human Genetics Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities*.

National Health and Medical Research Council *Medical Genetic Testing: Information for Health Professionals* (2010).

National Pathology Accreditation Advisory Council (NPAAC) *The Provision of Direct to Consumer Genetic Tests: Guiding Principles for Providers* (2014).

Nuffield Council on Bioethics *Genome Editing: An Ethical Review* (2016).

Nuffield Council on Bioethics *Genome Editing: An Ethical Review – A Short Guide* (2016).

Nuffield Council on Bioethics *Genome Editing: An Ethical Review – Key Points* (2016).

Nuffield Council on Bioethics *Genome Editing and Human Reproduction: Social and Ethical Issues* (2018).

Nuffield Council on Bioethics *Genome Editing and Human Reproduction: Social and Ethical Issues Short Guide* (2018).

Nuffield Council on Bioethics *Medical Profiling and Online Medicine: The Ethics of 'Personalised Healthcare' in a Consumer Age* (2010).

Nuffield Council on Bioethics *Medical Profiling and Online Medicine: The Ethics of 'Personalised Healthcare' in a Consumer Age – A Guide to the Report* (2010).

Nuffield Council on Bioethics *Submission to the House of Commons Science and Technology Committee Inquiry on Commercial Genomics* (2019).

Nuffield Council on Bioethics *Summary of Report: Genome Editing and Human Reproduction: Social and Ethical Issues* (2018).

Organisation for Economic Co-operation and *Development OECD Guidelines for Quality Assurance in Genetic Testing* (2007).

Organisation for Economic Co-operation and Development *OECD Guidelines on Human Biobanks and Genetic Research Databases* (2009).

PHG Foundation *A Common Framework of Principles for Direct-to-Consumer Genetic Testing Services: Response from the PHG Foundation* (2009).

Reinsurance Group of America (RGA) *Genetics and Insurance: Challenges and Opportunities II* (2019).

Ries, NM *Analysis of Policies and Practices of Direct-To-Consumer Genetic Testing Companies: Private Sector Databanks and Privacy Protection Norms* Health Law Institute, University of Alberta (2010).

SA Code of Practice for the Marketing of Health Products (2010).

Secretary of State for Health *Government Response to the House of Lords Science and Technology Committee Inquiry into Genomic Medicine* (2009).

South African Health Products Regulatory Agency (SAHPRA) *Classification of Medical Devices and IVDs* (2019).

South African Health Products Regulatory Authority (SAHPRA) *Guideline for a Licence to Manufacture, Import, Export or Distribute Medical Devices & IVDs* (2019).

South African Health Products Regulatory Authority (SAHPRA) *Registration of Medical Devices and Call-up Plan* (2018).

South African Law Reform Commission *Discussion Paper 112 Statutory Revision: Review of the Interpretation Act 33 of 1957 (Project 25)* (2006).

South African Medical Device Industry Association *An Overview of the Medical Device Code of Ethical Marketing and Business Practice* (2020).

South African Medical Device Industry Association *Medical Device Code of Ethical Marketing and Business Practice* (2017).

South African Medical Research Council *Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research* (2002).

South African Medical Research Council *The South African Medical Research Council Guidelines on the Responsible Conduct of Research* (2017).

South African Medical Technology Industry Association *Medical Device Code of Ethical Marketing and Business Practice, Version 7*.

Southern African Laboratory Diagnostics Association (SALDA) *SALDA In Vitro Diagnostics in South Africa* (2014).

Swiss Agency for Therapeutic Products *AW-Information Sheet: Genetic Tests* (2017).

United States Government Accountability Office, Testimony Before the Subcommittee on Oversight and Investigations, Committee on Energy and Commerce, House of

Representatives *Direct-To-Consumer Genetic Tests: Misleading Test Results are Further Complicated by Deceptive Marketing and Other Questionable Practices* (2010).

US Department of Health and Human Services 'Regulatory considerations for human cells, tissues, and cellular and tissue-based products: Minimal manipulation and homologous use: Guidance for Industry and Food and Drug Administration staff' (2017) *Food and Drug Administration* 1–24.

World Health Organization *Genomics and World Health: Report of the Advisory Committee on Health Research* (2002).

(c) Journal articles

Abul-Husn, NS, AO Obeng, SC Sanderson et al 'Implementation and utilization of genetic testing in personalized medicine' (2014) 7 *Pharmacogenomics Pers Med* 227–240.

Acharya, P & R Gautam 'Ethical perspectives of direct-to-consumer genetic testing' (2015) 11(1) *Online J Health Ethics* 1–12.

ACMG Board of Directors 'Direct-to-consumer genetic testing: A revised position statement of the American College of Medical Genetics and Genomics' (2016) 18(2) *Genet Med* 207–208.

Adams, R, F Adeleke, D Anderson et al 'POPIA code of conduct for research' (2021) 117(5/6) *S Afr J Sci* 1–12.

Adams, R, S Veldsman, M Ramsay et al 'Drafting a Code of Conduct for research under the Protection of Personal Information Act No. 4 of 2013' (2021) 117(5/6) *S Afr J Sci* 1–3.

Affdal, AO & V Ravitsky 'The best interest standard and the child's right to an open future' (2018) 18(8) *Am J Bioeth* 74–76.

Agurs-Collins, T, R Ferrer, A Ottenbacher et al 'Public awareness of direct-to-consumer genetic tests: Findings from the 2013 U.S. Health Information National Trends Survey' (2015) 30(4) *J Cancer Educ* 799–807.

Allen, L, M Hamidi & N Argintaru 'Direct-to-consumer genetic testing in Canada' (2011) 80(1) *UWOMJ*.

Allyse, M '23 and Me, we and you: Direct-to-consumer genetics, intellectual property and informed consent' (2013) 31(2) *Trends Biotechnol* 68–69.

Allyse, MA, DH Robinson, MJ Ferber et al 'Direct-to-consumer testing 2.0: Emerging models of direct-to-consumer genetic testing' (2018) 93(1) *Symposium on Precision Medicine* 113–120.

Altman, R 'Direct-to-consumer genetic testing: Failure is not an option' (2009) 86(1) *Clin Pharmacol Ther* 15–17.

American College of Medicine Genetics Board of Directors 'ACMG Statement on Direct-to-consumer Genetic Testing' (2004) 6(1) *Genet Med* 60.

American College of Obstatricians and Gynaecologists 'Ethical issues in genetic testing' (2008) *ACOG Committee Opinion No. 410* 1–8.

American Society of Human Genetics 'Advancing research and privacy: Achievements, challenges, and core principles' (2019) 105 *Am J Hum Genet* 445–447.

American Society of Human Genetics 'Statement on direct-to-consumer genetic testing in the United States' (2007) *Am J Hum Genet*.

Andorno, R 'The right not to know: An autonomy based approach' (2004) 30 *J Med Ethics* 435–439.

Annas, GJ, LH Glantz & PA Roche 'Drafting the Genetic Privacy Act: Science, policy and practical considerations' (1995) 23(4) *J Law Med Ethics* 360–366.

Annes, JP, MA Giovanni & MF Murray 'Risks of presymptomatic direct-to-consumer genetic testing' (2010) 363(12) *N Engl J Med* 1100–1101.

Antsaklis, P 'Autonomy and metrics of autonomy' (2020) 49 *Annu Rev Control* 15–26.

Apathy, NC, T Menser, LM Keeran et al 'Trends and gaps in awareness of direct-to-consumer genetic tests from 2007 to 2014' (2018) 54(6) *Am J Prev Med* 806–813.

Armstrong Holland, CM, EH Arbe-Barnes, EJ McGivern et al 'The 10th Oxbridge varsity medical ethics debate – Should we fear the rise of direct-to-consumer genetic testing?' (2018) 13(14) *Philos Ethics Humanit Med* 1–7.

Arribas-Ayllon, M 'Genetic testing and human subjectivity' (2015) 9(6) *Soc Personal Psychol Compass* 213–224.

Aspinall, M, SM Au, P Billings et al 'US system of oversight of genetic testing: A response to the charge of the Secretary of Health and Human Services' (2008) 5(5) *Per Med* 521–528.

Association for the Accreditation of Human Research Protections 'Privacy vs. confidentiality – What's the difference?' (2009) *University of Kentucky* 1–4.

Austin, J, A Semaka & G Hadjipavlou 'Conceptualising genetic counseling as psychotherapy in the era of genomic medicine' (2014) 23(6) *J Genet Counsel* 903–909.

Austin, SE & RA Hegele 'Clinical implications of direct-to-consumer genetic testing for cardiovascular disease risk' (2011) 27(6) *Can J Cardiol* 682–684.

Bair, S 'Direct-to-consumer genetic testing: Learning from the past and looking toward the future' (2012) 67 *Food & Drug LJ* 413–433.

Beard, M & JR Midgley 'Therapeutic privilege and informed consent: A justified erosion of patient autonomy' (2005) 51 *THRHR*.

Becker, F, CG van El, D Ibarreta et al 'Genetic testing and common disorders in a public health framework: How to assess relevance and possibilities' (2011) 19(1) *Eur J Hum Genet* 6–44.

Behrens, KG 'A critique of the principle of "respect for autonomy", grounded in African thought' (2018) 18(2) *Dev World Bioeth* 126–134.

Behrens, KG 'Towards an indigenous African bioethics' (2013) 6(1) *SAJBL* 32–35.

Benatar, D 'Confidentiality' (2003) 21 *CME* 11–14.

Benson, P 'Feminist second thoughts about free agency' (1990) 3 *Hypatia* 47–64.

Berg, C & K Fryer-Edwards 'The ethical challenges of direct-to-consumer genetic testing' (2008) 77(1) *J Bus Ethics* 17–31.

Bernstein, G 'Direct-to-consumer genetic testing: Gatekeeping the production of genetic information' (2010) 79(2) *UMKC L Rev* 1–12.

Berry, RM 'The genetic revolution and the physician's duty of confidentiality: The role of the old Hippocratic virtues in the regulation of the new genetic intimacy' (1997) 18(4) *J Leg Med* 401–441.

Biesecker, BB, MD Schwartz & TM Marteau ‘Enhancing informed choice to undergo health screening: A systematic review’ (2013) 37(3) *Am J Health Behav* 351–359.

Birks, D ‘How wrong is paternalism?’ (2018) 15(2) *J Moral Philos* 136–163.

Blell, M & M Hunter ‘Direct-to-consumer genetic testing’s red herring: “Genetic ancestry” and personalized medicine’ (2019) 6(48) *Front Med* 1–7.

Bloss, CS, BF Darst, EJ Topol et al ‘Direct-to-consumer personalized genomic testing’ (2011) 20(2) *Hum Mol Genet* 132–141.

Bloss, CS, NJ Schork & EJ Topol ‘Effect of direct-to-consumer genome-wide profiling to assess disease risk’ (2011) 364(6) *N Engl J Med* 524–534.

Bloss, CS, NE Wineinger, BF Darst et al ‘Impact of direct-to-consumer genomic testing at long term follow-up’ (2013) 50(6) *J Med Genet* 393–400.

Boddington, P ‘The ethics and regulation of direct-to-consumer genetic testing’ (2009) 1(71) *Genome Med* 1–2.

Bonomi, L, Y Huang & L Ohno-Machado ‘Privacy challenges and research opportunities for genomic data sharing’ (2020) 52 *Nat Genet* 646–654.

Borry, P ‘Europe to ban direct-to-consumer genetic tests?’ (2008) 26(7) *Nat Biotechnol* 736–737.

Borry, P, HC Howard, K Sénécal et al ‘Health-related direct-to-consumer genetic testing: A review of companies’ policies with regard to genetic testing in minors’ (2010) 9 *Fam Cancer* 51–59.

Borry, P, K Sénécal & BM Knoppers ‘Do it yourself newborn screening’ (2016) 170(6) *JAMA Pediatr* 523–524.

Borry, P, M Shabani & HC Howard ‘Nonpropositional content in direct-to-consumer genetic testing advertisements’ (2013) 13(5) *Am J Bioeth* 14–16.

Borry, P, MC Cornel & HC Howard ‘Where are you going, where have you been: A recent history of the direct-to-consumer genetic testing market’ (2010) 1(3) *J Community Genet* 101–106.

Borry, P, RE van Hellemondt, D Sprumont et al ‘Legislation on direct-to-consumer genetic testing in seven European countries’ (2012) 20 *Eur J Hum Genet* 715–721.

Botes, M, A Olckers & M Labuschaigne 'Data commercialisation in the South African health care context' (2021) 24 *PER/PELJ* 1–35.

Braverman, AM 'How the internet is reshaping assisted reproduction: From donor offspring registries to direct-to-consumer genetic testing' (2010) 11(2) *Minn JL Sci & Tech* 477–496.

Brett, GR, SA Metcalfe, DJ Amor et al 'An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice' (2012) 20 *Eur J Hum Genet* 825–830.

Brinegar, K, A Yetisen, S Choi et al 'The commercialization of genome-editing technologies' (2017) 37 *Crit Rev Biotechnol* 924–932.

Bunnik, EM, A Cecile, JW Janssens et al 'Informed consent in direct-to-consumer personal genome testing: The outline of a model between specific and generic consent' (2012) 28(7) *Bioethics* 343–351.

Bunnik, EM 'Do genomic tests enhance autonomy?' (2015) 41(4) *J Med Ethics* 315–316.

Bunnik, EM 'Why not order direct-to-consumer genetic testing for your children?' (2010) 6(3) *Genomics, Society and Policy* 68–70.

Burgermeister, J 'Switzerland has opened door to genetic discrimination, say ethicists' (2004) 329 *BMJ* 70.

Burton, A 'Are we ready for direct-to-consumer genetic testing?' (2015) 14(2) *Lancet Neurol* 138–139.

Cacchio, J 'What you don't know can hurt you: The legal risk of peering into the gene pool with direct-to-consumer genetic testing' (2018) 87(1) *UMKC L Rev* 219–244.

Callahan, D 'The puzzle of profound respect' (1995) 25(1) *Hastings Center Rep* 39–40.

Campbell, MC & SA Tishkoff 'African genetic diversity: Implications for human demographic history, modern human origins, and complex disease mapping' (2008) 9 *Annu Rev Genom Hum Genet* 403–433.

Cariati, F, V D'Argenio & R Tomaiuolo 'The evolving role of genetic tests in reproductive medicine' (2019) 17(267) *J Transl Med* 1–33.

Carter, S, D Taylor & I Bates 'Institutionalized paternalism? Stakeholders' views on public access to genetic testing' (2006) 11(3) *J Health Serv Res Policy* 155–161.

Caulfield, T & AL McGuire 'Direct-to-consumer genetic testing: Perceptions, problems, and policy responses' (2012) 63(1) *Annu Rev Med* 23–33.

Caulfield, T 'Direct-to-consumer testing: If consumers are not anxious, why are policymakers?' (2011) 130(1) *Hum Genet* 23–25.

Caulfield, T 'Genetic testing, liability, and regulatory policy: The Canadian situation' (2000) 41(1) *Jurimetrics* 7–21.

Caulfield, T, NM Ries, PN Ray et al 'Direct-to-consumer genetic testing: Good, bad or benign?' (2010) 77(2) *Clin Genet* 101–105.

Caulfield, T, S Chandrasekharan, Y Joly et al 'Harm, hype and evidence: ELSI research and policy guidance' (2013) 5(21) *Genome Med* 1–6.

Chandros Hull, S & K Prasad 'Reading between the lines: Direct-to-consumer advertising of genetic testing' (2001) 31(3) *Hastings Center Rep* 33–35.

Childress, JF 'The place of autonomy in bioethics' (1990) 20 *Hastings Center Rep* 12–17.

Cho, MK & P Sankar 'Forensic genetics and ethical, legal and social implications beyond the clinic' (2004) 36(11) *Nat Genet Supplement* 8–12.

Chokoshvili, D, DF Vears & P Borry 'Growing complexity of (expanded) carrier screening: Direct-to-consumer, physician-mediated, and clinic-based offers' (2017) 44 *Best Prac Res Cl Ob* 57–67.

Choudhury, A, M Ramsay, S Hazelhurst et al 'Whole-genome sequencing for an enhanced understanding of genetic variation among South Africans' 8 *Nat Commun* 1–12.

Chow-White, PA, M MacAulay, A Charters et al 'From the bench to the bedside in the big data age: Ethics and practices of consent and privacy for clinical genomics and personalized medicine' (2015) 17(3) *Ethics Inf Technol* 189–200.

Christman, J 'Relational autonomy and the social dynamics of paternalism' (2014) 17 *Ethic Theory Moral Prac* 369–382.

Christofides, E & K O'Doherty 'Company disclosure and consumer perceptions of the privacy implications of direct-to-consumer genetic testing' (2016) 35(2) *New Genet Soc* 101–123.

Clark, C 'Against confidentiality? Privacy, safety and the public good in professional communications' (2006) 6(2) *J Soc Work* 117–136.

Clarke, A & K Thirlaway '"Genomic counseling"? Genetic counseling in the genomic era' (2011) 3(7) *Genome Med* 1–3.

Cohen, IG, EY Adashi & V Ravitsky 'How bans on germline editing deprive patients with mitochondrial disease' (2019) 37(6) *Nat Biotechnol* 589–592.

Cohen-Almagor, R 'On the philosophical foundations of medical ethics: Aristotle, Kant, JS Mill and Rawls' (2017) 3 *Ethics Med Public Health* 436–444.

Cook-Deegan, R, C Derienzo, J Carbone et al 'Impact of gene patents and licensing practices on access to genetic testing for inherited susceptibility to cancer: Comparing breast and ovarian cancers with colon cancers' (2010) 12(4) *Genet Med* 15–38.

Cooke-Hubley, S & V Maddalena 'Access to genetic testing and genetic counseling in vulnerable populations: The d/deaf and hard of hearing population' (2011) 2 *J Community Genet* 117–125.

Copp, D 'The collective moral autonomy thesis' (2007) 38(3) *J Soc Philos* 369–388.

Cornel, MC, CG van El & P Borry 'The challenge of implementing genetic tests with clinical utility while avoiding unsound applications' (2014) 5(1) *J Community Genet* 7–12.

Cottrell, P 'Potential benefits and drawbacks to direct-to-consumer genetic testing' (2018) 1–5.

Covolo, L, S Rubinelli, E Ceretti et al 'Internet-based direct-to-consumer genetic testing: A systematic review' (2015) 17(12) *J Med Internet Res* 1–19.

Covolo, L, S Rubinelli, G Orizio et al 'Misuse (and abuse?) of the concept of empowerment. The case of online offer of predictive direct-to-consumer genetic tests' (2012) 1 *J Public Health Res* 7–10.

Critchley, C, D Nicol, M Otlowski et al 'Public reaction to direct-to-consumer online genetic tests: Comparing attitudes, trust and intentions across commercial and conventional providers' (2015) 24(6) *Public Underst Sci* 731–750.

Cummins, J & A Mak 'Direct-to-consumer gene testing in an Australian context' (2014) 4 *GenRe* 1–8.

Curnutte, M 'Challenging the boundaries of medicine: Consumer culture and genetic testing' (2012) 20(2) *Medic* 138–144.

Curnutte, M 'Regulatory controls for direct-to-consumer genetic tests: A case study on how the FDA exercised its authority' (2017) 36(3) *New Genet Soc* 209–226.

Curnutte, M & G Testa 'Consuming genomes: Scientific and social innovation in direct-to-consumer genetic testing' (2012) 31(2) *New Genet Soc* 159–181.

da Veiga, A, R Vorster, C Pilkington et al 'Compliance with the Protection of Personal Information Act and consumer privacy expectations: A comparison between the retail and medical aid industry' *16th International Information Security South Africa Conference* (August 2017 Johannesburg, South Africa) 16–23.

Daemmrich, AA '23andMe: The business and ethics of personal genetics testing' (2015) *University of Kansas School of Medicine* 1–15.

Dandara, C, J Greenberg, L Lambie et al 'Direct-to-consumer genetic testing: To test or not to test, that is the question' (2013) 103(8) *SAMJ* 510–512.

Dar-Nimrod, I & SJ Heine 'Genetic essentialism: On the deceptive determinism of DNA' (2011) 137(5) *Psychol Bull* 800–818.

De Vries, J, M Slabbert & MS Pepper 'Ethical, legal and social issues in the context of the planning stages of the Southern African human genome programme' (2012) 31(1) *Med Law* 119–152.

de Wet, S, H Oosthuizen & J Visser 'DNA profiling and the law in South Africa' (2011) 14(4) *PER/PELJ* 171–207.

Deber, RB, N Kraetschmer, S Urowitz et al 'Patient, consumer, client, or customer: What do people want to be called?' (2005) 8(4) *Health Expect* 345–351.

deBronkart, D 'From patient centred to people powered: Autonomy on the rise' (2015) *BMJ* 1–3.

Delaney, SK & MF Christman 'Direct-to-consumer genetic testing: Perspectives on its value in healthcare' (2016) 99(2) *Clin Pharmacol Ther* 146–148.

Deligiorgi, K 'Autonomy in bioethics' (2016) 3 *Symposion* 177–190.

Devisch, I 'Oughtonomy in healthcare: A deconstructive reading of Kantian autonomy' (2010) 13(4) *Med Health Care and Philos* 303–312.

Dhai, A 'Informed consent – 2008' (2008) 1(1) *SAJBL* 27–30.

Dick, HC ‘Risk and responsibility: State regulation and enforcement of the direct-to-consumer genetic testing industry’ (2012) 6(1) *St Louis UJ Health L & Pol’y* 167–200.

Dickenson, D ‘A corporate commons: Biobanking, patents and direct-to-consumer genetic tests’ (2012) *Conference: University of Maastricht* 1–7.

Dickenson, D ‘Testing times for the consumer genetics revolution’ (2014) 221(2951) *New Scientist* 26–27.

Domaradzki, J ‘Geneticization and biobanking’ (2019) *Polish Sociological Rev* 103–117.

Donner, MB, L VandeCreek, JC Gonsiorek et al ‘Balancing confidentiality: Protecting privacy and protecting the public’ (2008) 39(3) *Prof Psychol Res Pr* 369–376.

Dorfman, R, R Khan & G Mukerjee ‘Proposed regulatory framework for direct-to-consumer genetic testing: Diagnostics vs genetic screening’ (2014) 60(11) *Clin Chem* 1455–1456.

Dorsey, ER, KC Darwin, S Mohammed et al ‘Virtual research visits and direct-to-consumer genetic testing in Parkinson’s Disease’ (2015) *Digital Health* 1–18.

Dove, ES ‘Collection and protection of genomic data’ (2018) *University of Edinburgh School of Law, Research Paper Series No 2018/04* 1–10.

Dove, ES, SE Kelly, F Lucivero et al ‘Beyond individualism: Is there a place for relational autonomy in clinical practice and research?’ (2017) 12(3) *Clinical Ethics* 150–165.

Du, L & M Wang ‘Genetic privacy and data protection: A review of Chinese direct-to-consumer genetic test services’ (2020) 11(416) *Front Genet* 1–10.

Du, L & SI Becher ‘Genetic and genomic consultation: Are we ready for direct-to-consumer telegenetics?’ (2018) 9(550) *Front Genet* 1–8.

Ducournau, P, PA Gourraud, E Rial-Sebbag et al ‘Direct-to-consumer genetic testing through internet: Marketing, ethical and social issues’ (2011) 27(1) *Medecine Sciences* 95–102.

Dworkin, G ‘Acting freely’ (1970) 4(4) *Nous* 367–383.

Dworkin, G ‘Autonomy and behavior control’ (1976) 6 *Hastings Center Rep* 23–28.

Edelman, E & C Eng ‘A practical guide to interpretation and clinical application of personal genomic screening’ (2009) 339 *BMJ* 4253.

Edwards, KT & CJ Huang ‘Bridging the consumer-medical divide: How to regulate direct-to-consumer genetic testing’ (2014) 44(3) *Hastings Center Rep* 17–19.

Eissenberg, JC ‘Direct-to-consumer genomics: Harmful or empowering?’ (2017) 114(1) *Mo Med* 26–32.

Engelbrecht, SF ‘Can autonomy be limited – An ethical and legal perspective in a South African context?’ (2014) 32(1) *JFOS* 34–39.

Erlich, Y, JB Williams, D Glazer et al ‘Redefining genomic privacy: Trust and empowerment’ (2014) 12(11) *PLoS Biol* 1–5.

Esplin, E, E Haverfield, S Yang et al ‘Limitations of direct-to-consumer genetic screening for HBOC: False negatives, false positives and everything in between’ (2019) *Cancer Res*.

European Society of Human Genetics ‘Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes’ (2010) 18 *Eur J Hum Genet* 1271–1273.

Evans, JP & RC Green ‘Direct to consumer genetic testing: Avoiding a culture war’ (2009) 11(8) *Genet Med* 568–569.

Farham, B ‘Patient autonomy or patient confusion?’ (2014) 104(3) *SAMJ* 152.

Farmer, GD, H Gray, G Chandratillake et al ‘Recommendations for designing genetic test reports to be understood by patients and non-specialists’ (2020) 28 *Eur J Hum Genet* 885–895.

Farmer, MB, DC Bonadies, SM Mahon et al ‘Errors in genetic testing: The fourth case series’ (2019) 25(4) *Cancer J* 231–236.

Farrell, RM, PK Agatisa, M Mercer et al ‘Online direct-to-consumer messages about non-invasive prenatal genetic testing’ (2016) 1 *Reprod Biomed Soc Online* 88–97.

Fears, R & V ter Meulen ‘The perspective from EASAC and FEAM on direct-to-consumer genetic testing for health-related purposes’ (2013) 21(7) *Eur J Hum Genet* 703–707.

Feiler, T, K Gaitskell, T Maughan et al ‘Personalised medicine: The promise, the hype and the pitfalls’ (2017) 23(1) *New Bioeth* 1–12.

Felzmann, H ‘“Just a bit of fun”: How recreational is direct-to-customer genetic testing?’ (2015) 21(1) *New Bioeth* 20–32.

Fernandez Lynch, H, BE Bierer & IG Cohen ‘Confronting biospecimen exceptionalism in proposed revisions to the Common Rule’ (2016) 46(1) *Hastings Center Rep* 4–5.

Fieggen, KJ & NAB Ntusi 'Medical genetics, genomics and the future of medicine' (2019) 109(6) *SAMJ* 362–363.

Fieggen, KJ, LA Lambie & KA Donald 'Investigating developmental delay in South Africa: A pragmatic approach' (2019) 109(4) *SAMJ* 210–213.

Field, A, A Krokosky & SF Terry 'Direct-to-consumer marketing of genetic tests: Access does not reflect clinical utility' (2010) 14(6) *Genetic Alliance* 731–732.

Filipski, KK, JD Murphy & KJ Helzlsouer 'Updating the landscape of direct-to-consumer pharmacogenomic testing' (2017) 10 *Pharmgenomics Pers Med* 229–232.

Finnegan, T & A Hall 'Identification and genomic data' (2017) *PHG Foundation* 1–44.

Finney Rutten, LJ, SE Gollust, S Naveed et al 'Increasing public awareness of direct-to-consumer genetic tests: Health care access, internet use, and population density correlates' (2012) *J Cancer Epidemiol* 1–7.

Foster, MW & RR Sharp 'The contractual genome: How direct-to-consumer genomic services may help patients to take ownership of their DNA' (2008) 5(4) *Per Med* 399–404.

Fox, D 'Paying for particulars in people-to-be: Commercialisation, commodification and commensurability in human reproduction' (2008) 34(3) *J Med Ethics* 162–166.

Fox, D 'Reproductive negligence' (2017) 117(149) *Columbia L Rev* 149–242.

Fox, S 'Adolescents, graduated autonomy, and genetic testing' (2012) *Genet Res Int* 1–6.

Frank, L, J Wesson Ashford, PJ Bayley et al 'Genetic risk of Alzheimer's disease: Three wishes now that the genie is out of the bottle' (2018) 66(2) *J Alzheimers Dis* 421–423.

Frey, JA 'Against autonomy: Why practical reason cannot be pure' (2018) 41(4) *Manuscrito* 159–193.

Friend, L 'Direct-to-consumer genetic testing: Opportunities and risks in a rapidly evolving market' (2018) *KPMG* 1–12.

Frueh, FW, HT Greely, RC Green et al 'The future of direct-to-consumer clinical genetic tests' (2011) 12(7) *Nat Rev Genet* 511–515.

Fukuda, R & F Takada 'Legal regulations on health-related direct-to-consumer genetic testing in 11 countries' (2018) 48 *Kitasato Med J* 52–59.

Fulda, KG & K Lykens ‘Ethical issues in predictive genetic testing: A public health perspective’ (2006) 32(3) *J Med Ethics* 143–147.

Gammon, A & DW Neklason ‘Confidentiality & the risk of genetic discrimination: What surgeons need to know’ (2015) 24(4) *Surg Oncol Clin N Am*.

Garrison, NA & AL Non ‘Direct-to-consumer genomics companies should provide guidance to their customers on (not) sharing personal genomic information’ (2014) 14(11) *Am J Bioeth* 55–57.

Genzen, JR ‘Regulation of Laboratory-Developed Tests: A clinical laboratory perspective’ (2019) 152(2) *Am J Clin Pathol* 122–131.

Geransar, R & E Einsiedel ‘Evaluating online direct-to-consumer marketing of genetic tests: Informed choices or buyers beware?’ (2008) 12(1) *Genet Test* 13–23.

Gill, J, AJ Obley & V Prasad ‘Direct-to-consumer genetic testing: The implications of the US FDA’s first marketing authorization for BRCA mutation testing’ (2018) 319(23) *JAMA* 2377–2378.

Ginsburg, DH ‘Genetics and privacy’ (1999) 4(1) *Tex Rev Law Polit* 17–23.

Giovanni, MA, MR Fickie, LS Lehmann et al ‘Health-care referrals from direct-to-consumer genetic testing’ (2010) 14(6) *Genet Test Mol Biomarkers* 817–819.

Giraldi, L, M Colotto, R Pastorino et al ‘Medical student’s knowledge and attitude towards direct-to-consumer genetic tests’ (2016) 13(3) *Epidemiol Biostat Public Health* 1–8.

Giesen, D ‘From paternalism to self-determination and shared decision making’ (1988) 30 *Acta Juridica* 107–127.

Glen, S ‘Confidentiality: A critique of the traditional view’ (1997) 4(5) *Nurs Ethics* 403–406.

Gniady, JA ‘Regulating direct-to-consumer genetic testing: Protecting the consumer without quashing a medical revolution’ (2008) 76(5) *Fordham L Rev* 2429–2475.

Godard, B, L Kate, G Evers-Kiebooms et al ‘Population genetic screening programmes: Principles, techniques, practices, and policies’ (2003) 11 *Eur J Hum Genet* 49–87.

Goddard, KAB, D Duquette, A Zlot et al ‘Public awareness and use of direct-to-consumer genetic tests: Results from 3 state population-based surveys, 2006’ (2009) 99(3) *Am J Public Health* 442–445.

Goddard, KAB, J Robitaille, NF Dowling et al 'Health-related direct-to-consumer genetic tests: A public health assessment and analysis of practices related to internet-based tests for risk of thrombosis' (2009) 12(2) *Public Health Genom* 92–104.

Goldsmith, L, L Jackson, A O'Connor et al 'Direct-to-consumer genomic testing: Systematic review of the literature on user perspectives' (2012) 20(8) *Eur J Hum Genet* 811–816.

Goldsmith, L, L Jackson, A O'Connor et al 'Direct-to-consumer genomic testing from the perspective of the health professional: A systematic review of the literature' (2013) 4(2) *J Community Genet* 169–180.

Goldswain, GK 'The purposive approach to the interpretation of fiscal legislation – The winds of change' (2008) 16(2) *Meditari Accountancy Research* 107–121.

Gollust, SE, SW Gray, DA Carere et al 'Consumer perspectives on access to direct-to-consumer genetic testing: Role of demographic factors and the testing experience' (2017) 95(2) *Milbank Q* 291–318.

Gollust, SE, BS Wilfond & SC Hull 'Direct-to-consumer sales of genetic services on the internet' (2003) 5(4) *Genet Med* 332–337.

Gómez-Vírseda, C, Y de Maeseneer & C Gastmans 'Relational autonomy: What does it mean and how is it used in end-of-life care? A systematic review of argument-based ethics literature' (2019) 20 *BMC Med Ethics* 1–15.

Goodson, ML & BG Vernon 'A study of public opinion on the use of tissue samples from living subjects for clinical research' (2004) 57(2) *J Clin Pathol* 135–138.

Gostin, LO & JG Hodge 'Genetic privacy and the law: an end to genetic exceptionalism' (1999) 40 *Jurimetrics* 21–58.

Greaney, AM, DP O'Mathúna & PA Scott 'Patient autonomy and choice in healthcare: Self-testing devices as a case in point' (2012) 15(4) *Med Health Care and Philos* 383–395.

Green, RC & NA Farahany 'Regulation: The FDA is overcautious on consumer genomics' (2014) 505 *Nature* 286–287.

Greenleaf, G '2014-2017 update to Graham Greenleaf's *Asia Data Privacy Laws – Trade and Human Rights Perspectives*' (2017) 47 *UNSWLRS* 1–40.

Grimaldi, KA, MP Look, GA Scioli et al 'Personal genetics: Regulatory framework in Europe from a service provider's perspective' (2011) 19(4) *Eur J Hum Genet* 382–388.

Grishin, D, K Obbad & GM Church ‘Data privacy in the age of personal genomics’ (2019) 37(10) *Nat Biotechnol* 1115–1117.

Groves, C ‘Road-maps and revelations: On the somatic ethics of genetic susceptibility’ (2013) 32(3) *New Genet Soc* 264–284.

Guerrini, CJ, JK Wagner, SC Nelson et al ‘Who’s on third? Regulation of third-party genetic interpretation services’ (2020) 22 *Genet Med* 4–11.

Gupta, N ‘DNA extraction and Polymerase Chain Reaction’ (2019) 36(2) *J Cytol* 116–117.

Gurwitz, D & Y Bregman-Eschet ‘Personal genomics services: Whose genomes?’ (2009) 17(1) *Eur J Hum Genet* 883–889.

Gutmann Koch, V ‘PGTandMe: Social networking-based genetic testing and the evolving research model’ (2012) 22(1) *Health Matrix* 33–74.

Gutmann Koch, V & K Todd ‘Research revolution or status quo: The new common rule and research arising from direct-to-consumer genetic testing’ (2018) 56(1) *Hous L Rev* 81–121.

Gymrek, M, AL McGuire, D Golan et al ‘Identifying personal genomes by surname inference’ (2013) 339(6117) *Science* 321–324.

Haeusermann, T ‘Opinion: Get to know why people openly share genomic data’ (2017) *Scientist* 1–2.

Haeusermann, T, B Greshake, A Blasimme et al ‘Open sharing of genomic data: Who does it and why?’ (2017) 12(5) *PLoS One* 1–15.

Hall, DR ‘Avoiding paternalism but not moral perplexity’ (2018) 108(11) *SAMJ* 915–916.

Hall, JA, R Gertz, J Amato et al ‘Transparency of genetic testing services for “health, wellness and lifestyle”: Analysis of online prepurchase information for UK consumers’ (2017) 25(8) *Eur J Hum Genet* 908–917.

Hall, SA ‘Should public health respect autonomy?’ (1992) 18(4) *J Med Ethics* 197–201.

Hall, TO, AD Renz, KW Snapinn et al ‘Awareness and uptake of direct-to-consumer genetic testing among cancer cases, their relatives, and controls: The Northwest Cancer Genetics Network’ (2015) 16(7) *Genet Test Mol Biomarkers* 744–748.

Hamilton, JG, E Abdiwahab, HM Edwards et al 'Primary care providers' cancer genetic testing-related knowledge, attitudes, and communication behaviors: A systematic review and research agenda' (2017) 32(3) *J Gen Intern Med* 315–324.

Hanna, KE 'The ethical, legal, and social implications program of the National Center for Human Genome Research: A missed opportunity?' (1995) *National Academies Press*.

Harper, JC, J Geraedts, P Borry et al 'Current issues in medically assisted reproduction and genetics in Europe: Research, clinical practice, ethics, legal issues and policy' (2014) 29(8) *Hum Reprod* 1603–1609.

Harris, A, SE Kelly & S Wyatt 'Autobiologies on YouTube: Narratives of direct-to-consumer genetic testing' (2014) 33(1) *New Genet Soc* 60–78.

Harris, A, SE Kelly & S Wyatt 'Counseling customers: Emerging roles for genetic counselors in the direct-to-consumer genetic testing market' (2013) 22(2) *J Genet Counsel* 277–288.

Harris, A, S Wyatt & S Kelly 'Conceptualizing trust in digital environments health-e skepticism: Trust in the age of the internet' *A Decade in Internet Time: Presented at the panel contribution for the OII-iCS Symposium* (2011, Oxford) 1–11.

Harris, A, S Wyatt & S Kelly 'The gift of spit (and the obligation to return it): How consumers of online genetic testing services participate in research' (2013) 16(2) *Inf Commun Soc* 236–257.

Harvey, K & B Diug 'Editorial: Retail genetics' (2017) 40(3) *Australian Prescr* 86–87.

Hauskeller, C 'Direct to consumer genetic testing: Regulations cannot guarantee responsible use; an international industry certificate is needed' (2011) 342 *BMJ* 1163–1164.

Hawkins, AK & A Ho 'Genetic counseling and the ethical issues around direct to consumer genetic testing' (2012) 21(3) *J Genet Counsel* 367–373.

Haworth, L 'Review: Dworkin on autonomy' (1991) 102(1) *Ethics* 129–130.

Häyry, M & T Takala 'Genetic information, rights, and autonomy' (2001) 22(5) *Theor Med* 403–414.

Hazel, JW & C Slobogin 'Who knows what, and when? A survey of the privacy policies proffered by U.S. direct-to-consumer genetic testing companies' (2018) 28(1) *Cornell J Law Public Policy* 35–66.

Heidemann, S & K Schillhorn 'Implications of the German Human Genetic Examination Act on Laboratory Medicine' (2011) 35(5) *J Lab Med* 233–241.

Helgason, A & K Stefánsson 'The past, present, and future of direct-to-consumer genetic tests' (2010) 12(1) *Dialogues Clin Neurosci* 61–68.

Hendricks-Sturrup, RM & CY Lu 'Direct-to-consumer genetic testing data privacy: Key concerns and recommendations based on consumer perspectives' (2019) 9(2) *J Pers Med* 1–7.

Hendricks-Sturrup, RM, AER Prince & CY Lu 'Direct-to-consumer genetic testing and potential loopholes in protecting consumer privacy and nondiscrimination' (2019) 321(19) *JAMA* 1869–1870.

Hennen, L, A Sauter & E Van Den Cruyce 'Direct to consumer genetic testing: Insights from an internet scan' (2010) 29(2) *New Genet Soc* 167–186.

Herbst, MC 'Fact sheet on genetic testing for cancer' (2017) *CANSA* 1–15.

Herring, J & C Foster '"Please don't tell me" – The right not to know' (2012) 21(1) *Camb Q Healthc Ethics* 20–29.

Hoang, N, R Hayeems, J Davies et al 'Does personal genome testing drive service utilization in an adult preventive medicine clinic?' (2017) 8(3) *J Community Genet* 151–158.

Hock, KT, KD Christensen, BM Yashar et al 'Direct to consumer genetic testing: An assessment of genetic counselors' knowledge and beliefs' (2011) 13(4) *Genet Med* 325–332.

Hogarth, S & P Saukko 'A market in the making: The past, present and future of direct-to-consumer genomics' (2017) 36(3) *New Genet Soc* 197–208.

Hogarth, S 'Valley of the unicorns: Consumer genomics, venture capital and digital disruption' (2017) 36(3) *New Genet Soc* 250–272.

Hogarth, S, G Javitt & D Melzer 'The current landscape for direct-to-consumer genetic testing: Legal, ethical, and policy issues' (2008) 9 *Annu Rev Genom Hum Genet* 161–182.

Hogarth S & D Melzer 'The IVD Directive and genetic testing: Problems and proposals' *A briefing presented to the 20th meeting of Competent Authorities* (2007 Wellcome Trust, Lisbon) 1–22.

Hogarth, S, D Melzer & R Zimmern 'The regulation of commercial genetic testing services in the UK: A briefing for the Human Genetics Commission' (2005) *Genetic Services Sub-Group of the Human Genetics Commission* 1–23.

Hoglund-Shen, A 'Direct-to-consumer genetic testing, gamete donation, and the law' (2017) 55(3) *Fam Court Rev* 472–484.

Homer, N, S Szelinger, M Redman et al 'Resolving individuals contributing trace amounts of DNA to highly complex mixtures using high-density SNP genotyping microarrays' (2008) 4(8) *PLoS Genet* 1–9.

Horton, J 'Principles of biomedical ethics' (2002) 96(1) *Transactions of the Royal Society of Tropical Medicine and Hygiene* 107.

Horton, R & A Lucassen 'Consent and autonomy in the genomics era' (2019) 7 *Curr Genet Med Rep* 85–91.

Horton R & AM Lucassen 'The moral argument for heritable genome editing requires an inappropriately deterministic view of genetics' (2019) 45(8) *J Med Ethics* 526–527.

Horton RH & AM Lucassen 'Recent developments in genetic/genomic medicine' (2019) 133(5) *Clin Sci* 697–708.

Howard, HC & P Borry 'Direct-to-consumer genetic testing: The European regulatory landscape' *Centre for Biomedical Ethics and Law* 1–41.

Howard, HC & P Borry 'Direct-to-consumer genetic testing: More questions than benefits?' (2008) 5(4) *Per Med* 317–320.

Howard, HC & P Borry 'Direct-to-consumer pharmacogenomic testing' (2011) 12(10) *Pharmacogenomics* 1367–1370.

Howard, HC & P Borry 'Europe and direct-to-consumer genetic tests' (2011) 13 *Nat Rev Genet* 146.

Howard, HC & P Borry 'Survey of European clinical geneticists on awareness, experiences and attitudes towards direct-to-consumer genetic testing' (2013) 5(45) *Genome Med* 1–11.

Howard, HC & P Borry 'To ban or not to ban? Clinical geneticists' views on the regulation of direct-to-consumer genetic testing' (2012) 13(9) *EMBO Rep* 791–794.

Howard, HC, D Avard & P Borry 'Are the kids really all right? Direct-to-consumer genetic testing in children: Are company policies clashing with professional norms?' (2011) 19(11) *Eur J Hum Genet* 1122–1126.

Howard, HC, BM Knoppers & P Borry 'Blurring lines: The research activities of direct-to-consumer genetic testing companies raise questions about consumers as research subjects' (2010) 11(8) *EMBO Rep* 579–582.

Howes, R 'SALDA In Vitro Diagnostics in South Africa' (2014) *SALDA* 1–18.

Huang, H, H Yu, JE Andrews et al 'Uncovering hidden behavioral patterns in the era of "we media": Modeling the spatio-temporal dynamic for Twitter as a reporting tool for breaking news' *iConference* (2017) 1–4.

Huang, HY & M Bashir 'Direct-to-consumer genetic testing: Contextual privacy predicament' (2015) 52(1) *Proceedings of the Association for Information Science and Technology (ASIST)* 1–10.

Hudson, K, G Javitt, W Burke et al 'ASHG statement on direct-to-consumer genetic testing in the United States' (2007) 110(6) *Obstet Gynecol* 1392–1395.

Hull, SC, RR Sharp, JR Botkin et al 'Patients' views on identifiability of samples and informed consent for genetic research' (2008) 8(10) *Am J Bioeth* 62–70.

Hulshizer, R 'The impending impact of direct-to-consumer personal genomic services' (2010) 22(3) *ECRI Institute* 1–4.

Huml, AM, C Sullivan, M Figueroa et al 'Consistency of direct-to-consumer genetic testing results among identical twins' (2020) 133(1) *Am J Med* 143–146.

Idang, GE 'African culture and values' (2015) 16 *UNISA* 97–111.

Ikuenobe, P 'African communal basis for autonomy and life choices' (2018) 18(3) *Developing World Bioeth* 212–221.

Ikuenobe, P 'Relational autonomy, personhood, and african traditions' (2015) 65(4) *Philosophy East and West* 1005–1029.

Imai, K, LJ Kricka & P Fortina 'Concordance study of 3 direct-to-consumer genetic-testing services' (2011) 57(3) *Clin Chem* 518–521.

Irick, MD ‘Age of an information revolution: The direct-to-consumer genetic testing industry and the need for a holistic regulatory approach’ (2012) 49(1) *San Diego L Rev* 279–340.

Jackson, CS & MS Pepper ‘Opportunities and barriers to establishing a cell therapy programme in South Africa’ (2013) 4(54) *Stem Cell Res Ther* 1–7.

Jackson, L, L Goldsmith & H Skirton ‘Guidance for patients considering direct-to-consumer genetic testing and health professionals involved in their care: Development of a practical decision tool’ (2014) 31(3) *Fam Pract* 341–348.

Jacobs, W, P Stoop & R van Niekerk ‘Fundamental consumer rights under the Consumer Protection Act 68 of 2008: A critical overview and analysis’ (2010) 13(3) *PER/PELJ* 302–406.

Jamal, L, JC Sapp, K Lewis et al ‘Research participants’ attitudes towards the confidentiality of genomic sequence information’ (2014) 22(8) *Eur J Hum Genet* 964–968.

Janssens, ACJW, AAM Wilde & IM van Langen ‘The sense and nonsense of direct-to-consumer genetic testing for cardiovascular disease’ (2011) 19(2) *Neth Heart J* 85–88.

Janssens, ACJW & P Kraft ‘Research conducted using data obtained through online communities: Ethical implications of methodological limitations’ (2012) 9(10) *PLoS Med* 1–4.

Janssens, S, L Kalokairinou, D Chokoshvilli et al ‘Attitudes of cystic fibrosis patients and their parents towards direct-to-consumer genetic testing for carrier status’ (2015) 12(2) *Per Med* 99–107.

Jennings, B ‘Reconceptualizing autonomy: A relational turn in bioethics’ (2016) 46(3) *Hastings Center Rep* 11–16.

Jeong, G ‘Estimating the effects of consumer characteristics on the intention to use direct-to-consumer genetic testing’ (2018) 7(3) *Health Policy Technol* 233–238.

Jia, JL, DJ Polin & KY Sarin ‘Emerging technologies for health information in dermatology: Opportunities and drawbacks of web-based searches, social media, mobile applications, and direct-to-consumer genetic testing in patient care’ (2019) 38(1) *Sem Cutan Med Surg* 57–63.

Joh, EE ‘DNA theft: Recognizing the crime of nonconsensual genetic collection and testing’ (2010) 91(2) *B U L Rev, UC Davis Legal Studies Research Paper No. 232 91* 1–31.

Joly, Y, H Burton, BM Knoppers et al ‘Life insurance: Genomic stratification and risk classification’ (2014) 22(5) *Eur J Hum Genet* 575–579.

Jordaan, DW ‘The boy and his microscope: Interpreting section 56(1) of the National Health Act’ (2009) 2(1) *SAJBL* 12–19.

Jordaan, DW ‘Antipromethean fallacies: A critique of Fukuyama’s bioethics’ (2009) 28 *Biotechnol Law Rep* 577–590.

Jordaan, DW ‘Autonomy as an element of human dignity in South African case law’ (2009) 9(3) *Journal of Philosophy, Science & Law* 1–15.

Jordaan, DW ‘Human dignity and the future of the voluntary active euthanasia debate in South Africa’ (2017) 107(5) *SAMJ* 383–385.

Jordaan, DW ‘Science versus anti-science: The law on pre-embryo experimentation’ (2007) 124(3) *SALJ* 618–634.

Jordaan, DW ‘Social justice and research using human biological material: A response to Mahomed, Nöthling-Slabbert and Pepper’ (2016) 106(7) *SAMJ* 678–680.

Jordaan, DW, C Woodrow & MS Pepper ‘Banning private stem cell banks: A human rights analysis’ (2009) 25(1) *SAJHR* 126–151.

Jordaan, L ‘The legal validity of an advance refusal of medical treatment in South African law (part 1)’ (2011) 44(1) *De Jure* 32–48.

Joubert, E ‘Genetic testing and the insured’s right not to know’ (2009) 72 *THRHR* 17–25.

Käll, LF & K Zeiler ‘Bodily relational autonomy’ (2014) 21(9-10) *J Conscious Stud* 100–120.

Kalokairinou, L, HC Howard & P Borry ‘Current developments in the regulation of direct-to-consumer genetic testing in Europe’ (2015) 15(2-3) *Med Law Int* 97–123.

Kalokairinou, L, HC Howard, S Slokenberga et al ‘Legislation of direct-to-consumer genetic testing in Europe: A fragmented regulatory landscape’ (2018) 9(2) *J Community Genet* 117–132.

Kassirer, S, EE Levine & C Gaertig 'Decisional autonomy undermines advisees' judgments of experts in medicine and in life' (2020) 117(21) *Proc Natl Acad Sci USA* 11368–11378.

Kaye, J 'The regulation of direct-to-consumer genetic tests' (2008) 17(2) *Hum Mol Genet* 180–183.

Kaye, J 'The tension between data sharing and the protection of privacy in genomics research' (2012) 13 *Annu Rev Genomics Hum Genet* 415–431.

Kaye, J, EA Whitley, D Lund et al 'Dynamic consent: A patient interface for twenty-first century research networks' (2015) 23 *Eur J Hum Genet* 141–146.

Kent, A 'Consent and confidentiality in genetics: Whose information is it anyway?' (2003) 29(1) *J Med Ethics* 16–18.

Kessler, MD, L Yerges-Armstrong, MA Taub et al 'Challenges and disparities in the application of personalized genomic medicine to populations with African ancestry' (2016) 7(12521) *Nat Commun* 1–8.

Kevles, DJ 'From eugenics to patents: Genetics, law, and human rights' (2011) 75(3) *Ann Hum Genet* 326–333.

Keyter, A, S Banoo, S Salek et al 'The South African regulatory system: Past, present, and future' (2018) 9(1407) *Front Pharmacol* 1–13.

Khader, SJ 'The feminist case against relational autonomy' (2020) 17 *J Moral Philos* 499–526.

Khan, R & D Mittelman 'Consumer genomics will change your life, whether you get tested or not' (2018) 19(120) *Genome Biol* 120–124.

Kim, EE 'Personalized medicine: Promises and pitfalls' (2017) 58 *J Nucl Med* 861.

Kim, J 'Health technology update' (2017) 18 *CADTH* 1–17.

Kim, JW 'Direct-to-consumer genetic testing' (2019) 17(3) *Genomics Inform* 34.

Kim, JW 'Regulated sandbox and disease-related genetic tests as a direct-to-consumer test' (2019) 62(5) *J Korean Med Assoc* 240–242.

Kim, S 'Direct to consumer genetic tests and its social and ethical implications' (2013) 7 *Bioethics Pol'y Stud* 23–43.

Kin, CA ‘Coming soon to the “genetic supermarket” near you’ (1996) 48(6) *Stanford L Rev* 1573–1604.

King, JS ‘And genetic testing for all – The coming revolution in non-invasive prenatal genetic testing’ (2011) 42 *Rutgers LJ* 599–658.

Kirby, N ‘Gene editing and South African law’ (2016) 16(3) *Without Prejudice* 16–17.

Kishore, D ‘Test at your own risk: Your genetic report card and the direct-to-consumer duty to secure informed consent’ (2010) 59 *Emory LJ* 1553–1609.

Knapp, S & L VandeCreek ‘Balancing respect for autonomy with competing values with the use of principle-based ethics’ (2007) 44(4) *Psychother: Theory, Research, Practice, Training* 397–404.

Knapp van Bogaert, D & G Ogunbanjo ‘The human genome and gene “therapy”: Some ethical issues’ (2010) 52(1) *SA Fam Pract* 40–41.

Knoppers, BM, JR Harris, I Budin-Ljøsne et al ‘A human rights approach to an international code of conduct for genomic and clinical data sharing’ (2014) 133 *Hum Genet* 895–903.

Knoppers, BM, D Avard & HC Howard ‘Direct-to-consumer genetic testing: Driving choice?’ (2010) 10(8) *Expert Rev Mol Diagn* 965–968.

Knoppers, BM, R Isasi, T Caulfield et al ‘Human gene editing: Revisiting Canadian policy’ (2017) 2(3) *Regen Med* 1–2.

Kopinsky, SM ‘Communication in genetic counselling’ (1982) 62(21) *SAMJ* 758–764.

Kotzé, M ‘Liberal democracy and the genetic supermarket: Autonomy and freedom in Jürgen Moltmann’s political theology and biotechnology’ (2015) 1(1) *STJ* 201–215.

Krause, A ‘Benefits and pitfalls of genetic testing’ (2017) 17 *The Specialist Forum* 32–34.

Krause, A ‘New genetic testing technologies: Advantages and limitations’ (2019) 109(4) *SAMJ* 207–209.

KRISP ‘KRISP newsletter: Genomics Africa’ (2019) 3(7) *KRISP* 1–8.

Kromberg, JGR, EB Sizer & AL Christianson ‘Genetic services and testing in South Africa’ (2013) 4(3) *J Community Genet* 413–423.

Kromberg, JGR & A Krause 'Human genetics in Johannesburg, South Africa: Past, present and future (2013) 103(12) *SAMJ* 957–961.

Kromberg, JGR, TM Wessels & A Krause 'Roles of genetic counselors in South Africa' (2013) 22(6) *J Genet Counsel* 753–761.

Kulchak Rahm, A, H Spencer Feigelson, N Wagner et al 'Perception of direct-to-consumer genetic testing and direct-to-consumer advertising of genetic tests among members of a large managed care organization' (2005) 21(3) *J Genet Counsel* 448–461.

Kutz, G 'Direct-to-consumer genetic tests: Misleading test results are further complicated by deceptive marketing and other questionable practices' (2010) *US GAO* 1–29.

Lacaze, P, J Tiller & J Ryan 'The dangers of direct-to-consumer genetic testing for Alzheimer's Disease: Comment on "personal genomic testing, genetic inheritance, and uncertainty"' (2017) 14(4) *J Bioeth Inq* 585–587.

Laestadius, LI, JR Rich & PL Auer 'All your data (effectively) belong to us: Data practices among direct-to-consumer genetic testing firms' (2016) 19(5) *Genet Med* 513–520.

Lafferriere, JN 'Protecting privacy and family life in the era of recreational genetics and big data' (2016) 7 *Int J Juris Fam* 33–60.

Langford, AT, K Resnicow, JS Roberts et al 'Racial and ethnic differences in direct-to-consumer genetic tests awareness in HINTS 2007: Sociodemographic and numeracy correlates' (2012) 21(3) *J Genet Counsel* 440–447.

Lee, SSJ & L Crawley 'Research 2.0: Social networking and DTC genomics' (2009) 9(6-7) *Am J Bioeth* 35–44.

Lee, SSJ, S Vernez, KE Ormond et al 'Attitudes towards social networking and sharing behaviors among consumers of direct-to-consumer personal genomics' (2013) 3 *J Pers Med* 275–287.

Leighton, JW, K Valverde & BA Bernhardt 'The general public's understanding and perception of direct-to-consumer genetic test results' (2012) 15(1) *Public Health Genomics* 11–21.

Li, M & EP Diamandis (eds) 'Direct-to-consumer testing Q&A' (2017) 63 *Clin Chem* 635–641.

Lim, YA, TH Um, YH Chang et al 'A draft proposal for Korean classification and coding system for in vitro diagnostic medical devices' (2012) 2(3) *Lab Med Online* 148–165.

Lin, Z, AB Owen & RB Altman 'Genetics. Genomics research and human subject privacy' (2004) 305(5681) *Science* 183.

Lippi, G, EJ Favaloro & M Plebani 'Direct-to-consumer testing: More risks than opportunities' (2011) 65(12) *Int J Clin Pract* 1221–1229.

Lippman, A 'Led (astray) by genetic maps: The cartography of the human genome and health care' (1992) 35(12) *Soc Sci Med* 1469–1476.

Liu, Y & YE Pearson 'Direct-to-consumer marketing of predictive medical genetic tests: Assessment of current practices and policy recommendations' (2008) 27(2) *J Public Policy Mark* 131–148.

Loi, M 'Direct to consumer genetic testing and the libertarian right to test' (2016) 42(9) *J Med Ethics* 574–577.

Lombard, Z, F Baine, A Krause et al 'Implications of direct-to-consumer whole-exome sequencing in South Africa' (2016) 106(2) *SAMJ* 139–140.

Lu, M, CM Lewis & M Traylor 'Pharmacogenetic testing through the direct-to-consumer genetic testing company 23andMe' (2017) 10(47) *BMC Medical Genomics* 1–8.

Lucassen, A & M Parker 'Confidentiality and serious harm in genetics – Preserving the confidentiality of one patient and preventing harm to relatives' (2004) 12(2) *Eur J Hum Genet* 93–97.

Lucivero, F & B Prainsack 'The lifestylisation of healthcare? "Consumer genomics" and mobile health as technologies for healthy lifestyle' (2015) 4 *Appl Transl Genom* 44–49.

Lwoff, L 'Council of Europe adopts protocol on genetic testing for health purposes' (2009) 17(11) *Eur J Hum Genet* 1374–1377.

Lynch, J, A Parrott, RJ Hopkin et al 'Media coverage of direct-to-consumer genetic testing' (2011) 20(5) *J Genet Counsel* 486–494.

Magnus, D, MK Cho & R Cook-Deegan 'Direct-to-consumer genetic tests: Beyond medical regulation?' (2009) 1(2) *Genome Med* 1–4.

Mahesh, KP 'Laws and regulations associated with ownership of human biological material in South Africa' (2015) 8(1) *SAJBL* 11–18.

Mahmoud-Davis, SA 'Direct-to-consumer genetic testing: Empowering EU consumers and giving meaning to the informed consent process within the IVDR and GDPR frameworks' (2020) 19(1) *Wash U Global Stud L Rev* 1–52.

Mahomed, S, M Nöthling-Slabbert & MS Pepper 'Ownership and human tissue – The legal conundrum: A response to Jordaan's critique' (2017) 107(3) *SAMJ* 196–198.

Mahomed, S, M Nöthling-Slabbert & MS Pepper 'The legal position on the classification of human tissue in South Africa: Can tissues be owned?' (2013) 6(1) *SAJBL* 16–20.

Mainetti, R, S Oliveri, A Gorini et al 'Usability testing of two mini-games and one serious game to educate people about genetics' *pHealth 16th International Conference on Wearable, Micro & Nano Technologies for Personalized Health* (10-12 June 2019 Genoa, Italy) 82–87.

Majdik, ZP 'Judging direct-to-consumer genetics: Negotiating expertise and agency in public biotechnological practice' (2009) 12(4) *Rhetor Public Aff* 571–606.

Majumder, MA, CJ Guerrini & AL McGuire 'Direct-to-consumer genetic testing: Value and risk' (2021) 72 *Annu Rev Med* 155–166.

Malin, B, D Karp & RH Scheuermann 'Technical and policy approaches to balancing patient privacy and data sharing in clinical and translational research' (2010) 58(1) *J Investig Med* 11–18.

Malm, H 'Genetic privacy: Might there be a moral duty to share one's genetic information?' (2009) 9(6-7) *Am J Bioeth* 52–54.

Mangin, D, B Hudson & L Toop 'Direct-to-consumer genetic testing: Letting the genie out of the bottle?' (2012) 125(1364) *NZMJ* 7–10.

Manolio, TA 'Genomewide Association Studies and assessment of the risk of disease' (2010) 363(2) *New Eng J Med* 166–176.

Manrique de Lara, A, L Soto-Gómez, E Núñez-Acosta et al 'Ethical issues in susceptibility genetic testing for late-onset neurodegenerative diseases' (2019) 180(8) *Am J Med Genet* 609–621.

Manyonga, H, G Howarth, M Dinwoodie et al 'From informed consent to shared decision-making' (2014) 104(8) *SAMJ* 561–562.

Marietta, C & AL McGuire 'Direct-to-consumer genetic testing: Is it the practice of medicine?' (2009) 37(2) *J Law Med Ethics* 369–374.

Martin, K 'Understanding privacy online: Development of a social contract approach to privacy' (2016) 137 *J Bus Ethics* 551–569.

Martínez Otero, JM '"Better not to know?": Justifiable limits on the right to information in the realm of DTC genetic testing. An analysis of the European and Spanish legal framework' (2017) 24(2) *Eur J Health Law* 175–197.

Martínez Otero, JM 'Commercial speech in the realm of direct-to-consumer genetic testing. Are the current bans in the Spanish legal framework necessary and proportionate?' (2017) 46 *Law Human Genome Rev* 177–196.

Mathews, R, W Hall & A Carter 'Direct-to-consumer genetic testing for addiction susceptibility: A premature commercialisation of doubtful validity and value' (2012) 107(12) *Addiction* 2069–2074.

Mavroidopoulou, V, E Xera & V Mollaki 'Awareness, attitudes and perspectives of direct-to-consumer genetic testing in Greece: A survey of potential consumers' (2015) 60(9) *J Hum Genet* 515–523.

McBride, CM, CH Wade & KA Kaphingst 'Consumers' views of direct-to-consumer genetic information' (2010) 11 *Annu Rev Genom Hum Genet* 427–446.

McCabe, LL & ER McCabe 'Direct-to-consumer genetic testing: Access and marketing' (2004) 6(1) *Genet Med* 58–59.

McCarthy, JJ, HL McLeod & GS Ginsburg 'Genomic medicine: A decade of successes, challenges, and opportunities' (2013) 5(189) *Sci Transl Med* 1–17.

McCrudden, C 'Human dignity and judicial interpretation of human rights' (2008) 19(4) *EJIL* 655–724.

McGirr, J 'Quantifying moral & legal culpability with epigenetics: Case studies with PTSD, BPA, and Fetal Alcohol Syndrome' *Bioethics Symposium, Brown University* 1–13.

McGrath, SP, J Coleman, L Najjar et al 'Comprehension and data-sharing behavior of direct-to-consumer genetic test customers' (2016) 19(2) *Public Health Genomics* 116–124.

McGuire, AL & RA Gibbs 'Genetics. No longer de-identified' (2006) 312(5772) *Science* 370–371.

McGuire, AL & W Burke 'Raiding the medical commons: An unwelcome side effect of direct-to-consumer personal genome testing' (2008) 300(22) *JAMA* 2669–2671.

McGuire, AL, R Fisher, P Cusenza et al 'Confidentiality, privacy, and security of genetic and genomic test information in electronic health records: Points to consider' (2008) 10(7) *Genet Med* 495–499.

McGuire, AL, BJ Evans, T Caulfield et al 'Regulating direct-to-consumer personal genome testing' (2010) 330(6001) *Science* 181–182.

McGuire, AL, CM Diaz, T Wang et al 'Social networkers' attitudes toward direct-to-consumer personal genome testing' (2009) 9(6-7) *Am J Bioeth* 3–10.

McInerny, J 'Education in a genomic world' (2002) 27(3) *J Med Philos* 369–390.

McQuoid-Mason, D 'Medical ethics and the payment of fees before treatment' (2011) 101(11) *SAMJ* 798–799.

McQuoid-Mason, D 'Michael Jackson and the limits of patient autonomy' (2012) 5(1) *SAJBL* 11–14.

McQuoid-Mason, D & MA Dada 'The National Health Act: Some implications for family practice' (2006) 24(1) *CME* 12–14.

Meintjes-Van Der Walt, L 'Dealing with DNA-evidence: S v Maqhina 2001 (1) SACR 241 (T)' (2001) 14 *S Afr J Crim Just* 378–383.

Metz, T 'How the West was one: The Western as individualist, the African as communitarian' (2015) 47(11) *Educ Philos Theory* 1175–1184.

Meyers, DT 'Personal autonomy and the paradox of feminine socialization' (1987) 84 *J Philos* 619–628.

Middleton, A, A Mendes, CM Benjamin et al 'Direct-to-consumer genetic testing: Where and how does genetic counseling fit?' (2017) 14(3) *Per Med* 249–257.

Miller, MJ & P Freeman 'Legal issues relating to epigenetics in life insurance' *Presented at the 2018 Association of Life Insurance Counsel (ALIC) Annual Meeting* (7 May 2018) 1–12.

Mokgobi, M 'Understanding traditional African healing' (2014) 20 *Afr J Phys Health Educ Recreat Dance* 24–34.

Moodley, K, N Sibanda, K February et al '"It's my blood": Ethical complexities in the use, storage and export of biological samples: Perspectives from South African research participants' (2014) 15(4) *BMC Med Ethics* 1–7.

Morawetz, T 'Confidentiality and common sense: Insights from philosophy' (2011) 48 *San Diego L Rev* 357–378.

Morrison, AL 'Research revolution: Genetic testing consumers become research (and privacy) guinea pigs' (2011) 9(2) *J Telecommun High Technol Law* 573–605.

Moscarello, T, B Murray, CM Reuter et al 'Direct-to-consumer raw genetic data and third-party interpretation services: more burden than bargain?' (2019) 21(3) *Genet Med* 539–541.

Motloba, P & N Makwakwa 'Respecting autonomy (part 2)' (2018) 73(7) *S Afr Dent J* 460–462.

Motloba, PD 'Understanding of the principle of autonomy (part 1)' (2018) 73(6) *S Afr Dent J* 418–420.

Muhammad, R, C Ianuale, W Ricciardi et al 'Direct-to-consumer genetic testing: A systematic review of European guidelines, recommendations, and position statements' (2015) 19(10) *Genet Test Mol Biomarkers* 1–13.

Munung, NS, BM Mayosi & J de Vries 'Genomics research in Africa and its impact on global health: Insights from African researchers' (2018) 3 *Glob Health Epidemiol* 1–8.

Myers, MF & BA Bernhardt 'Direct-to-consumer genetic testing: Introduction to the special issue' (2012) 21(3) *J Genet Counsel* 357–360.

Mykitiuk, R 'Caveat emptor: Direct-to-consumer supply and advertising of genetic testing' (2004) 27(1) *Clin Invest Med* 23–32.

Nabholz, C & F Rechfeld 'Seeing the future? How genetic testing will impact life insurance' (2017) *Swiss Re* 1–16.

National Pathology Accreditation Advisory Council 'The provision of direct to consumer genetic tests: Guiding principles for providers' (2014) *NPAAC* 1–30.

National Planning Commission 'Our future – Making it work: National development plan 2020 executive summary' (2012) *National Planning Commission* 1–66.

Naveed, M, E Ayday, EW Clayton et al 'Privacy in the genomic era' (2015) 48(1) *ACM Comput Surv* 1–44.

Ndemo, B & T Weiss 'Making sense of Africa's emerging digital transformation and its many futures' (2017) 3(3-4) *Afr J Manag* 328–347.

Negley, G 'Philosophical views on the value of privacy' (1966) 31 *Law Contemp Probl* 319–325.

Nembaware, V, K Johnston, AA Diallo et al 'A framework for tiered informed consent for health genomic research in Africa' (2019) 51 *Nat Genet* 1566–1571.

Nicol, D, M Hagger, N Ries et al 'Time to get serious about privacy policies: The special case of genetic privacy' (2014) 42(1) *Fed L Rev* 149–179.

Nicol, D & M Hagger 'Direct-to-consumer genetic testing – A regulatory nightmare?' (2013) 198(9) *Med J Aust* 501–502.

Niemiec, E & HC Howard 'Ethical issues in consumer genome sequencing: Use of consumers' samples and data' (2016) 8 *Appl Transl Genom* 23–30.

Niemiec, E, P Borry, P Pinxten et al 'Content analysis of informed consent for whole genome sequencing offered by direct-to-consumer genetic testing companies' (2016) 37(12) *Hum Mutat* 1248–1256.

Niemiec, E, L Kalokairinou & HC Howard 'Current ethical and legal issues in health-related direct-to-consumer genetic testing' (2017) 14(5) *Pers Med* 433–445.

Nienaber, A 'Consent to and authorisation of the export and use of human biological specimens for future research – Perspectives from three African countries' (2011) 44(2) *CILSA* 225–254.

Nienaber, A & KN Bailey 'The right to physical integrity and informed refusal: Just how far does a patient's right to refuse medical treatment go?' (2016) 9(2) *SAJBL* 73–77.

Nnamuchi, O 'Confidentiality in medicine – A really decrepit concept in Africa' (2017) 36(1) *Med Law* 99–112.

Nordgren, A 'Designing preclinical studies in germline gene editing: Scientific and ethical aspects' (2019) 16(4) *Bioethical Inquiry* 559–570.

Norrgard, K 'DTC genetic testing for diabetes, breast cancer, heart disease and paternity' (2008) 1(1) *Nat Education* 86.

Nortvedt, P & F Vosman 'An ethics of care: New perspectives, both theoretically and empirically?' (2014) 21(7) *Nurs Ethics* 753–754.

Nöthling Slabbert, M & M Pepper '"A room of our own?" Legal lacunae regarding genomic sovereignty in South Africa' (2010) 73 *THRHR* 432–450.

Novy, MC 'Privacy at a price: Direct-to-consumer genetic testing & the need for regulation' (2010) 1 *JL Tech & Pol'y* 157–180.

O'Daniel, JM 'The prospect of genome-guided preventive medicine: A need and opportunity for genetic counsellors' (2010) 19(4) *J Genet Counsel* 315–327.

Ogbogu, U, A Zarzeczny, J Baltz et al 'Research on human embryos and reproductive materials: Revisiting Canadian law and policy' (2018) 13(3) *Healthc Policy* 10–19.

Ogunrin, O, K Woolfall, M Gabbay et al 'Relative solidarity: Conceptualising communal participation in genomic research among potential research participants in a developing Sub-Saharan African setting' (2018) 13(4) *PLoS ONE* 1–18.

Oh, B 'Direct-to-consumer genetic testing: Advantages and pitfalls' (2019) 17(3) *Genomics Inform* 1–3.

Oliveri, S & G Pravettoni 'The disclosure of direct-to-consumer genetic testing: Sounding out the psychological perspective of consumers' (2016) 8(5) *Biol Med* 1–4.

Oliveri, S, G Pravettoni, C Fioretti et al 'Let the individuals directly concerned decide: A solution to tragic choices in genetic risk information' (2016) 19(5) *Public Health Genomics* 307–313.

Oliveri, S, C Renzi, M Masiero et al 'Living at risk: Factors that affect the experience of direct-to-consumer genetic testing' (2015) 90(10) *Mayo Clin Proc* 1323–1326.

Olivier, B 'Foucault and individual autonomy' (2010) 40(3) *S Afr J Psychol* 292–307.

Ormond, KE 'From genetic counseling to "genomic counseling"' (2013) 1(4) *Mol Genet Genomic Med* 189–193.

Orth, M & P Luppa 'Direct-to-consumer-testing' (2015) 112 *Themen Der Zeit* 174–176.

Ortiz, AP, M López, LT Flores et al 'Awareness of direct-to-consumer genetic tests and use of genetic tests among Puerto Rican adults, 2009' (2011) 8(5) *Prev Chronic Dis* 1–11.

Ostergren, JE, MC Gornick, DA Carere et al 'How well do customers of direct-to-consumer personal genomic testing services comprehend genetic test results? Findings from the impact of personal genomics study' (2015) 18(4) *Public Health Genomics* 216–224.

P^{3}G Consortium, G Church, C Heeney et al 'Public access to genome-wide data: Five views on balancing research with privacy and protection' (2009) 5(10) *PLoS Genet* 1–4.

Palmer, JE 'Genetic gatekeepers: Regulating direct-to-consumer genomic services in an era of participatory medicine' (2012) 67(4) *Food & Drug LJ* 475–524.

Pàmpols Ros, T, JM García Sagredo, A Pérez Aytés et al 'Directed to consumer genetic testing: Perspective from the Ethics Commission of the Spanish Society for Human Genetics' (2019) 153(1) *Med Clin (Barc)* 35–40.

Paquin, RS, AS Richards, LM Koehly et al 'Exploring dispositional tendencies to seek online information about direct-to-consumer genetic testing' (2012) 2(4) *Behav Med Pract Policy Res* 392–400.

Park, JY, MT Risher T Caulfield et al 'Privacy in direct-to-consumer genetic testing' (2019) 65(5) *Clin Chem* 612–617.

Parker, Z, S Maslamoney, A Meintjes et al 'Building infrastructure for African human genomic data management' (2019) 18(47) *Data Sci J* 1–8.

Parliamentary Office of Science & Technology 'Consumer genetic testing' (2012) *POSTNOTE* 1–4.

Parthasarathy, S 'Assessing the social impact of direct-to-consumer genetic testing: Understanding sociotechnical architectures' (2010) 12(9) *Genet Med* 544–547.

Partlett, DF 'Misuse of genetic information: The common law and professionals' liability' (2003) 42(3) *Washburn LJ* 489–523.

Patch, C, J Sequeiros & MC Cornel 'Direct to consumer genetic tests' (2009) 17 *Eur J Hum Genet* 1111.

Patch, C, J Sequeiros & MC Cornel 'Erratum: Direct to consumer genetic tests' (2009) 17 *Eur J Hum Genet* 1204.

Patch, C, J Sequeiros & MC Cornel 'Genetic horoscopes: Is it all in the genes? Points for regulatory control of direct-to-consumer genetic testing' (2009) 17(7) *Eur J Hum Genet* 857–859.

Pearson, YE & Y Liu-Thompkins 'Consuming direct-to-consumer genetic tests: The role of genetic literacy and knowledge calibration' (2012) 31(1) *J Public Policy Mark* 42–57.

Pelto-Piri, V, K Engström & I Engström 'Paternalism, autonomy and reciprocity: Ethical perspectives in encounters with patients in psychiatric in-patient care' (2013) 14(49) *BMC Med Ethics* 1–8.

Pepper, MS 'Exporting DNA – Striking a balance between preventing exploitation and promoting innovation' (2017) 107(2) *SAMJ* 106–107.

Pepper, MS 'The human genome and molecular medicine – Promises and pitfalls' (2010) 100(11) *SAMJ* 722–724.

Pepper, MS 'The stem cell regulatory environment in South Africa – Cause for concern' (2009) 99(7) *SAMJ* 505–507.

Pepper, MS & MN Slabbert 'Human tissue legislation in South Africa: Focus on stem cell research and therapy' (2015) 8(2) *SAJBL* 4–11.

Pepper, MS, C Dandara, J de Vries et al 'An optimistic vision for biosciences in South Africa: Reply to Thaldar et al. (2019)' (2019) 115(7/8) *S Afr J Sci* 1.

Pepper, MS, C Dandara, J de Vries et al 'ASSAf consensus study on the ethical, legal and social implications of genetics and genomics in South Africa' (2018) 114(11/12) *S Afr J Sci* 1–3.

Phillips, AM 'Genomic privacy and direct-to-consumer genetics: Big consumer genetic data – What's in that contract?' (2015) *IEEE Symposium on Security and Privacy Workshops (SPW)* (21-22 May 2015 San Jose, California) 60–64.

Phillips, AM 'Reading the fine print when buying your genetic self online: Direct-to-consumer genetic testing terms and conditions' (2017) 36(3) *New Genet Soc* 273–295.

Phillips, KA, JR Trosman & MP Douglas 'Emergence of hybrid models of genetic testing beyond direct-to-consumer or traditional labs' (2019) 321 *JAMA* 2403.

Piehl, M 'Regulating hype and hope: A business ethics model approach to potential oversight of direct-to-consumer genetic tests' (2011) 16 *Mich St U J Med & L* 59–93.

Pieper, I 'Considering relational autonomy' *Austalasian Ethics Network Conference* (2014 Sydney, Australia).

Pieterse, M ‘The interdependence of rights to health and autonomy in South Africa’ (2008) 125 *SALJ* 553–572.

Pike, ER ‘Securing sequences: Ensuring adequate protections for genetic samples in the age of big data’ (2015) 37 *Cardozo L Rev* 1977–2033.

Plöthner, M, M Klora, D Rudolph et al ‘Health-related genetic direct-to-consumer tests in the German setting: The available offer and the potential implications for a solidarily financed health-care system’ (2017) 20 *Public Health Genomics* 203–217.

Politi, MC, DS Dizon, DL Frosch et al ‘Importance of clarifying patients’ desired role in shared decision making to match their level of engagement with their preferences’ (2013) 347 *BMJ*.

Popovsky, M ‘Exaggerated benefits and underestimated harms: The direct-to-market consumer genetic test market and how to manage it going forward’ (2010) 8(2) *Dartmouth LJ* 65–87.

Powell, KP, CA Christianson, WA Cogswell et al et al ‘Educational needs of primary care physicians regarding direct-to-consumer genetic testing’ (2012) 21(3) *J Genet Counsel* 469–478.

Powell, KP, WA Cogswell, CA Christianson et al ‘Primary care physicians’ awareness, experience and opinions of direct-to-consumer genetic testing’ (2011) 21(1) *J Genet Counsel* 113–126.

Prainsack, B ‘The power of prediction: How personal genomics became a policy challenge’ (2011) 40(4) *ÖZP* 401–415.

Prainsack, B & E Vayena ‘Beyond the clinic: “Direct-to-consumer” genomic profiling services and pharmacogenomics’ (2013) 14(4) *Pharmacogenomics* 403–412.

Prainsack, B, J Reardon, R Hindmarsh et al ‘Personal genomes: Misdirected precaution’ (2008) 456(7218) *Nature* 34–35.

Prystajecky, N, FSL Brinkman, B Auk et al ‘Personalized genetic testing and norovirus susceptibility’ (2014) 25(4) *Can J Infect Dis Med Microbiol* 222–224.

Pucheta, LL ‘Informed consent on direct to consumer genetic tests’ (2018) 85 *Prudentia Iuris* 137–150.

Rao, R ‘A veil of genetic ignorance? Protecting genetic privacy to ensure equality’ (2006) 51(4) *Villanova L Rev*.

Ravitsky, V ‘Non-Invasive Prenatal Testing (NIPT): Identifying key clinical, ethical, social, legal and policy issues’ (2015) *Nuffield Council on Bioethics Background Paper* 1–23.

Resnik, DB ‘Direct-to-consumer genomics, social networking, and confidentiality’ (2009) 9(6-7) *Am J Bioeth* 45–46.

Rheeder, R ‘Biobanks in South Africa: A global perspective on privacy and confidentiality’ (2017) 107(5) *SAMJ* 390–393.

Rhodes, R ‘Confidentiality, genetic information, and the physician-patient relationship’ (2001) 1(3) *Am J Bioeth* 26–28.

Richter, M, WDF Venter & A Gray ‘Home self-testing for HIV: AIDS exceptionalism gone wrong’ (2010) 100(10) *SAMJ* 636–642.

Roberts, JS, MC Gornick, DA Carere et al ‘Direct-to-consumer genetic testing: User motivations, decision making, and perceived utility of results’ (2017) 20(1) *Public Health Genomics* 36–45.

Roberts, ME, DL Riegert-Johnson & BC Thomas ‘Self diagnosis of Lynch Syndrome using direct to consumer genetic testing: A case study’ (2011) 20(4) *J Genet Counsel* 327–329.

Robertson, AS ‘Taking responsibility: Regulations and protections in direct-to-consumer genetic testing’ (2009) 24(1) *Berkeley Tech LJ* 213–243.

Robertson, JA ‘Procreative liberty in the era of genomics’ (2003) 29(4) *Am J Law Med* 439–487.

Robinson, M & G Glusman ‘Genotype fingerprints enable fast and private comparison of genetic testing results for research and direct-to-consumer applications’ (2018) 9(10) *Genes (Basel)* 1–13.

Roche, PA & GJ Annas ‘DNA testing, banking and genetic privacy’ (2006) 355(6) *N Engl J Med* 545–546.

Roos, A ‘Personal data protection in New Zealand: Lessons for South Africa?’ (2008) 11(4) *PER/PELJ* 62–109.

Rosemann, A, L Jiang & X Zhang 'The regulatory and legal situation of human embryo, gamete and germ line gene editing research and clinical applications in the People's Republic of China' (2017) *Nuffield Council on Bioethics Background Paper* 1–38.

Rosenbaum, S 'Insurance discrimination on the basis of health status: An overview of discrimination practices, federal law, and federal reform options' (2009) *O'Neill Institute for National and Global Health Law* 1–26.

Roth, SC 'What is genomic medicine?' (2019) 107(3) *J Med Libr Assoc* 442–448.

Rothstein, MA 'Precision medicine and the risk to privacy' (2018) 15(1) *SciTech Lawyer* 28–32.

Rothstein, MA 'Time to end the use of genetic test results in life insurance underwriting' (2018) 46(3) *J Law Med Ethics* 794–801.

Rothstein, MA 'Why treating genetic information separately is a bad idea' (1999) 4(1) *Tex Rev Law Polit* 33–37.

Rowe, K & K Moodley 'Patients as consumers of health care in South Africa: The ethical and legal implications' (2013) 14(15) *BMC Med Ethics* 1–9.

Rubin, IR & G Glusman 'Opportunities and challenges in interpreting and sharing personal genomes' (2019) 10(9) *Genes* 1–6.

Ruckenstein, M 'Keeping data alive: Talking DTC genetic testing' (2017) 20(7) *Inf Commun Soc* 1024–1039.

Ruhl, GL, JW Hazel, E Wright Clayton et al 'Public attitudes toward direct to consumer genetic testing' (2019) *AMIA Annu Symp Proc* 774–783.

Saelaert, M, H Mertes, T Moerenhout et al 'Ethical values supporting the disclosure of incidental and secondary findings in clinical genomic testing: A qualitative study' (2020) 21(1) *BMC Med Ethics* 1–12.

Saidi, T & T Douglas 'Medical device regulation in South Africa: The Medicines and Related Substances Amendment Act 14 of 2015' (2018) 108(3) *SAMJ* 168–170.

Salloum, RG, TJ George, N Silver et al 'Rural-urban and racial-ethnic differences in awareness of direct-to-consumer genetic testing' (2018) 18(1) *BMC Public Health* 1–6.

Sándor, J 'Genetic testing between private and public interests: Some legal and ethical reflections' (2018) 6(8) *Front Public Health* 1–8.

Sandroff, R ‘Direct-to-consumer genetic tests and the right to know’ (2010) 40(5) *Hastings Center Rep* 24–25.

Sanfilippo, PG, LS Kearns, P Wright et al ‘Current landscape of direct-to-consumer genetic testing and its role in ophthalmology: A review’ (2015) 43(6) *Clin Exp Ophthalmol* 578–590.

Sarata, AK ‘FDA regulation of Laboratory-Developed Tests (LDTs)’ (2019) *Congressional Research Service* 1–3.

Sarata, AK ‘Genetic exceptionalism: Genetic information and public policy’ (2011) *Congressional Research Service* 1–10.

Sarin, R ‘Ethics and clinical utility of direct-to-consumer genetic tests’ (2015) 11(1) *J Can Res Ther* 1–2.

Saukko, P ‘Shifting metaphors in direct-to-consumer genetic testing: From genes as information to genes as big data’ (2017) 36(3) *New Genet Soc* 296–313.

Saukko, P ‘State of play in direct-to-consumer genetic testing for lifestyle-related diseases: Market, marketing content, user experiences and regulation’ (2013) 72(1) *Proc Nutr Soc* 53–60.

Saunders, R ‘Legal implications of direct-to-consumer genetic testing for common diseases’ (2010) 1 *QMLJ* 71–79.

Savard, J, C Hickerton, SA Metcalfe et al ‘From expectations to experiences: Consumer autonomy and choice in personal genomic testing’ (2020) 11(1) *AJOB Empir Bioeth* 63–76.

Savulescu, J ‘Procreative beneficence: Why we should select the best children’ (2001) 15 *Bioethics* 413–426.

Savulescu, J & RW Momeyer ‘Should informed consent be based on rational beliefs?’ (1997) 23 *J Med Ethics* 282–288.

Scanlon ‘A theory of freedom of expression’ (1972) 1(2) *Philos Public Aff* 204–226.

Schaper, M & S Schicktanz ‘Medicine, market and communication: Ethical considerations in regard to persuasive communication in direct-to-consumer genetic testing services’ (2018) 19(56) *BMC Med Ethics* 1–11.

Schickedanz, AD & R Herdman 'Direct-to-consumer genetic testing: The need to get retail genomics right' (2009) 86(1) *Clin Pharmacol Ther* 17–20.

Schlanger, SJ 'Filling in the cracks: Improving the regulation of direct-to-consumer genetic tests' (2011) 14 *J Health Care L & Pol'y* 1–29.

Schleckser, K 'Physician participation in direct-to-consumer genetic testing: Pragmatism or paternalism' (2013) 26(2) *Harv J L & Tech* 695–730.

Schwartz, MD, K Rothenberg, L Joseph et al 'Consent to the use of stored DNA for genetics research: A survey of attitudes in the Jewish population' (2001) 98(4) *Am J Med Genet* 336–342.

Scott Roberts, J & J Ostergren 'Direct-to-consumer genetic testing and personal genomics services: A review of recent empirical studies' (2013) 1(3) *Curr Genet Med Rep* 182–200.

Scudder, N, D McNevin, SF Kelty et al 'Policy and regulatory implications of the new frontier of forensic genomics: Direct-to-consumer genetic data and genealogy records' (2019) 31(2) *Curr Issues Crim Justice* 194–216.

Segal, E, N Friedman, D Koller et al 'A module map showing conditional activity of expression modules in cancer' (2004) 36 *Nat Genet* 1090–1098.

Selemogo, M 'Evaluating the right to autonomy argument in the debate on coercive antenatal HIV testing in South Africa' (2010) 3(2) *SAJBL* 63–66.

Sénécal, K, P Borry, HC Howard et al 'Direct-to-consumer genetic testing: Overview of applicable norms and policies' (2009) 7 *GenEdit* 1–14.

Shabani, M & P Borry 'Rules for processing genetic data for research purposes in view of the new EU General Data Protection Regulation' (2018) 26(2) *Eur J Hum Genet* 149–156.

Shaer, O, O Nov, J Okerlund et al 'Informing the design of direct-to-consumer interactive personal genomics reports' (2015) 17(6) *J Med Internet Res* 1–22.

Sharkey, CM 'Direct-to-consumer genetic testing: The FDA's dual role as safety and health information regulator' (2019) 68(2) *Depaul L Rev* 343–384.

Sheehan, M 'The right to know and genetic testing' (2015) 41(4) *J Med Ethics* 287–288.

Sherman, K, LK Shaw, K Champion et al 'The effect of disease risk probability and disease type on interest in clinic-based versus direct-to-consumer genetic testing services' (2015) 38(5) *J Behav Med* 706–714.

Shi, X & X Wu 'An overview of human genetic privacy' (2017) 1387(1) *Ann NY Acad Sci* 61–72.

Shuff, J & JD Sims 'Communication perceptions related to life-threatening illness in a relationship: A Q methodology study' (2013) 41(2) *Fla Commun J* 81–96.

Sieber, JE 'Privacy and confidentiality: As related to human research in social and behavioral science' (2001) *Online Ethics Center at the National Academy of Engineering* 1–50.

Singh, SC 'Protection of human genetic information: Balancing interests in the use of personal genetic data' (2013) 55(2) *JILI* 175–200.

Singleton, A, LH Erby, KV Foisie et al 'Informed choice in direct-to-consumer genetic testing (DTCGT) websites: A content analysis of benefits, risks, and limitations' (2012) 21(3) *J Genet Counsel* 1–12.

Skirton, H 'Direct to consumer testing in reproductive contexts – Should health professionals be concerned?' (2015) 11(4) *Life Sci Soc Policy* 1–9.

Skirton, H, L Goldsmith, L Jackson et al 'Direct to consumer genetic testing: A systematic review of position statements, policies and recommendations' (2012) 82(3) *Clin Genet* 210–218.

Slabbert, M 'Donated human tissue: Is it a medical device or just tissue?' (2016) 37(3) *OBITER* 523–534.

Šlesingerová, E 'In risk we trust/editing embryos and mirroring future risks and uncertainties' (2019) 22(2) *Med Health Care Philos* 191–200.

Slokenberga, S 'Direct-to-consumer genetic testing: Changes in the EU regulatory landscape' (2015) 22(5) *Eur J Health Law* 463–480.

Smalley, E 'FDA warns public of dangers of DIY gene therapy' (2018) 36(2) *Nat Biotechnol* 119–120.

Smart, A, DA Bolnick & R Tutton 'Health and genetic ancestry testing: Time to bridge the gap' (2017) 10(3) *BMC Med Genomics* 1–9.

Smebye, KL, M Kirkevold & K Engedal 'Ethical dilemmas concerning autonomy when persons with dementia wish to live at home: A qualitative, hermeneutic study' (2015) 16(21) *BMC Health Serv Res* 1–12.

Soini, S 'Genetic testing legislation in Western Europe – A fluctuating regulatory target' (2012) 3(2) *J Community Genet* 143–153.

Somasundaram, J & L Wathieu 'Demand for genetic and medical testing: The role of anticipated emotions and probability distortion' (2016) *Working Paper Series 2016/50/DSC* 1–60.

Somogyi, AA & E Phillips 'Genomic testing as a tool to optimise drug therapy' (2017) 40(3) *Australian Prescriber* 101–104.

Spector-Bagdady, K 'Reconceptualizing consent for direct-to-consumer health services' (2015) 41(4) *Am J Law Med* 568–616.

Spector-Bagdady, K & E Pike 'Consuming genomics: Regulating direct-to-consumer genetic and genomic information' (2014) 92(4) *Nebraska L Rev* 677–745.

Spencer, DH, C Lockwood, E Topol et al 'Direct-to-consumer genetic testing: Reliable or risky?' (2011) 57(12) *Clin Chem* 1641–1644.

Stark, Z, L Dolman, TA Manolio et al 'Integrating genomics into healthcare: A global responsibility' (2019) 104(1) *Am J Hum Genet* 13–20.

Staunton, C & K Moodley 'Data mining and biological sample exportation from South Africa: A new wave of bioexploitation under the guise of clinical care?' (2016) 106(2) *SAMJ* 136–138.

Staunton, C, R Adams, ES Dove et al 'Ethical and practical issues to consider in the governance of genomic and human research data and data sharing in South Africa: A meeting report' (2019) 2(15) *AAS Open Res* 1–11.

Staunton, C, R Adams, M Botes et al 'Safeguarding the future of genomic research in South Africa: Broad consent and the Protection of Personal Information Act No. 4 of 2013' (2019) 109(7) *SAMJ* 468–470.

Sterckx, S, J Cockbain, HC Howard et al '"Trust is not something you can reclaim easily": Patenting in the field of direct-to-consumer genetic testing' (2013) 15(5) *Genet Med* 382–387.

Stewart, KF, A Wesselius, MAC Schreurs et al 'Behavioural changes, sharing behaviour and psychological responses after receiving direct-to-consumer genetic test results: A systematic review and meta-analysis' (2017) 9(1) *J Community Genet* 1–18.

Stewart, KFJ, D Kokole, A Wesselius et al 'Factors associated with acceptability, consideration and intention of uptake of direct-to-consumer genetic testing: A survey study' (2018) 21(1-2) *Public Health Genomics* 45–52.

Stirrat, GM 'Autonomy in medical ethics after O'Neill' (2005) 31 *J Med Ethics* 127–130.

Stoeklé HC, MF Mamzer-Bruneel, G Vogt et al '23andMe: A new two-sided data-banking market model' (2016) 17(19) *BMC Med Ethics* 1–11.

Su, P 'Direct-to-consumer genetic testing: A comprehensive view' (2013) 86(3) *Yale J Biol Med* 359–365.

Sunderman, SF 'The need for regulation of direct-to-consumer genetic testing in the United States: Assessing and applying the German policy model' (2013) 12(2) *Wash U Global Stud L Rev* 357–379.

Swales, L 'The Protection of Personal Information Act and data de-identification' (2021) 117(7/8) *S Afr J Sci* 1–3.

Swan, M 'Multigenic condition risk assessment in direct-to-consumer genomic services' (2010) 12 *Genet Med* 279–288.

Sweeny, K & AM Legg 'Predictors of interest in direct-to-consumer genetic testing' (2011) 26(10) *Psychol Health* 1259–1272.

Uhlmann, WR & RR Sharp 'Genetic Testing Integration Panels (GTIPs): A novel approach for considering integration of direct-to-consumer and other new genetic tests into patient care' (2012) 21 *J Genet Counsel* 374–381.

Talu, CK, MH Toper, Y Sahin et al 'Pathology and biobanking' (2020) 36(2) *Turkish J Pathol* 1–16.

Tamir, S 'Direct-to-consumer genetic testing: Ethical-legal perspectives and practical considerations' (2010) 18(2) *Med Law Rev* 213–238.

Tandy-Connor, S, J Guiltinan, K Krempely et al 'False-positive results released by direct-to-consumer genetic tests highlight the importance of clinical confirmation testing for appropriate patient care' (2018) 20(12) *Genet Med* 1515–1521.

Tarr, JA ‘Regulatory approaches to genetic testing in insurance’ (2002) 24(2) *Syd Law Rev* 189–206.

Tavani, HT ‘Philosophical theories of privacy: Implications for an adequate online privacy policy’ (2007) 38(1) *Metaphilosophy* 1–22.

Teare, HJA, M Prictor & J Kaye ‘Reflections on dynamic consent in biomedical research: the story so far’ (2021) 29 *Eur J Hum Genet* 649–656.

Thaldar, D ‘One material transfer agreement to rule them all? A call for revising South Africa’s new standard material transfer agreement’ (2020) 7(105) *Humanit Soc Sci Commun* 1–5.

Thaldar, D ‘Why POPIA does not apply to DNA’ (2021) 117(7/8) *S Afr J Sci* 1–2.

Thaldar, D & B Townsend ‘Protecting personal information in research: Is a code of conduct the solution?’ (2021) 117(3/4) *S Afr J Sci* 1–2.

Thaldar, D, J Kinderlerer & S Soni ‘An optimistic vision for biosciences in South Africa: A response to the ASSAf report on human genetics and genomics’ (2019) 115(7/8) *S Afr J Sci* 1.

Thaldar, D, M Botes, B Shozi et al ‘Human germline editing: Legal-ethical guidelines for South Africa’ (2020) 116(9/10) *S Afr J Sci* 1–7.

Thaldar, DW, M Botes & A Nienaber ‘South Africa’s new standard material transfer agreement: Proposals for improvement and pointers for implementation’ (2020) 21(85) *BMC Med Ethics* 1–13.

Thaldar, DW & B Shozi ‘The legal status of human biological material used for research’ *Forthcoming in SALJ*.

Thaldar, DW & B Townsend ‘Genomic research and privacy: A response to Staunton *et al*’ (2020) 110(3) *SAMJ* 172–174.

Thaldar, DW & BA Townsend ‘Exempting health research from the consent provisions of POPIA’ (2021) 24 *PER/PELJ* 1–30.

Thaldar, DW & M Steytler ‘Time for Cinderella to go to the ball: Reflections on the right to freedom of scientific research’ (2020) 138(2) *SALJ* 260–288.

The American College of Obstetricians and Gynecologists ‘Access to genetic testing’ (2010) 22 *ECRI Institute* 1.

The Lancet 'Direct-to-consumer genetic testing' (2012) 380(9837) *Lancet* 76.

The Lancet 'New guidelines for genetic tests are welcome but insufficient' (2010) 376(9740) *The Lancet* 488.

The Specialist Forum 'Limitations of DIY genetic testing' (2019) 19(6) *The Specialist Forum* 43–44.

Thiebes, S, PA Toussaint, J Ju et al 'Valuable genomes: Taxonomy and archetypes of business models in direct-to-consumer genetic testing' (2020) 22(1) *J Med Internet Res* 1–16.

Thomas, R 'Where to from Castell v De Greef? Lessons from recent developments in South Africa and abroad regarding consent to treatment and the standard of disclosure' (2007) 124(1) *SALJ* 188–215.

Thrush, SA & R McCaffrey 'Direct-to-consumer genetic testing: What the nurse practitioner should know' (2010) 6(4) *J Nurse Pract* 269–273.

Tiller, J & P Lacaze 'Regulation of internet-based genetic testing: Challenges for Australia and other jurisdictions' (2018) 6(24) *Front Public Health* 1–6.

Tkacik, M 'Characteristics of forms of autonomy' (2008) 15 *Int J Minor Group Rights* 369–401.

Tolouian, R & H Salameh 'Treat the patient not the lab value' (2010) 3 *NDT Plus* 81–83.

Tomlinson, T 'A CRISPR future for gene-editing regulation: A proposal for an updated biotechnology regulatory system in an era of human genomic editing' (2018) 87(1) *Fordham L Rev* 437–483.

Townend, D 'Conclusion: Harmonisation in genomic and health data sharing for research: An impossible dream?' (2018) 137(8) *Hum Genet* 657–664.

Townsend, B 'The lawful sharing of health research data in South Africa and beyond' (2021) *Inf Commun Technol Law* 1–18.

Townsend, BA 'Software as a medical device: Critical rights issues regarding artificial intelligence software-based health technologies in South Africa' (2020) 4 *TSAR* 747–762.

Townsend, BA & D Thaldar 'Informed consent in medical malpractice suits: An analysis of Beukes v Smith' (2020) 137(1) *SALJ* 13–25.

Townsend, BA & DW Thaldar 'Navigating uncharted waters: Biobanks and informational privacy in South Africa' (2019) 35(4) *SAJHR* 329–350.

Trent, R 'Direct-to-consumer DNA genetic testing and the GP' (2014) 43(7) *Aust Fam Physician* 436–439.

Turoldo, F 'Relational autonomy and multiculturalism' (2010) 19(4) *Camb Q Healthc Ethics* 542–549.

Turrini, M & B Prainsack 'Beyond clinical utility: The multiple values of DTC genetics' (2016) 8 *Appl Transl Genom* 4–8.

Turrini, M 'Online genomes: Problematizing the disruptiveness of direct-to-consumer genetic tests' (2018) 12(11) *Sociology Compass* 1–13.

Turrini, M 'Practicing the biomedicine to come: Direct-to-consumer genetic testing, healthism and beyond' (2015) 7(2) *eä Journal* 49–69.

Udesky, L 'The ethics of direct-to-consumer genetic testing' (2010) 376(9750) *The Lancet* 1377–1378.

van der Reyden, D 'The right to respect for autonomy: Part 1 – What is autonomy all about?' (2008) 38(1) *S Afr J Occup Ther* 27–31.

van Hellemondt, R, AC Hendriks & MH Breuning 'Regulating the use of genetic tests: Is Dutch Law an example for other countries with regard to DTC genetic testing?' (2011) 3(1) *Amsterdam Law Forum* 13–24.

van Hellemondt, RE, A Hendricks & M Breuning 'Which lessons can we learn from the European Union legal framework of medicines for the regulation of direct-to-consumer genetic tests?' (2012) 36 *Law and the Human Genome Rev* 119–135.

van Niekerk, B 'Torture – Our last hurdle towards civilization: Reflections on a recent conference' (1974) 91 *SALJ* 515–525.

Varelius, J 'The value of autonomy in medical ethics' (2006) 9(3) *Med Health Care Philos* 377–388.

Varga, EA 'You want to do what? My mother's choice to have direct-to-consumer genetic testing' (2012) 21(3) *J Genet Counsel* 382–385.

Vayena, E 'Direct-to-consumer genomics on the scales of autonomy' (2015) 41(4) *J Med Ethics* 310–314.

Verkerk, MA ‘The care perspective and autonomy’ (2001) 4 *Med, Health Care Philos* 289–294.

Vernon, R & M Concannon ‘Confidentiality and patient autonomy in a healthcare framework’ (2015) *Podiatry Now* 12–16.

Vlahovich, N, PA Fricker, MA Brown et al ‘Ethics of genetic testing and research in sport: A position statement from the Australian Institute of Sport’ (2016) 51(1) *Br J Sports Med* 5–11.

Vlahovich, N, DC Hughes, LR Griffiths et al ‘Genetic testing for exercise prescription and injury prevention: AIS-Athlome Consortium-FIMS joint statement’ (2017) 18 *BMC Genomics* 1–13.

Vrecar, I, B Peterlin, N Teran et al ‘Direct-to-consumer genetic testing in Slovenia: Availability, ethical dilemmas and legislation’ (2015) 25(1) *Biochemia Medica* 84–89.

Vybiral, D ‘Legal regulation for genetic testing in Austria’ (European Human Genetics Conference, Gothenburg, 14 June 2010) 1–20.

Wade, CH & BS Wilfond ‘Ethical and clinical practice considerations for genetic counselors related to direct-to-consumer marketing of genetic tests’ (2006) 142(4) *Am J Med Genet* 284–292.

Wagner, JK, JD Cooper, R Sterling et al ‘Tilting at windmills no longer: A data-driven discussion of DTC DNA ancestry tests’ (2012) 14(6) *Genet Med* 586–593.

Walker, RL ‘Medical ethics needs a new view of autonomy’ (2008) 33(6) *J Med Philos* 594–608.

Walker, RL ‘Respect for rational autonomy’ (2009) 19(4) *Kennedy Inst Ethics J* 339–366.

Wallace, SE, EG Gourna, V Nikolova et al ‘Family tree and ancestry inference: Is there a need for a “generational” consent?’ (2015) 16(1) *BMC Med Ethics* 1–9.

Walter, JK & L Friedman Ross ‘Relational autonomy: Moving beyond the limits of isolated individualism’ (2014) 133(1) *Pediatrics* 16–23.

Watt, E & E Kowal ‘What’s at stake? Determining indigeneity in the era of DIY DNA’ (2019) 38(2) *New Genet Soc* 142–164.

Weaver, M & TI Pollin ‘Direct-to-consumer genetic testing: What are we talking about?’ (2012) 21(3) *J Genet Counsel* 361–366.

Webborn, N, A Williams, M McNamee et al ‘Direct-to-consumer genetic testing for predicting sports performance and talent identification: Consensus statement’ (2015) 49(23) *Br J Sports Med* 1486–1491.

Webster, PC ‘Regulation of genetic tests unnecessary, government says’ (2010) 182(16) *CMAJ* 1715–1716.

Weiermiller, C ‘The future of direct-to-consumer genetic testing: Regulation and innovation’ (2015) 16(5) *NC JL & Tech* 137–171.

Weiner, K, P Martin, M Richards et al ‘Have we seen the geneticisation of society? Expectations and evidence’ (2017) 39(7) *Sociol Health Illn* 989–1004.

Weiss, MA & K Archick ‘U.S.-EU data privacy: From Safe Harbor to Privacy Shield’ (2016) *Congressional Research Service* 1–16.

Wertenbroch, K, RY Schrift, JW Alba et al ‘Autonomy in consumer choice’ (2020) 31(1-2) *Mark Lett* 429–439.

Wexler, A & PB Reiner ‘Oversight of direct-to-consumer neurotechnologies’ (2019) 363(6424) *Science* 234–235.

Wilde, A, B Meiser, PB Mitchell et al ‘Public interest in predictive genetic testing, including direct-to-consumer testing, for susceptibility to major depression: Preliminary findings’ (2010) 18(1) *Eur J Hum Genet* 47–51.

Wilks, AF ‘Kantian challenges for the bioenhancement of moral autonomy’ (2018) 83 *Roy Inst Philos Suppl* 121–143.

Williams, C, J Kitzinger & L Henderson ‘Envisaging the embryo in stem cell research: Rhetorical strategies and media reporting of the ethical debates’ (2003) 25(7) *Sociol Health Illn* 793–814.

Wils, JP ‘Autonomy and just health’ (2016) 50(1).

Wilson, TE & C Halverson ‘Home testing for COVID-19: Lessons from direct to consumer genetics’ (2020) 11(4) *J Community Genet* 497–499.

Witt, MM & MP Witt ‘Privacy and confidentiality measures in genetic testing and counselling: Arguing on genetic exceptionalism again?’ (2016) 57(4) *J Appl Genetics* 483–485.

World Health Organization & March of Dimes 'Management of birth defects and haemoglobin disorders: Report of a joint WHO-March of Dimes meeting' *World Health Organization* (Geneva, Switzerland, 17-19 May 2006) 1–27.

Wright, CF, A Hall & RL Zimmern 'Regulating direct-to-consumer genetic tests: What is all the fuss about?' (2011) 13(4) *Genet Med* 295–300.

Wright Clayton, E, BJ Evans, JW Hazel et al 'The law of genetic privacy: Applications, implications, and limitations' (2019) 6(1) *J Law Biosci* 1–36.

Yaneva-Deliverska, M 'Legal aspects of direct-to-consumer genetic tests' (2011) 17(1) *J of IMAB* 135–138.

Yim, SH & YJ Chung 'Reflections on the US FDA's warning on direct-to-consumer genetic testing' (2014) 12(4) *Genomics Inform* 151–155.

Zabel, J 'The killer inside us: Law, ethics, and the forensic use of consumer genetics' (2019) 24(2) *Berkeley J Crim L* 47–100.

Zawati, MH, P Borry & HC Howard 'Closure of population biobanks and direct-to-consumer genetic testing companies' (2011) 130(3) *Hum Genet* 425–432.

Zehnbauer, B 'Direct-to-consumer genetics testing – Fair comparisons?' (2011) 57(3) *Clin Chem* 369–371.

Zettler, PJ, JS Sherkow & HT Greely '23andMe, the Food and Drug Administration, and the future of genetic testing' (2014) 174(4) *JAMA Inter Med* 493–494.

Zuma, T, D Wight, T Rochat et al 'The role of traditional health practitioners in Rural KwaZulu-Natal, South Africa: Generic or mode specific?' (2016) 16(304) *BMC Complement Altern Med* 1–13.

(d) Theses and dissertations

Appanna, K 'The Doctrine of Therapeutic Privilege and its Place in South Africa' (Degree of Master of Laws, University of KwaZulu-Natal 2014).

Artz, M 'An Ethnography of Direct-to-consumer Genomics (DTCG): Design Anthropology Insights for the Product Management of a Disruptive Innovation' (Thesis for Degree of Master of Science, University of North Texas 2018).

Chen, FH ‘23andMe and the FDA: Negotiating Conceptions of Benefit in the Direct-to-consumer Genetic Testing Regulatory Debate’ (Master of Public Health, University of Washington 2014).

Curnutte, M ‘Consuming Genomes: The Coproduction of a New Scientific and Technological Order for Genetic Testing’ (PhD degree in Foundations of the Life Sciences and their Ethical Consequences, European School of Molecular Medicine (SEMM) and University of Milan 2012).

De Carvalho, CL ‘A Reflection on the Morality of Ownership of Genetic Material’ (Master of Science in Medicine, University of the Witwatersrand 2011).

Donnelly, DL ‘Privacy by (re)Design: A Comparative Study of the Protection of Personal Information in the Mobile Applications Ecosystem under United States, European Union and South African Law’ (Doctor of Laws thesis, University of KwaZulu-Natal, 2020).

Düsterwald, G ‘Understanding Doctors’ Knowledge and Attitudes Concerning Genetics and Genetic Services in South Africa’ (MSc Genetic Counselling, University of Cape Town 2015).

Egglestone, C ‘The Informational Aspects of Direct-to-Consumer Genetic Tests’ (Doctor of Philosophy, Loughborough University 2013).

Finlay, TM ‘UK Users’ and Genetics Clinicians’ Experiences of Direct-to-consumer Genetic Testing’ (Doctor of Philosophy, Cardiff University 2015).

Govender, S ‘The Protection of Genetic Privacy in South Africa: Towards a Legislative Response Based on a Cross-Jurisdictional Review of Legal Developments’ (Doctor of Philosophy, University of the Witwatersrand, 2012).

Johnson, R ‘Factors Affecting Intent to Use Consumer Genetic Tests: A Revised Technology Acceptance Model’ (Master of Business Administration, University of Pretoria 2010).

Jordaan, DW ‘Medical Biotechnology Law in South Africa: A Human Rights Analysis of Selected Topics’ (Doctor of Philosophy, University of Cape Town 2012).

Kamanga, VV ‘Product Labelling and Trade Description: Failure to Warn and the Consumer Protection Act 68 of 2008’ (LLM in Mercantile Law, University of Pretoria 2017).

Kinsley, N ‘The Use of Genetic Tests by the Individual Life Insurance Industry in South Africa’ (Master of Science in Medicine in Genetic Counselling, University of the Witwatersrand 2009).

Laurent, AC ‘A Critical Analysis of the Legality of Future Research and Experimentation of Germ-Line Gene Editing (GGE) in South African Law and Ethics’ (Legum Baccalaureus (LLB), University of Pretoria 2019).

Mahesh, KP ‘Genomic Sovereignty in South Africa: Ethico-Legal Issues’ (LLM Thesis, University of the Witwatersrand 2014).

Mitchell, CE ‘BRCA1 and BRCA2 Mutation Carrier Perspectives on Direct-To-Consumer Genetic Testing for Brca Mutations’ (Degree of Master of Science in Genetic Counseling, University of South Carolina 2019).

Moloi, M ‘The Influence of Online Consumer Reviews on Purchasing Intent’ (Master of Management in Strategic Marketing, University of the Witwatersrand 2016).

Moodley, S ‘The Involuntary Disclosure of Confidential Genetic Information to Related Affected Third Parties’ (Degree of Master of Laws in Medical Law, University of KwaZulu-Natal 2016).

Naidoo, L ‘Apart from Medical Emergencies, when is it Justified for Gynaecologists, and Obstetricians to Deviate from Informed Consent without Reverting Back to their Patients?’ (Degree of Master of Laws in Medical Law, University of KwaZulu-Natal 2018).

Naidoo, M ‘The CRISPR Patent Landscape: A South African Perspective’ (Degree of Master of Laws in Business Law, University of KwaZulu-Natal 2020).

Niemiec, E ‘Ethical, Legal and Social Issues Related to the Offer of Whole Exome and Whole Genome Sequencing’ (Doctoral Degree in Law, Science and Technology, 2018).

Pack, JK ‘The Impact of the Myriad Direct-To-Consumer Advertising Campaign for BRCA1/2 Genetic Testing in the Greater Cincinnati Area’ (Degree of Master of Science in Genetic Counseling, University of Cincinnati 2011).

Parrott, A ‘Media Coverage of Direct-To-Consumer Genetic Testing’ (Master of Science in Genetic Counselling, University of Cincinnati 2010).

Phillips, AM ‘Protecting the Rights of Consumers – Clickwrap Contracts and Direct-To-Consumer Genetic Testing’ (DPhil, St Hilda’s College 2015).

Ramakrishnan, S ‘Direct-to-consumer Advertisement of Predictive Genetic Tests: Influence on Consumer Attitudes, Intentions and Behavior’ (Doctor of Philosophy, University of Mumbai 2010).

Singh, A ‘The Impact of the Constitution on Transforming the Process of Statutory Interpretation in South Africa’ (Doctor of Philosophy, University of KwaZulu-Natal, 2014).

Tariyal, R ‘Finding Utility for Genetic Diagnostics in the Developing World’ (Masters of Science in Health Sciences and Technology, Massachusetts Institute of Technology 2010).

Townsend, BA ‘E-Health, Social Media and the Law in South Africa: Can Ethical Concerns in E-Health Practice be Addressed Through Regulation?’(Degree of Master of Laws, University of Cape Town 2013).

van der Walt, M ‘Underwriting Guidelines for Genetic Testing with Special Reference to the Relevant Ethical Aspects’ (MCom (Business Management), Rand Afrikaans University 2000).

Wagner, JK ‘Social and Legal Implications of DNA Ancestry Tests’ (Degree of Doctor of Philosophy, Pennsylvania State University 2010).

Zhang, P ‘Exploring the Potential of Direct-To-Consumer Genomic Test Data for Predicting Adverse Drug Events’ (Degree of Master of Science in the School of Engineering, Brown University 2018).

(e) Websites and online sources

23andMe ‘23andMe Genetic Health Risk Reports: What you should know’ (2020) available at *https://www.23andme.com/test-info/genetic-health/,* accessed on 23 June 2020.

23andMe ‘23andMe for healthcare professionals’ (2020) available at *https://medical.23andme.com/*, accessed on 5 October 2019.

23andMe ‘23andMe statement regarding FDA warning letter’ (25 November 2013) available at *https://mediacenter.23andme.com/press-releases/fda-letter-2013/,* accessed on 13 January 2020.

23andMe ‘23andMe takes first step toward FDA clearance’ (30 July 2012) available at *https://blog.23andme.com/news/23andme-takes-first-step-toward-fda-clearance/*, accessed on 7 December 2019.

23andMe ‘Becoming part of something bigger’ (2020) available at *https://www.23andme.com/research/?mkbanner=true*, accessed on 22 June 2020.

23andMe ‘Biobanking consent document’ (2020) available at *https://www.23andme.com/about/biobanking/*, accessed on 9 January 2021.

23andMe ‘Difference between DNA genotyping & sequencing’ available at *https://customercare.23andme.com/hc/en-us/articles/202904600-Difference-Between-DNA-Genotyping-Sequencing*, accessed on 17 January 2021.

23andMe ‘Health + ancestry service’ (2020) available at *https://www.23andme.com/dna-health-ancestry?mkpc=true*, accessed on 5 October 2019.

23andMe ‘How it works’ available at *https://www.23andme.com/howitworks/*, accessed on 20 November 2020.

23andMe ‘Individual data sharing consent’ (2020) available at *https://www.23andme.com/about/individual-data-consent/*, accessed on 9 January 2021.

23andMe ‘Privacy’ available at *https://www.23andme.com/privacy/*, accessed on 4 May 2020.

23andMe ‘Privacy highlights’ (1 January 2020) available at *https://www.23andme.com/about/privacy/*, accessed on 22 June 2020.

23andMe ‘Privacy is in our DNA’ (2020) available at *https://www.23andme.com/privacy/?mkbanner=true#privacy-drawer-three*, accessed on 22 June 2020.

23andMe ‘Providing saliva sample for DNA test kit’ available at *https://customercare.23andme.com/hc/en-us/articles/202904530-Providing-Saliva-Sample-for-DNA-Test-Kit*, accessed on 9 January 2021.

23andMe ‘Publications’ available at *https://www.23andme.com/publications/*, accessed on 9 January 2021.

23andMe ‘Research’ available at *https://www.23andme.com/en-int/research/*, accessed on 4 May 2020.

23andMe ‘Research consent document’ (2020) available at *https://www.23andme.com/about/consent/*, accessed on 9 January 2021.

23andMe ‘Returning DNA sample to the lab’ available at *https://customercare.23andme.com/hc/en-us/articles/202904570-Returning-DNA-Sample-to-the-Lab*, accessed on 25 June 2020.

23andMe ‘Terms of service’ (30 September 2019) available at *https://www.23andme.com/about/tos/*, accessed on 9 January 2021.

23andMe ‘What countries do you ship to?’ available at *https://int.customercare.23andme.com/hc/en-us/articles/214806628-What-countries-do-you-ship-to-*, accessed on 2 January 2020.

AAFP ‘GAO report critical of genetic tests marketed to consumers’ (11 August 2010) available at *https://www.aafp.org/news/health-of-the-public/20100811gaoreport-genetictests.html*, accessed on 5 November 2019.

Academy of Science of South Africa ‘POPIA: A code of conduct for research’ available at *https://www.assaf.org.za/index.php/2-uncategorised/798-popia-a-code-of-conduct-for-research*, accessed on 6 May 2021.

ACMG ‘About us’ available at *https://www.acmg.net/ACMG/About/About_ACMG/ACMG/About_ACMG/About_Us.aspx?hkey=51851f13-f452-4a91-8663-5dc1a19be91b*, accessed on 20 November 2019.

Allen, A ‘Privacy and medicine’ (28 January 2021) *Stanford Encyclopedia of Philosophy* available at *https://plato.stanford.edu/entries/privacy-medicine/*, accessed on 24 March 2020.

Allen, K ‘Africa should not be too quick to embrace the fourth industrial revolution’ (16 September 2019) *The Guardian* available at *https://www.theguardian.com/global-development/2019/sep/16/africa-should-not-be-too-quick-to-embrace-the-fourth-industrial-revolution*, accessed on 27 March 2020.

Alt Advisory ‘POPIA compliance’ available at *https://altadvisory.africa/popia/#applicationofpopia*, accessed on 9 October 2020.

Alzheimer Europe ‘Respect for autonomy’ (9 October 2009) available at *https://www.alzheimer-europe.org/Ethics/Definitions-and-approaches/The-four-common-bioethical-principles/Respect-for-autonomy*, accessed on 6 March 2020.

American College of Clinical Pharmacology ‘Genetic tests advertised directly to the consumer’ (21 July 2009) *EurekAlert!* available at *https://www.eurekalert.org/pub_releases/2009-07/sp-gta072109.php*, accessed on 11 October 2019.

American College of Medical Genetics and Genomics ‘ACMG responds to FDA’s approval for direct-to-consumer testing for three BRCA gene mutations’ (7 March 2018) *Cision* available at *https://www.prnewswire.com/news-releases/acmg-responds-to-fdas-approval-for-direct-to-consumer-testing-for-three-brca-gene-mutations-300610375.html*, accessed on 25 November 2019.

AncestryDNA ‘Your privacy’ (23 September 2020) available at *https://www.ancestry.com/cs/legal/privacystatement*, accessed on 9 January 2021.

AncestryDNA ‘Ancestry terms and conditions’ (23 September 2020) available at *https://www.ancestry.com/cs/legal/termsandconditions*, accessed on 9 January 2021.

AncestryDNA ‘AncestryDNA informed consent’ (24 July 2018) available at *https://www.ancestry.com/dna/lp/informedconsent-v4-en*, accessed on 9 January 2021.

AncestryDNA ‘Need to ship to another country?’ available at *https://www.ancestry.com/checkout/MLI?rtype=85&flow=3*, accessed on 2 January 2020.

AncestryHealth ‘DNA health testing & genetic health test’ available at *https://www.ancestry.com/health*, accessed on 6 October 2019.

AncestryHealth ‘Now your DNA reveals so much more with AncestryHealth’ (2020) available at *https://www.ancestry.com/health*, accessed on 5 October 2019.

Angelica, N ‘FDA 23andMe Personal Genome Service (PGS) Test – Evaluation of automatic class III designation – De novo request (memorandum)’ (24 October 2017) *Duke SciPol* available at *https://scipol.duke.edu/track/fda-23andme-personal-genome-service-pgs-test-%E2%80%93-evaluation-automatic-class-iii-designation-%E2%80%93-de*, accessed on 27 November 2020.

ASHG ‘Researchers quantify limitations of health reports from direct-to-consumer genetic tests’ (17 October 2019) available at *https://www.ashg.org/publications-news/press-releases/201910-limitations-direct-consumer/*, accessed on 27 November 2019.

Australian Government Department of Health 'IVD medical devices: Definitions & links' (3 December 2010) available at *https://www.tga.gov.au/ivd-medical-devices-definitions-links*, accessed on 11 January 2021.

Australian Law Reform Commission 'Genetic testing services provided directly to the public' (28 July 2010) available at *https://www.alrc.gov.au/publication/essentially-yours-the-protection-of-human-genetic-information-in-australia-alrc-report-96/11-regulating-access-to-genetic-testing/genetic-testing-services-provided-directly-to-the-public/*, accessed on 16 October 2019.

Barnett, S 'Caveat emptor – Beware of direct-to-consumer genetic testing' (29 August 2019) *ARK Invest* available at *https://ark-invest.com/analyst-research/caveat-emptor-beware-of-direct-to-consumer-genetic-testing/*, accessed on 1 February 2020.

Bates, M 'Direct-to-consumer genetic testing' (14 December 2018) *IEEE Pulse* available at *https://pulse.embs.org/november-2018/direct-to-consumer-genetic-testing/*, accessed on 11 September 2019.

Bazian 'New rules for home DNA tests' (4 August 2010) *NHS* available at *https://www.nhs.uk/news/genetics-and-stem-cells/new-rules-for-home-dna-tests/*, accessed on 18 October 2019.

Bhagattjee, P 'South Africa – Data protection overview' (July 2020) *Data Guidance* available at *https://www.dataguidance.com/notes/south-africa-data-protection-overview*, accessed on 9 October 2020.

Biology Dictionary 'DNA sequencing' (4 October 2019) available at *https://biologydictionary.net/dna-sequencing/*, accessed on 4 February 2021.

Biomedicine Agency 'I am tempted by a genetic test offered on the internet: Is it reliable?' (*Agence de la biomédecine*) available at *https://genetique-medicale.fr/la-genetique-medicale-et-vous/vous-etes-dans-cette-situation/article/je-suis-tente-par-un-test-genetique-propose-sur-internet-est-ce-que-c-est-fiable*, accessed on 18 October 2019.

Boodman, E 'Campaign in France to loosen ban on direct-to-consumer DNA testing gains traction' (21 November 2019) *Genetic Literacy Project* available at *https://geneticliteracyproject.org/2019/11/21/campaign-in-france-to-loosen-ban-on-direct-to-consumer-dna-testing-gains-traction/*, accessed on 3 January 2020.

Boodman, E ‘In France, activists fighting over lifting consumer genetic-testing ban’ (14 November 2019) *STAT* available at *https://www.statnews.com/2019/11/14/france-consumer-genetic-testing-ban/*, accessed on 4 January 2020.

Boodman, E ‘In France, it’s illegal for consumers to order a DNA spit kit. Activists are fighting over lifting the ban’ (14 November 2019) *STAT* available at *https://www.statnews.com/2019/11/14/france-consumer-genetic-testing-ban/*, accessed on 3 January 2020.

Botes, M ‘Direct-to-consumer genetic tests: A treacherous road’ (2015) *Caveat Legal* available at *http://www.caveatlegal.com/direct-to-consumer-genetic-tests-a-treacherous-road/*, accessed on 2 June 2020.

Bottomley, EJ ‘Less than 100 days before SA’s strict new privacy rules. Here’s how it will affect you’ (25 March 2021) *Business Insider South Africa* available at *https://www.businessinsider.co.za/popia-what-your-business-needs-to-do-2021-3*, accessed on 7 April 2021.

Boughtwood, T ‘Understanding direct-to-consumer genetic testing’ (12 April 2019) *Australian Genomics Health Alliance* available at *https://www.australiangenomics.org.au/understanding-direct-to-consumer-genetic-testing/*, accessed on 16 October 2019.

Bowser, AD ‘Direct-to-consumer genetic testing fraught with validity concerns’ (16 April 2019) *MD Edge* available at *https://www.mdedge.com/internalmedicine/article/199003/business-medicine/direct-consumer-genetic-testing-fraught-validity*, accessed on 6 January 2020.

Britannica ‘Eukaryote’ available at *https://www.britannica.com/science/eukaryote*, accessed on 18 February 2021.

Britannica ‘Software’ available at *https://www.britannica.com/technology/software*, accessed on 6 January 2021.

BSI ‘Medical devices technologies in vitro diagnostics’ available at *https://www.bsigroup.com/en-GB/medical-devices/technologies/ivd/*, accessed on 18 October 2019.

Burril & Company/Change Wave Research (2008) ‘Personalized medicine and wellness survey’ (2008) available at *http://www.burrillandco.com/content/CWSurvey_61708.pdf*, accessed on 15 March 2020.

Buthelezi, T ‘The Protection of Personal Information Act (POPIA) is here – A quick run down’ (7 August 2020) *Shepstone & Wylie Attorneys* available at *https://www.wylie.co.za/articles/the-protection-of-personal-information-act-popia-is-here-a-quick-run-down/*, accessed on 9 October 2020.

Buxton, J ‘Call to improve accuracy of predictive genetic tests’ (12 October 2009) *BioNews* available at *https://www.bionews.org.uk/page_91911*, accessed on 17 October 2019.

Cambridge Dictionary ‘Apparatus’ available at *https://dictionary.cambridge.org/dictionary/english/apparatus*, accessed on 6 January 2021.

Cambridge Dictionary ‘Appliance’ available at *https://dictionary.cambridge.org/dictionary/english/appliance*, accessed on 6 January 2021.

Cambridge Dictionary ‘Machine’ available at *https://dictionary.cambridge.org/dictionary/english/machine*, accessed on 6 January 2021.

Cambridge Dictionary ‘Pathology’ available at *https://dictionary.cambridge.org/dictionary/english/pathology*, accessed on 20 August 2020.

Cambridge Dictionary ‘Reagent’ available at *https://dictionary.cambridge.org/dictionary/english/reagent*, accessed on 6 January 2021.

Campbell, M ‘Limitations of health reports from direct-to-consumer genetic tests are identified’ (17 October 2019) *Genomics Research from Technology Networks* available at *https://www.technologynetworks.com/genomics/news/limitations-of-health-reports-from-direct-to-consumer-genetic-tests-are-identified-325877*, accessed on 5 January 2020.

Canadian College of Medical Geneticists 'Direct-to-consumer (DTC) genetic testing in this country' available at *https://www.ccmg-ccgm.org/news-article-11.html?id=230amp;catid=122*, accessed on 8 November 2019.

Canadian College of Medical Geneticists 'Letter to the Honourable Rona Ambrose, Health Canada' (2015) available at *https://www.ccmg-ccgm.org/images/pdf/DTC-letter-Final-April-15-2015_V6.pdf*, accessed on 12 January 2020.

Carnie, T 'Scientists call for tighter regulation of SA human genetics research' (7 December 2018) *Daily Maverick* available at *https://www.dailymaverick.co.za/article/2018-12-07-scientists-call-for-tighter-regulation-of-sa-human-genetics-research/*, accessed on 6 December 2019.

Cassidy, S 'Discovery to offer genetic testing' (23 September 2015) *IOL* available at *https://www.iol.co.za/lifestyle/health/discovery-to-offer-genetic-testing-1920337*, accessed on 28 August 2019.

Caulfield, T 'Direct to consumer genetic testing in Canada: should we be concerned?' (1 October 2014) *Healthy Debate* available at *https://healthydebate.ca/opinions/direct-consumer-genetic-testing*, accessed on 19 October 2019.

Caulfield, T 'The direct-to-consumer genetic testing fog' (25 May 2017) *Policy Options* available at *https://policyoptions.irpp.org/magazines/may-2017/the-direct-to-consumer-genetic-testing-fog-column-by-timothy-caulfield/*, accessed on 17 October 2019.

CBS News 'Behind at-home DNA testing companies sharing genetic data with third parties' (2 August 2018) *CBS News* available at *https://www.cbsnews.com/news/dna-privacy-at-home-tests-23andme-ancestrydna-sell-data-to-third-parties/*, accessed on 7 March 2020.

Cell Press 'Accessing your own genomic data is a civil right but requires strategies to manage safety' (4 January 2018) *Science Daily* available at *https://www.sciencedaily.com/releases/2018/01/180104131621.htm*, accessed on 29 December 2019.

Center for Devices and Radiological Health 'Medical devices' (2020) *FDA* available at *http://www.fda.gov/medical-devices*, accessed on 19 March 2020.

Chatterton, C ‘HGC publishes new report on direct-to-consumer genetic testing’ (9 August 2010) *BioNews* available at *https://www.bionews.org.uk/page_92504*, accessed on 3 January 2020.

Cheong, Q, S Chilukuri, D Quigley et al ‘Genetic testing: Opportunities to unlock value in precision medicine’ (31 October 2018) *McKinsey & Company* available at *https://www.mckinsey.com/industries/pharmaceuticals-and-medical-products/our-insights/genetic-testing-opportunities-to-unlock-value-in-precision-medicine#*, accessed on 7 January 2020.

Christman, J ‘Autonomy in moral and political philosophy’ (29 June 2020) *Stanford Encyclopaedia of Philosophy* available at *https://plato.stanford.edu/entries/autonomy-moral/*, accessed on 12 July 2020.

Clark, D ‘Genetic exceptionalism and paternalism themes in new German legislation’ (2 September 2009) *The Privacy Report* available at *https://theprivacyreport.com/2009/09/02/genetic-exceptionalism-and-paternalism-themes-in-new-german-legislation/*, accessed on 19 October 2019.

Clark, D ‘UK House of Lords issues report on genomic medicine’ (13 July 2009) *The Privacy Report* available at *https://theprivacyreport.com/2009/07/13/uk-house-of-lords-issues-report-on-genomic-medicine/*, accessed on 7 January 2020.

Clark, D ‘UK Human Genetics Commission proposes principles for DTC genetic testing services’ (15 October 2009) *The Privacy Report* available at *https://theprivacyreport.com/2009/10/15/u-k-human-genetics-commission-proposes-principles-for-dtc-genetic-testing-services/*, accessed on 19 October 2019.

Collins Dictionary ‘Body fluids’ available at *https://www.collinsdictionary.com/dictionary/english/body-fluids*, accessed on 3 August 2020.

Collins Dictionary ‘Instrument’ available at *https://www.collinsdictionary.com/dictionary/english/instrument*, accessed on 6 January 2021.

Collins Dictionary ‘Therapeutic’ available at *https://www.collinsdictionary.com/dictionary/english/therapeutic*, accessed on 6 May 2021.

Color 'Color informed consent' available at *https://www.color.com/informed-consent*, accessed on 9 January 2021.

Council of Europe 'Additional Protocol to the Convention on Human Rights and Biomedicine, Concerning Genetic Testing for Health Purposes' *Treaty Office* available at *https://www.coe.int/en/web/conventions/full-list*, accessed on 7 December 2019.

Council of Europe 'Details of Treaty No. 203' *Treaty Office* available at *https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/203*, accessed on 5 January 2020.

CRIN 'Bodily integrity' (2018) available at *https://archive.crin.org/en/home/what-we-do/policy/bodily-integrity.html*, accessed on 14 April 2021.

Crouse, C 'Genome pioneer Craig Venter is trying to decode death' (27 March 2018) *CNBC* available at *https://www.cnbc.com/2018/03/27/genome-pioneer-craig-venter-is-trying-to-decode-death.html*, accessed on 11 March 2020.

Cussins, J 'Direct-to-consumer genetic tests should come with a health warning' (15 January 2015) *The Pharmaceutical Journal* available at *https://www.pharmaceutical-journal.com/news-and-analysis/opinion/comment/direct-to-consumer-genetic-tests-should-come-with-a-health-warning/20067564.article*, accessed on 23 February 2021.

D'Olimpio, L 'Ethics explainer: Ethics of Care' (16 May 2019) *The Ethics Centre* available at *https://ethics.org.au/ethics-explainer-ethics-of-care/*, accessed on 21 February 2020.

DeCew, J 'Privacy' (18 January 2018) *Stanford Encyclopedia of Philosophy* available at *https://plato.stanford.edu/entries/privacy/*, accessed on 4 February 2020.

Department of Health 'SA delegation supports call for development of policy framework for traditional medicine' (2013) available at *http://www.doh.gov.za/show.php?id=360*, accessed on 25 February 2020.

Devlin, H 'Senior doctors call for crackdown on home genetic testing kits' (21 July 2019) *The Guardian* available at *https://www.theguardian.com/science/2019/jul/21/senior-doctors-call-for-crackdown-on-home-genetic-testing-kits*, accessed on 17 October 2019.

Dow, AE 'Genetic exceptionalism and the precautionary principle' (6 October 2009) *The Privacy Report* available at *https://theprivacyreport.com/2009/10/06/genetic-exceptionalism-and-the-precautionary-principle/*, accessed on 5 March 2020.

Dickey, MR 'The FDA wants 23andMe to stop marketing its genetic testing kits' (25 November 2013) *Business Insider* available at *https://www.businessinsider.com/fda-sends-warning-letter-to-23andme-2013-11*, accessed on 15 October 2019.

Division of Human Genetics 'MSc (Med) Genetic Counselling' *University of Cape Town* available at *http://www.humangenetics.uct.ac.za/hg/teaching/msc/counselling*, accessed on 16 February 2020.

DNAlysis 'About' available at *https://dnalysis.co.za/about/*, accessed on 22 June 2020.

DNAlysis 'Personalised healthcare starts here' (2020) available at *https://dnalysis.co.za/products/*, accessed on 22 June 2020.

DNAlysis 'Privacy policy' (2020) available at *https://dnalysis.co.za/privacy-policy/*, accessed on 22 June 2020.

DNAlysis 'Revolutionise your practice' (2020) available at *https://dnalysis.co.za/work-with-us/*, accessed on 22 June 2020.

Dryden, J 'Autonomy' *Internet Encyclopaedia of Philosophy* available at *https://iep.utm.edu/autonomy/*, accessed on 5 June 2020.

Dworkin, G 'Paternalism' (9 September 2020) *Stanford Encyclopedia of Philosophy* available at *https://plato.stanford.edu/entries/paternalism/*, accessed on 7 November 2020.

Dyer, L & C Kennedy 'Digital business in South Africa: Overview' (1 March 2021) *Thomson Reuters* available at *https://uk.practicallaw.thomsonreuters.com/w-007-8319?transitionType=Default&contextData=(sc.Default)&firstPage=true*, accessed on 5 April 2021.

EasyDNA 'About us' (2020) available at *https://www.easydna.co.za/about-us/*, accessed on 22 June 2020.

EasyDNA 'Clinical tests' (2020) available at *https://www.easydna.co.za/clinical-tests/*, accessed on 22 June 2020.

EasyDNA 'DNA testing tips' (2020) available at *https://www.easydna.co.za/dna-testing-tips/*, accessed on 22 June 2020.

EasyDNA 'Genetic counselling services' available at *https://www.easydna.co.za/genetic-counselling-services/*, accessed on 16 February 2020.

EasyDNA ‘Genetic glossary’ (2020) available at *https://www.easydna.co.za/genetic-glossary/*, accessed on 22 June 2020.

EasyDNA ‘Genetic predisposition test’ (2020) available at *https://www.easydna.co.za/genetic-predisposition-dna-testing/*, accessed on 22 June 2020.

EasyDNA ‘Home’ available at *https://www.easydna.co.za/*, accessed on 22 June 2020.

EasyDNA ‘Privacy policy’ (20 May 2018) available at *https://www.easydna.co.za/privacy-policy/*, accessed on 9 January 2021.

EasyDNA ‘Terms and conditions’ (24 May 2018) available at *https://www.easydna.co.za/terms-of-service/*, accessed on 22 June 2020.

EasyDNA ‘Useful websites’ (2020) available at *https://www.easydna.co.za/resources/useful-websites/*, accessed on 22 June 2020.

Educause ‘Guidelines for data de-identification or anonymization’ (July 2015) available at *https://www.educause.edu/focus-areas-and-initiatives/policy-and-security/cybersecurity-program/resources/information-security-guide/toolkits/guidelines-for-data-deidentification-or-anonymization*, accessed on 21 May 2021.

Eisenhart, S ‘New medical device registration requirements issued in South Africa’ (14 December 2016) *Emergo* available at *https://www.emergobyul.com/blog/2016/12/new-medical-device-registration-requirements-issued-south-africa*, accessed on 14 January 2020.

Eisenhart, S ‘South African medical device regulatory system set for implementation’ (22 August 2016) *Emergo* available at *https://www.emergobyul.com/blog/2016/08/south-african-medical-device-regulatory-system-set-implementation*, accessed on 18 June 2020.

Emergo ‘South Africa medical device regulations’ available at *https://www.emergobyul.com/resources/regulations-south-africa*, accessed on 12 January 2020.

Experian ‘What is the Safe Harbour Agreement?’ available at *https://www.experian.co.uk/business/glossary/safe-harbour-agreement/*, accessed on 13 May 2021.

Export.gov ‘South Africa – Medical devices’ (14 July 2019) available at *https://www.export.gov/apex/article2?id=South-Africa-medical-devices*, accessed on 6 March 2021.

Faculty of Health Sciences ‘MSc (Med) Genetic Counselling’ *University of the Witwatersrand* available at *https://www.wits.ac.za/pathology/divisions/human-genetics/academic-programmes/msc-med-genetic-counselling/*, accessed on 16 February 2020.

Farr, C ‘Consumer DNA testing hits a rough patch: Here’s how companies like Ancestry and 23andMe can survive’ (9 February 2020) *CNBC* available at *https://www.cnbc.com/2020/02/07/how-dna-testing-companies-like-ancestry-and-23andme-can-survive.html*, accessed on 7 April 2020.

Fogarty, P ‘Who’s making money from your DNA?’ (1 March 2019) *BBC* available at *https://www.bbc.com/worklife/article/20190301-how-screening-companies-are-monetising-your-dna*, accessed on 15 April 2020.

Furnival, J ‘The trouble with UNinformed consent in direct-to-consumer genetic testing’ (19 May 2021) *SoundRocket* available at *https://soundrocket.com/trouble-informed-consent-dtc-genetic-testing/*, accessed on 8 June 2021.

GEN ‘In warning letter, FDA orders 23andMe to stop selling saliva kit’ (25 November 2013) *Genetic Engineering and Biotechnology News* available at *https://www.genengnews.com/news/in-warning-letter-fda-orders-23andme-to-stop-selling-saliva-kit/*, accessed on 27 October 2019.

GenebyGene ‘Gene by Gene privacy policy’ available at *https://genebygene.com/wp-content/uploads/2019/08/GenebyGene_PrivacyPolicy.pdf*, accessed on 9 January 2021.

Genetic Alliance South Africa ‘Genetic counselling’ available at *https://www.geneticalliance.org.za/support/genetic-counselling/*, accessed on 16 February 2020.

Genetic Counselling ‘Genetic counselling’ available at *http://www.geneticcounselling.co.za/genetic-counselling.php*, accessed on 3 March 2020.

Genetics Education Canada – Knowledge Organization ‘Direct-to-consumer genetic testing’ (October 2017) *GECKO* available at *https://geneticseducation.ca/educational-*

resources/gec-ko-messengers/direct-to-consumer-genetic-testing/, accessed on 27 October 2019.

Genetics Generation 'Direct-to-consumer (DTC) genetics' (2015) *Genetics Generation* available at *https://knowgenetics.org/direct-to-consumer-dtc-genetics/*, accessed 8 October 2019.

Genetics Home Reference 'What are the benefits and risks of direct-to-consumer genetic testing?' (26 May 2020) *NIH* available at *https://ghr.nlm.nih.gov/primer/dtcgenetictesting/dtcrisksbenefits*, accessed on 28 May 2020.

GeneWatch UK 'Genes and marketing' available at *http://www.genewatch.org/sub-396520*, accessed on 8 October 2019.

GeneWatch UK 'GeneWatch slams voluntary gene test guidelines' (2010) available at *http://www.genewatch.org/article.shtml?als%5Bcid%5D=565806&als%5Bitemid%5D=566543*, accessed on 10 February 2020.

GeneWatch UK 'US gene test investigation' available at *http://www.genewatch.org/sub-566548*, accessed on 8 October 2019.

GeneWay 'Disclaimer' (2020) available at *https://geneway.co.za/disclaimer?width=600&height=700*, accessed on 22 June 2020.

GeneWay 'FAQ – Frequently asked questions' (2020) available at *https://www.geneway.co.za/faq-frequently-asked-questions*, accessed on 22 June 2020.

GeneWay 'GeneWay: The most cost-effective DNA test prices in South Africa' (2020 available at *https://geneway.co.za/geneway%E2%84%A2-most-cost-effective-dna-test-prices-south-africa?gclid=CjwKCAjwx9_4BRAHEiwApAt0zsggmRBOwBkvstNzjtgS3DWRe-_WGHzFLUDQWxhDOtZDUOb-es72kBoC-PsQAvD_BwE*, accessed on 22 June 2020.

GeneWay 'GeneWell' (2020) available at *https://www.geneway.co.za/tests/genewell%E2%84%A2*, accessed on 22 June 2020.

GeneWay 'Home' (2020) available at *https://geneway.co.za/*, accessed on 22 June 2020.

GeneWay 'How it works' available at *https://geneway.co.za/i-want-get-tested-how-does-it-work*, accessed on 21 November 2020.

GeneWay ‘I want to get tested, how does it work?’ (2020) available at *https://www.geneway.co.za/i-want-get-tested-how-does-it-work*, accessed on 25 June 2020.

GeneWay ‘Terms & conditions’ (2020) available at *https://geneway.co.za/terms-conditions?width=600&height=700*, accessed on 22 June 2020.

Genomes to People ‘The PGen Study’ available at *https://www.genomes2people.org/research/pgen/*, accessed on 23 October 2019.

GenomeWeb ‘Many DTC genetic testing firms fail to disclose info on privacy, confidentiality, research use’ (23 September 2016) available at *https://www.genomeweb.com/molecular-diagnostics/many-dtc-genetic-testing-firms-fail-disclose-info-privacy-confidentiality#.YQAVsY4zZPY*, accessed on 22 July 2020.

Genomics Africa ‘Human genomes’ available at *http://genomics.africa/human-genomes/*, accessed on 14 October 2019.

Genomics Education Programme ‘Direct-to-consumer genetic tests at an all-time high…’ (20 February 2018) available at *https://www.genomicseducation.hee.nhs.uk/blog/direct-to-consumer-genetic-tests-at-an-all-time-high/*, accessed on 23 February 2020.

Get Smarter ‘The 4th industrial revolution: Will South Africa be ready for the jobs of the future?’ (27 November 2019) available at *https://www.getsmarter.com/blog/career-advice/the-4th-industrial-revolution-will-south-africa-be-ready-for-the-jobs-of-the-future/*, accessed on 27 March 2020.

Gibbs, S ‘What is “safe harbour” and why did the EUCJ just declare it invalid?’ (6 October 2015) *The Guardian* available at *https://www.theguardian.com/technology/2015/oct/06/safe-harbour-european-court-declare-invalid-data-protection*, accessed 13 May 2021.

Giles, J ‘Must I comply with the POPI Act?’ (12 February 2020) *Michalsons* available at *https://www.michalsons.com/blog/must-i-comply-with -the-popi-act/41827*, accessed on 3 March 2021.

Gilligan, C ‘Care ethicists’ (16 July 2011) *Ethics of Care* available at *https://ethicsofcare.org/carol-gilligan/*, accessed on 15 February 2020.

Global Alliance for Genomics and Health ‘GA4GH’ available at *https://www.ga4gh.org/*, accessed on 6 January 2020.

Global Health Research ‘Consent, privacy, and confidentiality’ (2020) *Unite for Sight* available at *https://www.uniteforsight.org/research-course/module4*, accessed on 4 March 2020.

GMDN Agency ‘What is GMDN’ available at *https://www.gmdnagency.org/services/gmdn*, accessed on 7 December 2020.

Goldberg, RL ‘What is “healthism”?’ (30 July 2018) *Ask About Food* available at *https://askaboutfood.com/what-is-healthism-august-2018/*, accessed on 5 February 2020.

Government of Canada ‘Health Canada’s action plan on medical devices: Continuously improving safety, effectiveness and quality’ (December 2018) *Health Canada* available at *https://www.canada.ca/en/health-canada/services/publications/drugs-health-products/medical-devices-action-plan.html*, accessed on 2 February 2020.

Government of Canada ‘Legislation and guidelines – Medical devices’ available at *https://www.canada.ca/en/health-canada/services/drugs-health-products/medical-devices/legislation-guidelines.html*, accessed on 9 February 2020.

Government of Canada ‘Medical devices’ available at *https://www.canada.ca/en/health-canada/services/drugs-health-products/medical-devices.html*, accessed on 9 February 2020.

Grammarist ‘No man is an island’ available at *https://grammarist.com/phrase/no-man-is-an-island/*, accessed on 13 July 2020.

Griffiths, AJF ‘DNA sequencing’ *Britannica* available at *https://www.britannica.com/science/DNA-sequencing*, accessed on 4 February 2021.

Grothaus, M ‘How 23andMe is monetizing your DNA’ (5 January 2015) *Fast Company* available at *https://www.fastcompany.com/3040356/what-23andme-is-doing-with-all-that-dna*, accessed on 21 March 2020.

Gunning, E & N Gabryk ‘South Africa: Transferring personal information outside of South Africa: POPIA and the implications of Schrems II’ (13 August 2020) *Mondaq* available at *https://www.mondaq.com/southafrica/data-protection/973350/transferring-personal-information-outside-of-south-africa-popia-and-the-implications-of-schrems-ii*, accessed on 18 January 2021.

Gutierrez, A 'RE: 23andMe Personal Genome Service™' (10 June 2010) *FDA* available at *https://www.fda.gov/media/79205/download*, accessed on 6 December 2019.

Gutmann Koch, V 'Recreational genetics or research enterprise? Cloudy consent issues arising from direct-to-consumer genetic testing' (2 August 2018) *Bill of Health* available at *https://blog.petrieflom.law.harvard.edu/2018/08/02/recreational-genetics-or-research-enterprise-cloudy-consent-issues-arising-from-direct-to-consumer-genetic-testing/*, accessed on 26 June 2021.

Halim, S 'Should the EU establish a single market for direct-to-consumer genetic testing?' (6 August 2019) *Health Europa* available at *https://www.healtheuropa.eu/eu-genetic-testing/92762/*, accessed on 19 October 2019.

Hamilton, A 'Best inventions of 2008' (2008) *TIME* available at *http://content.time.com/time/specials/packages/article/0,28804,1852747_1854493_1854113,00.html*, accessed on 12 January 2020.

Harrison, TA & CA Lewis 'FDA actions smooth pathway to market for genetic health risk tests and vitamin D tests' (13 November 2017) *Venable* available at *https://www.venable.com/insights/publications/2017/11/fda-actions-smooth-pathway-to-market-for-genetic-h*, accessed on 12 February 2020.

Harvard Women's Health Watch 'Direct-to-consumer genetic testing kits' (September 2010) *Harvard Health Publishing* available at *https://www.health.harvard.edu/newsletter_article/direct-to-consumer-genetic-testing-kits*, accessed on 5 February 2021.

Health Direct 'Genetic testing' (July 2018) available at *https://www.healthdirect.gov.au/genetic-testing*, accessed on 15 September 2019.

Health Professions Council of South Africa (HPCSA) 'Medical technology' available at *https://www.hpcsa.co.za/?contentId=0&menuSubId=48&actionName=For%20Professionals*, accessed on 5 February 2020.

Hedgeman, B & A Shah 'Privacy concerns loom as direct-to-consumer genetic testing industry grows' (28 June 2019) *Epstein Becker Green* available at *https://www.healthlawadvisor.com/2019/06/28/privacy-concerns-loom-as-direct-to-consumer-genetic-testing-industry-grows/*, accessed on 3 March 2020.

Helix ‘Helix Laboratory Platform granted the first and only FDA authorization for a whole exome sequencing platform’ (11 January 2021) available at *https://www.helix.com/pages/helix-laboratory-platform-granted-fda-authorization*, accessed on 7 April 2021.

Helix ‘Helix platform consent’ (10 February 2020) available at *https://www.helix.com/pages/platform-consent*, accessed on 9 January 2021.

Helix ‘Helix privacy policy’ (30 November 2020) available at *https://www.helix.com/pages/privacy-policy*, accessed on 9 January 2021.

Helix ‘Helix terms of service’ (30 November 2020) available at *https://www.helix.com/pages/terms-of-service*, accessed on 9 January 2021.

Hill, R ‘Human Genetics Commission publish final report’ (7 June 2012) *BioNews* available at *https://www.bionews.org.uk/page_93623*, accessed on 17 October 2019.

Hinkley, E ‘What is DNA analysis?’ (21 March 2017) *DNA Testing Choice* available at *https://dnatestingchoice.com/en-us/news/what-is-dna-analysis*, accessed on 4 February 2021.

Hogan Lovells ‘FDA resumes enforcement relating to laboratory developed tests’ *Lexology* available at *https://www.lexology.com/library/detail.aspx?g=20fa976c-8380-454b-9b4d-0e24cf2aa609*, accessed on 13 January 2020.

Hogg, A ‘Welcome to the future: Discovery helping DNA mapping move from sci-fi to reality’ (14 October 2015) *BizNews* available at *https://www.biznews.com/innovation/2015/10/14/welcome-to-the-future-how-discovery-helped-dna-mapping-move-from-sci-fi-to-reality*, accessed on 16 March 2020.

Holmlund, J ‘The FDA’s warning letter to 23andMe’ (12 December 2013) *Bioethics at TIU* available at *https://blogs.tiu.edu/bioethics/2013/12/12/the-fdas-warning-letter-to-23andme/*, accessed on 15 October 2019.

Holpuch, A ‘FDA orders genetics company 23andMe to cease marketing of screening service’ (25 November 2013) *The Guardian* available at *https://www.theguardian.com/science/2013/nov/25/genetics-23andme-fda-marketing-pgs-screening*, accessed on 15 October 2019.

HomeDNADirect ‘About us’ available at *https://www.homednadirect.co.za/about-us/*, accessed on 22 June 2020.

HomeDNADirect ‘Genetic health testing’ (2020) available at *https://www.homednadirect.co.za/genetic-health-testing/*, accessed on 23 June 2020.

HomeDNADirect ‘Privacy policy’ (20 May 2018) available at *https://www.homednadirect.co.za/privacy-policy/*, accessed on 23 June 2020.

HomeDNADirect ‘Terms and conditions – General’ (24 May 2018) available at *https://www.homednadirect.co.za/terms-and-conditions/*, accessed on 23 June 2020.

Hopkins, SC ‘Op-Ed: Take it from a genetic counselor: 23andMe’s health reports are dangerously incomplete’ (26 October 2018) *Lose Angeles Times* available at *https://www.latimes.com/opinion/op-ed/la-oe-hopkins-23andme-genetic-testing-health-20181026-story.html*, accessed on 9 March 2020.

House of Lords Science and Technology Committee ‘Genomic medicine’ (2020) *UK Parliament* available at *https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10709.htm#a37*, accessed on 24 March 2020.

HPCSA ‘Statistics’ (1 October 2018) available at *https://hpcsa.co.za/?contentId=115*, accessed on 7 February 2020.

iSpot.tv ‘23andMe TV commercial’ (2013) available at *http://www.ispot.tv/ad/7qoF/23-and-me*, accessed on 2 November 2019.

Jaslow, R ‘FDA warns 23andMe, tells genetic testing firm to halt sales’ (25 November 2013) *CBS News* available at *https://www.cbsnews.com/news/fda-warns-23andme-tells-genetic-testing-firm-to-halt-sales/*, accessed on 25 October 2020.

Jones, L ‘FDA regulation defines business strategy in direct-to-consumer genetic testing’ (30 October 2017) *Biotech Connection* available at *https://biotechconnectionbay.org/viewpoint/fda-regulation-defines-business-strategy-in-direct-to-consumer-genetic-testing/#:~:text=The%20FDA%20regulates%20both%20genetic,different%20levels%20of%20FDA%20regulation*, accessed on 8 March 2020.

Kapdi, N, S Sulaiman & D Hofmeyr ‘South Africa: Transferring personal information across borders’ (2 February 2016) *Mondaq* available at *https://www.mondaq.com/southafrica/data-protection/462800/transferring-personal-information-across-borders*, accessed on 4 July 2020.

King, J '"It's not personal" – DNA, privacy, and direct to consumer genetic testing' (7 November 2019) *CIS* available at */blog/2019/11/%E2%80%9Cit%E2%80%99s-not-personal%E2%80%9D-%E2%80%94-dna-privacy-and-direct-consumer-genetic-testing,* accessed on 7 December 2019.

Kirby, N 'Commercialisation of healthcare in South Africa: Overview' (1 February 2021) *Thomson Reuters* available at *https://uk.practicallaw.thomsonreuters.com/9-618-5168?transitionType=Default&contextData=(sc.Default)&firstPage=true,* accessed on 6 March 2021.

KnowU 'About us' available at *https://knowudna.com/about-us/#labs,* accessed on 21 November 2020.

KnowU 'Home' available at *https://knowudna.com/,* accessed on 26 July 2020.

Kurth, HA 'South Africa's Protection of Personal Information Act, 2013, goes into effect July 1' (29 June 2020) *The National Law Review* available at *https://www.natlawreview.com/article/south-africa-s-protection-personal-information-act-2013-goes-effect-july-1,* accessed on 9 October 2020.

Laher, I 'The Consumer Protection Act and cross-border transactions' (9 November 2010) *Polity* available at *https://www.polity.org.za/article/the-consumer-protection-act-and-cross-border-transactions-2010-11-09,* accessed on 8 March 2021.

Lefebre, JJ, S Salgaonkar, G Pascutiu et al 'Genetics and insurance: Challenges and opportunities II' (15 April 2019) *RGA* available at *https://rgare.com/knowledge-center/media/research/genetics-and-insurance-challenges-and-opportunities-ii,* accessed on 25 August 2019.

Lendino, T 'The uncertain legal intersection of genetic tests and life insurance' (2018) *Lexology* available at *https://www.lexology.com/library/detail.aspx?g=0599cfaa-36cd-4e3a-b1dc-515964bab0a3,* accessed on 2 October 2019.

Leonelli, S 'Data – From objects to assets' (15 October 2019) *Nature* available at *https://www.nature.com/articles/d41586-019-03062-w,* accessed on 19 November 2019.

Letter from Canadian College of Medical Geneticists to the Honourable Rona Ambrose, Health Canada (19 July 2015) available at *https://www.ccmg-*

ccgm.org/images/pdf/DTC-letter-Final-April-15-2015_V6.pdf, accessed on 12 January 2020.

Levine, DS 'Improving access to genetic testing' (12 February 2018) *Global Genes* available at *https://globalgenes.org/2018/02/21/improving-access-to-genetic-testing/*, accessed on 12 September 2019.

Lewis, R 'Direct-to-consumer genetic testing: a new view' (8 November 2012) *DNA Science* available at *https://dnascience.plos.org/2012/11/08/direct-to-consumer-genetic-testing-a-new-view/*, accessed on 13 January 2020.

Lexico 'Consumer' (2020) available at *https://www.lexico.com/definition/consumer*, accessed on 14 March 2020.

Lexico 'Patient' (2020) available at *https://www.lexico.com/definition/patient*, accessed on 14 March 2020.

LexisNexis 'De minimis definition' available at *https://www.lexisnexis.co.uk/legal/glossary/de-minimis*, accessed on 10 May 2021.

Lister Hill National Center for Biomedical Communications 'Help me understand genetics: Direct-to-consumer genetic testing' (2019) available at *https://ghr.nlm.nih.gov/primer*, accessed on 3 October 2019.

Low, M '12 questions answered on the state of SAHPRA' (5 December 2018) *Spotlight* available at *https://www.spotlightnsp.co.za/2018/12/05/12-questions-answered-on-the-state-of-sahpra/*, accessed on 9 February 2021.

MacArthur, D 'UK House of Lords Report on Genomic Medicine: Implications for DTC genetic testing' (7 July 2009) *Wired* available at *https://www.wired.com/2009/07/uk-house-of-lords-report-on-genomic-medicine-implications-for-dtc-genetic-testing/*, accessed on 27 November 2019.

Maden, C 'Consumers don't necessarily know genetic risk can change' (3 August 2009) *BioNews* available at *https://www.bionews.org.uk/page_91744*, accessed on 17 October 2019.

Malherbe, J 'Long-awaited Medical Device Regulations unpacked' (18 January 2017) *Medical Brief* available at *https://www.medicalbrief.co.za/archives/long-awaited-medical-device-regulations-unpacked/*, accessed on 22 February 2020.

MapmyGenome ‘Terms and conditions’ available at *https://mapmygenome.in/terms-and-conditions/*, accessed on 9 January 2021.

Marquand, R ‘Bible reading altered history’ (13 November 1991) *The Christian Science Monitor* available at *https://www.csmonitor.com/1991/1113/13111.html*, accessed on 8 May 2020.

Martins, E ‘The philosophy of privacy’ (15 August 2018) *ODPA* available at *https://www.odpa.gg/news/news-article/?id=d1c5780d-4e0d-eb11-a813-000d3ad7da76*, accessed on 5 January 2021.

Mayo Clinic ‘Genetic testing’ (14 April 2020) available at *https://www.mayoclinic.org/tests-procedures/genetic-testing/about/pac-20384827*, accessed on 5 January 2021.

McDermott, A ‘Study uncovers new privacy worries for direct-to-consumer DNA testing’ (14 February 2020) *PNAS* available at *http://blog.pnas.org/2020/02/study-uncovers-new-privacy-worries-for-direct-to-consumer-dna-testing/*, accessed on 6 January 2021.

Merriam-Webster ‘Autonomy’ available at *https://www.merriam-webster.com/dictionary/autonomy*, accessed on 3 March 2021.

Merriam-Webster ‘Body fluid’ available at *https://www.merriam-webster.com/medical/body%20fluid*, accessed on 3 August 2020.

Merriam-Webster ‘Ejusdem generis rule’ available at *https://www.merriam-webster.com/legal/ejusdem%20generis%20rule*, accessed on 6 May 2021.

Merriam-Webster ‘Implement’ available at *https://www.merriam-webster.com/dictionary/implement*, accessed on 6 January 2021.

Merriam-Webster ‘Pathology’ available at *https://www.merriam-webster.com/dictionary/pathology*, accessed on 20 August 2020.

Merriam-Webster ‘Reagent’ available at *https://www.merriam-webster.com/dictionary/reagent*, accessed on 6 January 2021.

Mezher, M ‘UK introduces Medicines, Medical Devices Bill in post-Brexit overhaul’ (13 February 2020) *RAPS* available at *https://www.raps.org/news-and-articles/news-articles/2020/2/uk-introduces-medicines-medical-devices-bill-in-p*, accessed on 18 February 2020.

Miceli, S ‘At-home DNA tests still need the “human touch,” say panelists at genomics roundtable workshop’ (13 November 2019) available at *http://www8.nationalacademies.org/onpinews/newsitem.aspx?RecordID=11132019*, accessed on 19 March 2020.

Michel, F ‘“Healthism”: A neoliberal version of wellness’ (9 December 2012) *Solidarity* available at *https://solidarity-us.org/healthism/*, accessed on 6 February 2020.

Milligan, E & J Jones ‘Rethinking autonomy and consent in healthcare ethics’ (29 December 2016) *InTechOpen* available at *https://www.intechopen.com/books/bioethics-medical-ethical-and-legal-perspectives/rethinking-autonomy-and-consent-in-healthcare-ethics*, accessed on 2 March 2020.

Mitchell, C ‘IP address’ (23 June 2020) *Investopedia* available at *https://www.investopedia.com/terms/i/ip-address.asp*, accessed on 6 August 2020.

Molla, R ‘Why DNA tests are suddenly unpopular’ (13 February 2020) *Vox* available at *https://www.vox.com/recode/2020/2/13/21129177/consumer-dna-tests-23andme-ancestry-sales-decline*, accessed on 15 April 2020.

Molteni, M ‘Now you can sequence your whole genome for just $200’ (19 November 2018) *Wired* available at *https://www.wired.com/story/whole-genome-sequencing-cost-200-dollars/*, accessed on 11 October 2019.

Molteni, M ‘The US urgently needs new genetic privacy laws’ (1 May 2019) *Wired* available at *https://www.wired.com/story/the-us-urgently-needs-new-genetic-privacy-laws/*, accessed on 11 October 2019.

Monty, S ‘South Africa: Things to consider when packaging your products’ (7 November 2011) *Mondaq* available at *https://www.mondaq.com/southafrica/consumer-law/149662/things-to-consider-when-packaging-your-products*, accessed on 15 March 2021.

Moody, O ‘DNA testers tell dog to take up basketball’ (5 May 2018) *The Sunday Times* available at *https://www.thetimes.co.uk/article/doubts-over-dna-tests-after-company-fails-to-spot-its-client-was-a-dog-zpwhwr9m5*, accessed on 17 February 2020.

Munro, D ‘Class action law suit filed against 23andMe’ (2 December 2013) *Forbes* available at *https://www.forbes.com/sites/danmunro/2013/12/02/class-action-law-suit-filed-against-23andme/*, accessed on 19 April 2020.

Musée Protestant ‘The Reformers and the Bible: Sola scriptura’ *Musée Protestant* available at *https://www.museeprotestant.org/en/notice/the-reformation-and-the-bible-sola-scriptura/*, accessed on 8 May 2020.

MyHeritage ‘DNA testing’ available at *https://www.myheritage.com/health*, accessed on 19 March 2020.

MyHeritage ‘How do I collect and send back my DNA sample to the lab?’ available at *https://faq.myheritage.com/en/article/how-do-i-collect-and-send-back-my-dna-sample-to-the-lab*, accessed on 25 June 2020.

MyHeritage ‘Privacy policy’ (17 December 2020) available at *https://www.myheritage.com/privacy-policy*, accessed on 9 January 2021.

MyHeritage ‘Terms and conditions’ (2020) available at *https://www.myheritage.com/terms-and-conditions*, accessed on 5 October 2019.

Myriad ‘Myriad informed consent policies’ available at *https://myriadwomenshealth.com/consent-policies/*, accessed on 9 January 2021.

Nair, N ‘Gene testing of embryos in SA’ (7 October 2013) *Sunday Times* available at *https://www.timeslive.co.za/news/south-africa/2013-10-07-gene-testing-of-embryos-in-sa/*, accessed on 16 October 2019.

National Department of Health ‘Human genetics policy guidelines for the management and prevention of genetic disorders, birth defects and disabilities’ *Genetic Alliance* available at *http://www.geneticalliance.org.za/wp-content/uploads/2015/09/Policy-Guidelines-for-the-management-and-prevention-of-genetic-disorders-birth-defects-and-disabilities.pdf*, accessed on 22 November 2019.

National Human Genome Research Institute ‘A brief guide to genomics’ (7 November 2019) *NIH* available at *https://www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics*, accessed on 3 February 2020.

National Human Genome Research Institute ‘Privacy in genomics’ (24 February 2020) *NIH* available at *https://www.genome.gov/about-genomics/policy-issues/Privacy*, accessed on 3 March 2020.

National Practitioner Data Bank ‘Definitions’ (October 2018) *US Department of Health and Human Services* available at *https://www.npdb.hrsa.gov/guidebook/CDefinitions.jsp*, accessed on 6 May 2021.

National Society of Genetic Counselors 'Genetic counselors' available at *https://www.aboutgeneticcounselors.org/*, accessed on 23 June 2020.

National Society of Genetic Counselors 'NSGC Code of Ethics' (April 2017) available at *https://www.nsgc.org/p/cm/ld/fid=12*, accessed on 3 May 2020.

Nature Research 'Genetics research' available at *https://www.nature.com/subjects/genetics-research*, accessed on 7 January 2021.

NextBiosciences 'About us' (2020) available at *https://nextbio.co.za/about/#what*, accessed on 22 June 2020.

NextBiosciences 'CarrierScreen' (2020) available at *https://nextbio.co.za/carrierscreen/*, accessed on 22 June 2020.

NextBiosciences 'Terms and conditions' (2020) available at *https://nextbio.co.za/terms-conditions/*, accessed on 22 June 2020.

NHMRC 'Medical genetic testing: Information for health professionals' (2010) *National Health and Medical Research Council* available at *https://www.nhmrc.gov.au/about-us/publications/medical-genetic-testing-information-health-professionals*, accessed on 21 November 2019.

NIH '15 for 15: Direct-to-consumer genomic testing' (13 February 2019) *National Human Genome Research Institute* available at *https://www.genome.gov/dna-day/15-for-15/direct-to-consumer-genomic-testing*, accessed on 2 September 2019.

NIH 'Coverage and reimbursement of genetic tests' (15 August 2019) *National Human Genome Research Institute* available at *https://www.genome.gov/about-genomics/policy-issues/Coverage-Reimbursement-of-Genetic-Tests*, accessed on 13 September 2019.

NIH 'Direct-to-consumer genomic testing' (13 January 2020) *National Human Genome Research Institute* available at *https://www.genome.gov/dna-day/15-ways/direct-to-consumer-genomic-testing*, accessed on 23 September 2019.

NIH 'NIH scientists introduce British Lords to genomic medicine' *National Human Genome Research Institute* available at *https://www.genome.gov/27526872/house-of-lords-inquiry-on-genomic-medicine-visit-to-nih*, accessed on 24 September 2019.

NIH ‘Review of the ethical, legal and social implications research program and related activities (1990-1995)’ (1 October 2012) *National Human Genome Research Institute* available at *https://www.genome.gov/10001747/elsi-program-review-19901995*, accessed on 17 October 2019.

Noden, A ‘Council of Europe protocol on genetic testing enters into force’ (2 July 2018) *Health Europa* available at *https://www.healtheuropa.eu/council-europe-protocol-on-genetic-testing/86860/*, accessed on 20 November 2019.

Nogrady, B ‘How Indian biotech is driving innovation’ (12 December 2018) *Nature* available at *https://www.nature.com/articles/d41586-018-07671-9*, accessed on 5 January 2020.

Nordling, L ‘A new law was supposed to protect South Africans’ privacy. It may block important research instead’ (22 February 2019) *Science* available at *https://www.sciencemag.org/news/2019/02/new-law-was-supposed-protect-south-africans-privacy-it-may-block-important-research#:~:text=South%20Africa's%20current%2C%202015%20research,%E2%80%9Caware%20of%20the%20purpose.%E2%80%9D*, accessed on 17 February 2020.

Norton Rose Fulbright ‘South African perspective on ECJ Safe Harbour ruling’ (14 October 2015) available at *https://www.dataprotectionreport.com/2015/10/south-african-perspective-on-ecj-safe-harbour-ruling/*, accessed on 13 May 2021.

Nuffield Council on Bioethics ‘Personal genetic profiling services lack evidence for claims’ (12 October 2010) available at *https://www.nuffieldbioethics.org/news/personal-genetic-profiling-services-lack-evidence-for-claims*, accessed on 11 December 2019.

Nuffield Department of Population Health ‘Direct-to-consumer genetic testing (DTCGT)’ available at *https://www.ndph.ox.ac.uk/research/centre-for-health-law-and-emerging-technologies-helex/past-projects/direct*, accessed on 8 December 2019.

OECD ‘Regulatory developments in genetic testing in Austria’ available at *http://www.oecd.org/sti/emerging-tech/regulatorydevelopmentsingenetictestinginaustria.htm*, accessed on 18 October 2019.

OECD ‘Regulatory developments in genetic testing in Canada’ available at *http://www.oecd.org/sti/emerging-*

tech/regulatorydevelopmentsingenetictestingincanada.htm, accessed on 18 October 2019.

OECD 'Regulatory developments in genetic testing in Switzerland' available at *http://www.oecd.org/sti/emerging-tech/regulatorydevelopmentsingenetictestinginswitzerland.htm*, accessed on 18 October 2019.

Office of the Auditor General of Canada '2011 June Status Report of the Auditor General of Canada' (2011) *OAG* available at *https://www.oag-bvg.gc.ca/internet/English/parl_oag_201106_06_e_35374.html*, accessed on 3 December 2019.

Office of the Privacy Commissioner of Canada 'Direct-to-consumer genetic testing and privacy' (4 December 2017) *OPC* available at *https://www.priv.gc.ca/en/privacy-topics/health-genetic-and-other-body-information/02_05_d_69_gen/*, accessed on 21 October 2019.

Omni Accounts 'The fourth industrial revolution in South Africa' available at *https://www.omniaccounts.co.za/articles/is-sa-ready-for-4th-industrial-revolution*, accessed on 27 March 2020.

Oppenheim, J 'Medicines and Related Substances Amendment Acts come into force' (9 June 2017) *Bowmans* available at *https://www.bowmanslaw.com/insights/pharmaceuticals-healthcare/medicines-related-substances-amendment-acts-come-force/*, accessed on 22 July 2020.

Palmer, KM 'Another personal genetics company is sharing client data' (21 July 2015) *WIRED* available at *http://www.wired.com/2015/07/another-personal-genetics-company-selling-client-data*, accessed on 23 March 2020.

Parsons, M & L Crawford 'Malaysia publishes draft "White List" for personal data exports' (27 April 2017) *Hogan Lovells* available at *https://www.hoganlovells.com/en/publications/malaysia-publishes-draft-white-list-for-personal-data-exports*, accessed on 6 May 2021.

Paxman, R 'Direct-to-consumer genetic test results are questionable, research suggests' (6 June 2011) *BioNews* available at *https://www.bionews.org.uk/page_93001*, accessed on 17 October 2019.

Pear, R ‘States pass laws to regulate uses of genetic testing’ (18 October 1997) *The New York Times* available at *https://www.nytimes.com/1997/10/18/us/states-pass-laws-to-regulate-uses-of-genetic-testing.html*, accessed on 27 December 2019.

Pepper, MS, J Scholefield, M Slabbert et al ‘Why South Africa needs better laws for stem cell research and therapy’ (20 August 2015) *The Conversation* available at *https://theconversation.com/why-south-africa-needs-better-laws-for-stem-cell-research-and-therapy-45498*, accessed on 21 April 2020.

PHG Foundation ‘Ban on direct-to-consumer genetic tests in Germany’ (28 April 2009) *University of Cambridge* available at *https://www.phgfoundation.org/news/ban-on-direct-to-consumer-genetic-tests-in-germany*, accessed on 17 October 2019.

PHG Foundation ‘Concerns and prospects for pharmacogenomic testing’ (29 July 2009) *University of Cambridge* available at *https://www.phgfoundation.org/news/concerns-and-prospects-for-pharmacogenomic-testing*, accessed on 17 October 2019.

PHG Foundation ‘First prosecution under US Genetic Information Nondiscrimination Act’ (26 June 2015) *University of Cambridge* available at *https://www.phgfoundation.org/news/first-prosecution-under-us-genetic-information-nondiscrimination-act#:~:text=GINA%20has%20no%20equivalent%20legal,insurance%20purposes%20in%20most%20cases*., accessed on 11 March 2020.

PHG Foundation ‘House of Lords Report on Genomic Medicine’ (8 July 2009) *University of Cambridge* available at *https://www.phgfoundation.org/news/house-of-lords-report-on-genomic-medicine*, accessed on 7 January 2020.

PHG Foundation ‘Human Genetics Commission releases updated report on direct genetic tests’ (5 December 2007) *University of Cambridge* available at *https://www.phgfoundation.org/news/human-genetics-commission-releases-updated-report-on-direct-genetic-tests*, accessed on 18 October 2019.

Picard, A ‘Controversial genetic self-testing kits coming to Canada’ (2 October 2014) *The Globe and Mail* available at *https://www.theglobeandmail.com/life/health-and-fitness/health/genetic-self-testing-kits-to-come-to-canada/article20885678/?click=sf_globefb*, accessed on 7 December 2020.

Pierce, S 'Why you should talk to a genetic counselor before using an at-home DNA test' (3 September 2019) *TMC* available at *https://www.tmc.edu/news/2019/09/why-you-should-talk-to-a-genetic-counselor-before-using-an-at-home-dna-test/*, accessed on 8 March 2020.

Piper, M 'Autonomy: Normative' *Internet Encyclopaedia of Philosophy* available at *https://iep.utm.edu/aut-norm/#SH2c*, accessed on 4 June 2020.

Pollack, A 'FDA orders genetic testing firm to stop selling DNA analysis service' (25 November 2013) *The New York Times* available at *https://www.nytimes.com/2013/11/26/business/fda-demands-a-halt-to-a-dna-test-kits-marketing.html*, accessed on 15 October 2019.

Pomerantz, D 'I was fortunate to get post-23andMe genetic counseling. Everyone should have that option' (7 February 2020) *STAT* available at *https://www.statnews.com/2020/02/07/genetic-counseling-after-home-genetic-testing/*, accessed on 17 March 2020.

Privacy International 'DNA and genetic data' available at *https://privacyinternational.org/learn/dna-and-genetic-data#:~:text=DNA%20holds%20the%20key%20to,through%20DNA%20or%20RNA%20analysis.*, accessed on 8 January 2021.

Professional and Public Policy Committee of the European Society of Human Genetics 'Letter to the Human Genetics Commission' (2009) available at *https://www.eshg.org/fileadmin/www.eshg.org/documents/PPPC-ESHG-DTC-06122009.pdf*, accessed on 30 January 2020.

PWC 'Disrupting Africa: Riding the wave of the digital revolution' (2016) available at *https://www.pwc.com/gx/en/industries/technology/publications/disrupting-africa--riding-the-wave-of-the-digital-revolution.html*, accessed on 27 March 2020.

Pygma Consulting 'The Protection of Personal Information Act (POPIA): What it is, how to become compliant, and where it falls short' available at *https://pygmaconsulting.com/the-protection-of-personal-information-act-popia-what-it-is-how-to-become-compliant-and-where-it-falls-short/*, accessed on 9 October 2020.

Rakel, RE 'Therapeutics' (2021) *Britannica* available at *https://www.britannica.com/science/therapeutics*, accessed on 6 May 2021.

Ramirez, N 'The EU-US Privacy Shield invalidated: What this means for you' (1 June 2021) *Osano* available at *https://www.osano.com/articles/privacy-shield-invalidated*, accessed on 11 June 2021.

Raper, V 'Event review: Whose genome is it anyway?' (18 October 2010) *BioNews* available at *https://www.bionews.org.uk/page_92640*, accessed on 15 October 2019.

Ravenscraft, E 'How to protect your DNA data before and after taking an at-home test' (12 June 2019) *The New York Times* available at *https://www.nytimes.com/2019/06/12/smarter-living/how-to-protect-your-dna-data.html*, accessed on 6 March 2020.

Regalado, A 'J. Craig Venter to offer DNA data to consumers' (22 September 2015) *MIT Technology Review* available at *https://www.technologyreview.com/2015/09/22/166157/j-craig-venter-to-sell-dna-data-to-consumers/*, accessed on 9 February 2020.

RegDesk '6 major differences between EU's MDR/IVDR and MDD/IVDD' (25 December 2018) available at *https://www.regdesk.co/major-differences-eu-mdr-mdd/*, accessed on 8 February 2020.

RegDesk 'An overview of medical device regulations in the UK' (9 February 2019) available at *https://www.regdesk.co/overview-medical-device-regulations-united-kingdom/*, accessed on 8 February 2020.

Reisenberger, T 'Does the Consumer Protection Act apply to foreign retailers selling goods online to South Africa?' (20 November 2018) *Legalese* available at *https://legalese.co.za/does-the-consumer-protection-act-apply-to-foreign-retailers-selling-goods-online-to-south-africa/*, accessed on 8 March 2021.

Ries, J 'There's a catch that comes with taking a 23andme test' (12 March 2018) *Huffpost* available at *https://www.huffpost.com/entry/23andme-warnings-at-home-genetic-test_n_5c01b108e4b0606a15b54dae?guccounter=1*, accessed on 10 September 2019.

Roberts, M 'House of Lords Genomic Medicine Report calls for NHS overhaul to improve future healthcare' (9 July 2009) *BioNews* available at *https://www.bionews.org.uk/page_91701*, accessed on 24 March 2020.

Roberts, M '"DIY genetics tests" guidelines published' (4 August 2010) *BBC News* available at *https://www.bbc.com/news/health-10844495*, accessed on 20 August 2019.

Rosenbaum, E '5 biggest risks of sharing your DNA with consumer genetic-testing companies' (16 June 2018) *CNBC* available at *https://www.cnbc.com/2018/06/16/5-biggest-risks-of-sharing-dna-with-consumer-genetic-testing-companies.html*, accessed on 7 January 2020.

SAHPRA 'About us' available at *https://www.sahpra.org.za/who-we-are/*, accessed on 5 May 2020.

SAHPRA 'Medical devices' available at *https://www.sahpra.org.za/medical-devices/*, accessed on 5 May 2020.

SAPA 'Consumer Protection Act has international implications' (26 July 2010) *Sunday Times* available at *https://www.timeslive.co.za/sunday-times/business/2010-07-26-consumer-protection-act-has-international-implications/*, accessed on 8 March 2021.

Saunders, R 'Report review: Consumer genetic testing' (2 April 2012) *BioNews* available at *https://www.bionews.org.uk/page_93524*, accessed on 12 October 2019.

Science Learning Hub 'DNA extraction' (18 June 2009) available at *https://www.sciencelearn.org.nz/resources/2036-dna-extraction#:~:text=DNA%20extraction%20is%20a%20routine,see%20a%20stringy%20white%20mass.*, accessed on 4 February 2021.

Science Media Centre 'Experts react to House of Lords Report on Genomic Medicine' (6 July 2009) available at *https://www.sciencemediacentre.org/experts-react-to-house-of-lords-report-on-genomic-medicine-2/*, accessed on 24 March 2020.

Servick, K 'Frustrated U.S. FDA issues warning to 23andMe' (25 November 2013) *Science* available at *https://www.sciencemag.org/news/2013/11/frustrated-us-fda-issues-warning-23andme*, accessed on 15 October 2019.

Shanks, P '"Weak and meaningless" guidelines for UK DTC tests' (10 August 2010) *Center for Genetics and Society* available at *https://www.geneticsandsociety.org/biopolitical-times/weak-and-meaningless-guidelines-uk-dtc-tests*, accessed on 15 January 2020.

Shaprio, JK 'Regulation of Laboratory Developed Tests by FDA: Time for the agency to cease and desist until Congress enacts legislation' (21 October 2019) *FDA Law Blog* available at *http://www.fdalawblog.net/2019/10/regulation-of-laboratory-developed-tests-by-fda-time-for-the-agency-to-cease-and-desist-until-congress-enacts-legislation/*, accessed on 28 December 2019.

Shuren, JE & J Woodcock 'FDA announces collaborative review of scientific evidence to support associations between genetic information and specific medications' (20 February 2010) *FDA* available at *http://www.fda.gov/news-events/press-announcements/fda-announces-collaborative-review-scientific-evidence-support-associations-between-genetic*, accessed on 19 March 2020.

Singh, A 'Data without borders: How to manage cross-border data transfers in South Africa' (7 November 2017) *Alt Advisory* available at *https://altadvisory.africa/2017/11/07/data-without-borders-manage-cross-border-data-transfers-south-africa/*, accessed on 4 July 2020.

Smith, M 'Marketing, manufacturing, packaging & labelling, advertising' (12 October 2018) *Pharma Boardroom* available at *https://pharmaboardroom.com/legal-articles/marketing-manufacturing-packaging-labelling-advertising-south-africa/*, accessed on 16 January 2021.

Soini, S, H Kääriäinen & M Hietala 'National regulation of genetic counselling' (January 2008) *EuroGenTest* available at *http://www.eurogentest.org/index.php?id=679*, accessed on 18 October 2019.

Song, K '4 risks consumers need to know about DNA testing kit results and buying life insurance' (4 August 2018) *CNBC* available at *https://www.cnbc.com/2018/08/04/4--risks-consumer-face-with-dna-testing-and-buying-life-insurance.html*, accessed on 30 August 2019.

South African Revenue Service 'FAQ: Do I need to register as an importer if I buy personal goods from abroad e.g. from Amazon?' (9 March 2021) available at *https://www.sars.gov.za/faq/faq-do-i-need-to-register-as-an-importer-if-i-buy-personal-goods-from-abroad-e-g-from-amazon/*, accessed on 7 May 2021.

Southern African Development Community 'Member States' available at *https://www.sadc.int/member-states/*, accessed on 18 January 2021.

Southern African Society for Human Genetics 'Genetic counselling in South Africa' *SASHG* available at *http://sashg.org/about/focus-groups/genetic-counselling-south-africa/?s=*, accessed on 14 February 2020.

Stanford Encyclopedia of Philosophy 'Paternalism' (2017) available at *https://plato.stanford.edu/entries/paternalism/*, accessed on 2 February 2020.

Stevens, Y & G Marchant 'What's in it for me?' (8 April 2015) *SLATE* available at *https://slate.com/technology/2015/04/recruiting-people-for-genetics-studies-on-facebook.html*, accessed on 23 March 2020.

Stoljar, N 'Feminist perspectives on autonomy' (11 December 2018) *Stanford Encyclopaedia of Philosophy* available at *https://plato.stanford.edu/entries/feminism-autonomy/*, accessed on 5 May 2020.

Taylor, JS 'Autonomy: Ethics and political philosophy' (20 June 2017) *Encyclopaedia Britannica* available at *https://www.britannica.com/topic/autonomy*, accessed on 16 June 2020.

Taylor, SC 'Health care ethics' *Internet Encyclopedia of Philosophy* available at *https://iep.utm.edu/h-c-ethi/*, accessed on 6 October 2020.

The Local 'French ban on home DNA testing can't stop the Christmas craze' (20 December 2018) available at *https://www.thelocal.fr/20181220/french-ban-on-dna-testing-cant-stop-the-craze*, accessed on 19 October 2019.

The Medical Futurist 'An unmatchable list of direct-to-consumer genetic testing companies' (22 October 2018) *TMF* available at *https://medicalfuturist.com/an-unmatchable-database-of-direct-to-consumer-genetic-testing-companies/*, accessed on 9 September 2019.

The Medical Futurist 'The genomic data challenges of the future' (27 October 2018) *TMF* available at *https://medicalfuturist.com/the-genomic-data-challenges-of-the-future/*, accessed on 8 March 2020.

The Wellness Revolution 'FAQ' available at *https://thewellnessrevolution.co.za/faq/*, accessed on 22 June 2020.

The Wellness Revolution 'How it works' available at *https://thewellnessrevolution.co.za/how-it-works/*, accessed on 22 June 2020.

Tiwari, SS 'Capitalizing genome: The business of direct-to-consumer genetic testing in India' (1 April 2019) *Indian Academy of Sciences* available at *http://confluence.ias.ac.in/capitalizing-genome-the-business-of-direct-to-consumer-genetic-testing-in-india/*, accessed on 23 January 2020.

Tomlinson, C 'IN-DEPTH: The tangled web of medical device regulation in SA' (3 September 2020) *Spotlight* available at *https://www.spotlightnsp.co.za/2020/09/03/in-*

depth-the-tangled-web-of-medical-device-regulation-in-sa/, accessed on 5 December 2020.

TUV SUD 'FAQS: In Vitro Diagnostic Medical Device Regulation (IVDR)' available at *https://www.tuvsud.com/en/industries/healthcare-and-medical-devices/medical-devices-and-ivd/medical-device-market-approval-and-certification/eu-in-vitro-diagnostic-medical-device-regulation/faqs-in-vitro-diagnostic-medical-device-regulation-ivdr*, accessed on 10 February 2020.

UK Parliament 'Genomic Medicine – Science and Technology Committee' available at *https://publications.parliament.uk/pa/ld200809/ldselect/ldsctech/107/10709.htm#a37*, accessed on 14 December 2019.

Uren, C 'How South Africans can use their DNA to be good genomic citizens' (23 November 2020) *SABC* available at *https://www.sabcnews.com/sabcnews/how-south-africans-can-use-their-dna-to-be-good-genomic-citizens/*, accessed on 5 December 2020.

US Food and Drug Administration '510(k) premarket notification' *FDA* available at *https://www.accessdata.fda.gov/scripts/cdrh/cfdocs/cfPMN/pmn.cfm?ID=K182784*, accessed on 21 January 2020.

US Food and Drug Administration 'Approvals of FDA-regulated products' (12 June 2018) *FDA* available at *http://www.fda.gov/news-events/approvals-fda-regulated-products*, accessed on 26 January 2020.

US Food and Drug Administration 'Closeout letter - 23andMe' (25 March 2014) *FDA* available at *http://www.fda.gov/inspections-compliance-enforcement-and-criminal-investigations/warning-letters/23andme-inc-03252014*, accessed on 7 October 2019.

US Food and Drug Administration 'Device approvals, denials and clearances' (24 April 2019) *FDA* available at *http://www.fda.gov/medical-devices/products-and-medical-procedures/device-approvals-denials-and-clearances*, accessed on 19 December 2019.

US Food and Drug Administration 'Direct-to-consumer tests' (20 December 2019) *FDA* available at *http://www.fda.gov/medical-devices/vitro-diagnostics/direct-consumer-tests*, accessed on 28 October 2019.

US Food and Drug Administration 'FDA 101: An Overview of FDA's Regulatory Review and Research Activities' (29 October 2019) *FDA* available at

http://www.fda.gov/about-fda/what-we-do/fda-101-overview-fdas-regulatory-review-and-research-activities, accessed on 19 December 2019.

US Food and Drug Administration 'FDA allows marketing of first direct-to-consumer tests that provide genetic risk information for certain conditions' (6 April 2017) *FDA* available at *http://www.fda.gov/news-events/press-announcements/fda-allows-marketing-first-direct-consumer-tests-provide-genetic-risk-information-certain-conditions*, accessed on 19 March 2020.

US Food and Drug Administration 'FDA authorizes first direct-to-consumer test for detecting genetic variants that may be associated with medication metabolism' (20 February 2020) *FDA* available at *http://www.fda.gov/news-events/press-announcements/fda-authorizes-first-direct-consumer-test-detecting-genetic-variants-may-be-associated-medication*, accessed on 19 March 2020.

US Food and Drug Administration 'Federal Food, Drug, and Cosmetic Act (FD&C Act)' (11 March 2018) *FDA* available at *http://www.fda.gov/regulatory-information/laws-enforced-fda/federal-food-drug-and-cosmetic-act-fdc-act*, accessed on 15 January 2020.

US Food and Drug Administration 'Framework for Regulatory Oversight of Laboratory Developed Tests (LDTs)' (October 2014) *FDA* available at *https://www.fda.gov/regulatory-information/search-fda-guidance-documents/framework-regulatory-oversight-laboratory-developed-tests-ldts*, accessed on 2 February 2020.

US Food and Drug Administration 'Is it really "FDA approved?"' (17 January 2017) *FDA* available at *http://www.fda.gov/consumers/consumer-updates/it-really-fda-approved*, accessed on 21 January 2020.

US Food and Drug Administration 'Laws enforced by FDA' (12 April 2018) *FDA* available at *http://www.fda.gov/regulatory-information/laws-enforced-fda*, accessed on 15 January 2020.

US Food and Drug Administration 'Laboratory Developed Tests' (27 September 2018) *FDA* available at *https://www.fda.gov/medical-devices/vitro-diagnostics/laboratory-developed-tests*, accessed on 14 November 2019.

US Food and Drug Adminsitration 'Medical devices' (13 March 2020) *FDA* available at *http://www.fda.gov/medical-devices*, accessed on 19 March 2020.

US Food and Drug Administration ‘Overview of IVD regulation’ (16 September 2019) *FDA* available at *http://www.fda.gov/medical-devices/ivd-regulatory-assistance/overview-ivd-regulation*, accessed on 7 October 2019.

US Food and Drug Adminsitration ‘Recently-approved devices’ (4 May 2019) *FDA* available at *http://www.fda.gov/medical-devices/device-approvals-denials-and-clearances/recently-approved-devices*, accessed on 19 March 2020.

US Food and Drug Administration ‘Review memorandum: 23andMe Personal Genome Service (PGS) test’ (6 April 2017) *FDA* available at *https://www.accessdata.fda.gov/cdrh_docs/pdf16/den160026.pdf*, accessed on 3 October 2019.

US Food and Drug Administration ‘Table of pharmacogenetic associations’ (25 February 2020) *FDA* available at *http://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations*, accessed on 6 November 2019.

US Food and Drug Administration ‘What we do’ (11 March 2018) *FDA* available at *https://www.fda.gov/about-fda/what-we-do*, accessed on 18 October 2019.

van Harmelen, J ‘Ownership of genetic information and tissue under National Health Act’ (2020) *Lexology* available at *https://www.lexology.com/library/detail.aspx?g=6bd60fff-6db2-48c5-aaf1-adad605fdeff*, accessed on 24 March 2020.

van Harmelen, J, R Boda & R Dullabh ‘Genetic information: A new resource to be mined?’ (20 October 2020) *International Law Office* available at *https://www.internationallawoffice.com/Newsletters/Healthcare-Life-Sciences/South-Africa/ENSafrica/Genetic-information-a-new-resource-to-be-mined*, accessed on 16 February 2021.

Varga, O & J Sequeiros ‘Definitions of genetic testing in european and other legal documents’ (2004) *EuroGenTest* available at *http://www.eurogentest.org/index.php?id=732*, accessed on 18 October 2019.

Via, M ‘Big data in genomics: Ethical challenges and risks’ (24 June 2017) *Universitat de Barcelona* available at *https://www.redalyc.org/jatsRepo/783/78354511003/html/index.html*, accessed on 12 March 2020.

Werksmans Attorneys 'Local consumer law has international ramifications' (7 July 2010) available at *https://www.werksmans.com/legal-updates-and-opinions/local-consumer-law-has-international-ramifications/*, accessed on 8 March 2021.

Wild, S 'South Africa's science academy urges update to nation's genetics laws' (7 December 2018) *Nature* available at *https://www.nature.com/articles/d41586-018-07637-x*, accessed on 15 February 2020.

Williams, A & L Strom 'UK government publishes new draft legislation on medicines and medical devices' (20 February 2020) *Arnold & Porter* available at *https://www.biosliceblog.com/2020/02/uk-government-publishes-new-draft-legislation-on-medicines-and-medical-devices/*, accessed on 26 February 2020.

Wilson, BE 'IVDs and LDTs: Evolving visions of FDA oversight under the VALID Act' (29 July 2019) *Med Device Online* available at *https://www.meddeviceonline.com/doc/ivds-and-ldts-evolving-visions-of-fda-oversight-under-the-valid-act-0001*, accessed on 27 November 2019.

Wojcicki, A '23andMe provides an update regarding FDA's review' (5 December 2013) *23andMe Blog* available at *https://blog.23andme.com/news/fda-update/*, accessed on 24 March 2020.

Wong, G 'Why screening your genes is big business' (6 May 2009) *CNN* available at *http://edition.cnn.com/2009/HEALTH/05/05/consumer.genomics/index.html*, accessed on 3 October 2019.

World Health Organization 'Genetic research' available at *https://www.who.int/genomics/research/en/*, accessed on 7 January 2021.

World Health Organization 'Human genomics in global health' *WHO* (2019) available at *https://www.who.int/genomics/elsi/gentesting/en/*, accessed on 14 October 2019.

World Health Organization 'Quality & safety in genetic testing: An emerging concern' *WHO* available at *https://www.who.int/genomics/policy/quality_safety/en/index1.html*, accessed on 9 February 2020.

World Health Organization 'WHO definitions of genetics and genomics' *WHO* available at *https://www.who.int/genomics/geneticsVSgenomics/en/*, accessed on 4 February 2020.

Young, S 'Privacy versus confidentiality' (8 December 2017) *Office of the Saskatchewan Information and Privacy Commissioner* available at *https://oipc.sk.ca/privacy-versus-*

confidentiality/#:~:text=In%20terms%20of%20information%2C%20privacy,used%2C%20and%2For%20disclosed.&text=Confidentiality%20is%20the%20duty%20to,only%20to%20the%20extent%20possible., accessed on 20 April 2020.

Zhang, B 'Investigating disease risk through direct-to-consumer genetic testing' (2017) *Rare Disease Review* available at *https://www.rarediseasereview.org/publications/2017/5/2/investigating-disease-risk-through-direct-to-consumer-genetic-testing*, accessed on 24 March 2020.

Zheng, K, D Saeed, G Chan et al 'The genesis of direct-to-consumer genetic testing' available at *http://rgdoi.net/10.13140/RG.2.2.24470.75845*, accessed on 24 March 2020.

Zucker, S & B Harris 'The Protestant Reformation' *Khan Academy* available at *https://www.khanacademy.org/humanities/renaissance-reformation/reformation-counterreformation/beginner-guide-reforrmation/a/the-protestant-reformation*, accessed on 8 May 2020.

APPENDIX

UNIVERSITY OF KWAZULU-NATAL
INYUVESI YAKWAZULU-NATALI

Miss Amy Elizabeth Gooden (215031300)
School Of Law
Howard College

Dear Miss Amy Elizabeth Gooden,

Protocol reference number: 00013389
Project title Amended : Unregulated or not? A legal analysis of South Africa's legislative framework relevant to direct-to-consumer genetic testing.

Exemption from Ethics Review

In response to your application received on 23 September 2021 , your school has indicated that the protocol has been granted **EXEMPTION FROM ETHICS REVIEW.**
Any alteration/s to the exempted research protocol, e.g., Title of the Project, Location of the Study, Research Approach and Methods must be reviewed and approved through an amendment/modification prior to its implementation. The original exemption number must be cited.

For any changes that could result in potential risk, an ethics application including the proposed amendments must be submitted to the relevant UKZN Research Ethics Committee. The original exemption number must be cited.

In case you have further queries, please quote the above reference number.

PLEASE NOTE:
Research data should be securely stored in the discipline/department for a period of 5 years.

I take this opportunity of wishing you everything of the best with your study.

Yours sincerely,

[redacted]

Mr Simphiwe Peaceful Phungula
obo Academic Leader Research
School Of Law