THE INVOLUNTARY DISCLOSURE OF CONFIDENTIAL GENETIC INFORMATION TO RELATED AFFECTED THIRD PARTIES

A DISSERTATION PRESENTED

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Abbreviations

BMA: British Medical Association

DNA: Deoxyribose Nucleic Acid

GC-SA: Genetic Counsellors South Africa

GINA: Genetic Information Nondiscrimination Act

HGP: Human Genome Project

HPCSA: Health Professions Council of South Africa

HUGO: Human Genome Organisation

POPI: Protection of Personal Information Act

MRC: Medical Research Council

NHMRC: National Health and Medical Research Council

NHA: National Health Act

SASHG: Southern African Society for Human Genetics

UNESCO: United Nations Educational, Scientific and Cultural Organization

WHO: World Health Organization

WMA: World Medical Association

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CHAPTER 1

General Introduction

1.1 Background

The rapid advancement of medical technology has resulted in significant advantages for persons throughout the world. Genetic testing is an example of such advancement that has revolutionized medical science. However, it has also created several legal and ethical implications, many of which stem from the issue of confidentiality. There is unequivocally the need for such testing and thus there has been an almost seamless incorporation of human genetic technology into our healthcare system. The ramifications of such testing, however, have not been dealt with sufficiently by the South African legislature as well as South African hospitals in terms of their institutional ethics policies.

During the genetic testing process itself, an individual may be diagnosed with a specific genetic condition or disorder.[1] The information gained from such testing is used to prevent the onset of the condition.[2] It may also assist persons in making decisions on the treatment and management of the condition.[3] This information may be known as genetic information.[4] It must be stated, however, that genetic testing is not just an individual experience.[5] Unlike other medical tests, there is a familial nature to genetic tests in that it may produce information that is relevant to a patient's family. Genetic tests may indicate that biological relatives of the individual being tested are also at risk of a genetic condition or disorder.[6] Unwittingly, this places a clinician in a difficult legal and ethical predicament.

As a result, this study focuses on the privacy of genetic information in the context of an involuntary disclosure made by a medical practitioner to affected related third parties. In most cases, patients willingly warn affected individuals. The focal point of this study, however,

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[1] B Godard...et al 'Genetic information and testing in insurance and employment: technical, social and ethical issues' (2003) 11 (2) *European Journal of Human Genetics* 123.
[2] ibid 123.
[3] ibid 123.
[4] 'Genetic Information Discrimination' available at *http://www.eeoc.gov/laws/types/genetic.cfm*, accessed on 10 July 2014.
[5] M Van Riper 'Genetic Testing and the Family' (2005) 50 (3) *Journal of Midwifery & Women's Health* 227.
[6] S M Wolf & J P Kahn 'Genetic Testing and the Future of Disability Insurance: Ethics, Law & Policy' (2007) 35 (2) *Journal of Law, Medicine & Ethics* 9.

occurs when a patient refuses to provide their consent to the healthcare professional, preventing the healthcare professional from making the disclosure. Specifically, where healthcare professionals are placed in the unfortunate position of having to balance a patient's right to confidentiality, regarding their genetic information, against a duty to warn at-risk relatives. This dissertation questions whether or not healthcare practitioners have a duty to protect their patient's privacy at all costs.

Moreover, this study seeks to identify and scrutinize problems related to the confidentiality and disclosure of genetic information. The study will go on to provide solutions in order to minimise such problems between a patient, a medical practitioner and at-risk family members. In order to fully comprehend the dilemma that healthcare practitioners are faced with, the scope of the doctor-patient relationship must also be examined. It is important to establish whether or not healthcare professionals have an obligation to abide by their duty of confidentiality by not disclosing genetic information they have learnt as a result of carrying out their professional duties.

Furthermore, in order to understand the protection afforded to genetic information it is necessary to determine how genetic information is classified in South Africa. The two distinct views on how genetic information may be classified will be discussed later on. The manner in which genetic information is defined and classified in South Africa is crucial as it will determine how and to what degree genetic information is protected.

Although the above questions are of extreme importance, the ultimate purpose of this study is to question and accordingly establish whether or not South Africa has failed to adequately regulate the confidentiality and disclosure of genetic information. Any lack of guidance in this area would be disconcerting as healthcare professionals would be left to their own discretion when making decisions that may affect the health of several individuals. Consequently, if South African regulations and guidelines are found to be deficient, this study aims to provide recommendations that address the legal and ethical concerns of healthcare professionals.

At this juncture, it must be stated that any regulations relating to genetic information cannot merely be based on a legal outlook. An ethical perspective is required. Law often embodies the principles of ethics, and this dilemma deserves no less. There are several bioethics theories and biomedical ethics principles that will guide us as to what the best course of action is when faced with genetic information. It is my recommendation that a medical

practitioner must have regard for the ethical considerations involved in genetic testing before deciding whether or not to disclose genetic information to at-risk relatives.

Overall, the complexities involved in genetic sciences have affected countries across the world, which in turn has encouraged many countries to develop guidelines that govern the protection and disclosure of genetic information. It is apparent that the disclosure of genetic information is going to become an increasingly problematic issue in South Africa, unless steps are taken to provide unambiguous and ethically informed legislation and ethics policies. Accordingly, a detailed analysis on the different approaches countries have taken when regulating the involuntary disclosure and protection of genetic information must be discussed. It is vital that we draw on foreign and international governance documents in order to provide well informed recommendations for future genetics legislation and organizational ethics policies.

1.2 Breakdown of Chapters

Chapter 2 - provides an understanding of the science involved in the study of genetics. The chapter goes on to examine the history and development of genetic testing.

Chapter 3 - examines the classification of genetic information in South Africa; that is, whether or not genetic information falls within the definition of health information or whether such information is considered to be exceptional information.

Chapter 4 - is a general examination of confidentiality and disclosure in South Africa.

Chapter 5 - analyses current South African governance documents on the confidentiality and disclosure of genetic information

Chapter 6 - involves the discussion and application of ethical theories and biomedical principles to the act of involuntary disclosure.

Chapter 7 - examines foreign and international governance documents that have provided guidance on the confidentiality and disclosure of genetic information. It also examines the foreign ambit of the duty to warn.

Chapter 8 - provides recommendations for future genetics privacy legislation and organisational ethics policies in South Africa.

Chapter 9 - provides a final conclusion to this dissertation.

CHAPTER 2

The Science of Genetics: An Introduction to Genetic Testing

2.1 Introduction

With serious health related illnesses in South Africa requiring valuable resources and awareness, it is easy to overlook genetic conditions.[7] Nevertheless, approximately 5% of all pregnancies result in a child being born with a serious genetic condition, disability or congenital malformation.[8] It is also estimated that 1 in 19 South Africans have a genetic disorder.[9] Essentially, it is important to keep in mind that a genetic condition can occur in any person in any country.[10] This study attempts to demonstrate that with the prevalence of genetic conditions and genetic testing technology, issues of disclosure and confidentiality are bound to increase.

Consequently, in order to fully comprehend the difficulties linked to the disclosure of genetic information, it is necessary to understand the science behind genetics. Understanding the complexities of the field of genetics will provide insight as to how these complexities can be solved competently with the development of adequate regulations. Accordingly, this chapter sets out the science of genetics as well as the history and development of genetic testing.

2.2 Understanding Genetics

2.2.1 Cells, DNA and Genes

The human body is comprised of trillions of cells that contain hereditary material.[11] A cell is made up of several components, but of particular importance is the nucleus which contains deoxyribonucleic acid (DNA), a cell's hereditary material. A nucleus contains 23 pairs of

[7] World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003) 1.
[8] ibid 1.
[9] F Loubser...et al 'Support for Genetic Counselling services in the Western Cape' (25 February 2013) available at *https://gcnewssa.wordpress.com/*, accessed on 29 December 2014.
[10] World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003) 3.
[11] 'Handbook Help Me Understand Genetics' (7 July 2014) available at *http://ghr.nlm.nih.gov/handbook.pdf*, accessed on 14 July 2014.

chromosomes which is made up of DNA. A gene is made up of DNA and may be described as a functional unit of heredity. It has been estimated that a human being has between 20,000 and 25,000 genes. Each person has two copies of each gene which are essentially inherited from each parent (one copy from each parent). A map is used to determine a particular gene's location on a chromosome.[12]

2.2.2 Gene Mutations

'A gene mutation is a permanent change in the DNA sequence that makes up a gene'.[13] However, not all genetic mutations cause genetic disorders. There are two ways in which a gene mutation may occur. Firstly, there is the acquired mutation which occurs in DNA at some point in an individual's life. This mutation may be caused by environmental factors (e.g. ultraviolet radiation from the sun) or can occur as a result of a mistake made during cell division when DNA copies itself. An acquired mutation cannot be inherited. Secondly, a mutation may be inherited from a parent. In this instance, a gene mutation may result in a particular condition that affects multiple members of a family.[14]

2.2.3 Inheritance of Genetic Disorders

Even though a genetic condition may run in a family, it is difficult to predict that a family member will inherit the disorder as there are several factors that influence the chances of developing the disorder.[15] For instance, there are several ways in which the disorder may be inherited, the most relevant being the following:[16]

(i) Dominant Inheritance: Where an individual has a dominant disorder, the child has a 50 percent chance that he or she will inherit the mutated gene.

[12] ibid.
[13] ibid.
[14] ibid.
[15] ibid.
[16] ibid.

(ii) Recessive Inheritance: Where there are 'two unaffected people who each carry one copy of the mutated gene for'[17] a recessive disorder (carriers), the child has a 25 percent chance of developing the disorder.

(iii) X-linked Dominant Inheritance: Where a man has an X and a Y chromosome, a woman has two X chromosomes. Where a man has an X- linked dominant condition, his sons will not be affected, but all of his daughters will inherit the disorder. Where a woman has an X-linked dominant condition, there is a 50 percent chance that her child will inherit the condition.

(iv) X-linked Recessive Inheritance: Where a man has an X-linked recessive condition, his sons will remain unaffected, but 'his daughters will carry one copy of the mutated gene'.[18] Where a woman has an X-linked recessive condition, she 'has a 50 percent chance of having sons who are affected and a 50 percent chance of having daughters who carry one copy of the mutated gene'.[19]

2.2.4 Reduced Penetrance

When a person experiences the mutation of a particular gene and exhibits symptoms of a genetic condition, it is known as penetrance. Reduced penetrance occurs where a person has the mutation, but does not develop symptoms of the condition. For example, a person with the BRCA1 or BRCA2 gene mutation may or may not develop cancer during their life. A doctor is unable to predict if the individual with the mutation will eventually develop cancer.[20] This example demonstrates one of the complications a medical practitioner must consider when deciding whether or not they should make the disclosure.

[17] ibid.
[18] ibid.
[19] ibid
[20] ibid.

2.2.5 Genetic Conditions

Research has shown that the majority of conditions have a genetic element.[21] As individuals, we have a tendency of identifying genetic disorders such as Down Syndrome, breast cancer and cystic fibrosis, as its own illness, forgetting that they all share a common genetic component. The following are examples of well-known detectable genetic conditions:

(i) Colon Cancer: Affects both men and women and occurs when a malignant (cancerous) tumour develops in the large intestine. Most colon cancer cases are as a result of a genetic mutation that may happen to an individual. However, there is also a hereditary form, in which, a child may inherit the mutation from their parent.[22]

(ii) Huntington's Disease: This is an inherited neurological condition that ensures the loss of motor control, emotional problems and cognitive decline. The gene that causes Huntington's disease is a dominant gene and thus if an affected individual was to have a child, there is a 50 percent chance that the child will inherit the gene. If a child does, in fact, inherit the Huntington's disease gene, they will develop the disorder during their lifetime. However, if the child does not inherit the gene, the child will not develop Huntington's disease, nor will he or she pass on the gene to future children.[23]

2.3 Genetic Testing

2.3.1 The History and Development of Genetic Testing

The origins of genetic testing can be traced back to the nineteenth century when Gregor Mendel's experiment with peas introduced the basic mechanisms of inheritance to the world.[24] It was in 1953, however, when James Watson and Francis Crick made a discovery that revolutionised genetic science. In their well-known paper, Watson and Crick put forward an entirely different structure of deoxyribose nucleic acid (DNA) which has come to be

[21] ibid.
[22] 'Learning About Colon Cancer' available at *http://www.genome.gov/10000466*, accessed on 21 August 2014.
[23] 'Learning About Huntington's Disease' available at *https://www.genome.gov/10001215,* accessed on 22 August 2014.
[24] T H Morgan...et al *The Mechanism of Mendelian Hereditary* (1915) 1.

known as the double helix of DNA.[25] Their discovery has made a profound impression on the development of genetics by providing great insight on gene function.[26]

The 1980s saw the ideological origin for the Human Genome Project (HGP).[27] The HGP set out an expansive research effort to sequence and map all of the genes, collectively known as a genome, in the human species.[28] In 2000, the HGP announced that the majority of the human genome had been sequenced and in 2003 the project was completed, giving us the ability to understand the complete genetic blueprint of a human being.[29] Overall, the HGP has uncovered an abundance of information that has allowed for the development of genetic testing.

The emergence of genetic sciences in South Africa started soon after Watson and Crick's discovery of the double helix of DNA.[30] Any discussion of the history of genetics in South Africa, however, would not be complete without an examination of the eugenics movement. The term eugenics was coined by Francis Galton in 1883.[31] Eugenics advocated for the improvement of the human race by eliminating undesirable traits and multiplying desirable traits.[32]

Galton's campaign to improve the human race, or at the very least prevent its' supposed decline through selective breeding, spread rapidly throughout the world.[33] The progress of the eugenics movement, however, declined after the movement had been embraced by the Nazis.[34]

In a broader context, it may be said that human genetics stems from the idea of eugenics which advocates for the improvement of the physical, mental and behavioural characteristics of a human being by means of hereditary manipulation.[35] The fundamental similarity between genetics in the contemporary world and eugenics can be seen in one common goal: the

[25] F Crick & J Watson 'Molecular Structure of Nucleic Acids: a Structure for Deoxyribose Nucleic Acid' (1953) 171(4356) *Nature* 737.
[26] E Mayr *The Growth of Biological Thought: Diversity, Education, and Inheritence* (1982) 824.
[27] 'A Brief History of the Human Genome Project' available at *http://www.genome.gov/12011239*, accessed on 21 August 2013. The genome project was established in 1987 with the original goal of seeking information of gene mutations.
[28] ibid.
[29] ibid.
[30] J G R Kromberg & A Krause 'Human Genetics in Johannesburg, South Africa: Past, Present and Future' (2013) 103 (12) *SAMJ* 957.
[31] World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003) 10.
[32] D J Kevles 'Eugenics and human rights' (1999) 319 (7207) *BMJ* 435.
[33] D Wikler 'Can we Learn from Eugenics?' (1999) 25 (2) *Journal of Medical Ethics* 183.
[34] ibid 183.
[35] D J Kevles *In the Name of Eugenics: Genetics and the Uses of Human Hereditary* (1985) vii (preface).

elimination of 'inferior' genes in human beings.[36] Accordingly, we can see how far the medical world has come as a discussion of inferior genes today would involve the elimination of mutated or altered genes that may be connected to a particular condition or disease.[37]

From a South African perspective, eugenic ideas always seemed to be intrinsically linked to race.[38] The eugenics movement found great momentum in South Africa in 1920 when H B Fantham set up the Eugenics and Genetic Standing Committee of the South African Association for the Advancement of Science.[39] The Committee was against the mixing of racial groups as well as mixing between persons of the same racial groups who had different potentialities.[40] Accordingly, appropriate marriage and sterilization laws were to be used in order to prevent the birth of persons suffering from hereditary mental diseases, alcoholism and criminal tendencies.[41] The South African government, however, did not respond to the recommendations of the Committee and only went as far as suggesting voluntary sterilization.[42] In the coming years, it became apparent that certain sectors of the South African population did not provide support for the eugenics movement.[43]

More importantly, however, was the continued development of genetic services during this period. In the early 1960s, there was the establishment of informal genetic counselling services in both Johannesburg and Cape Town.[44] In 1972, Trefor Jenkins, Professor of Human Genetics at the University of Witwatersrand, attended the International Congress of Human Genetics in Vienna.[45] The knowledge he had gained on that trip led to the formalization and expansion of genetic counselling clinics in South Africa.[46] 1977 saw the growth and expansion of laboratory services and health personnel across the country.[47] This

[36] J Caplan & J Torpey *Documenting Individual Identity: The Development of State Practices in the Modern World* (2001) 280.
[37] ibid 280.
[38] S Dubow 'South Africa: Paradoxes In The Place Of Race' in A Bashford & P Levine (ed) *The Oxford Handbook of the History of Eugenics* (2010) 274. However, the apartheid regime in South African history alone allowed for white supremacy and thus such eugenic ideas were never of key importance.
[39] T Jenkins 'Medical Genetics in South Africa' (1990) 27 (12) *Med Genet* 762.
[40] ibid 762.
[41] ibid 762.
[42] ibid 762.
[43] ibid 762.
[44] J G R Kromberg & A Krause 'Human Genetics in Johannesburg, South Africa: Past, Present and Future' (2013) 103 (12) *SAMJ* 957.
[45] ibid 957.
[46] ibid 957.
[47] J G R Kromberg & E B Sizer & A L Christianson 'Genetic services and testing in South Africa' (2013) 4 (3) *Journal of Community Genetics* 416.

was as a result of the Minister of Health stating that genetic services formed a crucial part of South Africa's health system.[48]

At present, there are 12 main laboratories in South Africa that perform genetic tests.[49] There are also various other private laboratories throughout the country where certain genetic tests can be performed.[50] An example of such a private laboratory is Lancet Laboratories in Johannesburg which performed 3,292 genetic tests in 2008 alone.[51] Another example that shows the extent of genetic testing in South Africa concerns the four academic Human Genetics departments who performed 16,073 genetic tests in 2008.[52]

2.3.2 The Science of Genetic Testing

Genetic testing refers to the study of a person's genetic make-up[53] in order to establish whether or not that person has a genetic condition. The testing provides medical practitioners with the means to diagnose a genetic disorder. Genetic testing technology is currently able to detect numerous genetic conditions.[54] There are several different types of genetic tests that may be used to detect and confirm a genetic disorder. These tests may also determine the likelihood of an individual developing such a genetic disorder or the chances of them passing it on to potential children. Genetic testing may provide certainty where there are suggestions of possible conditions, diseases or disorders in a family's medical history.[55] The different types of genetic tests include:[56]

a) Newborn Screening - involves the screening of newborns for genetic disorders which enables medical practitioners to treat them as soon as possible.

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[48] ibid 416.
[49] ibid 418.
[50] ibid 418.
[51] ibid 418.
[52] ibid 418.
[53] 'Issues in the Insurance and Employment Settings: A Report ' available at *https://www.law.utoronto.ca/documents/Lemmens/Genetic%20Information%20and%20the%20Law.pdf* , accessed on 02 July 2014.
[54] 'Handbook Help Me Understand Genetics' (7 July 2014) available at *http://ghr.nlm.nih.gov/handbook.pdf*, accessed on 14 July 2014.
[55] Wolf (note 6 above; 8).
[56] 'Handbook Help Me Understand Genetics' (7 July 2014) available at *http://ghr.nlm.nih.gov/handbook.pdf*, accessed on 14 July 2014.

b) Diagnostic Testing - is performed at anytime during an individual's life in order to determine and ultimately confirm the presence of a particular genetic condition. This information will allow individuals to make decisions concerning the treatment and management of the condition.

c) Carrier Testing - involves testing persons with a family history of a genetic disorder. This testing is used to determine the likelihood of a couple 'having a child with a genetic condition'.[57]

d) Prenatal Testing - is used during pregnancy to determine if there is likelihood that the foetus will have a genetic condition.

e) Preimplantation Testing - is used to detect genetic abnormalities in an embryo before it is implanted through the process of in-vitro fertilization.

f) Predictive and Presymptomatic Testing - is testing that is used to identify gene mutations and accordingly the likelihood of an individual developing a genetic disorder in the future, that is, before there are actual signs of the disorder.

2.3.3 The Imperfect Science of Genetic Testing

Despite its predictive value, there is no denying that genetic testing still has a long way to go. For instance, although genetic testing is able to detect multiple genetic conditions, there are still genetic disorders for which no test has been developed in that the genetic cause of the disorder has not been found or a test to detect the condition has not been formed as yet.[58] In terms of conditions that they do have tests for, it is important to note that such tests do not provide certainty. These tests generally infer that there is a likelihood that the individual will develop the condition.[59] In these cases, genetic testing is said to be limiting.

There are also concerns relating to the practicalities involved in genetic testing, especially in a developing country such as South Africa. At present, it is evident that genetic testing is only available at a tertiary care level and thus such services are only available at private

[57] ibid.
[58] 'Handbook Help Me Understand Genetics' (7 July 2014) available at *http://ghr.nlm.nih.gov/handbook.pdf*, accessed on 14 July 2014.
[59] B Kuschke 'Access to genetic information and the insurer's duty of genetic data protection' (2007) available at *http://repository.up.ac.za/bitstream/handle/2263/5508/Kuschke_Access(2007).pdf?sequence=1*, accessed on 21 May 2014.

practices.[60] It is estimated that genetic tests in South Africa cost between R1,500 and R13,400 per test, depending on the specific type of genetic test required.[61] The exclusivity of the testing ensures that average South Africans will not be able to afford genetic testing. Accordingly, any legislation that is implemented in the future would focus primarily on the private sector.

Lastly, there are also problems surrounding the technicalities involved in genetic testing. As with any medical procedure, there is always the chance of an error occurring. In terms of genetic testing, errors in laboratories are not unheard off.[62] For example, a mistake may be made when a sample is being taken, labeled or examined. Other problems include the inadequate training of personnel, inappropriate test selection and misinterpreting test results.[63] In terms of these inadequacies, it is clear that future genetics legislation should also contain guidelines on good laboratory and testing practices.

2.4 Conclusion

Although genetic testing has come a long way, it is apparent that the study of genetics is not based on complete accuracy. Where genetic testing produces indecisive results, it would seem that medical practitioners would go through a much more difficult thought process when deciding whether an involuntary disclosure needs to be made. Accordingly, it is submitted that the lack of certainty involved in genetic testing justifies the need for exclusive genetics legislation that takes the uncertainty of genetics into account.

[60]Kromberg (note 47 above; 416).
[61] M C Herbst 'Fact Sheet on Genetic Testing for Cancer' (June 2014) available at *http://www.cansa.org.za/files/2014/06/Fact-Sheet-Genetic-Testing-Cancer-June-2014.pdf*, accessed on 15 December 2014.
[62] D Ravine & G Suthers 'Quality standards and samples in genetic testing' (2012) 65 (5) *Journal of Clinical Pathology* 1.
[63] 'Good Laboratory Practices for Molecular Genetic Testing for Heritable Diseases and Conditions' , available at *http://www.sashg.org/documents/CDC%20guidelines%20for%20genetic%20testing%20June%202009.pdf*, accessed on 16 December 2015.

CHAPTER 3

Genetic Information: Health Information verses Genetic Exceptionalism

3.1 Introduction

In order to determine the extent and ambit of the protection afforded to genetic information, it is necessary to establish how genetic information is defined and accordingly classified. The classification of genetic information is a controversial topic that has been debated throughout the world, resulting in two distinct schools of thought. Firstly, genetic information may be recognized as health information. On the other hand, we can recognise the uniqueness of genetic information and accordingly classify such information as distinctive from standard health information.[64] A determination is essential as such a classification will determine if special protection is required for such sensitive information.

3.2 The Definition of Genetic Information

In the United States, the Genetic Information Nondiscrimination Act (GINA) has defined genetic information as:[65]

> (4) Genetic information--
>
> (A) In general -- The term "genetic information" means, with respect to any individual, information about--
>
> (i) such individual's genetic tests,
>
> (ii) the genetic tests of family members of such individual, and
>
> (iii) the manifestation of a disease or disorder in family members of such individual.

[64] L O Gostin & J G Hodge 'Genetic privacy and the law: An end to genetics exceptionalism' (1999) 40 *Jurimetrics* 31.
[65] The Genetic Information Nondiscrimination Act of 2008 section 201.

(B) Inclusion of genetic services and participation in genetic research.--Such term includes, with respect to any individual, any request for, or receipt of, genetic services, or participation in clinical research which includes genetic services, by such individual or any family member of such individual.

(C) Exclusions -- The term “genetic information” shall not include information about the sex or age of any individual.

In 2010, final regulations were added under GINA which expanded the definition of genetic information to include information of the foetus carried by the individual as well as information on the individual's relative who is pregnant. The regulation goes on to state that genetic information also includes information on an embryo that is legally held by the individual or their relatives.[66]

Although broad, GINA has provided a comprehensive definition of genetic information. From this definition, we can state that genetic information includes information on the genetic tests of not only the relevant individual, but also their family members. It is also impressive that GINA goes further with its regulations by including information on future and potential children. For the purposes of this study, we can accept the extensive definition provided by GINA.

3.3 Health Information verses Genetic Exceptionalism

3.3.1 Health Information

From the above definition, the composition of genetic information is reasonably clear. However, the ultimate question asked by academics is whether or not genetic information is, in fact, special when compared to health information. Ethicist, Thomas Murray, contends that there is no justifiable reason for treating genetic information differently from other health information.[67] Dr Murray asserts that an acceptance for exceptionalism would result in a clear

[66] 'Regulations Under the Genetic Information Nondiscrimination Act of 2008' available at *https://www.federalregister.gov/articles/2010/11/09/2010-28011/regulations-under-the-genetic-information-nondiscrimination-act-of-2008#h-15*, accessed on 24 August 2014.

[67] 'Is genetic information truly exceptional?' available at *http://www.alrc.gov.au/publications/3-coming-terms-genetic-information/genetic-information-truly-%E2%80%98exceptional%E2%80%99?print*, accessed on 26 May 2014.

divide of diseases and disorders into separate groupings: genetic and non-genetic illness.[68] In support of this argument he has stated that there is not much in terms of our health that has no genetic element.[69] Dr Murray has gone on to state that much of our medical information is, in fact, genetic information, but is not defined as such because it is not as a direct result of a genetic test. [70]

Undeniably, there is merit in his argument as it is virtually impossible to completely separate health and genetic information. However, stating that genetic information should receive the same amount of protection as health information simply because there are similarities between the two should not be a compelling enough argument. In this instance, it is important to remember the exceptional information that can be derived solely from genetic tests.

Other writers that are against exceptionalism include Gostin and Hodge who have concluded that genetic exceptionalism is flawed for two reasons:[71]

> (i) strict protections of autonomy, privacy and equal treatment of persons with genetic conditions threaten the accomplishment of public goods; and
>
> (ii) there is no clear demarcation separating genetic data from other health data; other health data deserve protections in a national health information infrastructure.

Additionally, we may question the exceptionalism of genetic information as it may be relegated to just another means that is used to establish a medical condition.[72] In fact, it has been stated that certain medical tests provide more certainty than genetic testing.[73] A cholesterol test, for example, may predict and confirm poor health, whereas genetic testing, in certain cases, may only provide knowledge on increased susceptibility to a particular condition.[74] From the definition of genetic information provided by GINA, we can state that genetic information does not include any specific medical information, that is, the definition focuses on information that is as a result of genetic testing. Accordingly, in line with this

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[68] F Lagay 'Should Genetic Information Be Treated Separately?' (2001) 3 (1) *AMA Journal of Ethics* available at *http://journalofethics.ama-assn.org/2001/01/gnth1-0101.html*, accessed on 4 May 2015.
[69] ibid.
[70] ibid.
[71] Gostin & Hodge (note 64 above; 21).
[72] ibid 33.
[73] B Kuschke 'Access to genetic information and the insurer's duty of genetic data protection' (2007) available at *http://repository.up.ac.za/bitstream/handle/2263/5508/Kuschke_Access(2007).pdf?sequence=1*, accessed on 21 May 2014.
[74] ibid.

school of thought, it may be said that genetic information is not always as exceptional as one might believe.

3.3.2 Genetic Exceptionalism

In terms of genetic exceptionalism, genetic information is regarded as unique information, separate from standard health information.[75] Genetic exceptionalism has been defined as the 'societal practice of treating genetic data as different from other types of health data for the purposes of assessing privacy and security protections'.[76] Proponents of genetic exceptionalism, Annas, Glantz, and Roche state that there are three reasons as to why genetic information should be considered as distinctive private information:[77]

(i) it can predict an individual's likely medical future for a variety of conditions;

(ii) it divulges personal information about one's parents, siblings and children; and

(iii) it has historically been used to stigmatize and victimize individuals.

As noted above, genetic information may include information on genetic disorders that individuals may have already developed. However, such information is also of a predictive nature as it determines the likelihood of the individual actually developing the genetic condition.[78] In addition, genetic information may also be used to prevent genetic conditions that, at present, cannot be cured.[79] Genetic information also has the potential to influence individuals' decisions on reproduction, health and lifestyle.[80]

Another facet that sets genetic information apart is the fact that genetic tests produce information on whether or not the individual's family is at risk of certain genetic disorders.[81]

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[75] 'Is genetic information truly exceptional?' available at *http://www.alrc.gov.au/publications/3-coming-terms-genetic-information/genetic-information-truly-%E2%80%98exceptional%E2%80%99?print*, accessed on 26 May 2014.
[76] Gostin & Hodge (note 64 above; 31).
[77] G Annas & L H Glantz & P Roche 'Drafting the Genetic Privacy Act: Science, policy and practical considerations' (1995) 23 (4) *Journal of Law, Medicine and Ethics* 360.

[78] 'Handbook Help Me Understand Genetics' (7 July 2014) available at *http://ghr.nlm.nih.gov/handbook.pdf*, accessed on 14 July 2014.
[79] S A M McLean *Contemporary Issues in Law, Medicine and Ethics*(1996) 217.
[80] ibid 217.
[81] 'Handbook Help Me Understand Genetics' (7 July 2014) available at *http://ghr.nlm.nih.gov/handbook.pdf*, accessed on 14 July 2014.

By itself, this aspect is certainly exceptional as the affected family members may not have had this information previous to the testing of the individual.[82].

Overall, writers have also argued that genetic information is special, as where the gene is hereditary and not acquired, it provides information that is not dependent on a person's personal decisions surrounding their diets and lifestyle.[83] It is apparent that genetic testing can produce information that no other scientific testing can provide. The impact that this information can have on at-risk relatives, in my opinion, justifies why genetic information should be treated as exceptional. Accordingly, it is submitted that such distinct and sensitive information deserves a higher degree of protection than standard health information.

3.4 Conclusion

Overall, academics throughout the world continue to debate for and against genetic exceptionalism. Undoubtedly, genetic and health information, to a certain extent, is and will always be intrinsically linked. However, from the above discussion, it is clear that genetic testing produces information that is different from health information Therefore, it is submitted that in order to prevent the mismanagement of genetic information, there is a need to develop exclusive regulations that provide specific guidance on how to handle genetic information. For this reason, this study proposes that for the purposes of special protection, there is a need to lean towards genetic exceptionalism.

[82] F Lagay 'Should Genetic Information Be Treated Separately?' (2001) 3 (1) *AMA Journal of Ethics* available at *http://journalofethics.ama-assn.org/2001/01/gnth1-0101.html*, accessed on 4 May 2015.
[83] McLean (note 79 above; 222).

CHAPTER 4

Confidentiality, Privacy, Access to Information, and Disclosure in South Africa

4.1 Introduction

Medical practitioners are placed in a difficult position when they obtain knowledge, through their patient, that is helpful to third parties. In these situations, clinicians have the burdensome task of having to decide whether or not to breach confidentiality. Accordingly, this chapter examines the ambit of confidentiality and privacy laws in South Africa. It goes on to explore disclosure laws in South Africa and whether or not these laws apply to genetic information.

4.2 The Ethical Duty of Confidentiality

The trust and confidentiality involved in a doctor-patient relationship is not based purely on the law. There is an ethical duty that a medical professional has towards their patient. There are several sources of ethical codes in terms of confidentiality. For instance, there is the age-old Hippocratic Oath which is at the cornerstone of the ethical duty to maintain confidentiality in a doctor-patient relationship.[84] There is one section of the Oath that is of particular importance when discussing the concept of confidentiality:[85]

> Whatsoever things I see or hear concerning the life of men, in my attendance on the sick or even apart therefrom, which ought not be noised abroad, I will keep silence thereon, counting such things to be as sacred secrets.

The above words contained in the Hippocratic Oath captures the essence of the concept of confidentiality. This statement alone demonstrates the abundance of respect afforded to the principle of confidentiality. In order to emphasize the importance of the Hippocratic Oath, it

[84] M Davies *Textbook on Medical Law* (1996) 26.
[85] ibid 26.

must be mentioned that the World Medical Association (WMA) has updated the Oath in 1948 which has resulted in the Declaration of Geneva.[86]

The ethical principle of autonomy also plays a vital role in the confidentiality of a doctor-patient relationship.[87] In terms of this principle, personal information of an individual belongs to that individual and should not be disclosed to others, unless consent is obtained or in the instance 'where there is a legitimate requirement to breach confidentiality'.[88] Patients have an expectation of privacy when they share personal information with their medical practitioner, and thus in terms of their right to privacy,[89] they should be allowed autonomy over their private information.[90]

There is also the concept of trust to consider. A patient's expectation of confidence allows for trust which in turn ensures communication during the diagnosis and treatment process. Trust between a doctor and their patient ensures an openness and willingness from the patient to communicate. Where there is no understanding that a doctor has an ethical and legal duty to maintain the confidences of a patient, the doctor-patient relationship becomes inconsequential as in all probability, patients would withhold information that is necessary to make a correct diagnosis.[91]

Furthermore, in the event that a breach of confidentiality is justifiable, good clinical practice will dictate that patients should be advised before rather than later that their confidential information has been disclosed.[92] Even where the disclosure is necessary and justifiable, when a healthcare practitioner breaches confidentiality, they should be aware that they are likely going to lose the trust of a patient, irreversibly harming the doctor-patient relationship.[93] Accordingly, it must be mentioned that the HPCSA has predicted such harm to the doctor-patient relationship and has thus developed guidelines[94] which examines the concept of consent to testing. These guidelines state that there is only informed consent where healthcare practitioners have discussed certain matters with anyone considering

[86] World Medical Association Medical Ethics Manual 2 ed (2009) 24.
[87] A Dhai & DJ McQuoid-Mason *Ethics, Human Rights and Health Law* (2011) 87.
[88] ibid 87.
[89] Section 14 of the Constitution of the Republic of South Africa 1996. Section 14 states that everyone has the right to privacy.
[90] Dhai & McQuoid-Mason (note 87 above; 87).
[91] Dhai & McQuoid-Mason (note 87 above; 87).
[92] Dhai & McQuoid-Mason (note 87 above; 87).
[93] Dhai & McQuoid-Mason (note 87 above; 88).
[94] Health Professions Council of South Africa: Seeking Patients' Informed Consent: The Ethical Considerations, Booklet 9 (2008).

genetic testing.[95] This will include a discussion on the possibility of disclosure to affected related third parties. Essentially, it is hoped that the honesty provided by healthcare practitioners is sufficient to maintain a healthy doctor-patient relationship.

4.3 Confidentiality Laws in South Africa

Privacy and confidentiality laws in South Africa are currently governed by the Constitution,[96] legislation, and the common law. At the core of statutory law is the National Health Act[97] (NHA) which is arguably the most important and comprehensive health legislation that the South African legislature has ever passed. The NHA provides guidance and certainty on a variety of complex health issues relevant in South Africa. Of particular importance to this study, is the section on confidentiality. This section essentially recognizes and protects the confidentiality of health information.[98] Section 14 of the NHA regulates confidentiality between a healthcare professional and their patient. The section reads as follows:[99]

> (1) All information concerning a user, including information relating to his or her health status, treatment or stay in a health establishment, is confidential.
>
> (2) Subject to section 15, no person may disclose any information contemplated in subsection (1) unless-
>
> (a) the user consents to that disclosure in writing;
>
> (b) a court order or any law requires that disclosure; or
>
> (c) non-disclosure of the information represents a serious threat to public health.

[95] ibid 9.
[96] The Constitution of the Republic of South Africa,1996.
[97] 61 of 2003.
[98] National Health Act 61 of 2003 section 14.
[99]61 of 2003.

4.3.1 Confidentiality, Disclosure, and Genetic Information

Section 14 of the NHA[100] makes health information confidential and thus in order for genetic information to be confidential under the NHA it must fall under the classification of health information. Hypothetically, if genetic information were to be classified as health information, genetic information would remain confidential between a patient and their doctor, unless one of the above three exceptions applied. In the event that one of the above exceptions do not apply, the information remains confidential, unless the healthcare provider decides to involuntarily disclose such information. It is important to remember, however, that there is no indication from the NHA that genetic information does, in fact, fall under the scope of the Act.

Accordingly, section 15 of the NHA[101] regulates disclosures made by a healthcare provider:[102]

> (1) A health worker or any health care provider that has access to the health records of a user may disclose such personal information to any other person, health care provider or health establishment as is necessary for any legitimate purpose within the ordinary course and scope of his or her duties where such access or disclosure is in the interests of the user.

In terms of this section, a healthcare provider may only make a disclosure if it is in the interests of their patient.[103] However, this dissertation deals with the situation in which a medical practitioner makes a disclosure for the benefit of a third party. Accordingly, when a medical practitioner makes such a disclosure, it is apparent that it is against the wishes of their patient and thus contrary to section 15(1) of the NHA[104], that is, the disclosure is not in the interest of the patient. As a result, we can conclude that section 15(1) of the NHA[105] does not regulate the involuntary disclosure of genetic information.

[100] 61 of 2003.
[101] 61 of 2003.
[102] Section 15 of the National Health Act 61 of 2003
[103] Section 15 of the National Health Act 61 of 2003. Section 1 of the National Health Act 61 of 2003 states that "user means the person receiving treatment in a health establishment, including receiving blood or blood products...".
[104] 61 of 2003.
[105] 61 of 2003.

Accordingly, this dissertation sets out to determine whether the NHA[106] should be amended to unambiguously include guidance on the protection and involuntary disclosure of genetic information. This would ensure the confidentiality of genetic information. Alternatively, Parliament could create legislation that focuses exclusively on genetic information and its implications.

At this juncture, it is important to emphasize that this study is proposing that genetic exceptionalism should be accepted in South Africa. In line with this proposition, genetic information should have its own classification, rather than being classified as health information. This study asserts that genetic information is unique and distinctive information that warrants special protection. For this reason, in line with the concept of genetic exceptionalism (discussed in the previous chapter), this study proposes the need for exclusive legislation that will adequately regulate the confidentiality and involuntary disclosure of genetic information.

In support of this assertion, the Promotion of Access to Information Act[107] must be mentioned. Essentially, in terms of genetic information, it is apparent that this Act cannot provide relief for affected third parties in that a third party may only call upon this right if they are aware that there is genetic information relevant to them. Accordingly, this dissertation focuses on the instance where affected third parties are unaware that there is, in fact, information that is of interest to them. As a result, the right provided in the Promotion of Access to Information Act[108] is insufficient and thus exclusive effective genetics legislation is required.

4.3.2 Protection of Personal Information Act

In 2013, the president signed the Protection of Personal Information Act[109] (POPI). However, as of 11 April 2014 only certain sections of POPI have come into force.[110] POPI is relevant to this study in terms of section 32(5) of the Act which states that 'Personal information

[106] 61 of 2003.
[107] Act 2 of 2000.
[108] Act 2 of 2000.
[109] Protection of Personal Information Act 4 of 2013.
[110] G Teare 'South Africa: POPI Takes Effect' available at *http://www.mondaq.com/x/308172/Data+Protection+Privacy/POPI+Takes+Effect*, accessed on 28 August 2014.

concerning inherited characteristics may not be processed in respect of a data subject from whom the information concerned has been obtained...'

In this study, it is submitted that the NHA[111] deals exclusively with health information (which does not include genetic information). One of the reasons for this conclusion is that S32(51) of POPI specifically uses the words inherited characteristics. The use of such terminology infers a distinction between genetic information and health information as recognised by the NHA. Although recognition of this exceptional information is a positive step, there is still the need for the legislature to develop legislation that adequately deals with the concept of genetic information.

4.3.3 The Right to Privacy

In South Africa, the right to privacy is both a Constitutional and a common law right. The right to privacy is a fundamental right in our Constitution that all South African's are entitled too.[112] The concepts of privacy and confidentiality are intrinsically linked to one another. The concept of privacy, however, is clearly distinctive as it 'relates to aspects of a person's being into which no one else should intrude'.[113] Privacy is clearly a bigger notion than confidentiality and thus problems may arise when attempting to establish the ambit and scope of the right to privacy.

The right to confidentiality appears indirectly in section 14 of the Constitution in terms of the right to privacy, which states the following:[114]

> Everyone 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.

[111] Act 61 of 2003.
[112]The Constitution of the Republic of South Africa, 1996 section 14.
[113] Dhai & McQuoid-Mason (note 87 above; 86).
[114] The Constitution of the Republic of South Africa, 1996 section 14.

The section of particular importance is s 14(d) which makes reference to the privacy of their communications. Ultimately, we can infer that this section protects any communication between the individual undergoing genetic testing and his or her medical practitioner. Essentially, when a patient reveals personal information to their medical practitioner, they make the conscious decision to surrender control of certain aspects of their privacy in the belief that the information will remain solely with that practitioner. Even though they have relinquished a limited amount of control of their privacy, he or she should still have control as to how that information is shared.[115] Accordingly, a patient's privacy is violated when personal genetic information is shared with others, against their wishes.

It is crucial to point out that the Constitutional Court has also recognized the right to privacy as a common law right.[116] The Constitutional Court has stated that there is an intrinsic link between human dignity and privacy.[117] In terms of this dissertation, when a medical practitioner discloses a patient's genetic information to a third party, without their consent, the patient's privacy is violated. When such an infringement occurs, a patient's dignity[118] will also be affected. Accordingly, when a doctor decides to make an involuntary disclosure, he or she needs to be aware that the patient's right to privacy is not the only right that is being violated.

The common law right to privacy requires healthcare professionals to keep the confidence of their patients, except in certain circumstances. [119] The exception that is relevant to this study occurs where there is a threat to an endangered third party. A case that highlights this exception is the case of *Tarasoff v Regents of the University of California*[120] which essentially stated that there may be a breach of confidentiality where there is an endangered third party. Generally, in terms of this exception, a medical practitioner has a 'moral, social and legal duty to disclose certain information to an endangered third party that could save that person's

[115] Dhai & McQuoid-Mason (note 87 above; 87).
[116] Bernstein and others v Bester and others NNO 1996 (2) SA 751 (CC) at para 68.
[117]Khumalo v Holomisa 2002 5 SA 401 (CC) at para 27. The court went on to say the following ' *The right to privacy, entrenched in s 14 of the Constitution, recognises that human beings have a right to a sphere of intimacy and autonomy that should be protected from invasion. This right serves to foster human dignity. No sharp lines then can be drawn between reputation, dignitas and privacy in giving effect to the value of human dignity in our Constitution*.'.
[118] Section 10 of the Constitution of the Republic of South Africa 1996. Section 10 states that everyone has inherent dignity and the right to have their dignity respected and protected.
[119]Dhai & McQuoid-Mason (note 87 above; 88).
[120] Tarasoff v Regents of the University of California (1976).

life, or possibly prevent irreversible damage to his or her health'.[121] On the whole, it can be said that the disclosure of genetic information to at-risk relatives satisfies this criteria.

4.3.3.1 The Limitation Clause

As with all rights contained in the Constitution,[122] the right to privacy is not absolute. The right to privacy may be limited in terms of section 36 of the Constitution. Section 36(1) of the Constitution states that any right contained in the Bill of Rights may be limited in terms of the law of general application where it 'is reasonable and justifiable in an open and democratic society based on dignity, equality and freedom'. [123] Accordingly, even though a patient has a right to the privacy of their genetic information, this right may be limited where there is an affected related third party.

4.3.3.2 Access to Information

Section 32(1) of the Constitution[124] provides that every person has the right to access information. The Promotion of Access to Information Act[125] is national legislation that gives effect to the right to access information in terms of section 32(2). In terms of this Act, it is important to note that medical information falls under the definition of personal information. Accordingly, section 34(1)[126] states that a public body must refuse access to a record if the disclosure involves the unreasonable disclosure of personal information of a third party. Section 34(2)(d)[127] goes on to states that a record may not be refused where the information consists of a person's physical or mental health, or well-being. Consequently, it is apparent that affected related third parties should be allowed to access the genetic information of a relative as it concerns their health.

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[121] J A Singh 'The health professional and law in South Africa' in: K Moodley (ed). *Medical Ethics, Law and Human Rights: a South African Perspective* (2011) 138.
[122] The Constitution of the Republic of South Africa, 1996.
[123] The Constitution of the Republic of South Africa, 1996 section 36(1). Also see S v Makwanyane and Another 1995 (3) SA 391 (CC) at para 104. In Makwanyane, the Constitutional Court stated the limitation of a constitutional right involves the weighing of competing values and therefore the limitation of a right is an assessment based on proportionality.
[124]The Constitution of the Republic of South Africa 1996.
[125] Act 2 of 2000.
[126] The Promotion of Access to Information Act 2 of 2000.
[127] The Promotion of Access to Information Act 2 of 2000.

Furthermore, access to information is an important right that an affected related third party has. At the same time, however, the patient has a right to privacy of their genetic information. This conflict of rights experienced by the affected family member and the patient demand that a balancing of rights take place. Accordingly, the limitation clause will come into effect in order to establish which right takes precedence in the particular circumstances. The purpose of this study is not to disregard the right of privacy that all patients are entitled to and therefore there is the need to emphasize that the third party's right to information cannot consistently take preference over a patient's right to privacy. Each case must be considered individually.

Moreover, despite the significance of the right to access information and, it is important that we criticize the effectiveness of the right in this instance. Essentially, an affected third party may only exercise the right if they are aware of the relevant genetic information. Where a patient refuses to consent to the disclosure, it seems unlikely that affected individuals would realise that there is information that is of interest to them. Accordingly, the right to access information cannot come into play if the affected party is unaware of the existence of relevant information, thus making this provision inadequate in terms of the disclosure of genetic information.

4.3.4 The Privileged Relationship

The notion of privilege also forms an important part of the doctor-patient relationship. As a medical professional, there is a duty to ensure the confidence of your patients, and thus, in South Africa, the doctor- patient relationship is a privileged one.[128] However, it is important to note that in a doctor-patient relationship there is relative privilege, rather than absolute privilege which is associated with an attorney-client relationship.[129] In absolute privilege, under no circumstances is an attorney allowed to disclose information provided by their client. Relative privilege, however, refers to the situation, in which, for example, a court orders a medical professional, despite their refusal, to reveal confidential health information.[130] This provides an opportunity for a medical practitioner to make a disclosure when it is necessary. Furthermore, qualified privilege will exist where the medical

[128] Singh (note 121 above; 131).
[129] Singh (note 121 above; 131).
[130] Singh (note 121 above; 131).

practitioner making the disclosure has a 'moral, legal or social duty to make the disclosure to a person who has a reciprocal interest in receiving the information'.[131]

4.3.5 Implications of Breaching Confidentiality

Where a healthcare practitioner does, in fact, breach the confidentiality of a patient, he or she must be aware that the non-consensual patient has the following legal remedies:[132]

(i) A civil action against the healthcare professional for the invasion of privacy or even defamation.

(ii) The patient may make a complaint to the relevant regulatory body (e.g. the HPCSA).

(iii) The healthcare processional may also receive disciplinary action where a patient lays a complaint with the healthcare professional's employer.

Liability for breaching confidentiality is an important aspect. Presumably, it is on the minds of practitioners wanting to make a disclosure. The absence of guidance on disclosure of genetic information will affect the decision of the medical practitioner as he or she would most likely opt on the side of caution, choosing not to make the disclosure. On the other end of the spectrum, however, medical practitioners may feel that they will be liable for a failure to warn affected related third parties. Accordingly, it may be stated that the HPCSA have attempted to address this issue by providing guidelines that state certain circumstances in which a medical professional may disclose confidential information.[133] Overall, however, medical practitioners should be free from thoughts of liability when making such a crucial decision and thus there is the need for legislation that tackles this issue adequately.

4.4 Conclusion

Confidentiality between a clinician and their patient is not as straightforward as it once was. The modern phenomenon of genetic testing has shown us this by bringing confidentiality and

[131] D J McQuoid-Mason 'Interference with the clinical independence of doctors in hospitals faced with a shortage of resources: What should doctors do?' (2014) 104 (11) *SAMJ* 742.
[132] Singh (note 121 above; 140).
[133] Health Professions Council of South Africa: Confidentiality: Protecting and Providing Information, Booklet 10 (2008) 2-3.

disclosure issues to the forefront of modern medicine. In South Africa, much emphasis has been placed on the right to privacy and thus confidentiality. This, however, does not mean that patients are completely autonomous individuals. As shown above, disclosure has become a major part of our law. Balancing these rights, however, are not always easy for medical practitioners. Nevertheless, the situation should not be ignored by continuously ensuring the confidentiality of a patient.

CHAPTER 5

South African Governance on Genetics

5.1 Introduction

The uncertainty revolving around genetic information has left medical practitioners and academics scouring current South African health legislation and governance documents, searching for guidance on how to deal with the disclosure of genetic information. Accordingly, an analysis of all current regulations put forward in South Africa on this topic is necessary. It is important to determine whether or not South Africa lacks an adequate genetic sciences framework. Consequently, this chapter examines the progress that South Africa has already made in establishing a framework that focuses on the confidentiality and disclosure of genetic information.

5.2 South Africa Legislation on Genetics

5.2.1 The Human Tissue Act

Although repealed in 2012,[134] the Human Tissue Act[135] was a significant piece of legislation that has laid down the foundation for current health legislation in South Africa. Unfortunately, it is apparent that such legislation did not govern genetics in any form. The Act does not mention the confidentiality of genetic information or any other type of health related information. Fortunately, with the introduction of regulations, the complete disregard for matters concerning genetic information is slowly coming to an end. Lastly, it must be mentioned that many of the provisions in the Human Tissue Act that were not included in the NHA have now been included in the Regulations of the NHA.

[134] National Health Act 61 of 2003.
[135] Human Tissue Act 65 of 1983.

5.2.2 The National Health Act: Regulations

Previously, the comprehensive provisions contained in the NHA failed to provide any guidance on genetics and the privacy of genetic information. This changed in The Health Professions Council of South Africa 2012 when regulations concerning the storage and flow of genetic information were passed.[136] Unfortunately, there are no explicit provisions concerning disclosure to affected related third parties. However, the Government Gazette is still useful as it states that information will not be disclosed to a relevant person without the consent of a patient.[137] In essence, information is treated as confidential and the patient has complete control over the use of the information. At this point, it is important to note the contradiction between South African regulations and guidelines.

Essentially, while regulations state that genetic information may only be disclosed where the patient has provided their consent, HPCSA guidelines[138] (discussed in chapter 4) seem to provide a more flexible approach. Although no direct reference is made to genetic testing, the HPCSA has outlined detailed guidance that allows clinicians to disclose personal information where a patient's interest is outweighed by the public interest, that is, affected related third parties.[139] Contradictions between ethics guidelines and regulations only add confusion to an already complex situation. This inconsistency reinforces the need for a cohesive legislation that governs genetic information. Accordingly, where there is a conflict between NHA regulations and HPCSA guidelines, it is important to bear in mind that the regulations will take precedent.

Overall, the regulations provided for in the gazette are restrictive and limiting. Unlike HPCSA guidelines, the regulations fail to consider that genetic information affects more than one individual. The recommendations made in this dissertation allows for a flexible approach that attempts to reach decisions that are in the best interests of all parties involved.

[136] GN 9699 of GG 35099, 2/03/2012; 37
[137] ibid 37.
[138]Health Professions Council of South Africa: Confidentiality: Protecting and Providing Information, Booklet 10 (2008).
[139] ibid 6.

5.3 South African Guidelines

5.3.1 The Health Professions Council of South Africa

The Patient's Rights Charter states that information regarding a patient's health status is to remain confidential, unless a patient consents to the disclosure of such information.[140] The Health Professions Council of South Africa (HPCSA) has also developed guidelines on confidentiality between a doctor and their patient.[141] The HPCSA (Rule 13 of the Ethical Rules) provides that a medical practitioner may only disclose confidential information in certain circumstances, which includes:[142]

- In terms of a Statutory provision,
- At the instruction of a court,
- In the public interest,
- With the express consent of the patient,
- Disclosures in the public interest would include but not be limited to situations where the patient or other persons would be prone to harm as a result of risk related contact.

In terms of public interest, the guidelines state that where a patient refuses to consent, it is possible to make a disclosure in the public interest where the benefits to an individual or to society outweigh the patient's interest in keeping the information confidential.[143] The guidelines specifically use an example of an endangered third party in the context of a HIV patient.[144] It is submitted that a related third party who is affected by the genetic information of a patient qualifies as an endangered third party, thus making disclosure, in this instance, acceptable. The HPCSA guidelines go on to state that in these cases, medical professionals

[140] 'Patient's Rights Charter' available at *http://www.justice.gov.za/VC/docs/policy/Patient%20Rights%20Charter.pdf*, accessed on 18 July 2015.
[141] 'About HPCSA' available at *http://www.hpcsa.co.za/About*, accessed on 2 August 2014. The HPCSA is an amalgamation of 12 Professional Boards in the health field that is committed to ensuring professionalism and ethical conduct of all registered healthcare professionals.
[142] Health Professions Council of South Africa: Confidentiality: Protecting and Providing Information, Booklet 10 (2008) 2-3.
[143] ibid 6.
[144] ibid 6.

must weigh the possible harm against the benefits likely to occur from the disclosure of information.[145]

Furthermore, the guidelines state that the disclosure of personal information may be justified 'where third parties are exposed to a risk so serious that it outweighs the patient's right to confidentiality'.[146] An example of such an instance would be gaining pertinent information through genetic testing. Although the ethical duty of confidentiality is important, the above exceptions clearly indicate that such a duty is not absolute. In certain circumstances, a healthcare practitioner may have a duty to disclose confidential information even if it is contrary to the wishes of the patient. In considering such an involuntary disclosure, the ethical duty to society must be weighed against the ethical duty to a patient. [147] The overall approach taken by the HPCSA is in line with the recommendations made in this thesis. This study, however, wishes to take this approach further by providing a more detailed procedure that assists healthcare practitioners in making informed decisions.

5.3.1.1 HPCSA: The Duty to Disclose and Endangered Third Parties

The General Ethical Guidelines[148] provided by the HPCSA discuss disclosure by healthcare professionals. However, before a discussion on disclosure can take place, it is crucial to outline other relevant provisions in the HPCSA guidelines, specifically section 17 which examines consent to screening and testing in terms of genetics.[149] The HPCSA provision states that in order for there to be an informed decision a healthcare practitioner must discuss certain factors with anyone considering genetic testing. The factor of most relevance is provided in 17.2.4 which states that a clinician must explain 'any significant medical, social or financial implications of screening or testing for the particular condition or predisposition'.[150]

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[145] ibid 6.
[146] ibid 9.
[147] P Carstens & D Pearmain *Foundational Principles of South African Medical Law* (2007) 948.

[148]Health Professions Council of South Africa: General Ethical Guidelines for the Health Care Professions, Booklet 1 (2008).
[149]Health Professions Council of South Africa: Seeking Patients' Informed Consent: The Ethical Considerations, Booklet 9 (2008).
[150] ibid 12.

The concept of social implications in this instance could clearly indicate the involvement of affected third parties. In terms of genetic testing, it can be said that these guidelines have made it necessary for medical practitioners to inform patients on the possibility that the results of their tests may affect others and that disclosure to these related third parties may be necessary. Due to the vagueness of the provision, however, it is difficult to say with certainty if, in fact, section 17 requires a medical practitioner to discuss with their patients the issue of disclosure to affected related third parties. Nevertheless, it may be argued that the term "social implications" is wide enough to encompass the possibility of disclosure to affected related third parties.

Furthermore, it is important to remember that the HPCSA has also provided guidance on the actual duty to disclose. The guidelines contained in the General Ethical Guidelines allow healthcare practitioners to make disclosures when there is a compelling reason to do so.[151] For such a reason to exist, there must be 'the likelihood of serious harm to an identifiable third party'.[152] Accordingly, in terms of these guidelines, a medical practitioner may disclose vital genetic information when an affected third party is vulnerable to serious harm. Consequently, it may be said that South Africa, to a certain extent, has recognised a duty to disclose. Although not as evident as some would prefer, these guidelines provide some guidance in terms of the duty to disclose in South Africa.

There is also the concept of endangered third parties which is particularly relevant in the context of HIV infected persons. The HPCSA have provided guidelines on partner disclosure which state that it is good clinical practice for practitioners to encourage their HIV positive patients to inform their partner of their HIV status.[153] In the event that a patient refuses to consent to the disclosure, deciding whether or not to make a disclosure is at the discretion of the healthcare professional.[154] In this instance, the healthcare professional looks at the circumstances of the case.

[151] Health Professions Council of South Africa: General Ethical Guidelines for the Health Care Professions, Booklet 1 (2008) 7.
[152] Health Professions Council of South Africa: General Ethical Guidelines for the Health Care Professions, Booklet 1 (2008) 7.
[153] Health Professions Council of South Africa: Ethical Guidelines for Good Practice With Regard To HIV, Booklet 11 (2008) 5.
[154] ibid 5.

Accordingly, it is important to note that HPCSA guidelines[155] have allowed for disclosure to endangered third parties in the context of genetic research.[156] This reinforces the idea that the concept of confidentiality in the field of genetics in South Africa is not absolute. Overall, the approach taken by the HPCSA is similar to the one that this dissertation advocates for in the context of disclosure of genetic information. If the duty to disclose can be recognised in the context of endangered third parties, it can similarly apply in the genetic diagnostics context in relation to affected related third parties.

5.3.2 The Southern African Society for Human Genetics

The Southern African Society for Human Genetics (SASHG) is an organization that cannot be ignored when discussing guidelines in genetics. However, despite publishing several documents that provide guidance in relation to the field of genetics, the interest and focus of SASHG has not extended as far as disclosures of genetic information. Nevertheless, SASHG does understand the importance of genetic counselling and has thus examined the scope of counselling in addition to providing the following comprehensive definition of the concept:[157]

> Genetic counselling is a process whereby individuals and their families are assisted in addressing their concerns relating to the presence of or risk of a genetic disorder in themselves or a family member.

5.3.3 Genetic Counselling

Genetic counselling began to gain popularity when the Human Genome Project started to understand the genetic causes of many disorders as well as possible preventative methods and treatments for these disorders.[158] This study realises the importance of counselling and thus advocates for a non-directive approach to counselling. During non-directive counselling, a

[155] Health Professions Council of South Africa: General Ethical Guidelines for Biotechnology Research, Booklet 7 (2008).
[156] ibid 21.
[157] 'What is Genetic Counselling?' available at *http://www.sashg.org/documents/what-is-genetic-counselling-4-March-09.pdf*, accessed on 15 December 2015.
[158] J Greenberg...et al 'Genetic Counselling in South Africa' in Dhavendra Kumar (ed) *Genomics and Health in the Developing World* (2012) 533.

patient should be offered accurate and unbiased information by a genetic counsellor who supports a patient throughout the decision-making process.[159]

The work of a genetic counsellor includes examining a family's medical history, understanding and interpreting information about genetic conditions, discussing inheritance patterns of relevant genetic disorders, calculating risks of individuals and providing testing, treatment and management options to families.[160] Genetic counselling helps a patient make adjustments to their lifestyle in order for them to cope with the genetic disorder. In other words, a genetic counsellor assists a patient and their families in making informed decisions, which includes decisions on future reproduction.[161]

As of May 2014, they were 44 genetic counsellors that were registered with the HPCSA, in which, 2 are student genetic counsellors, 6 are intern genetic counsellors and 10 are student intern genetic counsellors.[162] According to mid-year statistics for 2015, the South African population is estimated at 54.96 million people.[163] Essentially, despite the South African government and global health organisations expressing a need for genetic counsellors, the actual number of counsellors we do have is inadequate, especially considering the size of our population.[164] The United States, for example, has a much better counsellor to patient ratio in that there is about 1 genetic counsellor for every 123,000 individuals.[165]

From the above figures it is easy to conclude that a large population of South African's are not receiving the support that genetic counselling can provide. This dissertation submits that the field of genetic counselling remains small because of a failure to create awareness of genetic counsellors.

Accordingly, Genetic Counsellors South Africa (GC-SA), a subgroup of Southern African Society for Human Genetics, is invaluable to the field of genetics. Their main goals include continuing the development of genetic counsellors; providing accessible genetic counselling

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[159] World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003) 31.
[160] F Loubser...et al 'Support for Genetic Counselling services in the Western Cape' (25 February 2013) available at *https://gcnewssa.wordpress.com/*, accessed on 29 December 2014.
[161] ibid.
[162] 'Statistics' (06 May 2014) available at *http://www.hpcsa.co.za/Publications/Statistics*, accessed on 29 December 2014.
[163] 'Mid-year Population Estimates 2015' available at *https://www.statssa.gov.za/publications/P0302/P03022015.pdf*, accessed on 1 February2016.
[164] R Macleod 'It's in the genes' available at *http://www.enca.com/south-africa/wha1t-sa-jolie-and-albino-share-need-genetic-counselling*, accessed on 28 December 2014.
[165] E Hammers 'Innovations In Service Delivery In the Age of Genomics: Workshop Summary' (2009) available at *http://www.ncbi.nlm.nih.gov/books/NBK26401/pdf/Bookshelf_NBK26401.pdf*, accessed on 1 February 2016.

to all communities in South Africa and advocating for patients and their support groups.[166] Groups such as GC-SA must become essential if South Africa is ever going to embrace the concept of genetic counselling.

Overall, this study recommends that counselling should be provided to a patient before he or she undergoes the genetic testing process. The idea behind such counselling should be to provide a level of comfort and knowledge to a patient who may not be adequately informed on the process. During this time, it would also be productive to prepare the individual for the possibility of a disclosure to affected related third parties.

5.3.3.1 Genetic Counselling: Ethical Guidelines

At this juncture, it is important to examine domestic ethics guidelines on genetic counselling that have a different stance to the regulations of the NHA. Two examples of such guidelines would be the recommendations produced by the Medical Research Council[167] (MRC) of South Africa and the guidelines produced by HPCSA.[168] These guidelines were developed for the purpose of obtaining a patient's informed consent for genetic testing.

Firstly, section 3.3.3 of MRC guidelines recommend that certain information be given to any patient undergoing genetic testing. [169] This information includes the seriousness of the condition, treatment options for the condition, reliability of results, probability of developing a genetic condition, and the implications for relatives.[170] The guidelines go on to state certain ethical principles that should be adhered to during the counselling process.[171] One such principle states that health professionals should inform their patients that the ethical duty to tell affected relatives of genetic risks lies with them.[172] Accordingly, it is clear that these guidelines focus on the individual at the expense of affected related third parties.

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[166] 'Making sense of Genetics' available at *http://www.geneticcounselling.co.za/gcsa.php*, accessed on 29 December 2014.
[167] Medical Research Council: Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research (2004).
[168] Health Professions Council of South Africa: General Ethical Guidelines for Biotechnology Research, Booklet 7 (2008).
[169] Medical Research Council: Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research (2004).
[170] ibid.
[171] ibid.
[172] ibid.

Another set of guidelines specific to the discipline of genetics are the HPCSA guidelines.[173] Although these guidelines are used for genetic testing in a research context, they may still be helpful in the clinical situation discussed in this study. In terms of these provisions, there is certain information that must be disclosed to a research participant in order for the participant to make an informed decision concerning the collection of genetic material and information.[174] This includes all the details of the conditions that are discovered during the research as well as information that may be relevant to future offspring or relatives.[175] Furthermore, the participant must be informed that where there is research information relevant to the health of relatives, no disclosure will be made to these affected relatives without the consent of the participant.[176]

Even though the HPCSA guidelines are in the context of research, it is important to remember that the core concepts of this study is confidentiality and disclosure, two problematic terms that have been identified and examined in these guidelines. Accordingly, we can learn how South African guidelines have dealt with these notions in the context of genetics. Consequently, it is apparent that these guidelines, similar to those developed by the MRC, prioritise confidentiality as the patient has the final decision of whether or not to disclose. Although placing a patient first is admirable, this approach can be rather simplistic in the context of a complicated situation. In contrast to the above guidelines, this study argues that medical professionals should inform a patient during counselling that there is, in fact, a chance that the disclosure will be made to affected related third parties without their consent.

5.3.4 Genetics Policy Guidelines in South Africa

In 2001, the Department of Health published Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities.[177] These guidelines state that genetic information must remain confidential, except where there is a risk of serious harm to relatives and the information could be used to prevent such harm.[178] The mention of

[173] Health Professions Council of South Africa: General Ethical Guidelines for Biotechnology Research, Booklet 7 (2008).
[174] ibid 36.
[175] ibid 36.
[176] ibid 37.
[177] Human Genetics Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (2001).
[178] ibid 50.

confidentiality is commendable, but there is still much to be desired in terms of disclosure. Firstly, there is no attempt to define "genetic information". In fact, South African guidelines in general have failed to provide a comprehensive definition of genetic information. In the absence of such a definition being spelled out in local guidelines, we may look to the definition provided by GINA for guidance. (Discussed in chapter 3).

Furthermore, there are no guidelines to indicate what constitutes "serious harm". Accordingly, there is also no mention of counselling and disclosure-related procedures that healthcare professionals should adhere to. Another shortfall is that these guidelines are not legally binding. Overall, this dissertation submits that this document is inadequate as it fails to provide sufficient guidance to healthcare professionals. Nevertheless, the one positive that can may be taken away from these guidelines is that in the case of genetic information, the right to confidentiality in South Africa is not absolute.

Another question that must be asked is how effective have these guidelines been in South Africa. Considering that this is the first comprehensive document that provides guidance on human genetics in South Africa, should there not have been the implementation of recommendations? For instance, the guidelines have recommended that positions for genetic counsellors should be made available throughout the country.[179] Even though the document states that 320 genetic counsellors are ideally required,[180] at present, there are only 26 fully qualified genetic counsellors that are registered with the HPCSA.[181] The failure to achieve this goal adds to the doubt that this document is, in fact, satisfactory.

5.4 Conclusion

From the above examination of guidance documents, it is apparent that even though there are certain regulations in South Africa that may be applied, there are no explicit regulations that sufficiently manage the confidentiality and disclosure of genetic information. This dissertation submits that due to the absence of adequate guidance on disclosure of genetic information, medical practitioners are reluctant to breach confidentiality. As a result of this reluctance, third parties may experience harm that could have been prevented, or at the very

[179] ibid 28.
[180] ibid 3.
[181] 'Statistics' (06 May 2014) available at *http://www.hpcsa.co.za/Publications/Statistics*, accessed on 4 January 2015.

least mitigated. Consequently, there is a need for comprehensive legislation that will be effective in practice. Accordingly, this dissertation provides recommendations (discussed in chapter 8) that may provide a starting point to any future legislation.

CHAPTER 6

Biomedical Principles of Ethics and Ethics Theories

6.1 Introduction

Theories in any field of study always seem to guide us to one particular perspective. The use of multiple theories, however, allow us to open our mind to various possibilities, with the understanding that there is not always one correct answer. The development and application of the principles of biomedical ethics and ethical theories have assisted healthcare practitioners in deciding what the best course of action is when faced with an ethical dilemma. This chapter will focus on the ethical implications involved in genetic testing and the disclosure of genetic information. It is submitted that these ethical considerations must be taken into account in order to effectively regulate the involuntary disclosure of genetic information.

6.2 The History and Development of Ethical Principles and Theories

The Belmont Report as well as philosophers Thomas Beauchamp and James Childress have influenced and accordingly shaped the way, in which, ethical principles are defined in the contemporary medical world.[182] After the Nuremberg trials, persons across the globe became aware of biomedical experiments that were conducted on human subjects in concentration camps, that is, the atrocities that were committed by doctors and scientists. The findings of the Nuremburg trials seemed to suggest that doctors needed a certain standard to aspire to and thus the Nuremberg Code was drafted. The ethical rules and principles contained in the code were insufficient to guide doctors in complex situations. As a result, in 1979, the National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research

[182] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 35.

was charged with the task of developing basic ethical principles that should guide biomedical and behavioral research that involve human subjects.[183]

This commission produced what has since become known as the Belmont Report. The Belmont Report stated that there are three basic principles that are particularly relevant to the ethics involved in the research of human subjects: the principles of respect for persons, beneficence and justice.[184] On the other hand, however, writers Beauchamp and Childress proposed that there are four principles that are relevant to the ethics involving human subjects: autonomy, beneficence, non-maleficence and justice.[185]

6.3 Ethical Theories

Ethical theories act as a theoretical framework in which individuals, and in this instance healthcare professionals, use when faced with an ethical dilemma. An ethical theory provides a moral standard that may be used to assess what is morally right and wrong regarding the actions of a healthcare professional. There are several different competing theories that can be used by a healthcare practitioner to justify an action as ethical. This dissertation, however, will be focusing on three theories in particular, namely, utilitarianism, virtue ethics and narrative ethics.[186] These particular theories were chosen in order to provide different perspectives as to what a medical professional ought to do and ought not to do. The application of these theories will demonstrate the benefits and limits of each approach taken by a clinician. It is hoped, that by using these specific theories, a clinician will find a balanced approach when facing a possible disclosure case.

[183] The Belmont Report: Ethical Principles and Guidelines for the Protection of Human Subjects of Research (1979).
[184] The Belmont Report: Ethical Principles and Guidelines for the Protection of Human Subjects of Research (1979).
[185] T L Beauchamp & J F Childress *Principles of Biomedical Ethics* 5 ed (2001).
[186] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 37.

6.3.1 Utilitarianism

Utilitarianism is said to be a form of the Consequential theories that focuses on the consequences of a decision.[187] In 1776, philosopher Jeremy Bentham developed a fundamental maxim that would become synonymous with the notion of utilitarianism: 'it is the greatest happiness of the greatest number that is the measure of right and wrong'.[188] In terms of Bentham's principle of utilitarianism, one must evaluate actions based on their consequences. From this, we can infer that Bentham's theory of utilitarianism means that one should perform the action that will bring about the best overall consequence by providing the greatest amount of happiness for the greatest number of people. [189]

Actions can be explained simplistically by stating that actions thought to produce good consequences are good actions, while actions thought to produce bad consequences are bad actions. In terms of a utilitarian approach, when faced with an ethical dilemma, the following must be done:[190]

(i) An examination of the possible and likely short- and long-term consequences that are as a result of the decision/action.

(ii) Consequences must be compared in order to determine how many people will be helped and to what degree, and how many harmed and to what degree.

According to utilitarianism, a clinician's decision to make the disclosure should be determined by the action that would provide the greatest amount of happiness. There are two options in this instance: keeping the confidentiality of their patient or making a disclosure to ensure the well-being of affected third parties. It is essential that we examine genetic information in a utilitarianism context as the patient is not the only individual that is affected by the results. In the instance where genetic testing reveals information that a patient's family are also at risk, a doctor's actions are of utmost importance.

The confidentiality of a doctor-patient relationship is always at the forefront of a medical practitioner's decision making process. However, where there are affected related third parties, medical practitioners must also ensure that there is thought for the well-being of these

[187] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 39.
[188] J H Burns 'Happiness and Utility: Jeremy Bentham's Equation' (2005) 17 (1) *Cambridge Journals* 46.
[189] 'Jeremy Bentham' available at *http://www.iep.utm.edu/bentham/*, accessed on 28 August 2014.
[190] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 39.

at-risk relatives. A medical practitioner must consider the number of affected relatives that they will be able to help. An early warning to the presence of a genetic condition can afford at-risk third parties an opportunity to seek out life saving treatment. Disclosure from a clinician will provide these at-risk third parties with time, giving them an opportunity to deal with the emotional and psychological strain associated with genetic conditions. It will also help them make informed decisions on the management and treatment of the condition.

In certain circumstances, a medical practitioner has an opportunity to help an affected third party take preventative measures ensuring that the at-risk relative does not develop the genetic condition. It seems senseless for a medical practitioner to have access to potential life saving information, yet not share it due to the constraints of a doctor-patient relationship. To take the utilitarian perspective further, it is not just existing family members that may benefit from the information as genetic information may also reveal information regarding future offspring. Individuals would have knowledge that if they were to have children, their child might be born with or eventually develop a genetic disorder. In this instance, disclosure is the responsible decision as it prevents parents from undergoing the financial burden and emotional stress associated with having a child who has a genetic condition.

Accordingly, a medical practitioner's decision or action to make the disclosure would cause the greatest amount of benefit for the greatest number of people and thus their action would be justified in terms of utilitarianism. At the same time, however, the utilitarianism approach in this instance may be criticized. Even though the involuntary disclosure of information will be beneficial to affected family members, when a doctor follows the utilitarianism theory, there is bound to be harm caused to others.[191] In this instance, it is the patient that is harmed.

The healthcare professional must keep in mind that once he or she breaches confidentiality, the patient, in all likelihood, will refuse to continue the doctor-patient relationship. Also, in the event that the patient seeks out other available healthcare practitioners, it is probable that the patient would continue their distrust for medical professionals. This distrust does not allow for an effective doctor-patient relationship, thus compromising patient care and treatment.

Another criticism of utilitarianism is that the prediction of consequence are not an exact science, and this is certainly true in terms of genetic testing. As stated earlier, genetic testing

[191] ibid 39.

cannot always predict with certainty that an individual will, in fact, develop the genetic condition. Here, a medical practitioner is basing their decision to take a certain course of action in anticipation of a consequence that may never materialize,[192] that is, the affected third parties may never develop the condition. In this instance, the harm caused to the doctor-patient relationship would have been futile.

6.3.2 Virtue Ethics

Inspiration for virtue ethics has been taken from the declarations of Greek philosopher, Aristotle who stated that a virtuous individual is someone who is in possession of ideal character traits that are derived from natural internal tendencies.[193] The main element that characterizes virtue ethics is an emphasis on the moral character of an agent and in this instance the healthcare practitioner. There has been much discussion as to how and why a healthcare professional should come to a decision when making a choice of whether or not to disclose information, but there has not been any explicit discussion on the healthcare practitioner himself.

In terms of virtue ethics, there is an assumption that a healthcare practitioner will only make morally appropriate decisions if he or she is a morally sensitive and skilled person. Virtue ethics focuses on the education, background, experience and development of the specific healthcare professional making the decision. A healthcare practitioner will make a decision for their patient based on their past experience in the belief that the decision they are making is the right one. Virtue ethicists believe that in order for a morally virtuous person to make a ethically correct decision, they require a good motive and the skills and practice that come with experience.[194]

In South Africa, it appears that a medical practitioner is the only person who is required to make the decision of whether or not to disclose. It is apparent that there are no safeguards in place to ensure an objective and thorough decision making process. In terms of virtue ethics, the healthcare practitioner will use their past experience to come to a decision of whether or

[192] ibid 39.
[193] 'Virtue Ethics' available at *http://www.iep.utm.edu/virtue/*, accessed on 5 September 2014.
[194] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 38.

not they should make the disclosure. It is disconcerting that practitioners are allowed to make a completely subjective decision with no thought of an objective standard.

A known criticism of virtue ethics is that a good motive combined with knowledge and experience does not guarantee that a good decision will be made in each instance.[195] Although experts in the medical field, it is apparent that most medical professionals dealing with genetic testing do not have the required expertise that is needed to make such a complex decision. Accordingly, it is probable that a medical practitioner's lack of experience in this area will make it difficult for him or her to come to a virtuous decision in every case.

Consequently, a major question that this study asks is whether such a decision should be made at the sole discretion of the medical practitioner involved. In answer to this question, it is important to emphasize that a healthcare practitioner's final decision may impact on more than just one person. As a result, this study recommends that a medical practitioner cannot be the only professional involved in making the decision. The decision to disclose must be done in consultation with others and with an objective standard in mind.

6.3.3 Narrative Ethics

The telling of a story has always been helpful in fleshing out a dilemma in medical ethics. Accordingly, by shining a light on the human perspective, we gain insight into a person's life and the complicated relationships they are apart off.[196] Narrative ethics is essentially a patient centered approach that requires time and resources and is thus of particular relevance in terms of genetic testing. Narrative ethics may only be effective in private practices and considering that genetic testing in South Africa generally occurs in these practices, it is apparent that there is the need to examine and involve a narrative approach.

Narrative ethics emphasizes a storytelling approach in which a patient's illness is the telling of a story that requires compassion and empathy. Allowing a patient an opportunity to outline their narrative provides the medical practitioner with a greater understanding of specific details of the case that is useful in the diagnostic process, details that the medical practitioner would not have known if not for the narrative of the patient. In order to assist the patient and

[195] ibid 39.
[196] A H Jones 'Narrative in medical ethics' (1999) 318 (7178) *BMJ* 253.

their family in coming to an appropriate course of action, a medical practitioner must understand, appreciate and be interested in the narrative of the patient.

When a clinician decides to disclose private genetic information to affected parties, they do so in the belief that it is in the best interests of the third party. In other words, the medical practitioner's approach seems to be one sided, focused on the well-being of affected third parties. While such actions by medical practitioners may be considered as admirable, it cannot continue. A more balanced decision-making process must take place which requires the use of a narrative ethics approach.

It is my submission that patients who refuse to consent to the disclosure should be given the opportunity to tell their story. Healthcare practitioners should allow patients to explain their decision as to why they do not wish for affected third parties to know that they are at risk. Since this dissertation is not advocating involuntary disclosure in every case, the reason as to why a patient refuses to consent, should play a pivotal role in the ultimate decision made by the healthcare professional.

For example, the instance where a patient refuses to consent because he or she does not want family members to know of their condition can be deemed to be an insufficient reason. In this situation, a patient would claim that their genetic condition is private, but at the same time the medical practitioner must be aware that their patient's right to privacy may be limited. However, a medical practitioner may experience problems where a patient's refusal to consent is more virtuous, based on the well-being of related affected third parties. In this instance, a more thorough decision-making process is needed.

6.3.3.1 Reasons to Refuse Consent

A patient's reason for refusing consent is another concept at the core of this dissertation. Accordingly, it is important to discuss certain scenarios that might occur in real life which have been brought to the attention of a medical practitioner by the patient through a narrative ethics approach. The following are examples of reasons a patient may have to refuse consent:

(i) The patient is of the belief that any disclosure would be unproductive as the condition is extremely serious or at such an advanced stage that medical intervention would not be able to

delay or alleviate the condition.[197] For example, medical interventions and treatment options for Huntington disease are minimal and at present, are only at the beginning stages.[198]

(ii) The patient is of the belief that if the related third party knew that he or she was going to develop the genetic condition, it would affect the third party's chances of obtaining life or health insurance at an affordable rate.[199]

(iii) A patient may believe that if the related third party were to have knowledge of the genetic condition, then he or she would have a negative outlook on life, affecting their overall quality of life. In this instance, an individual would go through severe emotional and psychological trauma upon discovering that they have a life-altering or fatal genetic condition. At the same time, however, it must be noted that such a reason is not sufficiently adequate. When a person experiences sudden traumatic news, it is only natural to go through a period of adjustment. However, where a third party will be particularly sensitive to such news (e.g. third party is at an advanced age), a medical practitioner must take the particular circumstances under consideration.

(iv) The patient believes that the related affected third party will not be able to afford medical treatment for the genetic condition. Disclosure in this instance would seem to be fruitless.

(v) The patient believes that a third party, as a result of the disclosure, will limit their quality of life by choosing not to get married or have children. Such restrictive decision making on the part of the third party is bound to lead that third party to a life based on frustration and isolation.[200]

(vi) A patient may also be concerned of becoming stigmatized by relatives who have knowledge of their condition. Essentially, a patient may be worried that they would be treated differently. In fact, a patient may even believe that family members may use such information for malicious purposes.[201]

(vii) A patient could also be estranged from their family as a result of a serious falling out or even past abuse. In this instance, the patient would fear being traced through the disclosure.

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[197] D C Wertz 'British Medical Association' (1998) 65 (6) *Human Genetics: Choice And Responsibility* 13.
[198] K Offit...et al 'The "Duty to Warn" a Patient's Family Members About Hereditary Disease Risk' (2004) 292 (12) *JAMA* 1470.
[199] D C Wertz 'British Medical Association' (1998) 65 (6) *Human Genetics: Choice And Responsibility* 13.
[200] K G Fulda & K Lykens 'Ethical issues in predictive genetic testing: a public health perspective' (2006) 32 (3) *Journal of Medical Ethics* 145.
[201] S Dheensa & A Fenwick & A Lucassen 'Is this knowledge mine and nobody else's? I don't feel that.' Patient views about consent, confidentiality and information-sharing in genetic medicine' (2015) 10 (1136) *BMJ* 3.

6.4 Biomedical Principles

6.4.1 Autonomy

As we can see from the above, there may be instances where a patient has a valid reason for not consenting to the disclosure. The patient's right to self-determination should also be evaluated in a clinician's decision making process and thus the ethical principle of autonomy must be examined. The literal meaning of autonomy is self-rule.[202] In terms of this principle, a patient must be given the opportunity to make the ultimate decision regarding their treatment after a healthcare practitioner has provided him or her with all the relevant information.[203] It is important for healthcare practitioners to respect autonomous individuals by giving weight to their considered opinions and choices, while abstaining from preventing or influencing their actions, unless these actions are detrimental to others. If a healthcare practitioner were to reject an autonomous individual's considered judgement, it would indicate a clear lack of respect for such person.[204]

Even though the patient may have the right to self-determination and thus the right to refuse the disclosure, it is important to remember that a third party may also feel as if they have a right to know of the existence of any genetic condition. The majority of related affected third parties would undoubtedly want to know if there is possibility that they may develop a genetic condition as such information would allow these individuals to make responsible and informed choices.[205]

6.4.2 Justice

The principle of justice deals with fairness in distribution. In terms of justice, if a person is denied a benefit they are entitled to without good reason, or if a burden is unduly imposed on a person, it is said that an injustice has occurred.[206] Parker and Lucassen state that the principle of justice underpins their notion of the joint account model. According to this

[202] K Moodley *Medical Ethics, Law and Human Rights: a South African Perspective* (2011) 42.
[203] ibid 42.
[204] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 36.
[205] K G Fulda & K Lykens 'Ethical issues in predictive genetic testing: a public health perspective' (2006) 32 *Journal of Medical Ethics* 145
[206] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 37.

model, 'genetic information is shared by more than just one person'[207] and should therefore be considered as familial in nature.[208] Parker and Lucassen have reasoned that there is no rationale as to why only one person in that family should be able to receive the benefit and accordingly exclude other relatives from the same benefit.[209] In this instance, the information will not be shared if the patient has a good reason to refuse consent.[210] Writers have gone so far as to suggest that genetic information may be regarded as collective or shared information.[211] Although this study advocates for the disclosure of genetic information to affected relatives, it is important to remember that disclosure can become a slippery slope for any healthcare practitioner. In this regard, clinicians should always be wary as it may be exceptionally easy to justify a case for disclosure of genetic information in every instance. A clinician should never forget the importance of confidentiality.

6.4.3 Non-Maleficence and Beneficence

At its very essence, non-maleficence prohibits healthcare practitioners from causing harm onto others.[212] Beauchamp and Childress have provided certain rules in terms of the principle of non-maleficience. For example, a healthcare practitioner should refrain from inflicting pain and suffering onto a patient.[213] It is interesting to note that harm is not restricted to physical harm alone as medical practitioners should also refrain from offending a patient and impeding their good quality of life.[214] From this we can infer that Beauchamp and Childress have included mental and emotional harm in their examination of non-maleficence.

Accordingly, it may be argued that the principle of non-maleficence is violated when a medical practitioner decides to make a disclosure against the wishes of their patient. In such a scenario, the decision to disclose could cause serious upheaval in a patient's life. Putting aside the harm done to the patient's emotional well-being, there is also the harm done to the doctor-patient relationship. As discussed earlier, under the theory of utilitarianism, the medical

[207] M Parker & A M Lucassen 'Genetic Information: a joint account?' (2004) 329 (7458) *BMJ* 166.
[208] ibid 166.
[209] ibid 166.
[210] ibid 166.
[211] A Davey & A Newson & P O'Leary 'Communication of Genetic Information within Families: The Case for Familial Comity' (2006) 3 (3) *Springer* 161.
[212] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 36.
[213] Moodley (note 202 above; 63).
[214] Moodley (note 202 above; 63).

practitioner's actions will irreversibly damage the doctor-patient relationship, negatively impacting the treatment of the patient. From the above, it is ostensible that any disclosure on the part of a clinician would be a contravention of the principle of non-maleficence.

Furthermore, there is the concept of beneficence which in essence is understood as acts of charity that go beyond a person's obligation.[215] This terms encompasses the moral essence of obligations healthcare practitioners owe to their patients.[216] Accordingly, beneficence is at the very core of the duty to warn. Essentially, even though a medical practitioner may not by law be required to make a disclosure to an affected related third party, he or she may feel they have a moral duty to that patient and thus the clinician performs an act of kindness by disclosing potentially life-saving genetic information.

6.5 Organizational Ethics

A medical practitioner should make every effort to ensure that access to the information is kept to a minimum by ensuring that those who are made aware of the information uphold its confidentiality. This is especially important in healthcare institutions and practices, where there is likely to be a breach of confidentiality, as certain persons (e.g. laboratory technicians) have access to a patient's confidential information.[217]

This is one of the reasons why this study suggests that health establishments develop and implement organizational ethics policies. Organizational ethics are concerned with ethical issues that healthcare establishments are faced with. In terms of organizational ethics, managers and governors of health facilities attempt to identify areas where values conflict in order to find solutions to these problems. When attempting to find a resolution, an ethics approach is used in which decision-making is based on certain values. Also, ethics policies would allow for a different understanding than the legal definition of confidentiality.[218]

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[215] J A Singh 'Ethical Decision-making' in MA Dada and DJ Mcquoid-Mason (eds.) *Introduction to Medico-Legal Practice* (2001) 36.
[216] ibid 36.
[217] Dhai & McQuoid-Mason (note 87 above; 87).
[218] J L Gibson...et al Organisational ethics. In Singer PA and Viens A (eds.) *The Cambridge Textbook of Bioethics* (2008) 243-244.

Whereas the law tells medical practitioners what they must do, ethics provides guidance by assisting a practitioner in determining what they should do. Where there is a conflict of interest, the ethical decision making process will assist in establishing the ethical legitimacy of decisions, demonstrating how decisions ought to be made. Overall, ethical policies and decision-making frameworks can be effective mechanisms for guiding ethical conduct in circumstances where interests tend to conflict.[219]

Despite its effectiveness, it is important to remember that organizational ethics polices are still inferior to the law as well as professional guidelines. When ethics policies are implemented in healthcare facilities, it is highly improbable that they would be found legally binding. A violation of such a policy is likely to result in the individual being disciplined internally, instead of being liable in terms of the law. Accordingly, the ideal solution would be the development of legislation that will ensure repercussions for ethically ill-advised conduct in the context of involuntarily disclosure.

6.6 Conclusion

From the above discussion of ethics, it is apparent that there is still no clear cut answer as to how medical practitioners should conduct themselves during and after the genetic testing process. However, the discussion of different theories and principles have provided us with various perspectives that will assist us in developing adequate regulations for medical practitioners. Essentially, in practice, after an application of ethical principles and theories to the relevant circumstances, a medical practitioner must balance the different theories and principles in order to come to the best possible solution for all parties. An understanding of bioethics has helped us to comprehend that healthcare professionals actions cannot be one methodical standard, but rather it must be based on a case by case basis.

[219] ibid 244-247.

CHAPTER 7

Foreign and International Guidance on Confidentiality and Disclosure

7.1 Introduction

Medical practitioners throughout the world have been facing issues surrounding patient confidentiality and at-risk relatives. The complexities involved in the disclosure of genetic information have forced numerous countries into taking action in an attempt to regulate these complexities. This chapter traces the development of governance documents that have governed the disclosure of genetic information. It goes on to analyze and discuss current governance documents of certain foreign jurisdictions and international organizations that have been effective in governing this matter. Lastly, this chapter explores the scope of the duty to warn in terms of foreign law.

7.2 The Development of Governance Documents

In 1983, the United State's President's Commission for the Study of Ethical Problems in Medicine and Biomedical and Behavioral Research correctly anticipated that genetic technology would be greatly magnified in future decades.[220] The commission concluded that confidentiality may be breached and genetic information may be disclosed to family members if the following conditions are met:[221]

(i) A reasonable effort to obtain consent to the disclosure has failed;

(ii) It is highly probable that the harm will materialize if the information is not disclosed. Also, the information that is withheld could be used to prevent the harm;

(iii) The harm that the third party would suffer would constitute serious harm; and

[220] 'National Information Resources on Ethics and Human Genetics' (February 1983) available at *http://kie.georgetown.edu/nrcbl/documents/pcemr/geneticscreening.pdf*, accessed on 25 August 2014.
[221] ibid.

(iv) There are adequate precautions taken to ensure that only the essential genetic information is disclosed.

In 1994, the American Institute of Medicine Committee compiled a report that assessed genetic risks. The committee suggested an approach called the "Miranda Warning". In terms of the Miranda warning, a patient would be informed in advance of the circumstances that would warrant the disclosure of genetic information to at-risk family members. Under these circumstances, the patient's consent would not be needed.[222] This approach is commendable in that it attempts to preserve the doctor-patient relationship.[223] On the other hand, however, a patient might be reluctant to undergo the testing in the first place, for fear that family members might become aware of their results.

7.3 International Governance Documents

7.3.1 World Health Organization

The only form of guidance that the World Health Organization (WHO) has provided on this matter takes the form of a document titled Review of Ethical Issues in Medical Genetics.[224] In terms of these guidelines, a clinician may inform affected persons if there is a likelihood of serious harm, despite the patient's wishes. Provided, however, that the following four conditions are met:[225]

i) Every effort that has been made to persuade the patient to make the disclosure has failed.

ii) If the relevant information is not disclosed, there is a high risk of harm to family members (including potential children). Also, there must be proof that the information could be used to prevent future harm.

iii) The harm to affected persons is serious.

iv) Only genetic information that is directly relevant to the affected party should be disclosed. Information that pertains to the individual must be kept confidential.

[222] B M Knoppers...et al 'Professional Disclosure of Familial Genetic Information (1998) 62 *Genet* 475-476.
[223] ibid 475-476.
[224] World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003).
[225] World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003) 51.

The document makes an impact by stating that if these conditions are met, clinicians should not be held liable if a disclosure is made.[226] Also, the fact that these guidelines have proposed pre- and post-test genetic counselling is an aspect that must be commended.[227] Overall, it is apparent that WHO has taken a concise and straightforward approach. It appears that WHO have produced guidelines that could be effective in balancing the rights of a patient and those of affected relatives.

7.3.2 UNESCO International Declaration on Human Genetic Data

The United Nations Educational, Scientific and Cultural Organization International Declaration[228] makes reference to the term genetic data instead of genetic information.[229] The Declaration affords genetic data a special status for several reasons. These reasons include the fact that genetic data can predict the genetic predispositions of individuals and that it may have a noticeable impact on family members, including potential children.[230] Accordingly, we can infer that genetic data contains similar information as genetic information and thus we may examine the declaration in context of this dissertation.

Article 14 of the Declaration deals with privacy and confidentiality. It essentially encourages countries to protect the confidentiality of genetic data of an individual and their family.[231] More importantly, however, Article 14 goes on to state that genetic data should not be disclosed to third parties, without the consent of a patient.[232] The declaration clearly supports a patient's right to confidentiality, failing to take into consideration the well-being of affected related third parties. Although admirable, such a straight forward approach is not practical when trying to resolve a conflict that affects several individuals.

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[226] ibid 51.
[227] ibid 72-75.
[228] International Declaration on Human Genetic Data (2003).
[229] 'Introducing UNESCO' available at *http://en.unesco.org/about-us/introducing-unesco*, accessed on 17 September 2014. The United Nations Educational, Scientific and Cultural Organization (UNESCO) is a special agency of the United Nations. One of the main purpose of UNESCO is to anticipate changes in the environment and accordingly create holistic policies that address global problems.
[230] International Declaration on Human Genetic Data (2003).
[231] International Declaration on Human Genetic Data (2003) Article 14(a).
[232] International Declaration on Human Genetic Data (2003) Article 14(b).

7.3.3 HUGO Ethics Committee

The Human Genome Organisation (HUGO) Ethics Committee, in a statement on DNA Sampling: Control and Access, has stated that 'shared biological risks [of family members] create special interests and moral obligations with respect to access, storage and destruction that may occasionally outweigh individual wishes'.[233] Although HUGO have not released express guidelines on the issue, from the above statement it is clear that they view disclosure as an acceptable course of action in certain circumstances.

7.3.4 World Medical Association

The WMA has made their stance on the topic known in the form of a statement on Genetics and Medicine.[234] According to paragraph 12 of this statement, a clinician may make a disclosure to affected third parties, without the consent of a patient, where not disclosing the genetic information would result in 'direct and imminent threat to the life or health'[235] of a third party. The WMA goes on to state that the clinician should generally discuss the situation with the patient first.[236]

Furthermore, unlike other guidelines, the WMA takes disclosure further by stating that it is preferable for a clinician, where possible, to consult an ethics committee before disclosing results to affected third parties.[237] This is an interesting provision as it doesn't allow for autonomous decision-making on the part of the physician. In fact, by consulting an ethics committee the physician, in all probability, is receiving the most well-balanced opinion in the matter. Despite the good intentions of this provision, it is crucial to criticize the practicalities of it. Continuously finding and accessing an ethics committee does not seem like a feasible option. However, what we can learn from these guidelines is that clinicians do not need to make the decision of whether or not to disclose by themselves. There is no reason as to why there cannot be an accessible an informed committee at a hospital or practice.

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[233] HUGO Ethics Committee 'Statement on DNA Sampling: Control and Access' (February 1998) available at *http://www.hugo-international.org/img/dna_1998.pdf* , accessed on 2 November 2015.
[234] World Medical Association 'WMA Statement on Genetics and Medicine' (October 2009) available at *http://www.wma.net/en/30publications/10policies/g11/*, accessed on 2 November 2015.
[235] ibid.
[236] ibid.
[237] ibid.

7.4 Regional Governance Documents

7.4.1 The European Convention on Human Rights and Biomedicine

Unfortunately, the European Convention on Human Rights and Biomedicine does not explicitly regulate the involuntary disclosure of genetic information. [238] Article 10 of the Convention on Human Rights and Biomedicine,[239] however, discusses private life and the right to information. Article 10 (1) of the Convention states that 'Everyone has the right to respect for private life in relation to information about his or her health'.[240] From this, we can deduce that the Convention advocates a patient's right to privacy. Furthermore, however, article 26 of the Convention states that rights may be restricted where it is necessary in the interest of public safety or for the protection of public health.[241] Accordingly, the Convention does not provide an absolute right of privacy for a patient. It may be argued that this restriction may allow the disclosure of genetic information to affected parties.

7.5 Professional Ethics Guidance Documents

7.5.1 United Kingdom

7.5.1.1 General Medical Council

The UK General Medical Council[242] has produced guidance on confidentiality for doctors who are registered with them. These guidelines state that when a patient refuses to consent to the disclosure, a doctor must balance their duty to take care of their patient with their duty to protect another person from serious harm. It goes on to state that if possible, a patient's identity should not be disclosed when informing others of the risk they are facing.[243]

[238] Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes (2008) Article 18. Article 18 in the Additional Protocol to the Convention concerning Genetic Testing for Health Purposes simply states that where an individual undergoes genetic testing that produces information relevant to the health of a relative, the individual tested should be informed of the results.
[239] Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (1997).
[240] Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (1997) Article 10(2).
[241] Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine (1997) Article 26.
[242] General Medical Council: Confidentiality (2009).
[243] ibid 27.

Although, these guidelines allow for the discretion of a doctor, it fails to lay out specific guidelines that will assist them through the decision-making process.

7.5.1.2 British Medical Association: Confidentiality and Disclosure of Health Information

In terms of British Medical Association (BMA) guidelines, healthcare professionals should play a vital role in advising patients on the implications of genetic information that affect family members. The guidelines also state that clinicians should encourage patients to share information with affected persons.[244] The guidelines go on to state that where a patient refuses to consent to the disclosure, a doctor should consider certain factors, such as:[245]

(i) The seriousness of the genetic disorder;

(ii) If informed, can affected relatives take appropriate action to protect themselves;

(iii) The patient's reason for refusing to consent to the disclosure; and

(iv) What is the degree of harm or benefit of sharing and withholding the information.

If after a consideration of these factors, a clinician is of the opinion that the disclosure should be made, he or she must first discuss the reasons for making the disclosure with the patient.[246] BMA clearly provides adequate guidelines, taking into consideration a patient's right to confidentiality as well as an endangered third party's right to know. The only area that may be problematic is the lack of explanations. Guidelines should provide more fleshed out regulations that assist a doctor in deciding how much weight to place on various circumstances or factors.

[244] British Medical Association: Confidentiality and disclosure of health information (1999) 17-18.
[245] ibid 17-18.
[246] ibid 18.

7.5.1.3 English Case Law: The Duty to Warn

A good example that may shed some light on the duty to warn occurs when a person tests positive for a mutation in the breast cancer gene. Essentially, if a woman was aware of the fact that she has the BRCA mutation, she would know that she has a higher risk of developing breast cancer or even ovarian cancer. With such knowledge, she would be able to take preventative measures. For example, a woman may have a mastectomy which is the removal of both breasts. Although extreme, a mastectomy has been known to reduce the risk of breast cancer by approximately 90%.[247]

In cases like these, it is important to question whether or not a clinician has a duty to warn an affected related third party. The duty to warn may be understood as a clinician's legal obligation to inform a third party of any imminent risk posed to their health, irrespective of a patient's right to confidentiality.[248] In terms of this duty, it is essential to question whether prevention measures may be taken if an individual is warned of their genetic predisposition. For the most part, it seems that the duty to warn has been recognized and developed in the courts of foreign countries. Accordingly, the ambit and extent of the duty to warn must be examined.

In the UK, there have been no cases that have specifically dealt with the disclosure of genetic information to affected third parties. However, there have been English cases that have considered the duty to warn. In the case of *W v Egdell*,[249] Dr *Egdell*, a psychiatrist provided a report on W, a patient who had been detained at a secure hospital after being convicted of manslaughter. The assessment provided did not support the removal of the patient to a less secure facility. W eventually brought an action against Dr *Egdell* for breach of confidentiality. The court held that Dr *Egdell* did owe his patient a duty of confidentiality. However, the court went on to say that disclosure in the public interest overrode the duty of confidentiality between a doctor and their patient.[250]

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[247] 'What to do if your Genetic Test Results are Positive?' available at *http://www.breastcancer.org/symptoms/testing/genetic/pos_results*, accessed on 20 September 2014.
[248] C Meyer-MacAulay ' Who's in Your Genes: A Physician's "Duty to Warn" Patients' Relatives about Genetic Risk' (2012) 78(3) *UWOMJ*M 38.
[249] W v Egdell [1990] 2 W.L.R. 471.
[250] A Chinn 'Confidentiality, privacy and the reporting by health professionals of a patient's unfitness to drive: legal dilemmas' (2002) available at *http://acrs.org.au/files/arsrpe/RS020068.PDF*, accessed on 22 December 2014.

In addition, the court identified the following three elements which a psychiatrist must consider when deciding whether or not to breach confidentiality:[251]

i) Disclosure is needed to protect the public interest.

ii) The risk must be real.

iii) The risk must involve physical harm.

Although this case involved a psychiatrist, we can still note that there is precedent in the English legal system for putting the duty to warn ahead of the right to confidentiality. Whether or not this approach will be taken in future cases in terms of disclosure of genetic information is yet to be seen. Of important note, however, is the development of the duty itself. It appears that English and foreign courts alike will continue to expand the scope of the duty to warn as it relates to the relevant situation.[252] South Africa should be drawing on the development of the duty in foreign law in order to establish the parameters of the duty to warn in South Africa.

7.5.2 United States

7.5.2.1 American Medical Association

The American Medical Association's Code of Medical Ethics[253] emphasizes the need for communication by recommending pre and post-test counselling for patients. The Code takes a proactive approach whereby a clinician discusses with their patient whether or not biological relatives should participate in the genetic testing process.[254] In essence, the code states that before a patient undergoes testing, a clinician should tell the patient the 'circumstances under which they would expect patients to notify biological relatives of the availability of information related to risk of disease'.[255] On a positive note, this governance document informs the patient of the possibility of disclosure before the tests are even

[251] M Turner & M Kennedy 'Tarasoff and the duty to warn third parties' (1997) 21 *Psychiatric Bulletin* 466.
[252] W v Egdell [1990] 2 W.L.R. 471. This case is an example of the English courts expanding the duty to warn.
[253] 'Opinion 2.131 - Disclosure of Familial Risk in Genetic Testing' (December 2003) available at *http://www.ama-assn.org/ama/pub/physician-resources/medical-ethics/code-medical-ethics/opinion2131.page?*, accessed on 25 April 2015.
[254] ibid.
[255] ibid.

conducted. The Code goes further by stating that clinicians should support their patient by assisting a patient in their communication with their relatives.[256]

Despite its proactive approach, the Code could become problematic as a result of its vagueness. Firstly, there does not appear to be a formal set of circumstances under which a clinician might expect disclosure. More importantly, however, is the proactive approach itself. Informing patients on the possibility of disclosure before the testing process begins could be unproductive. In fear of their confidentiality being breached, a patient may refuse to proceed with the test at all. Accordingly, the code should have provided clinicians with more detailed guidelines on this matter. For example, would the refusal to consent to the disclosure during the pre-test counselling mean that the clinician would refuse to conduct the genetic tests in question? Guidelines of this nature should answer such questions and hence this study finds that this Code, despite its promise, has a long way to go.

7.5.2.2 American Case Law: The Duty to Warn

An example of an endangered third party can be seen in the American case of *Tarasoff v Regents of the University of California*.[257] In this case, a male student told the university psychologist that he wanted to kill a female student, who had previously rejected his advances. The psychologist warned security services, but failed to inform the female student or her family. The male student eventually killed her and accordingly, the girl's family sued the University of California on the basis that the psychologist should have disclosed the threat to the girl so that she could have taken steps to protect herself. [258]

In the case, the defendant argued that the imposition placed on therapists to protect third parties was inconceivable as a therapist cannot adequately determine if a patient will in fact resort to violence.[259] This argument can be countered by this study as where there is involuntary disclosure of genetic information, in most cases, a medical practitioner will be able to predict the harm that the third party will experience. However, problems may arise where there is just the chance that someone might develop a genetic condition. Although the

[256] ibid.
[257] Tarasoff v Regents of the University of California (1976).
[258] Tarasoff v Regents of the University of California (1976) 17 Cal. 3d 431.
[259] Tarasoff v Regents of the University of California (1976) 17 Cal. 3d 438.

test does not provide absolute certainty in certain cases, the third party could pay the price when a doctor refuses to breach confidentiality.

The court in *Tarasoff* used the example of a therapist failing to warn the authorities that his patient had threatened to assassinate the President, because the therapist could not conclusively predict that the patient would in fact commit the crime.[260] Overall, the court realized the difficulty of predicting possible danger and thus stated that a therapist need only exercise 'that reasonable degree of skill, knowledge, and care ordinarily possessed and exercised by members of that professional specialty under similar circumstances'.[261]This study supports the approach taken in this case. Accordingly, it is submitted that this standard should be applied when a clinician is going through the decision making process.

The court stated that when a therapist establishes that their patient is a serious threat to another, a duty is placed on a therapist to take reasonable care to protect a third party from danger. The court went on to say that a therapist is required to take appropriate steps, depending on the circumstances of the case, in order to carry out their duty to warn. Furthermore, the court held that a therapist has an obligation to maintain the confidence of their patient, except where disclosure is necessary to prevent danger to others. In this instance, disclosure must be done in a manner that upholds the privacy of the patient. The court held that the university had a duty to warn the endangered third party and that 'protective privilege ends where the public peril begins'.[262] This is of particular relevance to the study. Essentially, when a medical professional decides to make a disclosure, he or she must do so by only sharing information relevant to the affected party. Any other information, including the identity of the patient (if possible), should remain confidential.

Although this decision is not binding in South Africa, the *Tarasoff* judgment clearly has persuasive value throughout the world. This case attempts to define the parameters of confidentiality in a doctor-patient relationship. From the case, we can infer that the duty of confidentiality is inferior when put up against the well-being of an endangered third party. Even though this case deals with confidentiality in the context of a doctor-patient relationship, in which the doctor is a therapist, there is no reason as to why the principles that flow from this case cannot be used in a conventional doctor-patient relationship. It is submitted that this case can and should apply to the involuntary disclosure of genetic

[260] Tarasoff v Regents of the University of California (1976) 17 Cal. 3d 441.
[261]Tarasoff v Regents of the University of California (1976) 17 Cal. 3d 439..
[262] Tarasoff v Regents of the University of California (1976) 17 Cal. 3d 442.

information. The confidentiality of a doctor-patient relationship needs to stop when a third party can be helped by the disclosure of genetic information.

On the other hand, however, it has been stated that genetic cases differ from cases involving a therapist. This aspect is worth examining as in genetic cases, the harm, to some extent, has already occurred as the affected third party would already have the gene mutation. Accordingly, no act on the part of the clinician can prevent the gene mutation from being present.[263] What must be remembered, however, is that there is always the possibility of preventative measures. For this reason, disclosure must be given serious consideration in most cases.

Another case of note is the 1995 American case of *Pate v Threlkel*.[264] In this case, Heidi Pate sued her mother's medical practitioner for failing to warn her mother that thyroid cancer could be hereditary. She claimed that if her mother had been informed, she could have prevented her own condition. The Supreme Court of Florida agreed with Pate in that the physician has a duty to warn the patient about the hereditary nature of the condition. [265]

In 1996, however, the court in *Safer v Estate of Pack*[266] broadened the duty to warn. In the case, Donna Safer sued the estate of the late Dr Pack who had treated her father for colorectal cancer. Donna Safer had eventually been diagnosed with colorectal cancer and claimed that Dr Pack had failed to warn those at risk. Contrary to *Pate v Threlkel*, the court stated that a doctor's duty to warn at-risk relatives may not always be met by informing the patient of the hereditary nature of the condition. The court held further, that a clinician must take reasonable steps to ensure that the immediate family members of the patient are warned.[267] This study supports the approach taken by this court. Where possible, a clinician should be taking reasonable steps to warn affected related third parties.

[263] M R King 'Physicians Duty to Warn a Patient's Offspring of Hereditary Genetic Defects: Balancing the Patient's Right to Confidentiality Against the Family Member's Right to Know- Can or Should Tarasoff Apply' (2000) 4(1) *Quinnipiac Health Law* 31.
[264] Pate v Threlkel 661 So 2d 278 (1995).
[265] Pate v Threlkel 661 So 2d 278 (1995).
[266] Safer v Estate of Pack 677 A 2d 1188 (1996).
[267] K Offit...et al 'The "Duty to Warn" a Patient's Family Members About Hereditary Disease Risk' (2004) 292 (12) *JAMA* 1470.

7.6 National Governance Documents

7.6.1 Australia

In 2006, Australia brought into operation the Privacy Legislation Amendment Act.[268] The amended Act allows a healthcare professional to disclose genetic information to affected related third parties 'where there is reasonable belief that doing so is necessary to lessen or prevent a serious threat to the life, health or safety'[269] of the third party. An important aspect of this amendment is that it is not necessary for the threat to be imminent.[270]

The amendments made provide a framework to allow for the disclosure of genetic information in the appropriate circumstances, rather than simply placing an obligation on healthcare practitioners to disclose information in all instances. It is also praiseworthy that the amendment introduced the requirement for the National Health and Medical Research Council (NHMRC) to develop these guidelines, for healthcare practitioners in the private sector. This requirement allowed the NHMRC to take into consideration ethical matters and accordingly contribute guidelines and practical guidance that will supplement the amendment. However, a glaring shortfall of these guidelines is that they do not regulate genetic information that reveals a serious threat to unborn children.[271] Also, it is important to keep in mind that these are not law, but rather non-binding guidelines.

Taking into consideration the wording of the amendment, the guidelines have established a framework that dictates when, by whom and in what manner the disclosure of the information may occur, without a patient's consent. There is also a test. The test provides for disclosure where there is:[272]

(i) a serious threat to life, health or safety of a genetic relative

(ii) the use or disclosure is necessary to lessen or prevent the threat.

When the above test is satisfied, a healthcare practitioner may disclose genetic information only where the disclosure is done in accordance with the guidelines. This study will only

[268] Privacy Legislation Amendment Act 2006 (No. 99, 2006).
[269] National Health and Medical Research Council: Guidelines for health practitioners in the private sector (2009) 1.
[270] Privacy Legislation Amendment Act 2006 (No. 99, 2006) Schedule 2.
[271] National Health and Medical Research Council: Guidelines for health practitioners in the private sector (2009) 1-2.
[272] ibid 8.

discuss the guidelines of most relevance. Guideline 1 states that when determining whether there is a serious threat, the healthcare practitioner should consider two factors. Firstly, what is the nature of the condition and what are the risks and treatment options associated with the condition. Secondly, what are the chances that a genetic family member will also have or develop the genetic disorder or be a carrier of the relevant gene mutation.[273]

Furthermore, if a medical practitioner concludes that there is, in fact, a serious threat, the practitioner should go on to determine if there is a possibility to lessen or prevent the threat. Here, a practitioner will consider whether the at-risk relative will be able to treat or prevent the condition if they had knowledge of the genetic condition. Also, if there is no cure for the disorder, will knowledge of the disorder assist a relative in managing the condition?[274]

Guideline 2 discusses relevant ethical considerations that a healthcare practitioner should take into account when deciding whether or not to disclose. The guidelines state that effective communication between a practitioner and a patient at the initial consultation may assist the patient in understanding the implications that may arise in genetic testing. Also, where a practitioner uses active listening techniques, he or she can fully understand the reasoning behind a patient's refusal to consent.[275]

The guidelines discuss communication in two instances. Firstly, there should be genetic counselling.[276] In this instance, however, the guidelines fail to make genetic counselling a compulsory undertaking for patients undergoing genetic testing. The second form of communication occurs where genetic tests confirm a genetic disorder or that there is a risk of a genetic disorder. In this situation, the practitioner should also discuss the implications for relatives with the patient. There should also be a discussion on the possible benefits of disclosing the information to affected relatives.[277] However, such a regulation may be problematic. Consequently, it is submitted that such a discussion should occur during genetic counselling, before genetic tests are conducted.

Guideline 3 states that a practitioner should first take reasonable steps to obtain the consent of the patient. According to this guideline, a patient should be given the necessary information in order to come to an informed decision. The guidelines provide a comprehensive list of

[273] ibid 9.
[274] ibid 9.
[275] ibid 17.
[276] ibid 24 .
[277] ibid 23.

what the information should include, for example, which relatives are at risk and what is the likelihood of that relative developing the genetic condition.[278]

7.7 Conclusion

An analysis of the above governance documents has shown that the majority of foreign jurisdictions seem inclined to maintain confidentiality. However, they are also of the opinion that confidentiality is not absolute, and thus disclosure should be allowed in instances where the harm to an affected party is serious. Overall, from the detailed nature of guidance provided, it is clear that a considerable amount of understanding and expertise has gone into the development of the above documents. Presumably, these countries have experienced the same difficulties with disclosure as South Africa and thus an opportunity to learn from such documents should not be overlooked.

Foreign guidelines seem to have understood the flexibility that is required when dealing with disclosure cases. For instance, Australian guidelines allow a medical practitioner to make a decision based on the facts and circumstances of each individual case. WHO has also produced guidelines that allow the clinician to view the circumstances from each individual's perspective. Accordingly, the main lesson that South Africa can learn from these guidelines lies in the fact that the above governance documents are by no means straightforward guidelines. South Africa must realize that there is no one course of action that will satisfy all circumstances. Any legislation created by South Africa in the future must account for the difficulties experienced by all parties involved, rather than just the patient. In particular, it is important for future legislation to recognise that affected related third parties may only receive treatment or manage the condition when they become aware that they may, in fact, have a specific genetic condition.

[278]ibid 24.

CHAPTER 8

Recommendations

8.1 Introduction

After an in depth analysis of South African law and ethics that surround the involuntary disclosure of genetic information, it is submitted that there is a need for Parliament to develop and implement legislation that regulates the confidentiality and disclosure of genetic information. This study wishes to propose recommendations that will assist health officials in developing informed legislation on genetics. The following recommendations are some of the essential matters that such legislation should contain. These recommendations attempt to balance a patient's right to privacy with an affected relative's right to know.

8.2 Recommendations

8.2.1 Genetic Information

The statute should include a comprehensive and clear definition of genetic information. The following definition is recommended:

Genetic information is information that is obtained as a result of:

(i) an individual's genetic tests;

(ii) the genetic tests of a genetic relative of the individual;

(iii) pre-natal screening of the foetus;

(iv) newborn screening of neonates;

(v) pre-implantation testing of an embryo;

(vi) the development of a genetic condition or disease in a family member or the individual; and

(vii) any other genetic services or genetic research that a family member or the individual has participated in.

Genetic information will exclude any information that is referred to as personal information.[279]

8.2.2 Pre-test Counselling

Pre-test counselling is essential in order to ensure that a patient fully grasps the complexities involved in genetic testing. It is recommended that only a genetic counsellor or a psychiatrist specializing in the complexities of genetic testing be responsible for conducting the pre-test counselling. The following should be discussed during pre-test counselling:

- A patient's family history should be conducted in order to recognize or establish the inheritance pattern of the genetic condition.[280]
- What can be learnt and observed from positive or negative genetic test results.[281]
- A discussion on the manner in which the test will be conducted and accordingly the accuracy of such tests which includes an explanation of the reliability, limitations and uncertainty of the test.[282]
- The genetic conditions that the individual is being tested for. In this explanation, there should be a discussion in which the patient understands the condition being tested for and the possible advantages of early detection.
- The possibility of the genetic tests producing information that is relevant to the patient's family members.[283] This situation occurs where the results may show that a relative of the patient will develop or is at risk of developing a genetic condition.
- The confidentiality of genetic information.

[279] The Promotion of Access of Information Act 2 of 2000 section 1 defines personal information. Information that falls within the definition of personal information will not be considered as genetic information.
[280] S Cummings 'The Genetic Testing Process: How Much Counselling is Needed?' (2000) 18 (21) *Journal of Clinical Oncology* 61.
[281] ibid 62.
[282] ibid 62.
[283] ibid 62.

- The possibility of disclosure to affected related third parties and how and by whom such a decision to disclose is made. It is important to discuss with the patient the possibility that disclosure to affected persons may be necessary. Having this discussion before the testing process promotes trust and honesty between counsellor and patient. The patient also has a chance to get used to the idea of disclosure to affected persons.

- Lastly, the emotional and psychological state of the patient prior to the testing.

Genetic pre-test counselling, however, should also afford a patient an opportunity to discuss their fears and reasons for non-disclosure to affected family members. Genetic counselling should be used to inform a patient that the genetic testing procedure is not just an individual process.[284] At the same time, however, the purpose of pre-test counselling is not to obtain consent for the disclosure of genetic information.

It is important that healthcare professionals do not decline to perform genetic tests on individuals who refuse to consent to disclosure. Such treatment cannot possibly be in the best interests of the patient. It seems harsh and to some extent senseless to obtain a patient's consent to disclose information that has not even being obtained yet. Accordingly, during the pre-test counselling, the psychiatrist should only make a patient aware that there is a possibility that their confidentiality may be breached.

8.2.3 Post-test Counselling

Once a patient is given the results of their genetic tests, there are several important decisions that must now be made. In post-test counselling, it is recommended that the same genetic counsellor or psychiatrist who conducted the pre-test counselling should be used as there is already a rapport established with the patient. With the presence of a medical practitioner, the following issues should be discussed in post-test counselling:

- The emotional well-being of the patient in the event that the patient does have a genetic condition.[285]

- Detailed information on the specific genetic condition.[286]

[284]World Health Organization 'Genetic Counselling Services' available at *http://www.who.int/genomics/professionals/counselling/en/*, accessed on 22 March 2016.
[285] ibid.

- Possible prevention, treatment or management options for the genetic condition.[287] This discussion should include possible support mechanisms that will assist a patient in coping with the condition.

- The risk of having a child with the particular condition should be evaluated and explained.[288]

- Also, where a test reveals information on relatives, the healthcare professional should discuss the importance of disclosing such results to affected related third parties. This discussion should include the benefits and drawbacks of disclosing the information.[289]

When genetic test results are obtained, a patient should be asked to consent to the disclosure of relevant genetic information to affected related third parties. During counselling, a patient should be encouraged to disclose the information. Where a patient refuses to consent, he or she should be informed that the confidential doctor-patient relationship is not automatically breached. It is the recommendation of this study that various factors should be considered, by a selected panel, before the decision to disclose is made.

8.2.4 Who is to make the decision?

It is the view of this study that a decision of whether or not to disclose cannot be made by a medical practitioner alone. This study recommends that the decision must be made by a select and informed panel. The panel should consist of a medical practitioner (preferably specializing in genetics), a psychiatrist specializing in human genetics and a genetic counsellor or social worker.

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[286] Medical Research Council: Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research (2004).
[287] Medical Research Council: Guidelines on Ethics for Medical Research: Reproductive Biology and Genetic Research (2004).
[288] World Health Organization 'Genetic Counselling Services' available at *http://www.who.int/genomics/professionals/counselling/en/,* accessed on 22 March 2016.
[289] J L Gold 'To Warn or Not to Warn? Genetic Information, Families, and Physician Liability' (2004) 8 (1) *MJM* 72.

8.2.5 Factors to consider when making the decision to disclose involuntarily

Before a panel decides whether or not genetic information should be disclosed to affected relatives, the following factors should be taken into consideration:

a) The degree of accuracy of the information that has been provided by the genetic test. Where the results are uncertain, in that, it states that there is only a risk of the person developing the condition, the panel should use reasonable skill and experience in coming to a decision. In this instance, the panel should have a reasonable conviction that the affected relative would in fact develop the condition. The panel should also consider whether or not the relative will be a carrier for the genetic condition.[290]

b) The severity of the genetic condition. In other words, does the genetic condition pose a serious threat to the health and safety of the individual. [291] Essentially, a medical professional must disclose pertinent genetic information to affected related parties where the harm of non-disclosure outweighs the harm of disclosure.[292]

c) Would the sharing of genetic information reduce the threat? Essentially, if the affected related third party has knowledge of the test results, is there an action that he or she could take to prevent the manifestation of the genetic condition. Also, is there effective treatment available for the genetic condition that would help the affected individual.[293]

d) The patient's reason for refusing to consent to the disclosure.[294]

e) Is the disclosure worth infringing a patient's right to privacy? The panel must consider the consequences of breaching confidentiality as the doctor-patient relationship could suffer irreparable harm. The disclosure could also create animosity between the patient and the affected relative.[295]

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[290] See National Health and Medical Research Council: Guidelines for health practitioners in the private sector (2009) 9. Australian guidelines state that medical practitioners must consider the probability of a relative having the condition as well as the probability of the relative being a carrier for the relevant gene mutation.
[291] See National Health and Medical Research Council: Guidelines for health practitioners in the private sector (2009) 8. Australian guidelines state that there must be a serious threat to affected relative's health and safety.
[292] National Health and Medical Research Council: Guidelines for health practitioners in the private sector (2009) 41.
[293] See World Health Organization 'Review of Ethical Issues in Medical Genetics' (2003) 51. WHO states that there must be proof that any disclosed information can be used to prevent harm to affected relatives.
[294] See British Medical Association: Confidentiality and disclosure of health information (1999) 18. BMA guidelines state that a patient's reason for refusing consent must be given due consideration.
[295] B M Knoppers...et al 'Professional Disclosure of Familial Genetic Information (1998) 62 *Genet* 475-476.

After a consideration of the above factors, if the panel believes that a disclosure to an affected related third party is justified, then the genetic information should be disclosed despite the lack of consent from the patient.

8.2.6 Further Recommendations

There are also general recommendations that need to be considered. Firstly, when a panel decides to make a disclosure, the panel should first inform the patient of their decision and the reasons as to why such a decision was made. Also, when a disclosure is made, if possible, the identity of the patient as well as the exact nature of their genetic condition should be kept confidential. Only relevant information should be disclosed.

8.3 Conclusion

At present, it is clear that South Africa's regulatory framework on the disclosure of genetic information is unsatisfactory. In order to deal with these challenges, this study has recommended comprehensive genetic privacy legislation. The above recommendations have been informed by governance documents of foreign jurisdictions as well as the basic components of South African law and ethics.

CHAPTER 9

Conclusion

The completion of the Human Genome Project has made scientists aware of the potential in the field of genetics. However, there is no denying that the current era of genetics imposes its own challenges to ethical and legal frameworks. The main area of law that seems to be tested in terms of genetics is the principle of confidentiality. Every patient walks into a clinician's office with the expectation that all health information will remain private. The value of confidentiality in the doctor-patient relationship is invaluable as it is essential to the treatment process. However, despite its importance, information of this nature cannot always remain confidential.

As a result, the involuntary disclosure of genetic information is a problem that is bound to become more prevalent in South Africa. Foreseeing the possible difficulty health professionals will experience in the coming years, this dissertation has questioned the adequacy of current South African regulations regarding genetic information. After an in depth examination of existing guidelines, it is apparent that regulations do, in fact, cover certain issues. Overall, however, this study has concluded that these regulations are not satisfactory in that there are still gaps in our law that needs to be addressed. Accordingly, this dissertation sets out to provide ethically informed legislation and organizational ethics policies that fill in those gaps.

Accordingly, the role of ethics policies should not be undervalued. The enactment of legislation is a lengthy process and thus there is a need for healthcare establishments, dealing with genetic testing, to develop their own institutional policies that regulate disclosure. Each individual health facility should establish ethical policies that are in sync with the values of the establishment. Those responsible for the governance of the health establishments will be able to produce policies that guide a clinician as to what they should do, rather than what they must do.

Overall, genetic information in clinical settings tend to raise several ethical and legal concerns and although genetic testing is focused primarily on the private sector, these concerns should not go unnoticed. Foreign jurisdictions have already attempted to address

these complexities by putting into operation regulatory frameworks and guidelines that will mitigate ethical and legal concerns.

In summary, it is submitted that the regulations put forward by South Africa have failed to address the concerns examined in this study. The failure to adequately define the duty of a medical practitioner has placed patients in uncomfortable positions and third parties at risk. Accordingly, it is submitted that the only way to effectively balance a patient's right to confidentiality and an endangered third party's right to know, is to develop legislation that encompasses the ethical and legal considerations discussed.

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