

GENOMIC DIVERSITY, RISK OF EXTINCTION AND CONSERVATION OF SOUTH AFRICAN INDIGENOUS AND VILLAGE PIG POPULATIONS

by

Mandisa Gugu Mbuyazi

**Submitted in fulfilment of the academic requirements of
Master of Science**

in Genetics

School of Life Sciences

College of Agriculture, Engineering and Science University of
KwaZulu-Natal Pietermaritzburg

South Africa



2025

PREFACE

The research contained in this dissertation was conducted by the candidate while based in South Africa at the Discipline of Genetics, School of Life Sciences of the College of Agriculture, Engineering and Science, University of KwaZulu-Natal in Pietermaritzburg, in collaboration with the Agricultural Research Council-Biotechnology Platform in Pretoria. The research was financially supported by the National Research Foundation (NRF) and the Department of Agriculture, Land Reform and Rural Development (DALRRD).

The contents of this work have not been submitted in any form to another university and, except where the work of others is acknowledged in the text, the results reported are due to investigations by the candidate.

Signed:



Mandisa Gugu Mbuyazi (Candidate)

Date: 25 June 2025



Dr Edgar Farai Dzomba (Supervisor)

Date: 23 June 2025



Dr Farai Catherine Muchadeyi (Co-supervisor)

Date: 23 June 2025



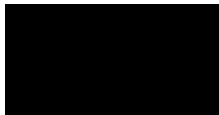
Dr Nompilo Lucia Hlongwane (Co-supervisor)

Date: 24 June 2025

DECLARATION: PLAGIARISM

I,Mandisa Gugu Mbuyazi..... declare that:

1. the research reported in this thesis, except where otherwise indicate, is my original work.
2. this thesis has not been submitted in full or in part for any degree or examination to any other university.
3. this thesis does not contain other persons' data, pictures, graphs or other information, unless specifically acknowledged as being sourced from other persons.
4. this thesis does not contain other persons' writing, unless specifically acknowledged as being sourced from other researchers. Where other written sources have been quoted, then:
 - a) their words have been re-written but the general information attributed to them has been referenced; and
 - b) where their exact words have been used, their writing has been placed inside quotation marks, and referenced.
5. this thesis does not contain text, graphics or tables copied and pasted from the Internet, unless specifically acknowledged, and the source being detailed in the thesis and in the References sections.



Signed: Mandisa Gugu Mbuyazi

Date: 25/06/2025

ABSTRACT

Genetic diversity, which includes variation within and among different populations, is vital for a population's ability to adapt to changing environments as it serves as the fundamental catalyst for evolution. South African indigenous (e.g., Kolbroek and Windsnyer) and village pig populations are valuable genetic resources, possessing unique adaptive traits and holding significant socio-economic importance for rural communities. However, these populations face genetic erosion and are at risk of extinction due to crossbreeding, inbreeding, and population decline, highlighting the urgent need for conservation efforts. This study initially explored the mating, breeding, and production systems of South African village pigs and their effects on genetic diversity. A total of 236 farmers were interviewed across six provinces. Limpopo had the highest number of respondents ($n = 70$), and pig samples collected ($n = 84$), mainly due to the greater availability and willingness of farmers to participate, as well as the province's high concentration of indigenous and village pig populations. Conversely, Mpumalanga had the fewest respondents ($n = 15$) and the lowest number of pig samples ($n = 37$), due to limited access to farmers in the area. The socio-economic characteristics of the households showed that most were male-headed (56.4%) and mainly aged between 41 and 60 years (47.1%), a substantial proportion (43.6%) were headed by females. Pig ownership was primarily among household heads (86.4%), with 7.2% shared with a spouse, and only 2.1% contributed by children. Among ethnic groups, Xhosa households were the most common (32.2%), followed by Pedi (23.7%) and Zulu (11.0%), while Venda, Ndebele, English, and Afrikaans groups combined made up less than 3.0%. Land ownership was mostly private, with 87.7% owning land, 7.2% leasing, and 5.1% using communal arrangements. Farm sizes varied significantly, but 44.9% managed between 0.25 and 2.0 hectares, and 33.9% had unspecified sizes. Pig farming was mainly semi-intensive (50.4%), with most farmers housing pigs in stalls or sheds (71.2%). Commercial breeds were predominantly kept, with the Large White being the most common (29.4%) and the Duroc the least. Most pig farmers in South African villages practice mixed livestock farming, where pigs are raised alongside other animals, such as cattle, goats, sheep, and poultry, within the same yard. Few households kept pigs exclusively. The feed and water sources used by farmers varied across regions. Morphometric traits analysed using PCA showed that PC1 and PC2 accounted for 32.6% and 20.6% of the total variance, respectively. Findings highlight the traditional practice of mixed livestock farming and the need for improved production systems. This study further examined genetic diversity, admixture, potential risk to extinction, functional genetic loss, inbreeding, and detected candidate genes in

the genomes of South African village pig populations. A total of 384 pig samples in South Africa, Eastern Cape ($n = 60$), Gauteng ($n = 84$), KwaZulu-Natal ($n = 63$), Limpopo ($n = 84$), Mpumalanga ($n = 37$), and North West ($n = 56$) were collected and genotyped using the Illumina PorcineSNP80K BeadChip. Additionally, 100 purebred (Duroc, Large White, South African Landrace, Kolbroek, and Windsnyer) pig genotypes were obtained from previous studies and used for genetic comparison. Genetic diversity parameters, including polymorphic information content (PIC), percentage of polymorphic loci ($\%P$), total (Na), effective allele numbers ($Ne^{alleles}$), P-value for the Global Hardy Weinberg test (P), observed (H_o), and expected heterozygosity (H_E), along with effective population sizes (Ne), were estimated in Rstudio using data.table and dplyr packages. Population structure, admixture, and ancestral analyses were performed using pairwise Fixation Index (F_{ST}), principal component analysis (PCA), ADMIXTURE, and TreeMix. Runs of homozygosity (ROH) and identity-by-descent (IBD) analyses examined inbreeding trends and detected candidate genes. Results indicated a rapid decline in effective population size (Ne) over generations, with low F_{ST} values (0.016 – 0.111) suggesting weak genetic differentiation. PCA revealed five genetic clusters, with PC1 and PC2 explaining 18.17% and 8.92% of total genetic variability, respectively. Optimal admixture clustering was revealed at $K = 14$. TreeMix analysis showed significant gene flow between commercial and indigenous breeds (i.e., gene flow between Windsnyer and South Landrace at $m = 1$). The KwaZulu-Natal – Harry Gwala population exhibited the highest percentage (23.55%) of long ROHs, indicating recent inbreeding, with the highest F_{ROH} estimates of 0.118. ROH islands were only detected on chromosome 7 (SSC7) from three populations (Gauteng - Tshwane, Gauteng - West Rand, and North West – Dr. Ruth Segomotsi). These ROH islands covered total 3.935 Mb, 2.930 Mb, and 1.145 Mb, respectively, of the total length of genomic regions. These regions harboured genes associated with economically important traits such as teat number ($ABCD4$), dressing percentage ($RPGRIP1$), lean meat percentage ($PSMCI$), and intramuscular fat content ($BDKRB2$). In addition, genes linked to reproductive traits, notably stillbirth ($PPP1R3E$, $PABPN1$), were also found. Both F_{ROH} and F_{HOM} values remained low, with all estimates below 0.20. The results of this study offer insights into the genetic diversity, structure, and inbreeding patterns of South African indigenous and village pig populations. By identifying regions of the genome associated with economically and reproductively important traits, this study highlights the potential for targeted genetic improvement while emphasizing the urgent need for conservation strategies. These findings contribute to a better understanding of how traditional farming and breeding practices

influence genetic resources and provide a foundation for sustaining adaptive potential and long-term resilience of local pig populations in South Africa.

Keywords: *genomic diversity, pigs, South Africa, Porcine SNP80K, indigenous, village pigs*

ACKNOWLEDGEMENTS

I sincerely thank my supervisors, Dr. Edgar F. Dzomba, Dr. Nompilo L. Hlongwane, and Dr. Farai C. Muchadeyi, for introducing me to genomics. I deeply appreciate your guidance and support throughout this study. The valuable contributions and mentorship of my supervisor, Dr. Nompilo L. Hlongwane, were crucial in helping me become a better scientist. I am grateful to her. I am immensely grateful to Dr. Magriet van der Nest for her input into my study, her great assistance with analysis, and for all she taught me in bioinformatics – I look forward to working with you again.

My heartfelt gratitude is extended to the University of KwaZulu-Natal and the Agricultural Research Council-Biotechnology Platform for providing the necessary infrastructure and research facilities. I am indebted to the National Research Foundation and the Department of Agriculture, Land Reform and Rural Development (Genetic Resources) for the financial support towards this study.

Thank you to the Department of Agriculture officials (Eastern Cape, Gauteng, KwaZulu-Natal, Limpopo, Mpumalanga, and North West) for their cooperation during sample collection. A heartfelt thank you to all the pig farmers for allowing us to sample.

I extend my sincere appreciation to the ARC-BTP Application Specialists, Alister Ngoben, for assisting with sample processing, and Maano Mmalima, for aiding in data analysis.

I thank my fellow postgraduate students at ARC-BTP for walking this journey with me and for their dedication during my sampling trips. I would also like to thank my lab sisters, Thubelihle, Vuyisile, and Andiswa, as well as my brother, Johannes, for consistently motivating me to remain committed throughout this journey and for requesting updates on the work and its progress. I would not have done it without love and support.

To my best friend, Londeka, thank you for always being there for me, for providing me with a second family, and for always being just one call away. Your kindness and support kept me sane during the hardest times. Ngiyabonga Mnganami.

To my parents, thank you for granting me life. Thank you, Mom (MaNdlazi), for your unconditional love and unwavering support. Without your consistent assistance, I would have given up. Most importantly, thank you for taking good care of my daughter when I was not around. I hope you are proud of me.

To my sister, deputy parent (Fanele), thank you for being my pillar and for your love and financial support over the years. My brother (Mlando) and my niece (Lisakhanya), I am forever grateful for your love and presence in my life, which always gave me strength to push harder.

To Baba kaThandolwethu, your unwavering love, patience, and support have been my greatest strength throughout this journey, from my undergraduate days to this moment. Your belief in me, constant encouragement, and ability to lift me during challenging times have made all the difference. I am truly grateful to have had you by my side.

And lastly, to my beautiful daughter, Thandolwethu, thank you for being the wonderful child you are and for your heartwarming smile. Even when my studies kept me away, your love and light never left my heart. The love I have for you has been my greatest motivation to keep going. Ngiyakuthanda ndodakazi yami!

DEDICATION

This thesis is dedicated to my daughter, Thandolwethu.

I did it all for you.

TABLE OF CONTENTS

GENOMIC DIVERSITY, RISK OF EXTINCTION AND CONSERVATION OF SOUTH AFRICAN INDIGENOUS AND VILLAGE PIG POPULATIONS	i
PREFACE	ii
DECLARATION: PLAGIARISM	iii
ABSTRACT	iv
ACKNOWLEDGEMENTS	vii
DEDICATION	ix
LIST OF TABLES	xiii
LIST OF FIGURES	xiv
ABBREVIATIONS	xvii
1 INTRODUCTION	1
1.1 Rationale for the research	1
1.2 Justification.....	2
1.3 Aims and Objectives.....	3
1.4 Outline of dissertation structure.....	3
1.5 References	4
2 LITERATURE REVIEW	7
2.1 Introduction	7
2.2 Pig Domestication.....	9
2.3 Overview of Pig Production in South Africa: Status and Challenges	9
2.4 Typology of South African Pig Production Systems	11
2.4.1 Intensive production.....	11
2.4.2 Semi-intensive production	12
2.4.3 Extensive production.....	12
2.5 Pig Breeds in South Africa	13
2.5.1 Commercial pig breeds	13
2.5.2 Indigenous pig breeds.....	16
2.5.3 Village pig populations.....	18
2.6 Crossbreeding of Indigenous with Exotic Breeds	18
2.7 Pig genetic diversity	19
2.8 Genetic Conservation of Indigenous and ‘Local’ Pig Breeds.....	19
2.8.1 Threats to Indigenous and Village Pig Populations	19
2.8.2 Importance of Conservation of Animal Genetic Resources.....	19
2.8.3 Conservation Strategies: In Situ and Ex Situ Approaches	20
2.9 Porcine Genomics and Statistical Tools	21

2.9.1 Single Nucleotide Polymorphisms	21
2.9.2 Porcine BeadChip.....	22
2.9.3 Genetic Diversity Indices	22
2.9.4 Population Structure and Pattern Analysis	22
2.9.5 Effective Population Size.....	23
2.9.6 Runs of Homozygosity	24
2.9.7 Identity by Descent.....	24
2.9.8 Selection Signatures	25
2.9.9 Copy Number Variations	25
2.11 Conclusion.....	26
2.12 References	26
3 PRODUCTION, BREEDING AND MATING SYSTEMS IN VILLAGE PIG FARMING: IMPLICATIONS FOR GENETIC DIVERSITY	37
3.1 Abstract.....	37
3.2 Introduction	37
3.3 Materials and Methods	39
3.3.1 Study sites and data collection	39
3.3.2 Data Analysis	41
3.4 Results	41
3.5 Discussion	47
3.6 Conclusion.....	51
3.7 References	51
4 GENETIC DIVERSITY, ADMIXTURE LEVELS, AND EXTINCTION RISK ASSESSMENT OF SOUTH AFRICAN INDIGENOUS AND VILLAGE PIG POPULATIONS RESULTING FROM CROSSBREEDING WITH EXOTIC BREEDS ..	55
4.1 Abstract.....	55
4.2 Introduction	55
4.3 Materials and Methods	59
4.3.1 Site Identification and Sample Collection	59
4.3.2 DNA Extraction, SNP Genotyping, and Quality Control	59
4.3.3 Data Analyses	60
4.4 Results	62
4.4.1 Genotyping and Quality Control	62
4.4.3 Effective Population Size.....	64
4.4.4 Population Genetic Differentiation, Structure, and Ancestry Estimation	65
4.5 Discussion	70
4.6 Conclusion.....	73
4.7 References	74

5 GENOME-WIDE IDENTIFICATION OF RUNS OF HOMOZYGOSITY AND ASSOCIATED GENES AND ESTIMATES OF GENOMIC INBREEDING IN SOUTH AFRICAN INDIGENOUS AND VILLAGE PIG POPULATIONS.....	78
5.1 Abstract.....	78
5.3 Materials and Methods	81
5.3.1 Sample Site and Data Collection.....	81
5.3.2 DNA Extraction, SNP Genotyping, and Quality Control	81
5.3.4 Identity by Descent.....	82
5.3.5 Genomic Inbreeding Coefficient Estimation	82
5.3.6 Runs of Homozygosity Detection	82
5.3.7 Detection of Common Runs of Homozygosity and Gene Annotation	83
5.4 Results	83
5.4.1 Identity by Descent.....	84
5.4.2 Genomic Inbreeding Coefficients	84
5.4.3 Distributions of Runs of Homozygosity	86
5.4.4 Common Runs Detected	90
5.5 Discussion	92
5.6 Conclusion.....	95
5.7 References	96
6 GENERAL DISCUSSION, CONCLUSIONS, CHALLENGES, AND RECOMMENDATIONS FOR FURTHER RESEARCH.....	101
6.1 Introduction	101
6.2 Aims and Objectives.....	101
6.3 General Discussion	102
6.4 Conclusion.....	104
6.5 Study Challenges & Recommendations	104
6.6 References	105
7 APPENDIX: SUPPLEMENTARY FILES.....	107

LIST OF TABLES

Table 2.1 Overview of South African pig production systems.....	13
Table 3.1 Morphometric traits and their categories are considered in the analysis of the physical characteristics of village pigs	40
Table 3.2 Socio-economic characteristics of households	42
Table 3.3 Livestock species kept in the household per province	44
Table 3.4 Frequencies (%) of household roles of pigs and management characteristics in the production system per province	45
Table 3.5 Boar selection, sourcing, and breeding practices	46
Table 4.1 Summary Results of SNP Quality Control	62
Table 4.2 Population genetic variability parameters.....	63
Table 5.1 Sampled South African village pig populations	81
Table 5.2 Mean and standard deviation of ROH based on inbreeding (F_{ROH}) and excess of homozygosity-based inbreeding (F_{HOM}) of South African village pi populations.....	86
Table 5.3 Mean length of ROH in Mb across 18 autosomes in South African village populations	89
Table 5.4 Top 50 percentile of runs of homozygosity	91

LIST OF FIGURES

Figure 2.1 Bayesian (MCMC) consensus tree of 122 <i>Sus</i> mtDNA control region haplotypes, rooted with a common warthog (<i>Phacochoerus aethiopicus</i>). The tree contains four major clades (1–4) and 14 clusters, each represented by a distinct color corresponding to regions on the Eurasian map. Tips from the island of Sulawesi correspond to the native wild boar (<i>Sus celebensis</i>), while all other tips represent wild <i>Sus scrofa</i> unless indicated by two-letter codes: sb (<i>Sus barbatus</i>), sv (<i>Sus verrucosus</i>). D1 to D6 indicate suggested centers of domestication, with D1–D3 representing areas where native wild boar haplotypes are identical to local domestic pigs. (Adapted from Larson <i>et al.</i>, 2005).....	8
Figure 2.2 Duroc boar (left) and Duroc sow with its piglets (right). Source: https://genetic.by/en/duroc (accessed 01 November 2025).	14
Figure 2.3 Large White sow. Source: https://farmingsouthafrica.co.za/best-pig-breeds-to-farm-with-in-south-africa/ (accessed 25 November 2025).	15
Figure 2.4 South African Landrace boar. Source: https://sasmallholder.co.za/2021/08/23/keeping-south-african-landrace-pigs/ (accessed 01 November 2025).	15
Figure 2.5 Kolbroek pig breed. Source: https://www.farmersweekly.co.za/app/uploads/2018/05/indigenous-Kolbroek-pig-Wessel-Pistorius.jpg (accessed 01 November 2025).	17
Figure 2.6 A Windsnyer pig breed (boar). Adapted from Moyo, 2017.....	17
Figure 2.7 Village pig breeds kept under a semi-intensive production system (Source: Photograph taken by the author, October 2023, Mpumalanga)	18
Figure 3.1 South African map displaying the geographical locations of the investigated pig populations	39
Figure 3.2 Morphological measurements taken from pigs in the study. The body measurements included height (H), length (L), girth (G), ear (E), and tail (T)	41
Figure 3.3 Number of pigs represented per province.....	43
Figure 3.4 Village pig breeds kept under different types of housing (Source: Photographs taken by the author, July 2023, Mpumalanga, and February 2024, North West).....	43
Figure 3.5 Distribution of pig production systems, housing types, and breeds kept by households.....	43
Figure 3.6 Principal Component Analysis using five quantitative traits (height, length, girth, ear length, and tail length). The ellipse represents the distribution of individuals across the PCA axis.....	47

Figure 4.1 South African map showing the locations of the population sample sites.....	59
Figure 4.2 Heterozygosity analysis (observed heterozygosity (H_O), expected heterozygosity (H_E), and heterozygote deficiency within population (F_{IS})).....	64
Figure 4.3 The estimated effective population size of 11 South African village pig populations over time with GONE (y-axis denotes the estimated N_e , while the x-axis represents generations ago. The different coloured lines indicate each population).....	64
Figure 4.4 Graphically representation of the pairwise F_{ST} values among the 13 village pig populations studied. Colours representing populations distances are defined on the scale at the top of the figure.	65
Figure 4.5 Principal Component Analysis Results of the eighteen pig populations including Hlongwane et al., (2020) SNP genotypes (21 084 SNPs)	66
Figure 4.6 Cross-validation plot of the admixture analysis	67
Figure 4.7 Admixture plot showing population clustering at $K = 14$. Each animal is represented by a single vertical line divided into K colours, where K is the number of assumed ancestral clusters that ranged from $K = 2$ to $K = 14$; the coloured segment shows the individual's estimated membership proportions in each cluster. Populations are grouped by type: the village/local populations (EC-ALN, EC-ORT, GP-JHB, GP-SBD, GP-TSW, GP-WRD, LP-CAP, LP-MOP, MP-GSD, NW-NMM, NW-RSM, ZN-HGL, ZN-UMG), commercial exotic breeds (PB-DUR, PB-LWT, PB-SAL), and indigenous breeds (PB-WIN, PB-KOL).	68
Figure 4.8 The output generated by OptM for the simulated dataset with $m = 6$ migration edges. (a) The mean and standard deviation (SD) across 10 iterations for the composite likelihood $L(m)$ (left axis, black circles) and the proportion of variance explained (right axis, red "x"s). The 99.8% threshold is shown as a horizontal dotted line. (b) The second-order rate of change (D_m) across different values of m	69
Figure 4.9 Population splits and mixture of the tested commercial, indigenous and village pig populations inferred using TreeMix. Six migrations were allowed as inferred by OptM. The migration edges are coloured according to their weight. The scale bar indicates ten times the average standard error of the values in the covariance matrix..	70
Figure 5.1 Heatmap representing genome-wide identity by descent (IBD) between pairs of South African village pigs estimated with the PI-HAT coefficient	84
Figure 5.2 Genomic inbreeding coefficients within each population (a) F_{ROH} distribution in the thirteen studied populations (b) F_{HOM} distribution in the thirteen studied populations	85
Figure 5.3 Individual patterns of ROH (<i>the number of ROH compared to the total length of ROH for individuals</i>). Each dot represents an individual.	87
Figure 5.4 ROH count per population in different classes.....	88
Figure 5.5 Distribution across chromosomes with the highest number of detected ROH (SSC1, SSC7, SSC13, and SSC14)	90

Figure 5.6 Manhattan plot displaying the distribution of runs of ROH hotspots in the studied South African village pigs. The x-axis represents the SNP genomic coordinate, while the y-axis shows the percentage (%) of overlapping ROH shared among individuals. (a) EC-ALN; (b) EC-ORT; (c) GP-JHB; (d) GP-SBD; (e) GP-TSW; (f) GP-WRD; (g) LP-CAP; (h) LP-MOP; (i) MP-GSD; (j) NW-NMM; (k) NW-RSM; (l) ZN-HGL; (m) ZN-UMG. The line parallel to the X-axis marks the top 50% of SNP frequency in a run of homozygosity (ROH). SNPs above that line correspond to ROH islands.....92

ABBREVIATIONS

AI - Artificial Insemination

AnGR - Animal Genetic Resources

ASF - African Swine Fever

CNV - Copy Number Variation

CV - Cross validation

DALLRD - Department of Agriculture, Land Affairs and Rural Development

FAO - Food and Agriculture Organisation

F_{IS} - Inbreeding coefficient within a breed

F_{HOM} - Inbreeding coefficient based on excess homozygosity

F_{ROH} - Inbreeding coefficient based on runs of homozygosity

F_{ST} - Fixation index

GRM - Genomic relationship matrix

HapFLK - Haplotype-based F-statistics using Local Kinship

H_E - Expected heterozygosity

H_O - Observed heterozygosity

HWE - Hardy-Weinberg equilibrium

IBD - Identity by descent

iHS - Integrated haplotype score

KZN - KwaZulu-Natal

LD - Linkage disequilibrium

MAF - Minor allele frequency

N_a - Mean allele number per locus

N_e - Effective population size

$N_e^{alleles}$ - Effective allele number

PCA - Principal component analysis

PIC - Polymorphic Information Content

QC - Quality control

QTL - Quantitative trait locus (loci)

REHH - Relative extended haplotype homozygosity

ROH - Runs of homozygosity

SAPPO - South African Pork Producers

SNP - Single Nucleotide Polymorphism

SSR - Simple Sequence Repeat

XP-EHH - Cross population extended haplotype homozygosity

1 INTRODUCTION

1.1 Rationale for the research

Pigs (*Sus scrofa domesticus*) are one of the earliest domesticated livestock species, originating about 9,000 years ago from their wild ancestor, the Eurasian wild boar (*Sus scrofa*), across multiple domestication centers such as the Near East, China, Europe, and Southeast Asia (Groenen *et al.*, 2012; Ruvinsky *et al.*, 2011). Domestication has led to a wide variety of pig breeds adapted to different environments and production systems around the world. In South Africa, two local breeds, Kolbroek and Windsnyer, are now recognized as indigenous and are taxonomically classified under *Sus indica* (Nicholas, 1999; Ramsay *et al.*, 2000). These breeds exhibit high adaptability to harsh environmental conditions, including limited feed availability and tropical disease pressure, and require minimal inputs for survival and reproduction (Marshall *et al.*, 2019; Swart *et al.*, 2010). As a result, they play an essential role in supporting rural communities by contributing to food security, poverty alleviation, and income generation (Antwi & Seahlodi, 2011; Halimani *et al.*, 2012; Madzimure *et al.*, 2012).

Despite their ecological and socio-economic importance, indigenous pig populations in South Africa are primarily managed under low-input, informal production systems characterized by limited access to resources and the absence of formal breeding programs (Babigumira *et al.*, 2021; Halimani *et al.*, 2012; Madzimure *et al.*, 2012). Due to economic constraints, many smallholder farmers maintain herds of fewer than five animals, which significantly increases the risk of inbreeding and genetic erosion over time (Lynch *et al.*, 1995; Mthi *et al.*, 2017; Nyamushamba *et al.*, 2017). Furthermore, the lack of pedigree records often leads to uncontrolled mating practices, such as intergenerational breeding, where parents mate with offspring or siblings with siblings. This results in increased homozygosity, which reduces genetic diversity and enhances the expression of harmful recessive alleles; a phenomenon called inbreeding depression (Ekue *et al.*, 2002; Illango *et al.*, 2002; Kardos *et al.*, 2017; Pekkala *et al.*, 2012, 2014).

In addition to extinction risks, indigenous pig populations are increasingly exposed to crossbreeding with high-yielding exotic breeds such as Large White, Landrace, and Duroc. While crossbreeding may offer short-term productivity gains such as improved growth rates, feed conversion efficiency, increased carcass yield, and enhanced reproductive performance, it poses a significant threat to the genetic integrity of indigenous breeds, leading to widespread admixture and potential loss of unique genetic traits (Hlongwane *et al.*, 2020). Moreover,

policies and market incentives often favor exotic breeds, promoting their adoption at the expense of local breeds, which are perceived as less productive (Chimonyo & Dzama, 2007). As a result, indigenous pigs remain underutilised and neglected despite their adaptive advantages in local environments.

1.2 Justification

Globally, livestock biodiversity is under serious threat, with approximately 35% pig breeds currently classified as endangered or already extinct (FAO, 2009). This alarming trend reflects broader challenges facing traditional livestock production systems, including habitat loss, displacement by high-performing breeds, and the homogenizing effects of globalization (FAO, 2009). In South Africa, these global pressures are compounded by recent outbreaks of African Swine Fever (ASF) in 2022, 2023, and 2024, particularly affecting provinces such as Eastern Cape, KwaZulu-Natal, Limpopo, and Mpumalanga (DALRRD, 2024). ASF is a highly contagious and often fatal viral disease that spreads rapidly among pig populations, causing devastating losses to both commercial and smallholder farmers (Mutua & Dione, 2021). The disease not only disrupts pork supply chains but also exacerbates the vulnerability of already fragile indigenous pig populations.

A critical challenge in conserving these breeds lies in the lack of comprehensive data on their genetic characteristics and population dynamics (FAO, 2000). Without accurate inventory and genetic profiling, effective conservation strategies cannot be developed or implemented. As global demand for livestock products continues to rise in response to population growth and changing dietary patterns (FAO, 2015), the gap between current pork production levels and projected consumption widens. Without targeted scientific and technological interventions, local producers may struggle to meet this growing demand.

However, indigenous and village pigs possess unique genetic traits that could serve as the foundation for developing new, resilient, and locally adapted breeds. Understanding the genetic makeup of indigenous and village pig populations is therefore essential for addressing agricultural, societal, and conservation challenges (Halimani *et al.*, 2020; Hlongwane *et al.*, 2020). Recent advancements in genomics provide powerful tools for assessing and preserving genetic diversity between livestock populations (Swart *et al.*, 2010). One such tool is single nucleotide polymorphism (SNP) genotyping array, which allows researchers to examine thousands of genetic markers across the genome simultaneously. With decreasing costs and increasing accessibility, SNP-based technologies have become central to studies on genetic variation, population structure, and breed relationships (Rothschild & Ruvinsky, 2011).

The use of high-density SNP arrays, such as the Porcine 80K SNP BeadChip, has been widely applied in global pig genetics research to investigate genetic diversity, admixture, and inbreeding levels (Feng *et al.*, 2022; Lin *et al.*, 2024; Xu *et al.*, 2023). Applying these genomic tools to South African indigenous and village pig populations will enable researchers to generate detailed insights into their genetic composition, breeding history, and levels of hybridization with exotic breeds. These findings will be instrumental in guiding future management, breeding, and conservation efforts aimed at preserving the country's valuable animal genetic resources.

1.3 Aims and Objectives

This study aims to undertake a comprehensive assessment of the current status of South African indigenous and village pig populations, recognizing them as unique and valuable components of the country's animal genetic resources. By integrating field surveys with advanced genomic techniques, this research seeks to inform sustainable management and conservation strategies that align with both agricultural development goals and biodiversity preservation priorities.

The objectives of this study were:

- (i) To investigate current mating systems, breeding practices, and production systems, and evaluate their impact on the maintenance of genetic diversity within indigenous and village pig populations.
- (ii) To characterize the genetic diversity, extent of admixture, and assess the potential risk of extinction among South African indigenous and village pig populations resulting from crossbreeding with prevalent exotic breeds.
- (iii) To examine inbreeding patterns and identify candidate genes using genomic approaches such as Identity by Descent (IBD) and Runs of Homozygosity (ROH) analyses, which detect regions of the genome potentially affected by inbreeding and that may harbour harmful recessive mutations.

1.4 Outline of dissertation structure

The dissertation contains a general introduction and literature review for the study, followed by three experimental chapters written in manuscript format. Conclusions and general discussions will be in Chapter 6. The dissertation chapters are as follows:

Chapter 1: General introduction of the thesis outlining the rationale, justification, and clearly defined aims and objectives of the research.

Chapter 2: Literature review encompassing the pig domestication process, pig breeds in South Africa, various production systems, conservation strategies, advances in porcine genomics, and relevant statistical tools.

Chapter 3: Production, breeding, and mating systems in village pig farming: implications for genetic diversity.

Chapter 4: Genetic diversity, admixture levels, and extinction risk assessment of South African indigenous and village pig populations resulting from crossbreeding with exotic breeds.

Chapter 5: Genome-wide identification of runs of homozygosity and associated genes and estimates of genomic inbreeding in South African indigenous and village pig populations.

Chapter 6: General discussion, conclusion, highlights challenges, and offers recommendations for further research.

1.5 References

- Antwi, M., & Seahlodi, P. (2011). Marketing Constraints Facing Emerging Small-Scale Pig Farmers in Gauteng Province, South Africa. *Journal of Human Ecology*, 36(1), 37–42. <https://doi.org/10.1080/09709274.2011.11906415>
- Babigumira, B. M., Sölkner, J., Mészáros, G., Pfeiffer, C., Lewis, C. R. G., Ouma, E., Wurzinger, M., & Marshall, K. (2021). A Mix of Old British and Modern European Breeds: Genomic Prediction of Breed Composition of Smallholder Pigs in Uganda. *Frontiers in Genetics*, 12(June), 1–12. <https://doi.org/10.3389/fgene.2021.676047>
- Broman, K. W., & Weber, J. L. (1999). Long homozygous chromosomal segments in reference families from the Centre d'Etude du Polymorphisme Humain. *American Journal of Human Genetics*, 65(6), 1493–1500. <https://doi.org/10.1086/302661>
- Bruford, M. W., Ginja, C., Hoffmann, I., Joost, S., Wengel, P. O., Alberto, F. J., Amaral, A. J., Barbato, M., Biscarini, F., Colli, L., Costa, M., Curik, I., Duruz, S., Ferencakovic, M., Fischer, D., Fitak, R., Groeneveld, L. F., Hall, S. J. G., Hanotte, O., ... Zhan, X. (2015). Prospects and challenges for the conservation of farm animal genomic resources, 2015-2025. *Frontiers in Genetics*, 6(OCT), 1–11. <https://doi.org/10.3389/fgene.2015.00314>
- Chimonyo, M., & Dzama, K. (2007). Estimation of genetic parameters for growth performance and carcass traits in Mukota pigs. *Animal*, 1(3), 317–323. <https://doi.org/10.1017/S1751731107661849>
- Dekkers J. C. (2004). Commercial application of marker- and gene-assisted selection in livestock: strategies and lessons. *J. Anim. Sci.* 82 E313–E328.
- Department of Agriculture, Land, Reform and Rural Development (DALRRD) African swine fever outbreak and surveillance update report, 2024. Available from: <https://www.dalrrd.gov.za/images/outbreaks/African%20Swine%20Fever/Reports/2024/african-swine-fever-report-update-2024-03-28.pdf>
- Ekue F.N. P.K.D., Mafeni M.J., Nfi A.N. & Njoya J. (2002) Survey of the traditional poultry production system in the Bamenda area, Cameroon. In: Characteristics and parameters of family poultry production in Africa. FAO/IAEA, Vienna, Austria, p. 15
- FAO. 2000. Secondary guidelines of farm animal genetic resources management plans. Management of small populations at risk. Rome: Food and Agriculture Organization of the United Nations.

- Feng, X., Diao, S., Liu, Y., Xu, Z., Wei, C., Yuan, X., & Li, J. (2022). Analysis of selection signatures for Hainan pigs across the whole genome based on SNP beadchips. <https://www.cabidigitallibrary.org/doi/full/10.5555/20220153870>
- Gibson, J., Morton, N., genetics, A. C.-H. molecular, & 2006, undefined. (2006). Extended tracts of homozygosity in outbred human populations. *Academic.Oup.ComJ Gibson, NE Morton, A Collins Human Molecular Genetics, 2006•academic.Oup.Com.* <https://doi.org/10.1093/hmg/ddi493>
- Groenen, M. A. M., Archibald, A. L., Uenishi, H., Tuggle, C. K., Takeuchi, Y., Rothschild, M. F., Rogel-Gaillard, C., Park, C., Milan, D., Megens, H. J., Li, S., Larkin, D. M., Kim, H., Frantz, L. A. F., Caccamo, M., Ahn, H., Aken, B. L., Anselmo, A., Anthon, C., ... Schook, L. B. (2012). Analyses of pig genomes provide insight into porcine demography and evolution. *Nature*, 491(7424), 393–398. <https://doi.org/10.1038/nature11622>
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2010). Pig genetic resource conservation: The Southern African perspective. *Ecological Economics*, 69(5), 944–951. <https://doi.org/10.1016/j.ecolecon.2010.01.005>
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2012a). Opportunities for conservation and utilisation of local pig breeds in low-input production systems in Zimbabwe and South Africa. *Tropical Animal Health and Production*, 45(1), 81–90. <https://doi.org/10.1007/S11250-012-0177-2/TABLES/9>
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2012b). Some insights into the phenotypic and genetic diversity of indigenous pigs in southern Africa. *South African Journal of Animal Science*, 42(5), 507–510. <https://doi.org/10.4314/SAJAS.V42I5.13>
- Hlongwane, N. L., Hadebe, K., Soma, P., Dzomba, E. F., & Muchadeyi, F. C. (2020). Genome Wide Assessment of Genetic Variation and Population Distinctiveness of the Pig Family in South Africa. *Frontiers in Genetics*, 11(May), 1–19. <https://doi.org/10.3389/fgene.2020.00344>
- Hlongwane, N. Lucia. (2022). Genome-wide characterization of South African pig breeds. <https://researchspace.ukzn.ac.za/handle/10413/22159>
- Kardos, M., Qvarnström, A., & Ellegren, H. (2017). Inferring individual inbreeding and demographic history from segments of identity by descent in *Ficedula* flycatcher genome sequences. *Genetics*, 205(3), 1319–1334. <https://doi.org/10.1534/GENETICS.116.198861/-/DC1>
- Illango J. E.A., Olupot H., Mabonga J. (2002) Rural poultry production in two agro-ecological zone of Uganda. In: Characteristics and parameters of family poultry production in Africa (p. 117. FAO/IAEA, Vienna, Austria.
- Jones G. F. (1998). “Genetic aspects of domestication, common breeds and their origin,” in *The Genetics of the Pig*, eds Rothschild M. F., Ruvinsky A. (Wallingford: CABI Publishing;).
- Lin, D., Qiu, Y., Zhou, F., Li, X., Deng, S., Yang, J., Chen, Q., Cai, G., Yang, J., Wu, Z., & Zheng, E. (2024). Genome-wide detection of multiple variants associated with teat number in French Yorkshire pigs. *BMC Genomics*, 25(1), 1–12. <https://doi.org/10.1186/S12864-024-10611-9/TABLES/4>
- Lukić, B., Ferencaković, M., Šalamon, D., Čačić, M., Orehovački, V., Iacolina, L., Curik, I., & Cubric-Curik, V. (2020). Conservation Genomic Analysis of the Croatian Indigenous Black Slavonian and Turopolje Pig Breeds. *Frontiers in Genetics*, 11(March). <https://doi.org/10.3389/fgene.2020.00261>
- Lynch, M., Conery, J., & Bu7rger3, R. (1995). MUTATION ACCUMULATION AND THE EXTINCTION OF SMALL POPULATIONS (Vol. 146, Issue 4). <http://www.journals.uchicago.edu/t-and-c>
- Madzimure, J., Chimonyo, M., Zander, K. K., & Dzama, K. (2012). Potential for using indigenous pigs in subsistence-oriented and market-oriented small-scale farming systems of Southern Africa. *Tropical Animal Health and Production*, 45(1), 135–142. <https://doi.org/10.1007/S11250-012-0184-3>
- Marshall, K., Gibson, J. P., Mwai, O., Mwacharo, J. M., Haile, A., Getachew, T., Mrode, R., & Kemp, S. J. (2019). Livestock genomics for developing countries - African examples in practice. *Frontiers in Genetics*, 10(APR), 1–13. <https://doi.org/10.3389/fgene.2019.00297>
- Masenya, M. B., Mphaphathi, M. L., Mapeka, M. H., Munyai, P. H., Makhafola, M. B., Ramukhithi, F. V., Malusi, P. P., Umesiobi, D. O., & Nedambale, T. L. (2011). Comparative study on semen characteristics of Kolbroek and Large White boars following computer aided sperm analysis®

- (CASA). *African Journal of Biotechnology*, 10(64), 14223–14229. <https://doi.org/10.5897/AJB11.2010>
- Mote B. E., Rothschild M. F. (2020). “Modern genetic and genomic improvement of the pig,” in *Animal Agriculture*, eds Bazer F. W., Lamb G. C., Wu G. (Cambridge: Academic Press;), 249–262. 10.1016/b978-0-12-817052-6.00014-8
- Mthi, S., Skenjana, A., of, P. F.-I. J., & 2017, undefined. (2017). Characteristics of small-scale sheep production systems in some communal areas of the Eastern Cape Province, South Africa. *Academicjournals.Org*, 8(12), 199–206. <https://doi.org/10.5897/IJLP2016.0326>
- Muchadeyi FC, Wollny CB, Eding H, Weigend S, Simianer H. Choice of breeding stock, preference of production traits and culling criteria of village chickens among Zimbabwe agro-ecological zones. *Trop Anim Health Prod.* 2009 Mar;41(3):403-12. doi: 10.1007/s11250-008-9204-8. Epub 2008 Jul 15. PMID: 18626780.
- Muñoz, M., Bozzi, R., García-Casco, J., Núñez, Y., Ribani, A., Franci, O., García, F., Škrlep, M., Schiavo, G., Bovo, S., Utzeri, V. J., Charneca, R., Martins, J. M., Quintanilla, R., Tibau, J., Margeta, V., Djurkin-Kušec, I., Mercat, M. J., Riquet, J., ... Óvilo, C. (2019). Genomic diversity, linkage disequilibrium and selection signatures in European local pig breeds assessed with a high density SNP chip. *Scientific Reports*, 9(1), 1–14. <https://doi.org/10.1038/s41598-019-49830-6>
- Nicholas, G. (1999). Kolbroek-the Unique Local Breed. *Farmer’s Weekly*, August
- Nyamushamba, G. B., Mapiye, C., Tada, O., Halimani, T. E., & Muchenje, V. (2017). Conservation of indigenous cattle genetic resources in Southern Africa’s smallholder areas: Turning threats into opportunities - A review. *Asian-Australasian Journal of Animal Sciences*, 30(5), 603–621. <https://doi.org/10.5713/ajas.16.0024>
- Pekkala, N., Knott, K. E., Kotiaho, J. S., Nissinen, K., & Puurtinen, M. (2014). The effect of inbreeding rate on fitness, inbreeding depression and heterosis over a range of inbreeding coefficients. *Evolutionary Applications*, 7(9), 1107–1119. <https://doi.org/10.1111/EVA.12145>
- Pekkala, N., Knott, K. E., Kotiaho, J. S., & Puurtinen, M. (2012). Inbreeding rate modifies the dynamics of genetic load in small populations. *Ecology and Evolution*, 2(8), 1791–1804. <https://doi.org/10.1002/ECE3.293>
- Ramsay, K., Smuts, M. and Els, H.C. 2000. Adding value to South African landrace breeds: Conservation through utilization. *Animal Genetic Resources Information* (27): 9-16
- Roder, W., Gratzer, G., & Wangdi, K. (2002). Cattle grazing in the conifer forests of Bhutan. *Mountain Research and Development*, 22(4), 368–374. [https://doi.org/10.1659/0276-4741\(2002\)022\[0368:CGITCF\]2.0.CO;2](https://doi.org/10.1659/0276-4741(2002)022[0368:CGITCF]2.0.CO;2)
- Ruvinsky, A., Rothschild, M. F., Larson, G., & Gongora, J. (2011). *Systematics and evolution of the pig. The Genetics of the Pig: Second Edition*, 1–13. <https://doi.org/10.1079/9781845937560.0001>
- SanCristobal M., Chevalet C., Haley C., Joosten R., Rattink A., Harlizius B., et al. (2006). Genetic diversity within and between European pig breeds using microsatellite markers. *Anim. Genet.* 37 189–198.
- Scandura M., Iacolina L., Apollonio M. (2011). Genetic diversity in the European wild boar *Sus scrofa*: phylogeography, population structure and wild × domestic hybridization. *Mamm. Rev.* 41 125–137. 10.1111/j.1365-2907.2010.00182.x
- Simon, D. L. (1984). Conservation of animal genetic resources - A review. *Livestock Production Science*, 11(1), 23–36. [https://doi.org/10.1016/0301-6226\(84\)90004-6](https://doi.org/10.1016/0301-6226(84)90004-6)
- Swart, H., Kotze, A., Olivier, P. A. S., & Grobler, J. P. (2010). Microsatellite-based characterization of Southern African domestic pigs (*Sus scrofa domestica*). In *South African Journal of Animal Science* (Issue 2). <http://www.sasas.co.za/sajas.asp>
- Taberlet, P., Coissac, E., Pansu, J., & Pompanon, F. (2011). Conservation genetics of cattle, sheep, and goats. *Comptes Rendus - Biologies*, 334(3), 247–254. <https://doi.org/10.1016/j.crv.2010.12.007>
- Xu, C., Zhang, W., Jiang, Y., Zhou, M., Liu, L., Su, S., Li, X., & Wang, C. (2023). Genome-Wide Detection and Analysis of Copy Number Variation in Anhui Indigenous and Western Commercial Pig Breeds Using Porcine 80K SNP BeadChip. *Genes*, 14(3). <https://doi.org/10.3390/genes14030654>

2 LITERATURE REVIEW

2.1 Introduction

Pigs (*Sus scrofa domestica*), the descendants of distinct domestication events in Asia and Europe that occurred over thousands of years (Chen *et al.*, 2007; Frantz *et al.*, 2015; Larson *et al.*, 2005; Larson *et al.*, 2014; Price & Hongo, 2020; Smith *et al.*, 2014; Zhang *et al.*, 2022), were introduced into South Africa through various historical channels, including trade routes. **Figure 2.1** illustrates the centers of pig domestication and the subsequent global spread (Larson *et al.*, 2005). Archaeological evidence suggests that the imported pigs were initially used by Portuguese travelers and later by Chinese trading ships as a means of barter with local communities along the South African coast (Ramsay *et al.*, 1994; Quin, 1959). Since then, pigs have played and continue to play a vital role in the country's socio-economic landscape and rural development. According to the Department of Agriculture, Land Reform, and Rural Development (DALRRD, 2023), South Africa's pig population is approximately 1,332,000, with the largest concentrations found in the Limpopo (24%) and North-West (20%) provinces. The country now maintains a diverse range of pig breeds, including commercial, indigenous, and non-descript types, although ruminants such as cattle, sheep, and goats remain the dominant livestock species (Gusha *et al.*, 2024; Khowa *et al.*, 2023; Taruvinga *et al.*, 2022).

The indigenous and village pig populations in South Africa play a crucial role in rural livelihoods. These pigs are well-adapted to local environments, resistant to diseases, and suited to low-input farming systems (Muchadeyi *et al.*, 2008; Dzama *et al.*, 2003; Munzhelele *et al.*, 2016; Madzimure *et al.*, 2012). In addition to playing a significant role in rural communities by providing protein and income, they also serve in important social and cultural functions, such as participating in traditional ceremonies, communal exchanges, and rituals (Adesehinwa *et al.*, 2024; Gcumisa *et al.*, 2016; Nahar *et al.*, 2015). However, despite their significance, most research often overlooks their contributions, and policy efforts continue to focus on commercial pig production, leaving them largely understudied (Chimonyo & Dzama, 2007). Recognising the cultural and economic importance of these pigs is key to creating sustainable and inclusive genetic conservation efforts.

This review aims to examine South African pig production systems and evaluate the genetic diversity and population divergence of indigenous and village pigs in South Africa. It highlights recent advances in pig genomics and examines the feasibility of conserving village and indigenous pig populations, along with the associated risks of extinction. Additionally, the review discusses the genomic tools used to study and preserve both neutral and functional

diversity within pig populations, concluding with an overview of the statistical genomic tools now employed to generate meaningful insights into these populations.

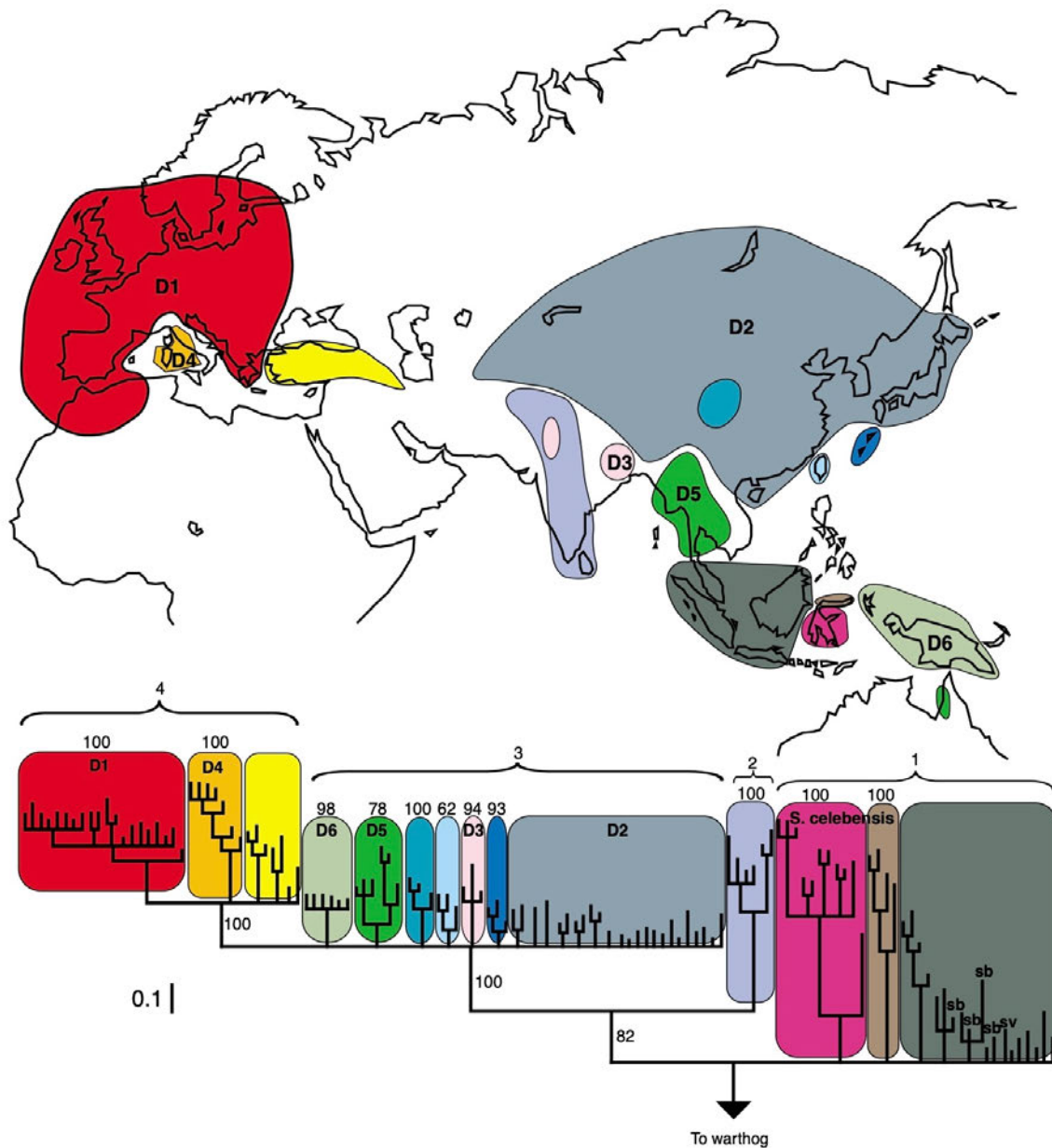


Figure 2.1 Bayesian (MCMC) consensus tree of 122 *Sus* mtDNA control region haplotypes, rooted with a common warthog (*Phacochoerus aethiopicus*). The tree contains four major clades (1–4) and 14 clusters, each represented by a distinct color corresponding to regions on the Eurasian map. Tips from the island of Sulawesi correspond to the native wild boar (*Sus celebensis*), while all other tips represent wild *Sus scrofa* unless indicated by two-letter codes: sb (*Sus barbatus*), sv (*Sus verrucosus*). D1 to D6 indicate suggested centers of domestication, with D1–D3 representing areas where native wild boar haplotypes are identical to local domestic pigs. (Adapted from Larson *et al.*, 2005)

2.2 Pig Domestication

Domestication is a genetic adaptation process where wild organisms are modified to better suit human needs and is achieved through controlled breeding and living conditions over multiple generations (Darwin, 1868). In pigs, this evolutionary process led to behavioural (*e.g.*, reduced aggression, watchfulness, and stress), morphological (*e.g.*, altered skull and teeth shape), and physiological (*e.g.*, increased growth and reproductive rates) changes all aimed to satisfy human needs. Also, the natural traits of *Sus Scrofa* allow them to interact with human cultures. For approximately 10,000 years, pigs have been interacting with humans, producing a large amount of animal-based protein for human diets (Gieling *et al.*, 2011). This mutual reliance between humans and domestic pigs is believed to have developed as early as 12,000 years ago (Redding, 2005). The origins of this relationship remain a mystery that may never be fully understood, but it is believed that wild boars were drawn to human settlements due to the availability of crops and waste for food (Zedder, 2012). The establishment of this interdependence between humans and animals resulted in priceless benefits for both as humans gained sustained access to animal food supplies, while domestic species were granted protection from predators and increased their population and geographical range far beyond that of their wild ancestors (Amills & Ramírez, 2010; Zeder *et al.*, 2006).

Domestic pigs are multipurpose animals that play a crucial role not only for people in rural villages but also in broader society through their contributions to food security and job creation. Beyond their agricultural significance, pigs are valuable in medical research, serving as models for human disease research (Gieling *et al.*, 2011). They are frequently used for the characterization of cardiovascular, diabetes, and dermatology drugs as their immune systems closely resemble that of humans (Ayuso *et al.*, 2020; Nunoya *et al.*, 2007; Pabst, 2020). Indeed, the domesticated pigs have significantly contributed to the development of agriculture, the widespread adoption of agricultural practices, and medicinal research globally. Understanding the evolution and dissemination of pigs can enhance our knowledge of the origins and expansion of livestock farming, as well as the emergence of early human civilizations.

2.3 Overview of Pig Production in South Africa: Status and Challenges

Production of livestock is of high economic importance, as it provides protein, solves unemployment, and earns foreign exchange (Taylor & Roese, 2006; Cole 1996). It enhances human nutrition, ensures food security, alleviates poverty, supports livelihoods, and generates employment opportunities for rural communities (Antwi & Seahlodi, 2011; Mergenthaler *et al.*, 2009). Of many livestock species, pig production plays a significant role, as pork is among the most consumed meats globally and the main source of animal-based protein for humans

(Ruan *et al.*, 2022). According to the FAO (2022), pork ranks as the second most consumed meat worldwide, accounting for 36% of total meat consumption, followed by poultry at 40% and beef at 22%, respectively (Kim *et al.*, 2023; Ruan *et al.*, 2022). However, with the ongoing increase in human population and economic development trends, animal protein demand, particularly pork, is anticipated to increase significantly. The World Economic Forum projects global meat consumption to double by 2050 (Faber *et al.*, 2020), and simultaneously, resource competition for feed, water, and land is likely to intensify due to environmental changes, requiring an increased pork supply. Against this global backdrop, the South African pig sector is a major contributor to the national agricultural economy. The country is home to approximately 1.3 million pigs, which together produce about 300,000 tons of pork (Adesehinwa *et al.*, 2024). The sector employs around 245,000 workers on commercial farms and in intensive units, supporting 1.45 million dependents (Meissner *et al.*, 2013). It has 170 formal sector pork producers, while 208,312 households participate in the informal pork sector (BFAP, 2020).

In terms of quality assurance, the country has made significant progress through initiatives such as the Pork 360 certification program. According to the Pork 360 annual report (2024), there are 151 certified units and 60 certified pig producers in South Africa. Pork 360 is a quality assurance program managed by the South African Pork Producers' Organisation (SAPPO) that ensures pork producers follow best practices throughout the production process, from farm to consumer. The program focuses on food safety, traceability, animal welfare, biosecurity, and environmental stewardship. While these structural improvements have greatly advanced the pig production sector, numerous challenges persist, particularly among smallholder and rural producers. Many smallholder farmers lack the necessary skills and resources, which results in poor management and elevated mortality rates (Munzhelele *et al.*, 2017). Pork quality and consumer acceptance are affected by pre-slaughter issues, emphasizing the necessity of better handling methods (Muchenje & Ndou, 2011).

Transboundary infectious diseases pose a major risk, with African swine fever being the most serious (Mokoele *et al.*, 2015). In rural areas, pigs are primarily raised for domestic use rather than for generating income, resulting in limited contribution to household income when compared to government grants (Gcumisa *et al.*, 2016). Productivity in these areas is further hindered by limited exposure to training, dependence on local breeds, swill-centered feeding methods, and poor access to veterinary services (Gcumisa *et al.*, 2016). Even where veterinary care is accessible, the associated costs are prohibitively high. To enhance the industry,

government incentives, improved disease surveillance, and control measures are required (Mokoele *et al.*, 2015; Munzhelele *et al.*, 2017).

2.4 Typology of South African Pig Production Systems

As modern domesticated pigs have been introduced and raised in numerous countries, including South Africa, pork is currently widely produced across South Africa (Visser, 2014). According to Visser (2014), the commercial pig industry in South Africa began in 1652 in the Cape of Good Hope. Today, three primary pig production systems are practiced in the country, which are intensive, semi-intensive, and extensive systems.

Pig production system includes buildings, equipment, production of feed ingredients, feed processing, and nutrient management. Most pig farmers in South Africa operate under intensive farming systems, while a smaller percentage practice semi-intensive or extensive farming. Extensive and semi-intensive systems are mainly practiced in rural populations, while the intensive system serves urban areas. **Table 2.1** summarizes the overview of the South African pig production system.

2.4.1 Intensive production

The intensive pig production system is the dominant commercial method of pig farming, with herd size and management determined by economic considerations. This system provides adequate housing with shade, pen space, and nutritionally balanced feed and clean water tailored to the specific requirements of pigs at different growth stages (Maes *et al.*, 2020; Matabane *et al.*, 2015). Farmers either grow or purchase feed, ensuring optimal nutrition to maximize productivity.

Selective breeding is a key component of intensive systems, used to improve desirable characteristics such as growth rate, feed conversion efficiency, and reproductive performance. Artificial insemination (AI) is frequently employed to enhance genetic quality (Krüger *et al.*, 2017). High-performance breeds or their crosses, such as Duroc, Large white, and South African Landrace, are used due to their superior productivity under intensive conditions (Hossain *et al.*, 2011). Indigenous pigs have traditionally been considered unsuitable for intensive commercial farming due to their slower growth and lower meat yields (Prolit, 2004).

Intensive pig production is costly, requires adequate capital, proper infrastructure, technology, and sufficient feed (Maes *et al.*, 2020). It is predominantly undertaken by commercial farmers and large agricultural businesses that supply the formal market.

2.4.2 Semi-intensive production

Semi-intensive production systems combine characteristics of both intensive and extensive systems. In this production system, pigs are provided with shelter (fence and roof) which protects them from extreme weather conditions, but have access to outside areas (Matabane *et al.*, 2015). This dual approach enables pigs to benefit from both controlled and natural environments, which may vary following fluctuations in climatic conditions, physiological state of the animal, or the type of market.

Semi-intensive production system typically keeps both commercial and indigenous pig breeds, and breeding systems are normally uncontrolled. Basic health care is practiced to prevent common diseases and protect them from outbreaks. However, due to having access to outside areas, biosecurity measures are compromised (Van't Klooster & Wingelaar, 2011). Feed in semi-intensive is usually a mix of commercial feed and kitchen waste, which assists in decreasing costs while ensuring sufficient nutrition (Van't Klooster & Wingelaar, 2011). This system is mostly used by smallholders and emerging farmers who have limited resources but aim to scale up production (Levy *et al.*, 2014; Munzhelele *et al.*, 2017).

2.4.3 Extensive production

The extensive pig production system is typically practiced by subsistence and smallholder farmers. It is characterized by low off-take and reproductive rates, high mortality rates, supplementary feeding, limited veterinary care, inadequate housing, and higher degrees of inbreeding (FAO, 2012).

In this system, pigs are left to roam freely, there is no shelter provision, and they are exposed to natural weather conditions with no protection from severe weather conditions (Matabane *et al.*, 2015; Van't Klooster & Wingelaar, 2011). Foraging on pastures and supplementing with the household kitchen waste is the primary feeding method (Van't Klooster & Wingelaar, 2011). Consequently, malnutrition of pigs is common, especially in sows (Lucović *et al.*, 2017).

Breeding and management practices in this system are uncontrolled, resulting in increased inbreeding. Indigenous pig breeds are commonly used due to their resilience and capability to withstand local environmental conditions (Halimani *et al.*, 2012). This system requires minimal and close to non-existent investment, making it accessible to resource-poor farmers in marginal areas (Mathobela *et al.*, 2024).

Table 2.1 Overview of South African pig production systems

Production System	Description
Intensive	Pigs are housed in proper housing with controlled temperatures, lighting and ventilation. They are sufficiently provided formulated feed and have access to proper veterinary care. Productivity and efficiency are prioritized (Madec <i>et al.</i> , 2010; Matabane <i>et al.</i> , 2015; Van't Klooster & Wingelaar, 2011).
Semi-intensive	Pigs are kept in housing structures with fence and roof. Temperature is not controlled. Feed is supplemented with kitchen waste, crop residue and limited commercial feed. Veterinary care is limited (Molotsi <i>et al.</i> , 2021; Munzhelele, 2015; Simbizi <i>et al.</i> , 2025).
Extensive	Pigs have no housing, roam freely. They forage for feed and sometimes supplemented with kitchen waste. Veterinary care is minimal, close to non-existence (Munzhelele <i>et al.</i> , 2017; Van't Klooster & Wingelaar, 2011).

2.5 Pig Breeds in South Africa

Over the past years, numerous pig breeds have been developed through both artificial and natural selection (Chen *et al.*, 2007). Currently, there are more than 730 pig breeds or lines worldwide, with approximately two-thirds located in China and Europe. Of those pig breeds, 58 are regarded as "transboundary" (found in more than one country), with 33 being international transboundary and 25 being regional transboundary breeds. Despite this diversity, the global distribution of pigs is dominated by five breeds originating from Europe or the United States (*i.e.*, Duroc, Hampshire, Landrace, Large white, and Pietrain) (FAO, 2006).

Pig breeds vary significantly in body colour, shape, size, ear carriage, prolificacy, behaviour, and other characteristics with which they are primarily classified. Morphological traits, such as tail shape and length, as well as the presence or absence of physical features like tusks, are also considered. Moreover, classifications are also based on the geographic regions where they are developed or raised (Brown *et al.*, 1996).

2.5.1 Commercial pig breeds

In South Africa, the primary commercial pig breeds include the Duroc, Large White, and Landrace. These breeds are predominantly utilised in the formal production sector due to their

desirable growth performance and high fecundity. In contrast, indigenous and undefined pigs tend to underperform compared to exotic breeds in terms of reproduction and growth traits, including litter size, litter and birth weight, weaning weight, and average daily weight gain. Additionally, they often have limited access to veterinary care (Halimani *et al.* 2010). Moreover, there are existing policy disincentives that tend to favour commercial breeds and discourage the production of indigenous pig breeds (Chimonyo & Dzama, 2007).

2.5.1.1 Duroc

The Duroc pig breed is believed to have been introduced to America by Christopher Columbus from Spain or Portugal. Its development took place in the United States (New England) in the early 19th century and late 1800s, through the crossing of four strains of red pig breeds. It was officially recognised as a distinct breed in the late 1800s (Taylor & Roese, 2005). The breed was first imported from Canada into South Africa around 1980/81 (Plug & Badenhorst, 2001; Halimani *et al.*, 2020).

The Duroc is a large-framed, late-maturing breed ideal for heavy carcass production. The forequarters, especially the head and neck, are light, and it has small, drooping ears. Its coat colour is a uniform rusty red, with shades ranging from dark red to golden red. They have a medium length and a slight dish of the face (Taylor & Roese, 2005). The breed is not very susceptible to stress. Duroc pigs are commonly used in commercial pig breeding worldwide due to their desirable traits of rapid growth, good meat quality, and high feed conversion efficiency (Chen *et al.*, 2022; DIAO *et al.*, 2018; Wang *et al.*, 2019).



Figure 2.2 Duroc boar (left) and Duroc sow with its piglets (right). Source: <https://genetic.by/en/duroc> (accessed 01 November 2025).

2.5.1.2 Large White

Large White pig breed originated from England, United Kingdom in the late 1700s and was officially acknowledged in England as a distinct breed in 1868 (Briggs, 1983; Jones, 1998; Taylor & Roese, 2005). The British first introduced it into South Africa in the 1890s. It is large-bodied and has pink skin with a white coat colour, a short snout, erect ears, and a curly tail.

Large White sows produce many piglets on average, and they make good mothers (Bosse *et al.*, 2014; Taylor & Roese, 2005). They are famous for their exceptional growth rates and feed efficiencies (Bosse *et al.*, 2014). These breeds are considered the ultimate dual-purpose breed, and when crossed with any other pure breed, will produce progeny of excellent bacon type.

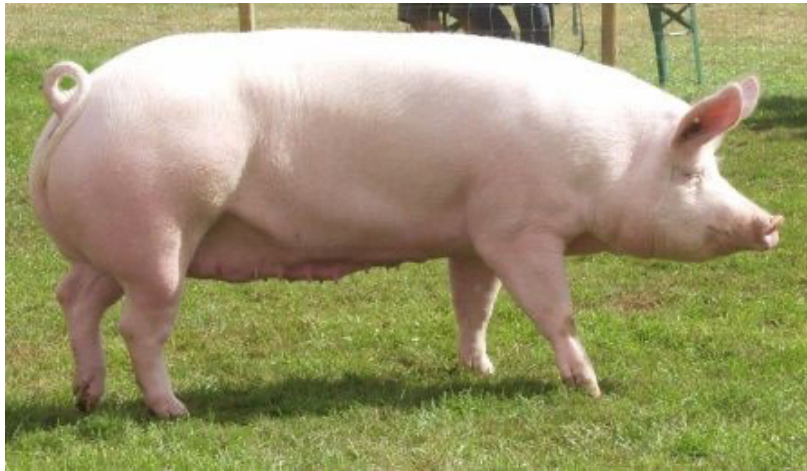


Figure 2.3 Large White sow. Source: <https://farmingsouthafrica.co.za/best-pig-breeds-to-farm-with-in-south-africa/> (accessed 25 November 2025).

2.5.1.3 South African Landrace

The South African Landrace ancestors were initially developed in Denmark through the crossbreeding of native Danish pigs and Large White pigs and began being transported to other countries from 1949 (Treacy, 1976). It was first brought to South Africa from Holland, Netherlands in 1952 (Briggs, 1983). It is currently the second most prominent commercial pig breed in the country (Visser *et al.*, 1993). It has an elongated body with a soft bristle white coat, dense and broad hams, straight back, and a small head with lopped ears (Smith *et al.*, 2014). It closely resembles its progenitor, the Large White pig breed. This breed is famous for its rapid growth, feed efficiency, and for regularly producing large litters.



Figure 2.4 South African Landrace boar. Source: <https://sasmallholder.co.za/2021/08/23/keeping-south-african-landrace-pigs/> (accessed 01 November 2025).

2.5.2 Indigenous pig breeds

South African indigenous pig breeds are generally classified as *Sus indica*, a typical Chinese pig (Nicholas, 1999). In South Africa, the two main recognized indigenous pig breeds are the Windsnyer and the Kolbroek, both which serve as a source of food and income in rural areas (Madzimore et al., 2013).

Indigenous pig breeds are hardy, can survive with minimal resources, and possess stronger immunity (Motsa'a *et al.*, 2021; Munzhelele *et al.*, 2017). They require less veterinary care and feed, and they can withstand harsh environmental conditions better, ideal for outdoor production in hot climates owing to their greater thermotolerance (Wilson, 2009). Consequently, these pigs are primarily kept by economically vulnerable groups in resource-poor households within marginal areas, where they serve as essential sources of food, income, and security. These households often cannot afford expensive vaccines and medicine and have limited access to veterinary care and extension services (Chiduwa *et al.*, 2008; Chikwanha *et al.*, 2007). Additionally, indigenous pigs are better able to ingest and digest fibrous feeds compared to exotic breeds (Kanengoni *et al.*, 2002, 2004).

Despite their advantages, they remain underutilised in the mainstream economy of South Africa. Indigenous pigs have long been considered not ideal for intensive commercial breeding due to their slow growth rate and poor meat yield (Prolit, 2004). However, the programme manager of the Pig Performance Scheme in Irene, South Africa, Visser (as described by Nicolas, 1999), proposed that breeding the indigenous breeds such as Kolbroek with exotic breeds like Duroc or Large White could result in reduced superfluous fat. Therefore, it is necessary to explore whether it is feasible to produce pork meat suitable for fresh consumption using indigenous pig breeds from South Africa.

2.5.2.1 Kolbroek

The exact origin of South Africa's indigenous pig, the Kolbroek, remains unclear; however, it exhibits some traits similar to those of traditional Chinese pig breeds. The name 'Kolbroek' is believed to have originated from Coalbrook, a British East India Company ship that was wrecked at Cape Hangklip near Betty's Bay in 1778. The ship had been carrying pigs on board that survived the wreck, and it is thought that the name Kolbroek came from those animals (Hoffman *et al.*, 2005). The Kolbroek pig is characterized by its "spotted" colour pattern, brownish coloration around the hindquarters (often referred to as "trousers"), and long snout. It is recognized for its adaptability, docile temperament, and notable resistance to stress (Nicolas, 1999). Kolbroek pigs are resilient, hardy and have a docile nature, scavenge outside

of homesteads for survival, thrive on high fibre diets, and have high disease tolerance (Visser, 2004).



Figure 2.5 Kolbroek pig breed. Source: <https://www.farmersweekly.co.za/app/uploads/2018/05/indigenous-Kolbroek-pig-Wessel-Pistorius.jpg> (accessed 01 November 2025).

2.5.2.2 Windsnyer

The Windsnyer pig breed is indigenous in Southern Africa. It is called Windsnyer (wind-cutter) because of its long nose and narrow body (Halimani *et al.*, 2010). It is black, has a solid muscular body (Smith *et al.*, 2014). Windsnyer pigs mature early with a mature weight of approximately 100 kg but grow at a slower rate compared to exotic breeds (Madzimure, 2011; Ncube *et al.*, 2003). It is known for its hardiness, scavenging abilities, and its capacity to survive on low-nutrient food through efficient conversion (Chimonyo *et al.*, 2010; Ndindana *et al.*, 2002).



Figure 2.6 A Windsnyer pig breed (boar). Adapted from Moyo, 2017

2.5.3 Village pig populations

The village pig populations are distinct to their geographic regions where they are raised, often possessing genetic traits that contribute valuable production and survival qualities to future breeding programs (Hlongwane *et al.*, 2020). According to Barnes & Fleischer (1998), these traits include adaptability to severe or harsh environments, disease resistance, and the ability to thrive under harsh production systems in developing countries. In South Africa, village pig breeds exemplify such pigs, possessing the qualities mentioned above. These pigs are of an unidentified lineage source, no selection is occurring amongst them, and their genetic pool is heterogeneous. There is minimal research done on village pig breeds; thus, they lack formal breed names, leading to them being classified as ecotypes.

Village pigs provide meat for household consumption and generate income (Gcumisa *et al.*, 2016). Despite their importance, they lack structured production, mating, and breeding systems. They are often inbred, resulting in elevated mortality rates. Southern African village pig farmers rely on locally accessible feed sources for nourishing their animals (Chikwanha *et al.*, 2007). However, these feed supplies are seasonal and insufficient to meet the year-round nutritional requirements and support their maintenance and production.



Figure 2.7 Village pig breeds kept under a semi-intensive production system (Source: Photograph taken by the author, October 2023, Mpumalanga)

2.6 Crossbreeding of Indigenous with Exotic Breeds

To improve the production performance of indigenous breeds, farmers often crossbreed them with exotic breeds to combine resilience and hardiness with superior reproductive and production traits (Halimani *et al.*, 2012; Visser, 2014). Indeed, research has demonstrated that breeding indigenous pig breeds with exotic, commercial pig breeds introduce advantageous genetic variants for reproduction, growth, and carcass quality among village pigs (Hlongwane *et al.*, 2024; Yang *et al.*, 2024). This admixture has the potential to improve economic traits in

village pigs, which are typically underutilised in the formal production sector (Hlongwane *et al.*, 2024). However, this crossbreeding practice raises concerns about conserving the genetic resources of indigenous pigs, as it may result to outbreeding depression, leading to reduced production and reproductive traits (Kirkpatrick, 2017; Madzimure *et al.*, 2010).

2.7 Pig genetic diversity

Genetic diversity refers to the variation in alleles and genotypes within a population, providing the basis for adaptation and evolutionary processes (Frankham *et al.*, 2002). Evolutionary forces, including mutation, adaptation, isolation, selective breeding, and genetic drift, have contributed significantly to the enormous diversity within local populations (Groeneveld *et al.*, 2010). Pigs have adapted to thrive in diverse environments through domestication and selection.

The genetic diversity of various animal breeds has declined over the past few decades, resulting to many breeds becoming lost, extinct, or endangered (Barker, 2001). The loss of genetic diversity within and across populations may lead to a possible loss of future economically significant genes in farm animals (Gerrits *et al.*, 2005). The development of effective programmes and methods for the genetic conservation and improvement of locally adapted indigenous populations is essential and requires an understanding of their genetic makeup.

2.8 Genetic Conservation of Indigenous and ‘Local’ Pig Breeds

2.8.1 Threats to Indigenous and Village Pig Populations

There are various threats to populations of indigenous and village pig breeds that could lead to extinction. These include replacing indigenous breeds with exotics, unbalanced admixture through uncontrolled crossbreeding, unclear, non-existent, or ill-defined policies, poverty, and lack of information (FAO, 2007; Halewood and Mugabe, 2002; Mendelsohn, 2003; Safaloah, 2001). The loss of a breed leads to the disappearance of unique adaptive traits, which are governed by complex interacting genes (Giuffra *et al.*, 2000).

2.8.2 Importance of Conservation of Animal Genetic Resources

Conservation of Animal Genetic Resources (AnGR) is crucial. AnGR refers to the total of human efforts (such as strategies, planning, management, policies, and actions) implemented to maintain the diversity of the farm’s animal genetic resources, ensuring preservation for both current and future agricultural and food production needs (Rege *et al.*, 2003). Conservation of indigenous and village pig breeds is a key point whenever animal production system development for smallholder farmers is discussed. The limited market participation of most pig farmers is one of the primary obstacles to the conservation of pig genetic resources in Southern Africa (Halimani *et al.*, 2020). Their economic importance primarily influences the

conservation of local breeds but also relies on other factors such as their socio-cultural value, ability to adapt to local agro-climatic conditions, role in developing local communities and marginal areas, and their scientific significance (Muñoz *et al.*, 2019). Although these breeds show economic and social relevance, they are threatened by inefficient utilisation (Muñoz *et al.*, 2018). This inefficiency is mainly due to local breeds being unable to compete with faster-growing exotic breeds, leading to indiscriminate crossbreeding that has reduced the genetic diversity of local breeds. However, many small-scale farmers do recognise the value and importance of local pigs and opt to preserve them (Halimani *et al.*, 2012a).

Indigenous pig breeds have exceptional qualities, including adaptability to harsh environmental conditions, disease resistance, and thrive under challenging production systems in developing countries (Motsa'a *et al.*, 2021). Indigenous populations may provide unique genetic variations that could be advantageous for producers in enhancing the diversity of commercial lines (Swart *et al.*, 2010). Conservation of indigenous and village pig populations is done to prevent genomic diversity loss that may occur from population decline, reducing the ability of pigs to adapt, survive, and thrive. Population size reduction leads to inbreeding and consequently results in loss of genomic diversity (Bosse & van Loon, 2022; Frankham *et al.*, 2002; Pinto *et al.*, 2023). Populations with higher genomic diversity are more equipped to withstand environmental changes and adapt successfully. Therefore, it is imperative to work on the conservation of indigenous pig breeds and the advancement of small-scale farmers' production systems to conserve their genetic potential. Moreover, to support breed promotion and sustainability, comprehensive phenotypic and genetic characterization is required to have a better understanding of their potential contributions to agricultural and commercial breeding practices.

2.8.3 Conservation Strategies: In Situ and Ex Situ Approaches

For conservation, two main strategies are used, which are *in situ* and *ex situ* conservation techniques (Bayer, 2006; Holt and Pickard, 1999). *In situ* conservation refers to the conservation of pigs through ensuring their use by farmers within their natural environments, allowing them to undergo ongoing dynamic breed development and evolution, and the maintenance of both agro-ecosystems and ecological balance. This conservation approach provides the opportunity for pigs to preserve their distinct adaptations like disease resistance, harsh environmental conditions tolerance and use of available local feed resources (Gandini & Oldenbroek, 2007). Due to its natural management methods, the small farmers play an essential role in this strategy.

The *ex-situ* conservation has two major methods, *in vivo* and cryoconservation. *Ex-situ in vivo* conservation refers to conserving live pig populations outside their natural environment, where they evolved. Cryo-conservation is the widely used *ex situ* conservation method, which refers to the keeping of biological samples such as semen and embryos to maintain their viability for decades. Gene banks serve as repositories for this type of conservation method. The *ex-situ* conservation method is important to guarantee long-term preservation of genetic material that could be lost through population decline, inbreeding, and environmental changes (Oldenbroek & Waaij, 2014). Both conservation strategies are critical for supporting the sustainable use of indigenous and local pig breeds in agriculture and for maintaining their potential to contribute to global food security and genetic improvement programs.

The decline of genetic diversity amongst and in animal breeds has led to the necessity for conservation of certain breeds (Ramsay *et al.*, 2000). Therefore, conservation efforts are required to minimise further loss of genetic diversity and the extinction of certain breeds. Phenotypic and genetic characterization, along with sustainable use of genetic resources and the implementation of structured, tailored breeding programs, have been proposed as complementary strategies to conserve threatened genetic diversity (Molotsi *et al.*, 2017).

AnGR are essential for food security and livelihoods globally, particularly in developing countries where approximately a billion people rely solely on livestock production (FAO, 2009). Preventing extinction and conserving livestock breeds are essential for the long-term sustainability and health of the global animal industry. The well-preserved animals are a useful resource for genes that can allow sustained genetic improvement as well as enable adaptation to varying breeding objectives and environments (Notter, 1999).

2.9 Porcine Genomics and Statistical Tools

2.9.1 Single Nucleotide Polymorphisms

The development and advancement of computational tools and high-throughput sequencing technologies have significantly advanced the study of livestock genomics. Researchers can now employ different genetic markers to investigate genetic diversity, reveal population structure and evolutionary histories, identify selection signatures, loci affecting quantitative and qualitative traits, and ascertain genomic regions linked to economically and environmentally significant traits due to these advancements. Researchers have utilised a variety of genetic markers, from microsatellites to high-throughput SNP data, to investigate diversity and inbreeding levels (Ba *et al.*, 2020; Ciobanu *et al.*, 2001; Lukić *et al.*, 2020; Van Ba *et al.*, 2020; Zhao *et al.*, 2021). Microsatellites, or Single Sequence Repeats (SSRs), have been widely used in numerous genetic studies of South African livestock, including cattle,

goats, chickens, and sheep (Mtileni *et al.*, 2011; Qwabe *et al.*, 2013; van der Westhuizen *et al.*, 2020; Visser *et al.*, 2011). However, the evolution of genotyping technologies and advancements in high-throughput sequencing have replaced SSRs with SNPs in animal genetic research over the past decade.

2.9.2 Porcine BeadChip

The Porcine BeadChip was initially introduced in 2009 as the Porcine SNP60K BeadChip, a powerful genome-wide SNP genotyping array containing 62,163 SNPs, specifically designed for pigs. It was later updated to the Porcine SNP80K BeadChip (Illumina, San Diego, CA, USA) around 2018, which has 68,528 SNPs and offers enhanced genomic coverage, with an average inter-SNP gap of 38 kb across porcine chromosomes (Ramos *et al.*, 2009; Xu *et al.*, 2023). The pigs' commercial SNP chips have been primarily created using the most heterogenous SNPs between a wild boar and four cosmopolitan breeds, but to separate breeds of interest, it is essential that a more specific subset of these SNPs must be chosen (Muñoz *et al.*, 2020). The SNP array has been widely employed to identify loci associated with both quantitative and qualitative traits, to reveal evolutionary history and population structure, and to assess the extent of linkage disequilibrium across the pig genome. Breed-specific informative SNP selection can eliminate redundant and noisy SNPs and lower the cost of breed identification when compared to commercial SNP chips.

2.9.3 Genetic Diversity Indices

Genetic diversity within a population is commonly assessed using molecular marker data through measures such as the proportion of polymorphic loci, effective population size (N_e), observed (H_O) and expected (H_E) heterozygosity (Boettcher *et al.*, 2015).

2.9.4 Population Structure and Pattern Analysis

For genetic datasets, it is especially important to have a clear understanding of the underlying structure and properties of the data (Patterson *et al.*, 2006). Population structure refers to the organization of genetic variation and is often characterized by variations in allele frequencies due to combined evolutionary processes that include gene flow, mutation, genetic drift, selection and demographic history (Andam *et al.*, 2017). Studying population structure is important as it assesses the genetic variation patterns within species across populations, identifies genetic differences that are geographically distributed, and how they are influenced by factors like migration, natural selection, and geographic barriers. Population structure analysis reveals the structure of the population based on its genetic makeup and is crucial for understanding evolutionary processes and conservation efforts (Andam *et al.*, 2017). Different methods are utilised in both animals and humans to study the genetic population structures.

Widely used methods include genomic principal component analysis (PCA) (Luca, 2007), Bayesian clustering methods (ADMIXTURE and STRUCTURE) (Patterson *et al.*, 2006), and population graph approaches implemented in software like TreeMix, which infers population splits and admixture events (Pickrell & Pritchard, 2012).

PCA has become a standard tool in genetics, and its application to genetic data is believed to provide a natural way of revealing population structure. The multivariate data in the majority of PCA applications has an unknown covariance, and the PCA attempts to select a subspace to project the data into to capture the majority of relevant information. PCA is now applied to analyse haplotype frequencies, in addition to microsatellites and SNPs, to study population structure (Patterson *et al.*, 2006). ADMIXTURE and STRUCTURE estimate allele frequencies and admixture proportions by assuming that the genotypes in the samples originate from one of "*K*" ancestral populations. Treemix uses large sets of SNPs to infer the historical relationships between populations, using a graph-based approach that allows migration events and population splits. The study conducted by Ocampo-Gallego *et al.* (2024) is an example of genetic research that integrated all the methodological approaches mentioned: the PCA, ADMIXTURE, and the population-graph framework implemented in TreeMix. In their analysis of four Colombian pig breeds and their relationships with other species, the authors detected moderate genetic differentiation and observed clear separation of animal groups driven by geographical isolation and distinct founding effects. Their findings also showed that Colombian Creole pigs share a strong genetic relationship with the Iberian pig, along with varying degrees of relatedness to other breeds.

Pairwise F_{ST} has emerged as the basis for accurately predicting population structure. Sewall Wright and Gustave Malécot, working independently, introduced F -statistics in the 1940s and 1950s, as a tool for describing the partitioning of genetic diversity within and among populations (Malécot, 1948; Wright, 1951). Wright's F -statistics, particularly F_{ST} , offer valuable insights into the evolutionary processes shaping genetic variation within and among populations. As one of the most widely used descriptive statistics in population and evolutionary genetics, F_{ST} can be used in identifying genomic regions under selection. Furthermore, comparing F_{ST} estimates across different genomic regions can reveal aspects of a population's demographic history, including patterns of divergence, migration, and genetic drift (Wright, 1951).

2.9.5 Effective Population Size

The concept of effective population size (N_e), originally introduced by Sewall Wright, remains central to animal and plant breeding, conservation genetics, molecular variation studies, and

evolutionary biology, as it quantifies the effects of genetic drift and inbreeding in populations (Allendorf et al., 2012; Fedorca *et al.*, 2024; Wang *et al.*, 2016). According to Jasielczuk *et al.* (2020), N_e is a genetic statistic that quantifies the level of genetic diversity and genetic drift within a population. Estimated N_e is the widely recommended estimator for assessing genetic threats in livestock (FAO, 1992; Gandini *et al.*, 2004) and is estimated based on the number of breeding animals, boars and sows within a population. Historically, N_e was calculated based on pedigree information; however, due to advancements in genomics technology, it can now also be measured from genetic information (Fabbri *et al.*, 2020). By understanding N_e and its patterns, one may monitor and control the genetic variation loss in populations.

2.9.6 Runs of Homozygosity

Runs of Homozygosity (ROH) are consecutive homozygous segments of the DNA sequence in diploid genomes (Gibson *et al.*, 2006). ROH has been used to quantify an individual's autozygosity and identify inbreeding levels as well as genetic relationships, providing support in estimating the true level of autozygosity at the individual and population levels (Ferencakovic *et al.*, 2011; Ferenčaković *et al.*, 2013; Kim *et al.*, 2015; Zavarez *et al.*, 2015). Autozygosity occurs when the progeny inherits shared chromosomal segments from their parents, who share a common ancestor. An individual inherits identical by descent (IBD) chromosomal fragments from both parents (Wright, 1922), resulting in homozygous segments in the genome of the progeny, causing ROH (Broman *et al.*, 1999). This can result from a variety of factors such as natural and artificial selection, population bottleneck, genetic drift, and mating of closely related individuals (Curik *et al.*, 2014; Keller *et al.*, 2011). Xie *et al.* (2019) used the Illumina Porcine SNP60K BeadChip to scan for ROHs and identify the candidate genes and genomic inbreeding coefficient in pig genomes (Landrace, Songliao black, and Yorkshire).

2.9.7 Identity by Descent

Identity by Descent (IBD) refers to two alleles or genomic segments at a specific chromosomal region inherited from a shared ancestor (Browning & Browning, 2012; Gusev *et al.*, 2009; Thompson, 2013). IBD analysis is a valuable genomic tool for identifying shared ancestry and estimating genetic relatedness within and between populations (Broman & Weber, 1999; Palamara *et al.*, 2012; Szpiech *et al.*, 2013). In livestock genetics, particularly in conservation efforts, IBD aids in detecting inbreeding patterns, measuring genomic diversity, and uncovering demographic histories, such as population bottlenecks or founder effects (Browning & Browning, 2012; Gusev *et al.*, 2009; Thompson, 2013). IBD-based metrics can reveal how closely related animals are in small, isolated, or poorly managed herds (Browning

& Browning, 2012; Palamara *et al.*, 2012). This information is essential for assessing the risk of inbreeding depression, evaluating genetic erosion, and guiding breeding and conservation strategies to maintain genetic diversity (Browning & Browning, 2012; Han & Abney, 2011; Thompson, 2013). Despite its usefulness, IBD analysis has rarely been applied to indigenous and village pig populations in South Africa, creating a significant research gap that this study aims to address.

2.9.8 Selection Signatures

Signatures positive of selection are genomic regions that have become more common and fixed in a population because of their functional significance in certain biological processes. These regions are identified by their lower genetic variability and distinct regional LD patterns (Pérez O'Brien *et al.*, 2014). Information on selection signatures is valuable for informing management strategies aimed at enhancing both production traits and adaptability in livestock populations. A series of methods are available to identify genomic regions under selection (Lewontin & Krakauer, 1973; Rubin *et al.*, 2010; Tajima, 1983), including F_{ST} (Weir & Cockerham, 1984), integrated haplotype score (iHS) (Sabeti *et al.*, 2007; Voight *et al.*, 2006), relative extended haplotype homozygosity (REHH), cross-population extended haplotype homozygosity (XP-EHH) (Sabeti *et al.*, 2007), and Haplotype-based F-statistics using Local Kinship correction (HapFLK) (Fariello *et al.*, 2013). The iHS test is a powerful method for the detection and characterization of genomic regions that have experienced recent selection within a population, particularly when the selected allele has risen to moderate frequencies but has not yet reached fixation (Tang *et al.*, 2007; Voight *et al.*, 2006). Selection signatures have been detected in pig populations linked to key traits, such as adaptation to high altitudes (Ai *et al.*, 2013), muscle growth (Wang *et al.*, 2014), and body size (Rubin *et al.*, 2012). Moreover, several researchers have highlighted selection in domestic pigs for productivity, tolerance, and disease resistance (D'Alessandro *et al.*, 2022; Floridia *et al.*, 2023; Sutera *et al.*, 2023).

2.9.9 Copy Number Variations

With advancements in DNA sequencing technologies and molecular biology techniques, genetic variations can be studied at the DNA level, including SNPs, copy number variations (CNVs), and large structural variations (Iafrate *et al.*, 2004). CNVs were first introduced in the early 2000s and results from mutations (insertion, deletion, duplication, translocation and inversion) of DNA fragments that are 1 kb or larger sizes in length available at variable copy number in comparison to the reference genome (Feuk *et al.*, 2006; Redon *et al.*, 2006). These mutations range in size from kilobases (kb) to megabases (Mb) in length and are commonly observed in both genomes of human and domestic animals (Feuk *et al.*, 2006). These genetic

variants serve as the foundation of biological evolution and are key contributors to genetic and phenotypic diversity that leads to the development of distinctive traits (Jia *et al.*, 2019). Investigating CNVs present in the genome is crucial to understanding the genetic diversity of the genomes, genomic structure, and differences (Xu *et al.*, 2023). Numerous CNVs are being identified with various genome analysis platforms. Computational programming tools like PennCNV (Wang *et al.*, 2007), QuantiSNP (Colella *et al.*, 2007), cnvPartition (Illumina) and CNV Workshop (Gai *et al.*, 2010) have been developed to identify CNVs from SNP array data. PennCNV was reported by (Winchester *et al.*, 2009) as the most accurate program for detecting CNVs for the Illumina platform. Recently, Xu *et al.* (2023) identified autosomal CNVs from indigenous and commercial pigs using Illumina Porcine 80K SNP BeadChip and Penn CNV software, analysing the distribution of CNVs within the pig genome and across different populations to thoroughly assess their population genetic background variations and genetic diversity. In pigs, CNV regions harbouring candidate genes responsible for many complex traits related to meat production and meat quality have been identified (Chen *et al.*, 2012).

2.11 Conclusion

Genetic diversity in pigs plays a crucial role in sustainable breeding and conservation efforts, and advances in genomic technologies have provided deeper insights into genetic variation, population structure, and breed relationships. However, several knowledge gaps remain in understanding the mating, breeding, and production systems of South African indigenous and village pigs, particularly under low-input, smallholder conditions. Additionally, comprehensive genomic analyses, including Identity by Descent (IBD) patterns, inbreeding trends, and candidate genes associated with economically and adaptively important traits, are scarce for these populations. The impacts of uncontrolled crossbreeding and population decline on genetic diversity and extinction risk have also been insufficiently quantified. Collectively, these gaps underscore the need for an integrative assessment combining socio-economic, phenotypic, and genomic approaches. This study addresses these gaps by analysing genetic diversity, admixture levels, extinction risk, functional genetic loss, inbreeding patterns, and candidate genes using genome-wide approaches, providing insights essential for the management, breeding, and conservation of South African village pig populations.

2.12 References

- Adesehinwa, A. O. K., Boladuro, B. A., Dunmade, A. S., Idowu, A. B., Moreki, J. C., & Wachira, A. M. (2024). Pig production in Africa: current status, challenges, prospects and opportunities. *Animal Bioscience*, 37(4), 730. <https://doi.org/10.5713/AB.23.0342>
- Ai, H., Huang, L., & Ren, J. (2013). Genetic Diversity, Linkage Disequilibrium and Selection Signatures in Chinese and Western Pigs Revealed by Genome-Wide SNP Markers. *PLOS ONE*, 8(2), e56001. <https://doi.org/10.1371/JOURNAL.PONE.0056001>

- Allendorf, F. W., Luikart, G., Aitken, S. N., & John Wiley, W. A. (2012). Conservation and the genetics of populations. <https://books.google.com/books?hl=en&lr=&id=VVploydG8jYC&oi=fnd&pg=PR1&ots=wEq3VrMblg&sig=dvTvPg--4wQjcb1Gq32TZDYU5Mo>
- Amills, M., & Ramírez, O. (2010). Origin and Genetic Diversity of Pig Breeds. <https://doi.org/10.1002/9780470015902.a0022884>
- Andam, C. P., Challagundla, L., Azarian, T., Hanage, W. P., & Robinson, D. A. (2017). Population Structure of Pathogenic Bacteria. In *Genetics and Evolution of Infectious Diseases: Second Edition* (pp. 51–70). Elsevier Inc. <https://doi.org/10.1016/B978-0-12-799942-5.00003-2>
- Antwi, M., & Seahlodi, P. (2011). Marketing Constraints Facing Emerging Small-Scale Pig Farmers in Gauteng Province, South Africa. *Journal of Human Ecology*, 36(1), 37–42. <https://doi.org/10.1080/09709274.2011.11906415>
- Ayuso, M., Buyssens, L., Stroe, M., Valenzuela, A., Allegaert, K., Smits, A., Annaert, P., Mulder, A., Carpentier, S., Van Ginneken, C., & Van Cruchten, S. (2020). The Neonatal and Juvenile Pig in Pediatric Drug Discovery and Development. *Pharmaceutics*, 13(1), 44. <https://doi.org/10.3390/PHARMACEUTICS13010044>
- Ba, N. V., Arakawa, A., Ishihara, S., Nam, L. Q., Thuy, T. T. T., Dinh, N. C., Ninh, P. H., Cuc, N. T. K., Kikuchi, K., Pham, L. D., & Taniguchi, M. (2020). Evaluation of genetic richness among Vietnamese native pig breeds using microsatellite markers. *Animal Science Journal*, 91(1), e13343. <https://doi.org/10.1111/ASJ.13343>
- Barnes, A., & Fleischer, J. (1998). Growth rate and carcass characteristics of indigenous (Ashanti Dwarf) pig. *Ghana Journal of Agricultural Science*, 31(2). <https://doi.org/10.4314/GJAS.V31I2.1935>
- Bayer, W. (2006). Deep frozen or alive and kicking? Different approaches to the conservation of farm animal diversity. In *Issue Papers: People, Food and Biodiversity*.
- BFAP. (2020). Agriculture and Agro-processing Master Plan - Market-led inclusive agriculture transformation. Pretoria: BFAP.
- Bosse, M., Megens, H. J., Madsen, O., Frantz, L. A. F., Paudel, Y., Crooijmans, R. P. M. A., & Groenen, M. A. M. (2014). Untangling the hybrid nature of modern pig genomes: a mosaic derived from biogeographically distinct and highly divergent *Sus scrofa* populations. *Molecular Ecology*, 23(16), 4089. <https://doi.org/10.1111/MEC.12807>
- Bovula, N., & Honours, B. (n.d.). Response in fertility of Windsnyer boars to increasing α -tocopherol supplementation.
- Briggs, H.M., 1983. International Pig Breed Encyclopedia. Elanco Animal Health, Indianapolis, USA.
- Broman, K. W., & Weber, J. L. (1999). Long homozygous chromosomal segments in reference families from the Centre d'Etude du Polymorphisme Humain. *American Journal of Human Genetics*, 65(6), 1493–1500. <https://doi.org/10.1086/302661>
- Browning, S. R., & Browning, B. L. (2012). Identity by descent between distant relatives: detection and applications. *Annual Review of Genetics*, 46, 617–633. <https://doi.org/10.1146/ANNUREV-GENET-110711-155534>
- Charlesworth, B., & Charlesworth, D. (2010). Elements of evolutionary genetics. <https://www.research.ed.ac.uk/en/publications/elements-of-evolutionary-genetics>
- Chen, C., Qiao, R., Wei, R., Guo, Y., Ai, H., Ma, J., Ren, J., & Huang, L. (2012). A comprehensive survey of copy number variation in 18 diverse pig populations and identification of candidate copy number variable genes associated with complex traits. *BMC Genomics*, 13(1), 1–10. <https://doi.org/10.1186/1471-2164-13-733/TABLES/2>
- Chen, K., Baxter, T., Muir, W. M., Groenen, M. A., & Schook, L. B. (2007). Genetic Resources, Genome Mapping and Evolutionary Genomics of the Pig (*Sus scrofa*). *International Journal of Biological Sciences*, 3(3), 153–165. <https://doi.org/10.7150/IJBS.3.153>
- Chen, Z., Teng, J., Diao, S., Xu, Z., Ye, S., Qiu, D., Zhang, Z., Pan, Y., Li, J., Zhang, Q., & Zhang, Z. (2022). Insights into the architecture of human-induced polygenic selection in Duroc pigs. *Journal of Animal Science and Biotechnology*, 13(1), 1–12. <https://doi.org/10.1186/S40104-022-00751-X/FIGURES/4>
- Chiduwa, G., Chimonyo, M., Halimani, T. E., Chisambara, S. R., & Dzama, K. (2008). Herd dynamics and contribution of indigenous pigs to the livelihoods of rural farmers in a semi-arid area of

- Zimbabwe. *Tropical Animal Health and Production*, 40(2), 125–136. <https://doi.org/10.1007/S11250-007-9071-8/METRICS>
- Chikwanha, O. C., Halimani, T. E., Chimonyo, M., Dzama, K., & Bhebhe, E. (2007). Seasonal changes in body condition scores of pigs and chemical composition of pig feed resources in a semi-arid smallholder farming area of Zimbabwe. *African Journal of Agricultural Research*, 2(9), 468–474. <http://www.academicjournals.org/AJAR>
- Chimonyo, M., & Dzama, K. (2007). Estimation of genetic parameters for growth performance and carcass traits in Mukota pigs. *Animal*, 1(3), 317–323. <https://doi.org/10.1017/S1751731107661849>
- Chimonyo, M., Bhebhe, E., Dzama, K., Halimani, T. E., & Kanengoni, A. (2005). Improving smallholder pig production for food security and livelihood of the poor in Southern Africa. In *African Crop Science Conference Proceedings* (Vol. 7, No. pt. 2 of 3, pp. 569-573).
- Chimonyo, M., Dzama, K., & Mapiye, C. (2010). Growth performance and carcass characteristics of indigenous Mukota pigs of Zimbabwe. *Tropical Animal Health and Production*, 42(5), 1001–1007. <https://doi.org/10.1007/S11250-009-9522-5>
- Ciobanu, D. C., Day, A. E., Nagy, A., Wales, R., Rothschild, M. F., & Plastow, G. S. (2001). Genetic variation in two conserved local Romanian pig breeds using type 1 DNA markers. *Genetics Selection Evolution*, 33(4), 417–432. <https://doi.org/10.1051/GSE:2001125>
- Colella, S., Yau, C., Taylor, J. M., Mirza, G., Butler, H., Clouston, P., Bassett, A. S., Seller, A., Holmes, C. C., & Ragoussis, J. (2007). QuantiSNP: an Objective Bayes Hidden-Markov Model to detect and accurately map copy number variation using SNP genotyping data. *Nucleic Acids Research*, 35(6), 2013–2025. <https://doi.org/10.1093/NAR/GKM076>
- Curik, I., Ferenčaković, M., & Sölkner, J. (2014). Inbreeding and runs of homozygosity: A possible solution to an old problem. *Livestock Science*, 166(1), 26–34. <https://doi.org/10.1016/J.LIVSCI.2014.05.034>
- D'Alessandro, E., Arfuso, F., Floridia, V., Tardiolo, G., Fazio, F., Giannetto, C., Piccione, G., & Zumbo, A. (2022). Different genotype and a liquid whey-supplemented diet influence the resilience of pigs through immune-modulation and anti-inflammatory response. *Frontiers in Veterinary Science*, 9, 1046101. <https://doi.org/10.3389/FVETS.2022.1046101/BIBTEX>
- Darwin, C. (1868). Darwin, C. R. 1868. The variation of animals and plants under domestication. London: John Murray. First edition, first issue. Volume 2. https://darwin-online.org.uk/converted/published/1868_Variation_F877/1868_Variation_F877.2.html
- Darwin C. On the Origins of the Species by Means of Natural Selection, Or the Preservation of Favoured Races in the Struggle for Life. UK: John Murray. 1859
- DIAO, S. qi, LUO, Y. yu, MA, Y. long, DENG, X., HE, Y. ting, GAO, N., ZHANG, H., LI, J. qi, CHEN, Z. mou, & ZHANG, Z. (2018). Genome-wide detection of selective signatures in a Duroc pig population. *Journal of Integrative Agriculture*, 17(11), 2528–2535. [https://doi.org/10.1016/S2095-3119\(18\)61984-7](https://doi.org/10.1016/S2095-3119(18)61984-7)
- Fabbri, M. C., Dadousis, C., & Bozzi, R. (2020). Estimation of Linkage Disequilibrium and Effective Population Size in Three Italian Autochthonous Beef Breeds. *Animals* 2020, Vol. 10, Page 1034, 10(6), 1034. <https://doi.org/10.3390/ANI10061034>
- Faber, I., Castellanos-Feijoó, N. A., Van de Sompel, L., Davydova, A., & Perez-Cueto, F. J. A. (2020). Attitudes and knowledge towards plant-based diets of young adults across four European countries. *Exploratory survey. Appetite*, 145. <https://doi.org/10.1016/J.APPET.2019.104498>
- FAO. (2006). Domestic Animal Diversity Information System (DAD-IS) | Food and Agriculture Organization of the United Nations. <https://www.fao.org/dad-is/en/>
- FAO. 1992. The role of ruminant livestock in food security in developing countries. FAO Committee on World Food Security, 17th Session, 23 to 27 March 1992, Rome, Italy.
- FAOSTAT. Crops and livestock products [Internet] Rome, Italy: Food and Agriculture Organization of the United Nations; c2023.
- Fariello, M. I., Boitard, S., Naya, H., SanCristobal, M., & Servin, B. (2013). Detecting signatures of selection through haplotype differentiation among hierarchically structured populations. *Genetics*, 193(3), 929–941. <https://doi.org/10.1534/GENETICS.112.147231>
- Ferencakovic, M., Hamzic, E., Gredler, B., ... I. C.-A., & 2011, undefined. (2011). Runs of homozygosity reveal genome-wide autozygosity in the Austrian Fleckvieh cattle. *Hrcak.Srce.HrM Ferencakovic, E Hamzic, B Gredler, I Curik, J SölknerAgriculturae Conspectus Scientificus, 2011•hrcak.Srce.Hr.* <https://hrcak.srce.hr/72108>

- Ferenčaković, M., Hamzić, E., Gredler, B., Solberg, T. R., Klemetsdal, G., Curik, I., & Sölkner, J. (2013). Estimates of autozygosity derived from runs of homozygosity: Empirical evidence from selected cattle populations. *Journal of Animal Breeding and Genetics*, 130(4), 286–293. <https://doi.org/10.1111/JBG.12012>
- Feuk, L., Carson, A. R., & Scherer, S. W. (2006). Structural variation in the human genome. *Nature Reviews. Genetics*, 7(2), 85–97. <https://doi.org/10.1038/NRG1767>
- Feuk, L., Marshall, C. R., Wintle, R. F., & Scherer, S. W. (2006). Structural variants: changing the landscape of chromosomes and design of disease studies. *Human Molecular Genetics*, 15 Spec No 1. <https://doi.org/10.1093/HMG/DDL057>
- Floridia, V., Giuffrè, L., Giosa, D., Arfuso, F., Aragona, F., Fazio, F., Chen, C., Song, C., Romeo, O., & D'Alessandro, E. (2023). Comparison of the Faecal Microbiota Composition Following a Dairy By-Product Supplemented Diet in Nero Siciliano and Large White × Landrace Pig Breeds. *Animals*, 13(14), 2323. <https://doi.org/10.3390/ANI13142323/S1>
- Frankham, R., Briscoe, D., & Ballou, J. (2002). Introduction to conservation genetics. [https://books.google.com/books?hl=en&lr=&id=F-XB8hqZ4s8C&oi=fnd&pg=PR13&dq=Frankham,+R.,+Briscoe,+D.+A.,+%26+Ballou,+J.+D.+\(2002\).+Introduction+to+conservation+genetics.+New+York,+NY:+Cambridge+University+Press.&ots=CNDhy14N4P&sig=cmJwJcPqfTwUMrNQ2KXtHSlfzXQ](https://books.google.com/books?hl=en&lr=&id=F-XB8hqZ4s8C&oi=fnd&pg=PR13&dq=Frankham,+R.,+Briscoe,+D.+A.,+%26+Ballou,+J.+D.+(2002).+Introduction+to+conservation+genetics.+New+York,+NY:+Cambridge+University+Press.&ots=CNDhy14N4P&sig=cmJwJcPqfTwUMrNQ2KXtHSlfzXQ)
- Frantz, L. A., Schraiber, J. G., Madsen, O., Megens, H.-J., Cagan, A., Bosse, M., Paudel, Y., M A Crooijmans, R. P., Larson, G., & M Groenen, M. A. (2015). Evidence of long-term gene flow and selection during domestication from analyses of Eurasian wild and domestic pig genomes. <https://doi.org/10.1038/ng.3394>
- Frantz, L., Meijaard, E., Gongora, J., Haile, J., Groenen, M. A. M., & Larson, G. (2016). The evolution of suidae. *Annual Review of Animal Biosciences*, 4(Volume 4, 2016), 61–85. <https://doi.org/10.1146/ANNUREV-ANIMAL-021815-111155/CITE/REFWORKS>
- Gai, X., Perin, J. C., Murphy, K., O'Hara, R., D'arcy, M., Wenocur, A., Xie, H. M., Rappaport, E. F., Shaikh, T. H., & White, P. S. (2010). CNV Workshop: An integrated platform for high-throughput copy number variation discovery and clinical diagnostics. *BMC Bioinformatics*, 11. <https://doi.org/10.1186/1471-2105-11-74>
- Gandini, G., & Oldenbroek, K. (2007). Strategies for moving from conservation to utilisation. In *Utilisation and conservation of farm animal genetic resources* (pp. 29-54). Wageningen Academic.
- Gcumisa, S. T., Oguttu, J. W., & Masafu, M. M. (2016). Pig farming in rural South Africa: A case study of uthukela district in KwaZulu-Natal. *Indian Journal of Animal Research*, 50(4), 614–620. <https://doi.org/10.18805/IJAR.11175>
- Gerrits, R. J., Lunney, J. K., Johnson, L. A., Pursel, V. G., Kraeling, R. R., Rohrer, G. A., & Dobrinsky, J. R. (2005). Perspectives for artificial insemination and genomics to improve global swine populations. *Theriogenology*, 63(2), 283–299. <https://doi.org/10.1016/J.THERIOGENOLOGY.2004.09.013>
- Gibson, J., Morton, N., genetics, A. C.-H. molecular, & 2006, undefined. (2006). Extended tracts of homozygosity in outbred human populations. *Academic.Oup.ComJ Gibson, NE Morton, A CollinsHuman Molecular Genetics*, 2006•academic.Oup.Com. <https://doi.org/10.1093/hmg/ddi493>
- Gieling, E. T., Schuurman, T., Nordquist, R. E., & van der Staay, F. J. (2011). The pig as a model animal for studying cognition and neurobehavioral disorders. *Current Topics in Behavioral Neurosciences*, 7(1), 359–383. https://doi.org/10.1007/7854_2010_112
- Giuffra, E., Kijas, J., Amarger, V., Carlborg, Ö., Genetics, J. J., & 2000, undefined. (2000). The origin of the domestic pig: independent domestication and subsequent introgression. *Academic.Oup.ComE Giuffra, JMH Kijas, V Amarger, Ö Carlborg, JT Jeon, L AnderssonGenetics*, 2000•academic.Oup.Com. <https://academic.oup.com/genetics/article-abstract/154/4/1785/6047989>
- Groeneveld, L. F., Lenstra, J. A., Eding, H., Toro, M. A., Scherf, B., Pilling, D., Negrini, R., Finlay, E. K., Jianlin, H., Groeneveld, E., & Weigend, S. (2010). Genetic diversity in farm animals – a review. *Animal Genetics*, 41(SUPPL. 1), 6–31. <https://doi.org/10.1111/J.1365-2052.2010.02038.X>

- Gusev, A., Lowe, J. K., Stoffel, M., Daly, M. J., Altshuler, D., Breslow, J. L., Friedman, J. M., & Pe'er, I. (2009). Whole population, genome-wide mapping of hidden relatedness. *Genome Research*, 19(2), 318–326. <https://doi.org/10.1101/GR.081398.108>
- Gusha, B., Palmer, A. R., & Renato. (2024). A Stochastic Frontier Analysis of the Performance of Livestock in the North-Eastern Cape Communal Rangelands, South Africa. *J. Agric. Ext. Gusha, Palmer &*, 52(1), 90–115. <https://doi.org/10.17159/2413-3221/2024/v52n1a14307>
- Halewood, M., & Mugabe, J. (2001, September). Policy incentives for on-farm conservation and use of Africa's biodiversity. In *Incentive Measures for Sustainable Use and Conservation of Agrobiodiversity. Experiences and Lessons from Southern Africa. Proceedings of a Workshop*, Lusaka, Zambia (pp. 73-88).
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2010). Pig genetic resource conservation: The Southern African perspective. *Ecological Economics*, 69(5), 944–951. <https://doi.org/10.1016/j.ecolecon.2010.01.005>
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2012a). Opportunities for conservation and utilisation of local pig breeds in low-input production systems in Zimbabwe and South Africa. *Tropical Animal Health and Production*, 45(1), 81–90. <https://doi.org/10.1007/S11250-012-0177-2/TABLES/9>
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2012b). Some insights into the phenotypic and genetic diversity of indigenous pigs in southern Africa. *South African Journal of Animal Science*, 42(5), 507–510. <https://doi.org/10.4314/SAJAS.V42I5.13>
- Halimani, T. E., Mapiye, O., Marandure, T., Januarie, D., Imbayarwo-Chikosi, V. E., & Dzama, K. (2020). Domestic Free-Range Pig Genetic Resources in Southern Africa: Progress and Prospects. *Diversity 2020*, Vol. 12, Page 68, 12(2), 68. <https://doi.org/10.3390/D12020068>
- Han, L., & Abney, M. (2011). Identity by Descent Estimation With Dense Genome-Wide Genotype Data. *Genetic Epidemiology*, 35(6), 557. <https://doi.org/10.1002/GEPI.20606>
- Hlongwane, N. L., Dzomba, E. F., Hadebe, K., van der Nest, M. A., Pierneef, R., & Muchadeyi, F. C. (2024). Identification of Signatures of Positive Selection That Have Shaped the Genomic Landscape of South African Pig Populations. *Animals*, 14(2), 236. <https://doi.org/10.3390/ANI14020236/S1>
- Hlongwane, N. L., Hadebe, K., Soma, P., Dzomba, E. F., & Muchadeyi, F. C. (2020). Genome Wide Assessment of Genetic Variation and Population Distinctiveness of the Pig Family in South Africa. *Frontiers in Genetics*, 11(May), 1–19. <https://doi.org/10.3389/fgene.2020.00344>
- Hoffman, L. C., Styger, W. F., Brand, T. S., & Muller, M. (2005). A comparison between the growth, yield and carcass characteristics of 2 South African indigenous pig breeds. 6. <http://www.sasas.co.za/Popular/Popular.html>
- Holt, W. V., & Pickard, A. R. (1999). Role of reproductive technologies and genetic resource banks in animal conservation. *Reviews of reproduction*, 4, 143-150.
- Hossain, M., Chakma, S., Khatun, M., Hasanuzzaman, M., Miah, M., & Biswas, M. (2011). Production systems of swine in the rural areas of Rangamati and Khagrachari districts of Bangladesh. *Bangladesh Journal of Animal Science*, 40(1–2), 28–33. <https://doi.org/10.3329/BJAS.V40I1-2.10787>
- Iafate, A. J., Feuk, L., Rivera, M. N., Listewnik, M. L., Donahoe, P. K., Qi, Y., Scherer, S. W., & Lee, C. (2004). Detection of large-scale variation in the human genome. *Nature Genetics*, 36(9), 949–951. <https://doi.org/10.1038/NG1416>
- Jasielczuk, I., Gurgul, A., Szmatoła, T., Semik-Gurgul, E., Pawlina-Tyszko, K., Szyndler-Nędza, M., Blicharski, T., Szulc, K., Skrzypczak, E., & Bugno-Poniewierska, M. (2020). Comparison of linkage disequilibrium, effective population size and haplotype blocks in Polish Landrace and Polish native pig populations. *Livestock Science*, 231, 103887. <https://doi.org/10.1016/J.LIVSCI.2019.103887>
- Jia, C., Wang, H., Li, C., Wu, X., Zan, L., Ding, X., Guo, X., Bao, P., Pei, J., Chu, M., Liang, C., & Yan, P. (2019). Genome-wide detection of copy number variations in polled yak using the Illumina BovineHD BeadChip. *BMC Genomics*, 20(1), 1–14. <https://doi.org/10.1186/S12864-019-5759-1/TABLES/3>
- Jones, G.F. (1998) Genetic Aspects of Domestication, Common Breeds and Their Origin. In: Rothschild, M.F. and Ruvinsky, A., Eds., *The Genetics of the Pig*, CAB International, Wallingford, 17-50.

- Kanengoni, A. T., Dzama, K., Chimonyo, M., Kusina, J., & Maswaure, S. M. (2002). Influence of level of maize cob meal on nutrient digestibility and nitrogen balance in Large White, Mukota and LW × M F1 crossbred pigs. *Animal Science*, 74(1), 127–134. <https://doi.org/10.1017/S1357729800052292>
- Kanengoni, A. T., Dzama, K., Chimonyo, M., Kusina, J., & Maswaure, S. M. (2004). Growth performance and carcass traits of Large White, Mukota and Large White × Mukota F1 crosses given graded levels of maize cob meal. *Animal Science*, 78(1), 61–66. <https://doi.org/10.1017/S1357729800053844>
- Keller, M. C., Visscher, P. M., & Goddard, M. E. (2011). Quantification of Inbreeding Due to Distant Ancestors and Its Detection Using Dense Single Nucleotide Polymorphism Data. *Genetics*, 189(1), 237–249. <https://doi.org/10.1534/GENETICS.111.130922>
- Khowa, A. A., Tsvuura, Z., Slotow, R., & Kraai, M. (2023). The utilisation of domestic goats in rural and peri-urban areas of KwaZulu-Natal, South Africa. *Tropical Animal Health and Production*, 55(3), 1–10. <https://doi.org/10.1007/S11250-023-03587-3/FIGURES/4>
- Kim, E. S., Sonstegard, T. S., Van Tassell, C. P., Wiggans, G., & Rothschild, M. F. (2015). The relationship between runs of homozygosity and inbreeding in Jersey cattle under selection. *PLoS ONE*, 10(7). <https://doi.org/10.1371/JOURNAL.PONE.0129967>
- Kim, S. W., Gormley, A., Jang, K. B., & Duarte, M. E. (2023). Current status of global pig production: an overview and research trends. *Animal Bioscience*, 37(4), 719. <https://doi.org/10.5713/AB.23.0367>
- Kirkpatrick, F. D. (2017). Crossbreeding in Beef Cattle. 471.
- Krüger, D. A., van Marle-Koster, E., & Theron, H. E. (2017). Comparison of on-farm progeny performances from local and imported boar semen used in the South African pig industry. *South African Journal of Animal Science*, 47(5), 688–696. <https://doi.org/10.4314/SAJAS.V47I5.12>
- Larson, G., Dobney, K., Albarella, U., Fang, M., Matisoo-Smith, E., Robins, J., Lowden, S., Finlayson, H., Brand, T., Willerslev, E., Rowley-Conwy, F., Andersson, L., & Cooper, A. (2005). Worldwide phylogeography of wild boar reveals multiple centers of pig domestication. *Science*, 307(5715), 1618–1621. https://doi.org/10.1126/SCIENCE.1106927/SUPPL_FILE/LARSON.SOM.PDF
- Larson, G., ecology, D. F.-A. review of, evolution, undefined, and, undefined, & 2014, undefined. (2014). The evolution of animal domestication. *Annualreviews.Org*, 66, 115–151. <https://doi.org/10.1146/annurev-ecolsys-120213-091620>
- Levy, M., Dewey, C., Weersink, A., Mutua, F., Carter, N., & Poljak, Z. (2014). Evaluating critical factors to the economic feasibility of semi-intensive pig rearing in western Kenya. *Tropical Animal Health and Production*, 46(5), 797–808. <https://doi.org/10.1007/S11250-014-0568-7/TABLES/7>
- Lewontin, R. C., & Krakauer, J. (1973). DISTRIBUTION OF GENE FREQUENCY AS A TEST OF THE THEORY OF THE SELECTIVE NEUTRALITY OF POLYMORPHISMS. *Genetics*, 74(1), 175–195. <https://doi.org/10.1093/GENETICS/74.1.175>
- Luca, D. (2007). Cluster analysis and PCA for modeling population structure.
- Lukić, B., Ferencaković, M., Šalamon, D., Čačić, M., Orehovački, V., Iacolina, L., Curik, I., & Cubric-Curik, V. (2020). Conservation Genomic Analysis of the Croatian Indigenous Black Slavonian and Turopolje Pig Breeds. *Frontiers in Genetics*, 11(March). <https://doi.org/10.3389/fgene.2020.00261>
- Luković, Z., D. Skorput, and D. Karolyi. 2017. Pig welfare at different production systems. In: *Proceedings of the 11th International Symposium Modern Trends in Livestock Production*, October 11-13 Belgrade Serbia, 169-177.
- Madec, F., Hurnik, D., Porphyre, V., & Cardinale, E. (2010). Good practices for biosecurity in the pig sector : issues and options in developing and transition countries.
- Madzimure, J. (2011). Climate change adaptation and economic valuation of local pig genetic resources in communal production systems of South Africa.
- Madzimure, J., Chimonyo, M., Zander, K., & Dzama, K. (2010). Breeding Practices Of Indigenous Pig Genetic Resources In Communal Production Systems Of South Africa. <https://www.researchgate.net/publication/265490610>
- Maes, D. G. D., Dewulf, J., Piñeiro, C., Edwards, S., & Kyriazakis, I. (2020). A critical reflection on intensive pork production with an emphasis on animal health and welfare. *Journal of Animal Science*, 98(Supplement_1), S15–S26. <https://doi.org/10.1093/JAS/SKZ362>

- Malécot, G. *Les Mathématiques de l'Hérédité* (Masson, Paris, 1948).
- Matabane, M. B., Nethenzheni, P., Thomas, R., Netshirovha, T. R., Norris, D., Nephawe, K. A., & Nedambale, T. L. (2015). *Applied Animal Husbandry & Rural Development. Appl. Anim. Husb. Rural Develop*, 8, 19–25.
- Matabane, M., Nethenzheni, P., & Thomas, R. (2015). Status of the smallholder pig farming sector in Gauteng Province of South Africa. <https://www.cabidigitallibrary.org/doi/full/10.5555/20163008053>
- Mathobela, R. M., Molotsi, A. H., Marufu, M. C., Strydom, P. E., & Mapiye, C. (2024). Transitioning opportunities for sub-Saharan Africa's small-scale urban pig farming towards a sustainable circular bioeconomy. *International Journal of Agricultural Sustainability*, 22(1). <https://doi.org/10.1080/14735903.2024.2315918>
- Meissner, H. H., Scholtz, M. M., & Engelbrecht, F. A. (2013). Sustainability of the South African Livestock Sector towards 2050 Part 2: Challenges, changes and required implementations. *South African Journal of Animal Science*, 43(3), 289–319. <https://doi.org/10.4314/SAJAS.V43I3.6>
- Mendelsohn, R. (2003). The challenge of conserving indigenous domesticated animals. *Ecological Economics*, 45(3), 501–510.
- Mergenthaler, M., Weinberger, K., & Qaim, M. (2009). The food system transformation in developing countries: A disaggregate demand analysis for fruits and vegetables in Vietnam. *Food Policy*, 34(5), 426–436. <https://doi.org/10.1016/J.FOODPOL.2009.03.009>
- Mokoale, J. M., van Rensburg, L. J., van Lochem, S., Bodenstein, H., du Plessis, J., Carrington, C. A. P., Tom Spencer, B., & Fasina, F. O. (2015). Overview of the perceived risk of transboundary pig diseases in South Africa. *Journal of the South African Veterinary Association*, 86(1), 9. <https://doi.org/10.4102/JSAVA.V86I1.1197>
- Molotsi, A. H., Cupido, M., & Hoffman, L. C. H. (2021). Characterization of smallholder pig production systems in the Western Cape, South Africa. *Tropical Animal Health and Production*, 53(2). <https://doi.org/10.1007/S11250-021-02649-8>
- Motsa'a, J. S., Defang, H. F., Hako, T. B. A., Ojong, E. T., Mube, K. H., Nguekem, C. L., Tagning, Z. P. D., Mouchili, M., & Keambou, T. C. (2021). *Applied Animal Husbandry & Rural Development. Appl. Anim. Husb. Rural Develop*, 14, 32–39.
- Moyo, D. (2017). Comparison of febrile responses, thermoregulation and skin morphology in the local Kolbroek, Windsnyer and exotic Large White breeds of pigs in South Africa.
- Muchenje, V., & Ndou, S. P. (2011). How pig pre-slaughter welfare affects pork quality and the pig industry. News and Industry Article, South African Pork Producers Organization (SAPPO), 1–44.
- Muñoz, M., Bozzi, R., García, F., Núñez, Y., Geraci, C., Crovetto, A., García-Casco, J., Alves, E., Škrlep, M. S., Charneca, R., Martins, J. M., Quintanilla, R., Tibau, J., Kušec, G., Djurkin-Kušec, I., Mercat, M. J., Riquet, J., Estellé, J., Zimmer, C., ... Nazionale Allevatori Suini, A. (2018). Diversity across major and candidate genes in European local pig breeds. <https://doi.org/10.1371/journal.pone.0207475>
- Muñoz, M., Bozzi, R., García-Casco, J., Núñez, Y., Ribani, A., Franci, O., García, F., Škrlep, M., Schiavo, G., Bovo, S., Utzeri, V. J., Charneca, R., Martins, J. M., Quintanilla, R., Tibau, J., Margeta, V., Djurkin-Kušec, I., Mercat, M. J., Riquet, J., ... Óvilo, C. (2019). Genomic diversity, linkage disequilibrium and selection signatures in European local pig breeds assessed with a high density SNP chip. *Scientific Reports*, 9(1), 1–14. <https://doi.org/10.1038/s41598-019-49830-6>
- Muñoz, M., García-Casco, J. M., Alves, E., Benítez, R., Barragán, C., Caraballo, C., Fernández, A. I., García, F., Núñez, Y., Óvilo, C., Fernández, A., Rodríguez, C., & Silió, L. (2020). Development of a 64 SNV panel for breed authentication in Iberian pigs and their derived meat products. *Meat Science*, 167. <https://doi.org/10.1016/J.MEATSCI.2020.108152>
- Munzhelele, P. 2015. Evaluation of the production systems and constraints of smallholder pig farming in three agro-ecological zones of Mpumalanga province, South Africa. MSc (Agric) thesis, University of South Africa.
- Munzhelele, P., Oguttu, J., Fasanmi, O. G., & Fasina, F. O. (2017). Production constraints of smallholder pig farms in agro-ecological zones of Mpumalanga, South Africa.

- Nahar, N., Uddin, M., Gurley, E. S., Jahangir Hossain, M., Sultana, R., & Luby, S. P. (2015). Cultural and Economic Motivation of Pig Raising Practices in Bangladesh. *EcoHealth*, 12(4), 611. <https://doi.org/10.1007/S10393-015-1046-Z>
- Ncube, M., Dzama, K., Chimonyo, M., Kanengoni, A., & Hamudikuwanda, H. (2003). Effect of boar genotype on reproductive performance of the local sows of Zimbabwe. <https://hdl.handle.net/10568/66703>
- Ndindana, W., Dzama, K., Ndiweni, P. N. B., Maswaure, S. M., & Chimonyo, M. (2002). Digestibility of high fibre diets and performance of growing Zimbabwean indigenous Mukota pigs and exotic Large White pigs fed maize based diets with graded levels of maize cobs. *Animal Feed Science and Technology*, 97(3–4), 199–208. [https://doi.org/10.1016/S0377-8401\(01\)00345-5](https://doi.org/10.1016/S0377-8401(01)00345-5)
- Nicholas, G., 1999. Kolbroek – the unique local breed. *Farmer's Weekly*, August.
- Nunoya, T., Shibuya, K., Saitoh, T., Yazawa, H., Nakamura, K., Baba, Y., & Hirai, T. (2007). Use of Miniature Pig for Biomedical Research, with Reference to Toxicologic Studies. *J Toxicol Pathol*, 20, 125–132.
- Ocampo-Gallego, R. J., Vargas, Y. G., & Flórez, J. C. R. (2024). Genome-wide diversity, linkage disequilibrium, and admixture in the main Colombian Creole pig breeds. *Tropical Animal Health and Production*, 56(8), 336. <https://doi.org/10.1007/S11250-024-04140-6/TABLES/5>
- Oldenbroek, K., & Waaij, L. van der. (2014). Animal breeding and genetics for BSc students: Textbook animal breeding. Groen Kennisnet. <https://research.wur.nl/en/publications/animal-breeding-and-genetics-for-bsc-students-textbook-animal-bre>
- Pabst, R. (2020). The pig as a model for immunology research. *Cell and Tissue Research*, 380(2), 287. <https://doi.org/10.1007/S00441-020-03206-9>
- Palamara, P. F., Lencz, T., Darvasi, A., & Pe'er, I. (2012). Length Distributions of Identity by Descent Reveal Fine-Scale Demographic History. *American Journal of Human Genetics*, 91(5), 809. <https://doi.org/10.1016/J.AJHG.2012.08.030>
- Patterson, N., Price, A. L., & Reich, D. (2006). Population structure and eigenanalysis. *PLoS Genet*, 2(12), 190. <https://doi.org/10.1371/journal.pgen.0020190>
- Pérez O'Brien, A. M., Utsunomiya, Y. T., Mészáros, G., Bickhart, D. M., Liu, G. E., Van Tassell, C. P., Sonstegard, T. S., Da Silva, M. V., Garcia, J. F., & Sölkner, J. (2014). Assessing signatures of selection through variation in linkage disequilibrium between taurine and indicine cattle. *Genetics, Selection, Evolution : GSE*, 46(1), 19. <https://doi.org/10.1186/1297-9686-46-19>
- Pickrell, J. K., & Pritchard, J. K. (2012). Inference of population splits and mixtures from genome-wide allele frequency data. *Nature Precedings* 2012, 1–1. <https://doi.org/10.1038/npre.2012.6956.1>
- Plug, I & Badenhorst, S. (2001). The distribution of macromammals in Southern Africa over the past 30 000 Years: as reflected in animal remains from archaeological sites. Present and historical distributions of macromammals in southern Africa. *Order-Artiodactyla*. Transvaal Museum Monographs, 12(1)
- Pork 360, 2024 - The assured mark of South African pork. Available from: <https://pork360.co.za/>
- Price, M., & Hongo, H. (2020). The Archaeology of Pig Domestication in Eurasia. *Journal of Archaeological Research*, 28(4), 557–615. <https://doi.org/10.1007/S10814-019-09142-9/METRICS>
- Prolit, 2004. Pig Farming. Project Literacy Productions, Cape Town, South Africa
- Ramos, A. M., Crooijmans, R. P. M. A., Affara, N. A., Amaral, A. J., Archibald, A. L., Beever, J. E., Bendixen, C., Churcher, C., Clark, R., Dehais, P., Hansen, M. S., Hedegaard, J., Hu, Z. L., Kerstens, H. H., Law, A. S., Megens, H. J., Milan, D., Nonneman, D. J., Rohrer, G. A., ... Groenen, M. A. M. (2009). Design of a High Density SNP Genotyping Assay in the Pig Using SNPs Identified and Characterized by Next Generation Sequencing Technology. *PLOS ONE*, 4(8), e6524. <https://doi.org/10.1371/JOURNAL.PONE.0006524>
- Ramsay, K.A., Reed, D.S., Bothma, A.J. & Lepen, J.M., 1994. Profitable and environmentally effective farming with early domesticated livestock in Southern Africa, Department of Agriculture, Pretoria.
- Rangoma, M. (2013). Some credit facilities available to livestock farmers in Kenya. Research Institute, India
- Redding RW. (2005). Breaking the mold, a consideration of variation in the evolution of animal domestication. In: Vigne JD, Peters J, Helmer D, (eds) *The First Steps of Animal Domestication*. Oxbow Books: Oxford. pp 41–48.

- Redon, R., Ishikawa, S., Fitch, K. R., Feuk, L., Perry, G. H., Andrews, T. D., Fiegler, H., Shapero, M. H., Carson, A. R., Chen, W., Cho, E. K., Dallaire, S., Freeman, J. L., González, J. R., Gratacòs, M., Huang, J., Kalaitzopoulos, D., Komura, D., MacDonald, J. R., ... Hurles, M. E. (2006). Global variation in copy number in the human genome. *Nature*, 444(7118), 444–454. <https://doi.org/10.1038/NATURE05329>
- Reed, C. A. (2017). The pattern of animal domestication in the prehistoric Near East. *The Domestication and Exploitation of Plants and Animals*, 361–380. <https://doi.org/10.4324/9781315131825-33>
- Rege, J., economics, J. G.-E., & 2003, undefined. (2003). Animal genetic resources and economic development: issues in relation to economic valuation. ElsevierJEO Rege, JP GibsonEcological Economics, 2003•Elsevier. https://www.sciencedirect.com/science/article/pii/S0921800903000879?casa_token=r7nn0cI1cVEAAAAA:VtwRX-dn_ZmTnDf4djSpCBIDYi6jHhq2g5VbSb4nN9hqmBhNQMxo1Vsus4iP722WqmYqffg6iA
- Ruan, D., Yang, J., Zhuang, Z., Ding, R., Huang, J., Quan, J., Gu, T., Hong, L., Zheng, E., Li, Z., Cai, G., Wang, X., & Wu, Z. (2022). Assessment of Heterozygosity and Genome-Wide Analysis of Heterozygosity Regions in Two Duroc Pig Populations. *Frontiers in Genetics*, 12, 812456. <https://doi.org/10.3389/FGENE.2021.812456/BIBTEX>
- Rubin, C. J., Megens, H. J., Barrio, A. M., Maqbool, K., Sayyab, S., Schwochow, D., Wang, C., Carlborg, Ö., Jern, P., Jørgensen, C. B., Archibald, A. L., Fredholm, M., Groenen, M. A. M., & Andersson, L. (2012). Strong signatures of selection in the domestic pig genome. *Proceedings of the National Academy of Sciences of the United States of America*, 109(48), 19529–19536. https://doi.org/10.1073/PNAS.1217149109/SUPPL_FILE/ST05.DOCX
- Rubin, C. J., Zody, M. C., Eriksson, J., Meadows, J. R. S., Sherwood, E., Webster, M. T., Jiang, L., Ingman, M., Sharpe, T., Ka, S., Hallböök, F., Besnier, F., Carlborg, R., Bedhom, B., Tixier-Boichard, M., Jensen, P., Siegel, P., Lindblad-Toh, K., & Andersson, L. (2010). Whole-genome resequencing reveals loci under selection during chicken domestication. *Nature* 2010 464:7288, 464(7288), 587–591. <https://doi.org/10.1038/nature08832>
- Ruvinsky, A., Rothschild, M. F., Larson, G., & Gongora, J. (2011). Systematics and evolution of the pig. *The Genetics of the Pig: Second Edition*, 1–13. <https://doi.org/10.1079/9781845937560.0001>
- Sabeti, P. C., Varilly, P., Fry, B., Lohmueller, J., Hostetter, E., Cotsapas, C., Xie, X., Byrne, E. H., McCarroll, S. A., Gaudet, R., Schaffner, S. F., Lander, E. S., Frazer, K. A., Ballinger, D. G., Cox, D. R., Hinds, D. A., Stuve, L. L., Gibbs, R. A., Belmont, J. W., ... Stewart, J. (2007). Genome-wide detection and characterization of positive selection in human populations. *Nature* 2007 449:7164, 449(7164), 913–918. <https://doi.org/10.1038/nature06250>
- Safalaoh, A. C. L. (2001). Village chicken upgrading programme in Malawi. *World's Poultry Science Journal*, 57(2), 179–188.
- Simbizi, V., Moerane, R., & Gummow, B. (2025). The Role of Smallholder Pig Farmers in the Biosecurity of Pig Diseases in the Eastern Cape Province of South Africa. *Veterinary Medicine International*, 2025(1), 4755096. <https://doi.org/10.1155/VMI/4755096>
- Smith, D. B., Simmonds, P., Jameel, S., Emerson, S. U., Harrison, T. J., Meng, X. J., Okamoto, H., Van der Poel, W. H. M., & Purdy, M. A. (2014). Consensus proposals for classification of the family Hepeviridae. *The Journal of General Virology*, 95(Pt 10), 2223–2232. <https://doi.org/10.1099/VIR.0.068429-0>
- Sutera, A. M., Arfuso, F., Tardiolo, G., Riggio, V., Fazio, F., Aiese Cigliano, R., Paytuví, A., Piccione, G., & Zumbo, A. (2023). Effect of a Co-Feed Liquid Whey-Integrated Diet on Crossbred Pigs' Fecal Microbiota. *Animals*, 13(11), 1750. <https://doi.org/10.3390/ANI13111750/S1>
- Swart, H., Kotze, A., Olivier, P. A. S., & Grobler, J. P. (2010). Microsatellite-based characterization of Southern African domestic pigs (*Sus scrofa domestica*). In *South African Journal of Animal Science* (Issue 2). <http://www.sasas.co.za/sajas.asp>
- Szpiech, Z. A., Xu, J., Pemberton, T. J., Peng, W., Zöllner, S., Rosenberg, N. A., & Li, J. Z. (2013). Long runs of homozygosity are enriched for deleterious variation. *American Journal of Human Genetics*, 93(1), 90–102. <https://doi.org/10.1016/j.ajhg.2013.05.003>
- Tajima, F. (1983). EVOLUTIONARY RELATIONSHIP OF DNA SEQUENCES IN FINITE POPULATIONS. *Genetics*, 105(2), 437–460. <https://doi.org/10.1093/GENETICS/105.2.437>

- Tang, K., Thornton, K. R., & Stoneking, M. (2007). A New Approach for Using Genome Scans to Detect Recent Positive Selection in the Human Genome. *PLOS Biology*, 5(7), e171. <https://doi.org/10.1371/JOURNAL.PBIO.0050171>
- Taruvunga, A., Kambanje, A., Mushunje, A., & Mukarumbwa, P. (2022). Determinants of livestock species ownership at household level: Evidence from rural OR Tambo District Municipality, South Africa. *Pastoralism*, 12(1), 1–11. <https://doi.org/10.1186/S13570-021-00220-6/TABLES/3>
- Taylor, G., & Roese, G. (2005). Breeds of pigs - Duroc. www.dpi.nsw.gov.au
- Treacy, D. A. (1976). A genetic analysis of the pedigree Landrace pig breed in Australia. *Australian Journal of Experimental Agriculture*, 16(78), 76–81. <https://doi.org/10.1071/EA9760076>
- Van Ba, N., Nam, L. Q., Do, D. N., Van Hau, N., & Pham, L. D. (2020). An assessment of genetic diversity and population structures of fifteen Vietnamese indigenous pig breeds for supporting the decision making on conservation strategies. *Tropical Animal Health and Production*, 52(3), 1033–1041. <https://doi.org/10.1007/S11250-019-02090-Y/FIGURES/3>
- van't Klooster, J. H., & Wingelaar, A. (2011). Pig farming in the tropics: from free range to small-scale intensive production systems.
- Van der Bank, F. H., Groenewald, M., & Kotze, A. (1997). Biochemical polymorphism in five pig breeds in South Africa. *South African Journal of Animal Science*, 27(3).
- Visser, D. (2014). Modern Pig Production - Danie Visser - Google Books. <https://books.google.co.za/books?id=mpXorQEACAAJ>
- Visser, D.P., Browne, A. & Botes, F.DuT., 1993. Breeds and breeding programs. In: Pig Production in South Africa. Ed. Kemm, E.H., Agricultural Research Council. V & R Printers, Pretoria. pp. 19-20.
- Voight, B. F., Kudaravalli, S., Wen, X., & Pritchard, J. K. (2006). A Map of Recent Positive Selection in the Human Genome. *PLOS Biology*, 4(3), e72. <https://doi.org/10.1371/JOURNAL.PBIO.0040072>
- Wang, J., Ren, Q., Hua, L., Chen, J., Zhang, J., Bai, H., Li, H., Xu, B., Shi, Z., Cao, H., Xing, B., & Bai, X. (2019). Comprehensive Analysis of Differentially Expressed mRNA, lncRNA and circRNA and Their ceRNA Networks in the Longissimus Dorsi Muscle of Two Different Pig Breeds. *International Journal of Molecular Sciences* 2019, Vol. 20, Page 1107, 20(5), 1107. <https://doi.org/10.3390/IJMS20051107>
- Wang, K., Li, M., Hadley, D., Liu, R., Glessner, J., Grant, S. F. A., Hakonarson, H., & Bucan, M. (2007). PennCNV: An integrated hidden Markov model designed for high-resolution copy number variation detection in whole-genome SNP genotyping data. *Genome Research*, 17(11), 1665–1674. <https://doi.org/10.1101/GR.6861907>
- Wang, Z., Chen, Q., Yang, Y., Yang, H., He, P., Zhang, Z., Chen, Z., Liao, R., Tu, Y., Zhang, X., Wang, Q., & Pan, Y. (2014). A genome-wide scan for selection signatures in Yorkshire and Landrace pigs based on sequencing data. *Animal Genetics*, 45(6), 808–816. <https://doi.org/10.1111/AGE.12229>
- Weir, B. S., & Cockerham, C. C. (1984). Estimating F-Statistics for the Analysis of Population Structure. *Evolution*, 38(6), 1358. <https://doi.org/10.2307/2408641>
- Wilson, R. T. (2009). Fit for purpose - the right animal in the right place. *Tropical Animal Health and Production*, 41(7), 1081–1090. <https://doi.org/10.1007/S11250-008-9274-7/FIGURES/5>
- Winchester, L., Yau, C., & Ragoussis, J. (2009). Comparing CNV detection methods for SNP arrays. *Briefings in Functional Genomics*, 8(5), 353–366. <https://doi.org/10.1093/BFGP/ELP017>
- Wright, D. (1922). Coefficients of Inbreeding and Relationship. *The American Naturalist*, 56(645), 330–338. <https://doi.org/10.1086/279872>
- Wright, S. The genetical structure of populations. *Ann. Eugen.* 15, 323–354 (1951).
- Xie, R., Shi, L., Liu, J., Deng, T., Wang, L., Liu, Y., & Zhao, F. (2019). Genome-Wide Scan for Runs of Homozygosity Identifies Candidate Genes in Three Pig Breeds. *Animals* 2019, Vol. 9, Page 518, 9(8), 518. <https://doi.org/10.3390/ANI9080518>
- Xu, C., Zhang, W., Jiang, Y., Zhou, M., Liu, L., Su, S., Li, X., & Wang, C. (2023). Genome-Wide Detection and Analysis of Copy Number Variation in Anhui Indigenous and Western Commercial Pig Breeds Using Porcine 80K SNP BeadChip. *Genes*, 14(3). <https://doi.org/10.3390/genes14030654>
- Yang, R., Jin, S., Fang, S., Yan, D., Zhang, H., Nie, J., Liu, J., Lv, M., Zhang, B., & Dong, X. (2024). Genetic introgression from commercial European pigs to the indigenous Chinese Lijiang breed

- and associated changes in phenotypes. *Genetics Selection Evolution*, 56(1), 1–15. <https://doi.org/10.1186/S12711-024-00893-8/FIGURES/4>
- Zavarez, L. B., Utsunomiya, Y. T., Carmo, A. S., Neves, H. H. R., Carneiro, R., Ferencakovic, M., Pérez O'Brien, A. M., Curik, I., Cole, J. B., Van Tassell, C. P., da Silva, M. V. G. B., Sonstegard, T. S., Sölkner, J., & Garcia, J. F. (2015). Assessment of autozygosity in Nellore cows (*Bos indicus*) through high-density SNP genotypes. *Frontiers in Genetics*, 5(JAN). <https://doi.org/10.3389/FGENE.2015.00005/FULL>
- Zeder, M. A., Emshwiler, E., Smith, B. D., & Bradley, D. G. (2006). Documenting domestication: the intersection of genetics and archaeology. *Trends in Genetics: TIG*, 22(3), 139–155. <https://doi.org/10.1016/J.TIG.2006.01.007>
- Zeder MA. (2012). Pathways to domestication. In: Gepts P, Famula RL, Bettinger RL, Brush SB, Damania AB, McGuire PE, Qualset CO, (eds) *Biodiversity in Agriculture: Domestication, Evolution, and Sustainability*. Cambridge University Press: Cambridge. pp 227–259.
- Zhang, M., Liu, Y., Li, Z., Lü, P., Gardner, J. D., Ye, M., Wang, J., Yang, M., Shao, J., Wang, W., Dai, Q., Cao, P., Yang, R., Liu, F., Feng, X., Zhang, L., Li, E., Shi, Y., Chen, Z., ... Fu, Q. (2022). Ancient DNA reveals the maternal genetic history of East Asian domestic pigs. *Journal of Genetics and Genomics*, 49(6), 537–546. <https://doi.org/10.1016/J.JGG.2021.11.014>
- Zhao, Y., Zhu, X., Jiang, Y., Li, Z., Li, X., Xu, W., Wei, H., Li, Y., & Li, X. (2021). Genetic diversity and variation of seven Chinese grass shrimp (*Palaemonetes sinensis*) populations based on the mitochondrial COI gene. *BMC Ecology and Evolution*, 21(1), 1–10. <https://doi.org/10.1186/S12862-021-01893-8/FIGURES/4>

3 PRODUCTION, BREEDING AND MATING SYSTEMS IN VILLAGE PIG FARMING: IMPLICATIONS FOR GENETIC DIVERSITY

3.1 Abstract

Pork is consumed worldwide and contributes to the primary agricultural sector and national economy. In South Africa, pork production is practiced across all nine provinces, including village communities. However, village pig production in South Africa is neglected and understudied due to its minimal contribution to the formal economy and market systems. This study examined the mating, breeding, and production systems of village pigs across six provinces. Data were collected from 236 households in Eastern Cape ($n = 69$), Gauteng ($n = 24$), KwaZulu-Natal ($n = 30$), Limpopo ($n = 70$), Mpumalanga ($n = 15$), and North West ($n = 28$) using semi-structured questionnaires, recorded in Excel, and analysed with SPSS and RStudio. Poor record-keeping was a major challenge, making data collection dependent on farmers' knowledge. Descriptive statistics revealed that most households were male-headed (56.4%) with household heads predominantly aged 41–60 years (47.1%). Only 18.2% of households kept pigs exclusively, while most practiced mixed livestock farming. Pigs were primarily managed under semi-intensive production (50.4%), with 71.2% using stalls or sheds, 14.4% using kraals, 11.4% using yards, and 3.0% providing no housing. Pigs were mainly kept for household meat consumption and sales, with Large White (29.4%) and Windsnyer (29.4%) being the most common breeds. Breeding practices included hand-mating (47.6%) and uncontrolled mating (38.0%), while boars were primarily kept for breeding (61.9%). Principal Component Analysis of five quantitative morphometric traits (height, length, girth, ear length, tail length) explained 53.2% of total variation, with PC1 and PC2 accounting for 32.6% and 20.6%, respectively, revealing provincial clustering and overlaps indicative of crossbreeding and regional genetic similarity. Overall, village pigs in South Africa are relatively homogeneous and non-descript, reflecting production systems that include poor record-keeping, variable feed quality, and inconsistent water supply, which can negatively affect productivity and genetic diversity. The study highlights the need for improved management practices, farmer training, and integration of smallholder farmers into formal markets to enhance production efficiency and conserve South African village pig genetic resources.

Keywords: *production systems, mating, breeding, village pigs, morphometric traits, phenotypic*

3.2 Introduction

In South Africa, pig production is one of the underrepresented industries in the overall agricultural sector, and it contributes about 2.2% to the primary agricultural sector (DALRRD,

2022). In South Africa, pork consumption is lower than that of poultry and beef, despite global trends showing high pork consumption (BFAP, 2021). The consumption of pork has steadily increased over the past few years, resulting in an increase in pork slaughtering to cater to the high demand, thus leading to a rise in pork production. Pig husbandry in South Africa is of considerable economic importance and is distributed across all nine provinces. There are approximately 400 commercial pork producers and 19 stud breeders in South Africa (DAFF, 2012). In 2019, the number of pigs was estimated to be 1,454 million, with Limpopo province contributing the highest share at 24%, followed by North West at 20%. Gauteng, KwaZulu-Natal, and Western Cape accounted 11% each. The province with the lowest pig numbers is Northern Cape at 1% of pork production (DAFF, 2021).

Pig farming represents a valuable component of AnGR, as pigs are highly prolific, have a relatively short gestation period of about 114 days, which is longer than rabbits, whose gestation period is 28 to 34 days, but shorter than many other livestock species (Bazer & Johnson, 2014; Yunusov *et al.*, 2024). Pigs require little space and can be integrated with other forms of subsistence agriculture where land resources are scarce (DAFF, 2012). Pigs are regarded as crucial livestock species in the livelihoods of rural farmers. Pig production enhances self-sufficiency, improves food security for rural households, and boosts incomes.

Pig mating and breeding systems influence the genetic background of pig populations and play a significant role in performance and meat quality. In rural areas of South Africa, backyard pig farming and emerging small-scale farming are the most prevalent farming practices (Matabane *et al.*, 2015; Mpalala & Obokoh, 2024). These farmers raise most indigenous and village pigs in marginal and landless areas with no animal records. The lack of animal performance records has long been observed to negatively affect genetic improvement programs in marginal areas of many developing countries (Kahi *et al.*, 2003; Roessler *et al.*, 2008). Tada *et al.* (2012) reported that the lack of performance records, especially for indigenous and village pig breeds, may result in undefined breeding practices and uncontrolled mating. Uncontrolled mating then results in the production of non-uniform animals, the emergence of undesirable traits, genetic defects, and inbreeding depression (Leroy, 2014; Muñoz *et al.*, 2019; Schiavo *et al.*, 2021).

Crossbreeding of indigenous and village pigs with exotic pig breeds is now common (Hlongwane *et al.*, 2020; Okoro *et al.*, 2017). The mating and breeding systems practiced for rural populations under the low-output production system include crossbreeding. Crossbreeding of indigenous and village pig populations with exotic breeds is practiced, making exotic breeds adapt to marginal areas and for indigenous breeds to improve their productivity (Aryee *et al.*, 2024; Hlongwane *et al.*, 2024; Madzimure *et al.*, 2012).

Crossbreeding has been reported to ensure rapid genetic progress with desired traits, hence complementarily of traits and exploitation of heterosis in animal performance (Imbayarwo-Chikosi, 2009), but it leads to the loss of indigenous animal genetic resources; thus, it is not recommended.

The overall objective of this study was to establish the type of mating, breeding and production systems utilised by South African village pig farmers and their implications on pig genetic diversity.

3.3 Materials and Methods

3.3.1 Study sites and data collection

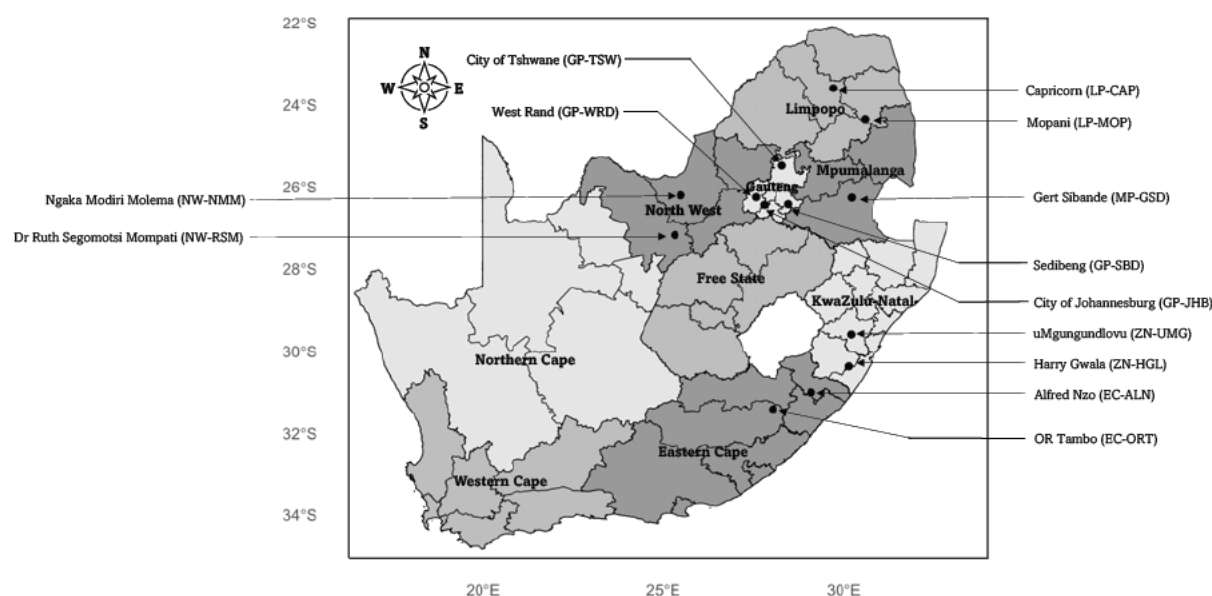


Figure 3.1 South African map displaying the geographical locations of the investigated pig populations

Six pig-farming provinces in South Africa were visited for data collection. Farmers were interviewed in the following districts: Eastern Cape – Alfred Nzo (EC-ALN) and OR Tambo (EC-ORT); Gauteng – Tshwane (GP-TSW), West Rand (GP-WRD), Johannesburg (GP-JHB), and Sedibeng (GP-SBD); KwaZulu-Natal – Harry Gwala (ZN-HGL) and uMgungundlovu (ZN-UMG); Limpopo – Capricorn (LP-CAP) and Mopani (LP-MOP); Mpumalanga – Gert Sibande (MP-GSD); and North West – Ngaka Modiri Molema (NW-NMM) and Dr. Ruth Segomotsi Mompati (NW-RSM) (**Figure 3.1**). Provincial officials assisted in identifying suitable pig farmers for the study.

Data was collected through semi-structured questionnaires, observations, and interviews. The questionnaire included information on general household details, feeding management, breeding management, and phenotypic descriptions. The questionnaires were used to

individually interview the household heads; other family members were allowed to provide additional relevant information. The questions were asked in the farmer's native language with the help of translators to improve the quality of the data captured. Most pig farmers interviewed did not keep records. Therefore, due to a lack of recording systems, answers were based on farmers' knowledge and memory recall. A total of 236 farmers were interviewed, and 535 adult pigs, including both males and females, were sampled for morphometric trait measurements. Five qualitative traits and five quantitative morphometric body measurements were taken and observed, respectively. The types of morphological traits collected are explained in **Table 3.1**. Linear body measurements were recorded in centimetres using a flexible measuring tape (**Figure 3.2**). All the measurements were taken by one person to avoid between-individual variations.

Table 3.1 Morphometric traits and their categories are considered in the analysis of the physical characteristics of village pigs

Traits	Description
Quantitative	
Body Length (L)	The distance between point of shoulder and the pin bone
Height at Withers (H)	The vertical distance between ground and the point of withers
Chest Girth (G)	The circumference of the body behind the base and the hump and just behind the elbows of the front legs
Ear Length (E)	The base of the ear to the pointed end of the ear
Tail Length (T)	The distance from the base l to the tail tip
Qualitative	
Coat Pattern	Uniform – One Colour, Uniform – Two Colours, Spotted, Pied
Ear Orientation	Drooping, Erect, Lateral
Tail Presentation	Curled, Straight
Body Colour	Black, Grey, White, Brown
Hoof Colour	Black, Grey, White, Brown



Figure 3.2 Morphological measurements taken from pigs in the study. The body measurements included height (H), length (L), girth (G), ear (E), and tail (T)

3.3.2 Data Analysis

The data collected was captured in a Microsoft Excel spreadsheet. Descriptive statistics (mean, frequency, and percentage) for qualitative and quantitative data were estimated using Statistical Package for the Social Sciences (SPSS version 28.0.1.0) (Murugan, 2022). The results obtained were presented in frequency and percentage tables and charts.

The Principal Component Analysis (PCA) was computed using Factorial Analysis of Mixed Data (a multivariate analysis method suitable for both quantitative and qualitative data) to cluster individuals using their morphological traits. The R packages FactoMineR (version 2.11), ggplot2 (version 3.5.1), factoextra (version 1.0.7), and gridExtra (version 2.3) were used for data visualization (Auguie, 2017; Kassambara & Mundt, 2020; Lê *et al.*, 2008; Wickham, 2016). Populations kept had no clear phenotypic or genetic evidence to show their names; rather, they were represented by their geographical locations and possible breed names proposed by farmers.

3.4 Results

The socio-economic characteristics reveal that most of the households are male headed, accounting for 56.4%, while 43.6% are female headed. The majority of household heads are aged between 41-60 years (47.1%). The Xhosas dominated among the ethnic groups with 32.2%, followed by Pedi (23.7%) and Zulu (11.0%), while Venda, Ndebele, English, and Afrikaans groups together accounted for less than 3.0%. Land ownership is mainly private, with 87.7% owning land, 7.2% leasing, and 5.1% using communal arrangements. Farm sizes vary greatly, 44.9% hold between 0.25 – 2.0 hectares, and 33.9% have unspecified sizes. Pig ownership is predominantly concentrated with the household head (86.4%), while joint

ownership with a spouse account for 7.2%, and minimal contributions (2.1%) come from sons and daughters (**Table 3.2**).

Table 3.2 Socio-economic characteristics of households

	Descriptors					
	Household head					
	Male headed			Female headed		
Percentage	133 (56.4)			103 (43.6)		
	Household head's age					
	≤30	31-40	41-50	51-60	61-70	≥70
Percentage	34 (14.4)	23 (9.7)	53 (22.5)	58 (24.6)	46 (19.5)	22 (9.3)
	Ethnic Group					
	Zulu	Swati	Xhosa	Tswana	Sotho	Pedi
Percentage	26 (11.0)	8 (3.4)	76 (32.2)	30 (12.7)	8 (3.4)	56 (23.7)
	Tsonga	Venda	Ndebele	English	Afrikaans	Coloured
	21 (8.9)	2 (0.8)	1 (0.4)	2 (0.8)	2 (0.8)	4 (1.7)
	Land ownership					
	Own		Lease		Communal	
Percentage	207 (87.7)		17 (7.2)		12 (5.1)	
	Land holding/Farm size					
	0.25 – 2.0	2.25 – 4.0	4.25-6.0	6.25 – 8.0	>8	Unknown
Percentage	106 (44.9)	11 (4.7)	14 (5.9)	3 (1.3)	22 (9.3)	80 (33.9)
	Pig Ownership					
	Head	Spouse	Head & Spouse		Sons	Daughters
Percentage	204 (86.4)	10 (4.2)	17 (7.2)		2 (0.8)	3 (1.3)

The number of pigs owned by pig farmers in South African villages is illustrated in **Figure 3.3**. Generally, the study shows that more sows (39.87%) than boars (22.71%) were sampled, reflecting the emphasis of rural pig farmers on maintaining breeding sows to sustain and expand their herds.

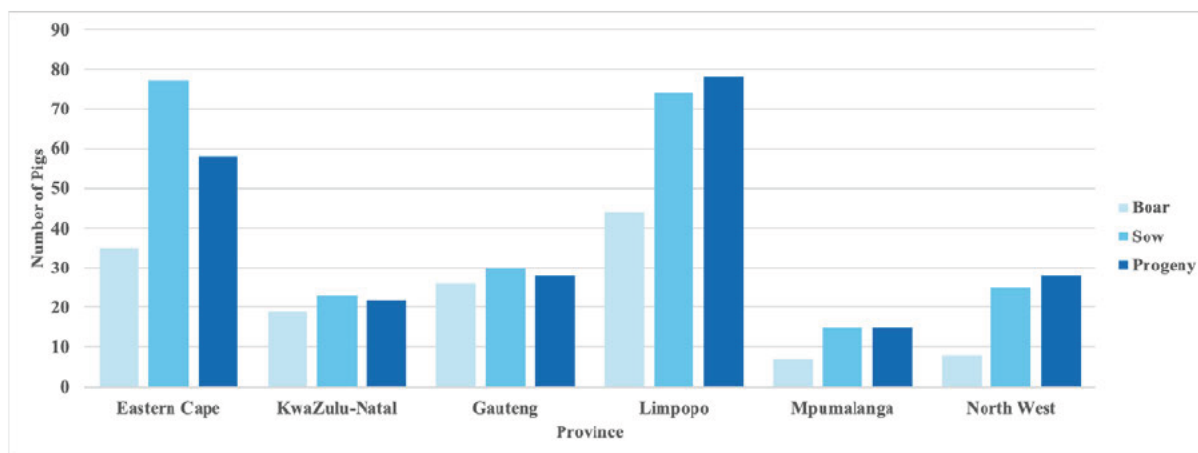


Figure 3.8 Number of pigs represented per province



Figure 3.9 Village pig breeds kept under different types of housing (Source: Photographs taken by the author, July 2023, Mpumalanga, and February 2024, North West)

Pig production systems were primarily semi-intensive (50.4%), with many farmers housing pigs in stalls or sheds (71.2%) (**Figure 3.4a**). Others used kraals (14.4%) (**Figure 2.6**) or yards (11.4%) (**Figure 3.4b**) and some farmers providing no housing (3.0%) (**Figure 3.4c**). The most kept breeds (29.4%) were Large White and Windsnyer followed by crossbreeds (15.3%) with Duroc being the least kept breed (2.0%) (**Figure 3.5**). Very few households kept pigs exclusively and were predominantly in Limpopo ($n = 24$; 34.3%). The majority of the farmers practiced mixed livestock farming, and pigs were reared with other livestock animals such as cattle, goats, sheep, and chickens in the same yard (**Table 3.3**).

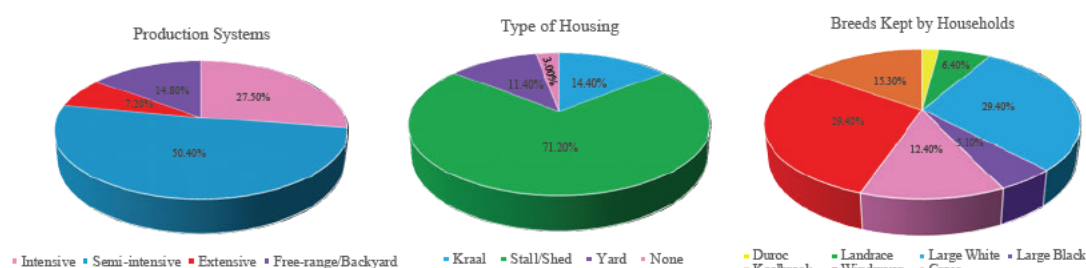


Figure 3.10 Distribution of pig production systems, housing types, and breeds kept by households

Table 3.3 Livestock species kept in the household per province

Household Livestock	Provinces					
	Eastern Cape (<i>n</i> =69)	KwaZulu- Natal (<i>n</i> =24)	Gauteng (<i>n</i> =30)	Limpopo (<i>n</i> =70)	Mpumalanga (<i>n</i> =15)	North West (<i>n</i> =28)
Pigs only	11 (15.9)	5 (20.8)	7 (23.3)	24 (34.3)	4 (26.7)	12 (42.9)
Cattle	32 (46.4)	12 (50.0)	11 (36.7)	28 (40.0)	8 (53.3)	8 (28.6)
Sheep	24 (34.8)	6 (25.0)	12 (40.0)	7 (10.0)	1 (6.7)	4 (14.3)
Goats	35 (50.7)	11 (45.8)	11 (36.7)	29 (41.4)	5 (33.3)	8 (28.6)
Chickens	47 (68.1)	17 (70.8)	18 (60.0)	31 (44.3)	8 (53.3)	10 (35.7)
Other	4 (5.8)	0 (0)	3 (10.0)	4 (5.7)	0 (0)	0 (0)

This analysis highlighted regional differences in pig production and management practices across the provinces (**Table 3.4**). The pigs were primarily kept for household meat consumption, ceremonies, and selling. Most pig farmers mix feed their pigs, especially in Mpumalanga (*n* = 12; 80.0%) and North West (*n* = 20; 71.4%). Gauteng was dominated by farmers buying formally formulated feed for their pigs (*n* = 20; 66.7%). Kitchen waste usage was most common in the Eastern Cape (*n* = 27; 39.1%). The majority of the pigs accessed water from rivers in the Eastern Cape (*n* = 42; 60.9%), whilst boreholes were the main water sources in Gauteng (*n* = 24; 80.0%). Municipal water was used heavily in Mpumalanga (*n* = 13; 86.7%). All households in Gauteng were providing pigs with good, clear water (*n* = 30; 100%), and it was freely available in 73.3% of the households. Over 85% of households in Limpopo, Mpumalanga, and North West also provided good, clear water to their pigs. Limpopo reported a small percentage of salty water (*n* = 6; 8.6%), while greenish water was reported in low proportions in Eastern Cape (*n* = 11; 15.9%) and KwaZulu-Natal (*n* = 2; 8.3%).

Table 3.4 Frequencies (%) of household roles of pigs and management characteristics in the production system per province

		Study Provinces					
		Eastern Cape	KwaZulu- Natal	Gauteng	Limpopo	Mpumal- anga	North West
Uses	Meat & sales	40 (58.0)	13 (54.2)	8 (26.7)	43 (61.4)	13 (86.7)	16 (57.1)
	Meat	24 (34.1)	0 (0)	1 (3.3)	13 (18.6)	0 (0)	6 (21.4)
	Sales	3 (4.3)	11 (45.8)	21 (70.0)	14 (20.0)	2 (13.3)	6 (21.4)
	Ceremonies	1 (1.4)	0 (0)	0 (0)	0 (0)	0 (0)	0 (0.0)
	Investment	1 (1.4)	0 (0)	0 (0)	0 (0)	0 (0)	0 (0.0)
Feed source	Mix	33 (47.8)	16 (66.7)	9 (30.0)	43 (61.4)	12 (80.0)	20 (71.4)
	Kitchen	27 (39.1)	3 (12.5)	0 (0)	15 (21.4)	0 (0)	3 (10.7)
	Waste						
	Bought-in	9 (13.0)	5 (20.8)	20 (66.7)	12 (17.1)	3 (20.0)	5 (17.9)
	Other	0 (0)	0 (0)	0 (0)	0 (0)	0 (0)	0 (0.0)
Water source	Borehole	5 (7.2)	7 (29.2)	24 (80.0)	27 (38.6)	0 (0)	10 (35.7)
	Dam	0 (0)	4 (16.7)	0 (0)	4 (5.7)	0 (0)	1 (3.6)
	River	42 (60.9)	6 (25.0)	0 (0)	2 (2.9)	2 (13.3)	1 (3.6)
	Municipal	21 (30.4)	7 (29.2)	6 (20.0)	34 (48.6)	13 (86.7)	16 (57.1)
	Water well	1 (1.4)	0 (0)	0 (0)	3 (4.3)	0 (0)	0 (0.0)
Water frequency	Freely	11 (15.9)	8 (33.3)	22 (73.3)	18 (25.7)	1 (6.7)	9 (32.1)
	Once/day	9 (13.0)	1 (4.2)	1 (3.3)	13 (18.6)	1 (6.7)	4 (14.3)
	Twice/day	31 (44.9)	11 (45.8)	5 (16.7)	30 (42.9)	11 (73.3)	7 (25.0)
	Thrice/day	18 (26.1)	4 (16.7)	2 (6.7)	9 (12.9)	2 (13.3)	8 (28.6)
Water quality	Good/clear	28 (40.6)	14 (58.3)	30 (100)	61 (87.1)	13 (86.7)	26 (92.9)
	Muddy	13 (18.8)	4 (16.7)	0 (0.0)	2 (2.9)	0 (0)	2 (7.1)
	Salty	0 (0)	0 (0)	0 (0.0)	6 (8.6)	0 (0)	0 (0.0)
	Smelly	17 (24.6)	4 (16.7)	0 (0.0)	0 (0.0)	2 (13.3)	0 (0.0)
	Greenish	11 (15.9)	2 (8.3)	0 (0.0)	1 (1.4)	0 (0)	0 (0.0)

Analysis revealed that the majority of the pig farmers primarily keep boars for breeding purposes (61.9%) while a few maintain them for socio-cultural purposes (7.2%) and for fattening (1.8%). Size (60.2%) and shape (48.8%) are the most important factors influencing the selection of boars. Colour (27.1%), performance (22.3%), availability (24.7%), and temperament (3.0%) were the least considered factors. Most boars are bred by the farmers, only a small proportion of boars are shared (1.8%), borrowed (10.2%), or donated (1.2%). Among

the boars, Large White (42.8%) breed was the most dominant, followed by Windsnyer (15.1%). Hand mating was the most practiced method (47.6%), followed by uncontrolled (38.0%).

Table 3.5 Boar selection, sourcing, and breeding practices

Primary reason for keeping boar									
Breeding			Socio-cultural			Fattening		No boar	
146 (61.9)			17 (7.2)			3 (1.8)		70 (29.7)	
Reason for choice of boar									
Size		Shape		Temperament		Colour		Performance	Availability
100 (60.2)		81 (48.8)		5 (3.0)		45 (27.1)		37 (22.3)	41 (24.7)
Source of boar									
Bred		Bought		Donated		Borrowed		Sharing	Unknown
66 (39.8)		66 (39.8)		2 (1.2)		17 (10.2)		3 (1.8)	12 (7.2)
Breed Name									
Duroc	L.Black	L.White	Landrace	Kolbroek	Warthog	Windsnyer	Cross	Unknown	
10 (6.0)	3 (1.8)	71 (42.8)	12 (7.2)	17 (10.2)	1 (0.6)	25 (15.1)	16 (9.6)	11 (6.6)	
Mating									
Uncontrolled			Hand Mating			Group Mating		Artificial Insemination	
63 (38.0)			79 (47.6)			22 (13.3)		2 (1.2)	

PC1 and PC2 accounted for 32.6% and 20.6% of the total variation, respectively (**Figure 3.6**). The Eastern Cape population formed a distinct cluster that was separate from other populations, with few outliers. Limpopo indicated high variability, spreading largely across all populations. KwaZulu-Natal and Mpumalanga overlap, and Gauteng and North West suggest similarities in their pig breeds. The overlaps might be due to geographical proximity as these provinces border each other, which could allow for easier movement of pigs, genetic mixing, and similar breeding practices. Similar breeds clustered closely across all the populations.

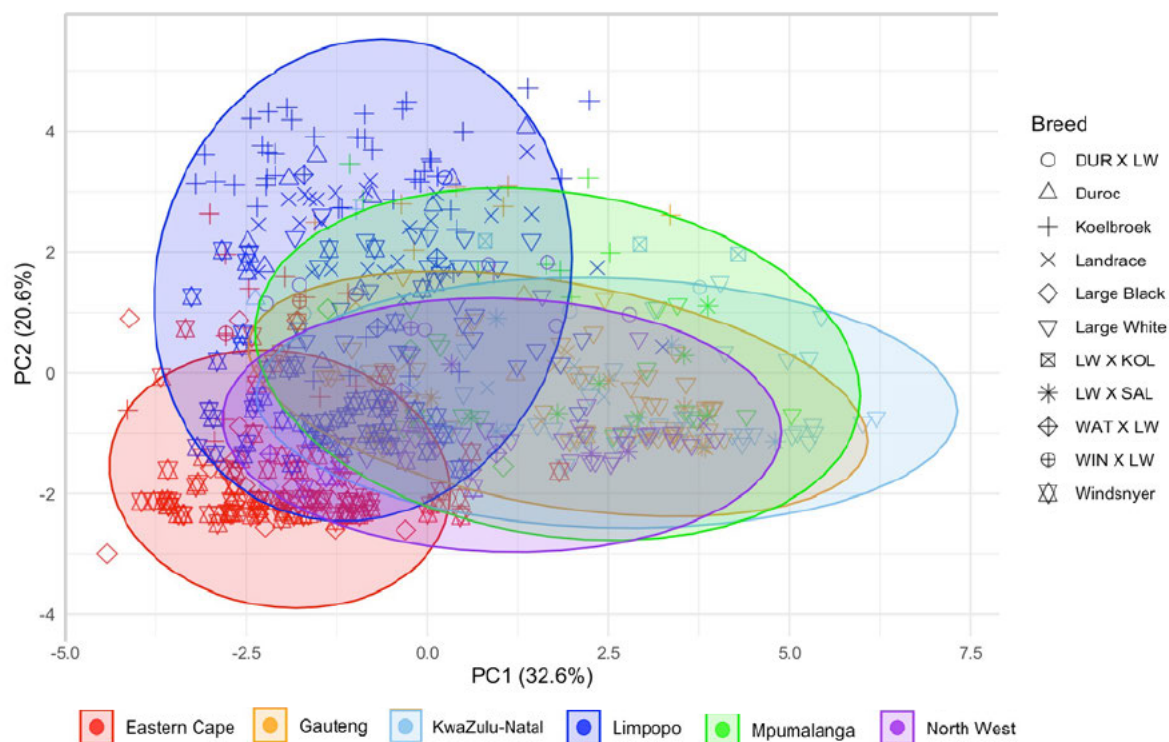


Figure 3.11 Principal Component Analysis using five quantitative traits (height, length, girth, ear length, and tail length). The ellipse represents the distribution of individuals across the PCA axis

3.5 Discussion

The results and analysis of this study highlighted some shortcomings. Collecting crucial production data is challenging when there are no records to support the information gathered, and the only methods of validation are the farmers' knowledge and memories. Poor record-keeping among smallholder pig farmers has been previously reported by other researchers in the Eastern Cape, Gauteng, KwaZulu-Natal, Mpumalanga, and Western Cape (Gcumisa *et al.*, 2016; Madzimure *et al.*, 2012; Matabane *et al.*, 2015; Molotsi *et al.*, 2021; Munzhelele *et al.*, 2017). To enhance data reliability, observational analyses were used to validate some of the data collected from the farmers. Despite these challenges, the present study revealed majority of the pig owners were the household heads, men, but the day-to-day care and management is primarily handled by women (Rindia Wati *et al.*, 2022; Sillitoe, 2001), since adult females are liable for any homestead-related activities (Guèye, 2003) of which pigs are a part of. These types of economic alienation at the family level are made by the distinctive ability of pigs to thrive on household waste and garden crops, which are often the purview of women. Most farmers were 41-50 years of age (22.5%), which indicates that indeed the younger generation would rather go to cities in pursuit of salaried employment than engage in agriculture (Brooks *et al.*, 2013). This finding can also highlight a delayed entry into animal agriculture. Schembri

et al. (2013) reported similar observations in Australia. Moreover, it is firmly believed that rural farming is a low-return activity, and only the poor engage in it. However, this perception is largely driven by low productivity, poor market access, and limited financial and technical support faced by smallholder farmers (FAO, 2019). In South Africa, rural pig farming often relies on household labour, low-quality feed, and minimal veterinary input, which restrict profitability (Tada *et al.*, 2012; Adesehinwa *et al.*, 2024). Despite these challenges, village pig production plays a vital role in food security, nutrition, and livelihood resilience, and with improved institutional support, it holds potential for sustainable rural development (FAO, 2019).

The dominance of Xhosa (32.2%), Pedi (23.7 %) among the farmer respondents reflects the ethnic composition in certain rural areas of South Africa. While direct scientific studies examining how ethnicity influences pig breeding practices and genetic diversity in these communities are limited, previous research highlights that local pig genetic resources across Africa are under-characterised, and sociocultural factors, including traditional management practices, play a critical role in shaping breed diversity (Pius *et al.*, 2024). Understanding this ethnic distribution, therefore, offers useful context for interpreting how cultural practices might indirectly influence genetic diversity, despite the absence of explicit studies linking ethnicity and pig breed structure in South Africa.

Land ownership was similar to the findings of Marcus (2022) and Matabane *et al.* (2015) in North West and Gauteng provinces, respectively. The studies also reported that most of the smallholders owned their land, while others leased or used communal land. Most pig farmers primarily practiced intensive or semi-intensive farming systems (77.9%), which is beneficial as it increases production, weight gain, and profit, and reduces pre-weaning mortalities than in free range or backyard farming systems (Dione *et al.*, 2014; Madzimore *et al.*, 2012; Mutua *et al.*, 2010). This finding aligns with observations by Munzhelele *et al.* (2017) in Mpumalanga and Molotsi *et al.* (2021) in the Western Cape, who also reported high numbers of farmers using the intensive or semi-intensive systems. Most farmers used stalls or sheds for pig housing. The housing was made out of treated or untreated wood, wire, iron sheets, bricks, concrete, and mud. Some households provided shelter with roof, whereas others did not. Dione *et al.* (2014) highlighted that housing built and provided by farmers largely depends on their financial status. This trend was evident across the three study areas, where most farmers used materials that were easily repurposed and either free or cheaper. Not all farmers provided housing for their pigs. Similar findings were reported by Kagira *et al.* (2010), who also noted that not all

smallholder farmers provided housing to their pigs. However, Manchidi (2009) advised that pigs should be protected from direct sunlight and wind, as this can lead to stress.

Village pig production primarily relies on unimproved indigenous pigs that vary widely in morphological appearance and body size. Consistent with this, our findings showed that most farmers preferred to keep improved pig breeds, with 41.8% maintaining indigenous breeds, while others kept crossbred pigs at 15.3%, and the rest kept exotic pigs. This is similar to the results reported by other studies that farmers still keep indigenous pig breeds (Chiduwa *et al.*, 2008; Madzimure *et al.*, 2012), however, in contrast to the reports of Gcumisa *et al.* (2016). This is due to the goal to match the available resources to production, since indigenous pigs are more miniature and are more capable of foraging to supplement their diets (Lekule & Kyvsgaard, 2003). Most farmers who specifically mentioned breeding crossbreeds reported having Large White and Landrace hybrids known as “Camborough”.

The main reason for keeping pigs in the study areas was mostly for household meat consumption and selling. The findings that keeping pigs is for sales and household meat consumption are in line with the report of Madzimure *et al.* (2012). Farmers keeping pigs for ceremonies were only found in the Eastern Cape. The low numbers of farmers keeping pigs as ceremonial animals are expected, as most people normally slaughter cattle, sheep, goats, and chickens for the nature of the ceremony (Strolin *et al.*, 2001; Gwaze *et al.*, 2009). Less than 45% of households kept pigs across each province, and they were mostly in the North West.

The majority of the pig farmers primarily kept boars for breeding purposes; however, 38.1% farmers did not own boars, which may limit opportunities for controlled mating and genetic improvement. The lack of a boar in a herd is common among small herds, and farmers borrow or share boars for breeding. The borrowing and sharing of boars in these villages enable boars and sows to intermingle among the herds, which reduces inbreeding levels (Jaitner *et al.*, 2001). Using boars from their own herd for breeding may result in inbreeding, which can lower the animals' health and production efficiency, thereby significantly reducing their diversity. The purpose of keeping boars in these households influences the specific traits that farmers prioritize in their selection. In these study areas, size and shape are the most important factors influencing the selection, as they were mainly selected for breeding. A boar should have the right size and body conformation to successfully mate with a sow. When farmers were asked about breeding, 47.8% practised hand mating, and 38.0% indicated that they do not control the breeding of their pigs.

Feed is one of the most essential components in livestock production (Khan *et al.*, 2005), and poor-quality feed provided by village farmers is often a constraint to production (Kosgey *et al.*, 2008). In our study, the majority of the farmers provided bought-in feed to their pigs, especially in Gauteng. Bought-in feed is feed that is formulated and produced by animal feeding companies. This type of feed is typically more costly, comes in pellet form, and is made specially for pigs and their different development stages. Some farmers are unable to afford this feed (Dione *et al.*, 2014), especially within the smallholder sector. Thus, other farmers stated that they do not solely provide this feed; they mix it with kitchen waste, swill to save money. In this study, the swill used to feed pigs consisted of kitchen waste and kitchen waste mixed with sorghum brewers' grains (amavovo). Swill consisted of leftovers from schools, hospitals, and hotels. Feeding practices observed in the present study are similar to those previously reported by Dione *et al.* (2014), Kagira *et al.* (2010), Matabane *et al.* (2015) Munzhelele *et al.* (2017), and Ström *et al.* (2017), that farmers did not exclusively provide their pigs with commercial feed due to affordability. Unfortunately, the practice of feeding swill can introduce exotic diseases, including foot-and-mouth disease (Relun *et al.*, 2015). Most households provided good, clear water to their pigs more than two times a day. However, some households provided bad quality water fetched from natural water resources (rivers, dams, and water ponds). Access to water in most rural areas of South Africa has been reported as a challenge. According to Statistics South Africa's (Stats SA) Census (2022), only 82.4% of households in South Africa had access to piped water either inside their dwelling or inside their yard. Thus, some farmers depend on unstable water sources.

Linear body measurements and phenotypic traits can be used to characterize different breeds of livestock and can also reveal each individual's origin, relationship, size, and shape (Panda *et al.*, 2020; Traoré *et al.*, 2008). Earlier research has employed multivariate analysis of morphological traits to reveal population structure, assist in breed classification, and assess genetic variation among native pig populations across various agro-ecological zones (Takele *et al.*, 2021). Principal Component Analysis (PCA) is particularly useful for summarising correlated characteristics. For instance, Okoro *et al.* (2015) applied PCA to study morphometric traits in crossbred pigs in Nigeria, while Tolenkomba *et al.* (2013) used PCA to characterize Mizo local pigs. These studies demonstrate the successful application of PCA for differentiating pig populations based on morphological traits, providing a comparative framework and supporting the use of PCA in our study. In our analysis, PCA clustered individuals according to their provinces, suggesting distinct patterns of variation associated with geographical origins, likely influenced by regional genetic similarities or shared environmental factors. Overlapping groups might suggest inter-provincial similarities or mixed

breeding practices or the use of the same breeds across different provinces, leading to genetic similarities and reduced differentiation between regions. Geographical location plays a significant role in breed differentiation. Moreover, different breeds clustering together might be due to crossbreeding practices. Crossbreeding is a common practice in the pig industry (Steyn *et al.*, 2021). Hlongwane *et al.* (2020) reported that significant admixture of different pig breeds occurs in rural areas, often due to indiscriminate crossbreeding among local populations.

3.6 Conclusion

The South African village pig populations are relatively homogeneous in terms of morphometric traits but remain undefined and often kept under traditional low-input systems. This study revealed several challenges in smallholder pig farming practices, including poor recordkeeping, uncontrolled breeding, and adapting farming systems to their available resources. Observed variation in morphometric traits indicates a need for further research on their genetic make-up and the adaptability of the village populations. Future research and development efforts should prioritise integrating smallholder village farmers into the formal market sector and implementing policies that address these challenges to ensure the long-term viability and sustainability of village pigs. Additionally, improving access to quality extension services and seeking solutions to constraints using locally available means is essential.

3.7 References

- Adesehinwa, A. O. K., Boladuro, B. A., Dunmade, A. S., Idowu, A. B., Moreki, J. C., & Wachira, A. M. (2024). Pig production in Africa: current status, challenges, prospects and opportunities. *Animal Bioscience*, 37(4), 730. <https://doi.org/10.5713/AB.23.0342>
- Aryee, S. N. D., Owusu-Adjei, D., Osei-Amponsah, R., Skinner, B. M., Amuzu-Aweh, E. N., Ahunu, B., Enright, A., & Sargent, C. A. (2024). Population Structure and Genomic Characterisation of the Ashanti Dwarf Pig of Ghana. *Animals*, 14(5). <https://doi.org/10.3390/ANI14050792>
- Auguie, B. (2017). Miscellaneous Functions for “Grid” Graphics [R package gridExtra version 2.3]. CRAN: Contributed Packages. <https://doi.org/10.32614/CRAN.PACKAGE.GRIDEXTRA>
- Bazer, F. W., & Johnson, G. A. (2014). Pig blastocyst–uterine interactions. *Differentiation*, 87(1–2), 52–65. <https://doi.org/10.1016/J.DIFF.2013.11.005>
- Brooks, K., Zorya, S., Gautam, A., & Goyal, A. (2013). Agriculture as a sector of opportunity for young people in Africa. Policy Research Working Paper Series. <https://ideas.repec.org/p/wbk/wbrwps/6473.html>
- Census 2022 shows South Africa’s population grew to 62 million | Statistics South Africa <https://www.statssa.gov.za/?p=16711> Accessed: 2025-03-11
- Chiduwa, G., Chimonyo, M., Halimani, T. E., Chisambara, S. R., & Dzama, K. (2008). Herd dynamics and contribution of indigenous pigs to the livelihoods of rural farmers in a semi-arid area of Zimbabwe. *Tropical Animal Health and Production*, 40(2), 125–136. <https://doi.org/10.1007/S11250-007-9071-8/METRICS>
- DAFF, 2021 A Profile of The South African Pork Market Value Chain. <http://www.daff.gov.za/images/Branches/Economica%20Development%20Trade%20and%20Marketing/marketing/annual-publications/livestock/pork-market-value-chain-profile-2021.pdf> Accessed: 2025-03-18
- DALRRD. (2022). A profile of the South African Beef market value chain 2021. 1–36.

- Department of Agriculture, Forestry and Fisheries (DAFF), 2012, A profile of the South African pork market value chain, pp 38. Available from: <http://www.nda.agric.za/docs/AMCP/Pork2012.pdf>
- Dione, M. M., Ouma, E. A., Roesel, K., Kungu, J., Lule, P., & Pezo, D. (2014). Participatory assessment of animal health and husbandry practices in smallholder pig production systems in three high poverty districts in Uganda. *Preventive Veterinary Medicine*, 117(3–4), 565–576. <https://doi.org/10.1016/J.PREVETMED.2014.10.012>
- FAO. (2019). The State of Food and Agriculture 2019: Moving forward on food loss and waste reduction. Rome: Food and Agriculture Organization of the United Nations.
- Gcumisa, S. T., Oguttu, J. W., & Masafu, M. M. (2016). Pig farming in rural South Africa: A case study of uthukela district in KwaZulu-Natal. *Indian Journal of Animal Research*, 50(4), 614–620. <https://doi.org/10.18805/IJAR.11175>
- Guèye, E. F. (2003). Gender issues in family poultry production systems in low-income food-deficit countries. *American Journal of Alternative Agriculture*, 18(4), 185–195. <https://doi.org/10.1079/AJAA200350>
- Gwaze, F. R., Chimonyo, M., & Dzama, K. (2009). Variation in the functions of village goats in Zimbabwe and South Africa. *Tropical Animal Health and Production*, 41(7), 1381–1391. <https://doi.org/10.1007/S11250-009-9326-7>
- Hlongwane, N. L., Hadebe, K., Soma, P., Dzomba, E. F., & Muchadeyi, F. C. (2020). Genome Wide Assessment of Genetic Variation and Population Distinctiveness of the Pig Family in South Africa. *Frontiers in Genetics*, 11(May), 1–19. <https://doi.org/10.3389/fgene.2020.00344>
- Hlongwane, N. L., Dzomba, E. F., Hadebe, K., van der Nest, M. A., Pierneef, R., & Muchadeyi, F. C. (2024). Identification of Signatures of Positive Selection That Have Shaped the Genomic Landscape of South African Pig Populations. *Animals*, 14(2), 236. <https://doi.org/10.3390/ANI14020236/S1>
- Jaitner, J., Sowe, J., Secka-Njie, E., & Dempfle, L. (2001). Ownership pattern and management practices of small ruminants in The Gambia — implications for a breeding programme. *Small Ruminant Research*, 40(2), 101–108. [https://doi.org/10.1016/S0921-4488\(00\)00221-2](https://doi.org/10.1016/S0921-4488(00)00221-2)
- Kagira, J. M., Kanyari, P. W. N., Maingi, N., Githigia, S. M., Ng'ang'a, J. C., & Karuga, J. W. (2010). Characteristics of the smallholder free-range pig production system in western Kenya. *Tropical Animal Health and Production*, 42(5), 865–873. <https://doi.org/10.1007/S11250-009-9500-Y>
- Karamurugan, S., & Govindarajan, B. (2023). Statistical package for the social science. *Int J Bus Econ Res*, 8(4), 616–8.
- Kassambara, A., & Mundt, F. (2020). Extract and Visualize the Results of Multivariate Data Analyses [R package factoextra version 1.0.7]. CRAN: Contributed Packages. <https://doi.org/10.32614/CRAN.PACKAGE.FACTOEXTRA>
- Khan, R., Journal, R. U.-P. V., & 2005, undefined. (n.d.). Characteristics of rural subsistence small holder livestock production system in mountainous areas of NWFP, Pakistan. Core.Ac.UkRN Khan, RH UsmaniPakistan Veterinary Journal, 2005•core.Ac.Uk, 25(3), 2005. Retrieved March 11, 2025, from <https://core.ac.uk/download/pdf/26916993.pdf>
- Kosgey, I. S., Rowlands, G. J., Van Arendonk, J. A. M., & Baker, R. L. (n.d.). Small ruminant production in the tropics: a study of smallholder and pastoral/extensive farming systems in Kenya. Biometrics.Ilri.OrgIS Kosgey, GJ Rowlands, JAM van Arendonk, RL BakerSmall Ruminant Research, 2004•biometrics.Ilri.Org. Retrieved March 15, 2025, from <https://biometrics.ilri.org/Publication/Full%20Text/Chapter%203-26th.pdf>
- Lê, S., Josse, J., & Husson, F. (2008). FactoMineR: An R Package for Multivariate Analysis. *Journal of Statistical Software*, 25(1), 1–18. <https://doi.org/10.18637/JSS.V025.I01>
- Lekule, F. P., & Kyvsgaard, N. C. (2003). Improving pig husbandry in tropical resource-poor communities and its potential to reduce risk of porcine cysticercosis. *Acta Tropica*, 87(1), 111–117. [https://doi.org/10.1016/S0001-706X\(03\)00026-3](https://doi.org/10.1016/S0001-706X(03)00026-3)
- Leroy, G. (2014). Inbreeding depression in livestock species: review and meta-analysis. *Animal Genetics*, 45(5), 618–628. <https://doi.org/10.1111/AGE.12178>
- Madzimure, J., Chimonyo, M., Zander, K. K., & Dzama, K. (2012). Potential for using indigenous pigs in subsistence-oriented and market-oriented small-scale farming systems of Southern Africa. *Tropical Animal Health and Production*, 45(1), 135–142. <https://doi.org/10.1007/S11250-012-0184-3>
- Manchidi, M. J. (2009). An evaluation of small-scale piggeries in Limpopo Province, South Africa.

- Marcus, K. S. (2022). Production analysis of commercial and small holder pig farmers in the North West province of South Africa.
- Matabane, M., Nethenzheni, P., & Thomas, R. (2015). Status of the smallholder pig farming sector in Gauteng Province of South Africa. <https://www.cabidigitallibrary.org/doi/full/10.5555/20163008053>
- Molotsi, A. H., Cupido, M., & Hoffman, L. C. H. (2021). Characterization of smallholder pig production systems in the Western Cape, South Africa. *Tropical Animal Health and Production*, 53(2). <https://doi.org/10.1007/S11250-021-02649-8>
- Mpalala, N., & Obokoh, L. O. (2024). Small-Scale Pig Farming as a Tool for Unemployment Alleviation in Buffalo City, Eastern Cape of South Africa. *South African Journal of Agricultural Extension*, 52(3), 85–102. <https://doi.org/10.17159/2413-3221/2024/V52N3A15544>
- Muñoz, M., Bozzi, R., García-Casco, J., Núñez, Y., Ribani, A., Franci, O., García, F., Škrlep, M., Schiavo, G., Bovo, S., Utzeri, V. J., Charneca, R., Martins, J. M., Quintanilla, R., Tibau, J., Margeta, V., Djurkin-Kušec, I., Mercat, M. J., Riquet, J., ... Óvilo, C. (2019). Genomic diversity, linkage disequilibrium and selection signatures in European local pig breeds assessed with a high density SNP chip. *Scientific Reports*, 9(1), 1–14. <https://doi.org/10.1038/s41598-019-49830-6>
- Munzhelele, P., Oguttu, J., Fasanmi, O. G., & Fasina, F. O. (2017). Production constraints of smallholder pig farms in agro-ecological zones of Mpumalanga, South Africa.
- Murugan, K. S. (2022). Statistical Package for the Social Science. In *Journal of Business and Economics Research (IJBER)* (Vol. 8). <https://www.researchgate.net/publication/367127751>
- Mutua, F., Arimi, S., Ogara, W., Dewey, C., & Schelling, E. (2010). Farmer Perceptions on Indigenous Pig Farming in Kakamega District, Western Kenya. *Nordic Journal of African Studies*, 19(1), 15–15. <https://doi.org/10.53228/NJAS.V19I1.213>
- Okoro, V. M., Ogundu, U. E., Okani, M., Oziri, I., Eneowo, O., Olisenekwu, O. T., Kadurumba, O., Ogbuewu, I. P., Onyemauwa, S., Ukwu, H. O., & Ibe, S. N. (2015). Principal Component Analysis of Conformation and Blood Marker Traits at Pre- and Post-Weaning Stages of Growth in F2 Crossbred Nigerian Indigenous X Landrace Pigs. *Animal Biotechnology*, 26(4), 243–250. <https://doi.org/10.1080/10495398.2015.1014043>
- Okoro, V., Research, C. M.-A. E. & E., & 2017, undefined. (2017). Estimates of crossbreeding parameters for growth and conformation traits in Nigerian Indigenous and exotic pig breeds. Aloki.HuVMO Okoro, CA MbajiorguApplied Ecology & Environmental Research, 2017•aloki.Hu. https://doi.org/10.15666/aeer/1504_117128
- Panda, S., Gaur, G., Sahoo, N., ... P. B.-I. J. of, & 2020, undefined. (2020). Principal component analysis of morphometric and growth traits in crossbred piglets. *Epubs.Icar.Org.InS Panda, GK Gaur, NR Sahoo, PK Bharti, J KarIndian Journal of Animal Sciences*, 2020•epubs.Icar.Org.In, 90(8), 1168–1171. <https://doi.org/10.56093/ijans.v90i8.109303>
- Pius, L., Huang, S., Wanjala, G., Bagi, Z., & Kusza, S. (2024). African Local Pig Genetic Resources in the Context of Climate Change Adaptation. *Animals* 2024, Vol. 14, Page 2407, 14(16), 2407. <https://doi.org/10.3390/ANI14162407>
- Quantification of the South African Pork Value Chain: Phase 1 Bureau for Food and Agricultural Policy (BFAP) in collaboration with The South African Pork Producers' Association. (2021) <https://www.bfap.co.za/wp-content/uploads/2023/04/Pork-Value-Chain-Study.pdf> Accessed: 2025-03-18
- Relun, A., Charrier, F., Trabucco, B., Maestrini, O., Molia, S., Chavernac, D., Grosbois, V., Casabianca, F., Etter, E., & Jori, F. (2015). Multivariate analysis of traditional pig management practices and their potential impact on the spread of infectious diseases in Corsica. *Preventive Veterinary Medicine*, 121(3–4), 246–256. <https://doi.org/10.1016/J.PREVETMED.2015.07.004>
- Rindia Wati, N. K., Amrawaty, A. A., & Sri Lestari, V. (2022). Gender roles at pig farming. *Advances in Environmental Biology*, 16(11), 1–5. <https://doi.org/10.22587/AEB.2022.16.11.1>
- Schembri, N., Hernandez-Jover, M., Toribio, J. A., & Holyoake, P. K. (2013). Demographic and production practices of pig producers trading at saleyards in eastern Australia. *Australian Veterinary Journal*, 91(12), 507–516. <https://doi.org/10.1111/AVJ.12131>
- Schiavo, G., Bovo, S., Muñoz, M., Ribani, A., Alves, E., Araújo, J. P., Bozzi, R., Čandek-Potokar, M., Charneca, R., Fernandez, A. I., Gallo, M., García, F., Karolyi, D., Kušec, G., Martins, J. M., Mercat, M. J., Núñez, Y., Quintanilla, R., Radović, ... Fontanesi, L. (2021). Runs of

- homozygosity provide a genome landscape picture of inbreeding and genetic history of European autochthonous and commercial pig breeds. *Animal Genetics*, 52(2), 155–170. <https://doi.org/10.1111/AGE.13045>
- Sillitoe, P. (2001). Pig men and women, big men and women: Gender and production in the New Guinea highlands. *Ethnology*, 40(3), 171–192. <https://doi.org/10.2307/3773964>
- Steyn, Y., Lourenco, D. A., Chen, C. Y., Valente, B. D., Holl, J., Herring, W. O., & Misztal, I. (2021). Optimal definition of contemporary groups for crossbred pigs in a joint purebred and crossbred genetic evaluation. *Journal of Animal Science*, 99(1), 1–8. <https://doi.org/10.1093/JAS/SKAA396>
- Strolin, Laura, Jacqueline Studer, and Michele Degli Esposti. "3.3| Animals and Ceremonies." In *Archaeozoology of Southwest Asia and Adjacent Areas XIII: Proceedings of the Thirteenth International Symposium*, University of Cyprus, Nicosia, Cyprus, June 7-10, 2017, vol. 3, p. 255. Lockwood Press, 2021.
- Ström, G., Djurfeldt, A. A., Boqvist, S., Albiñ, A., Sokerya, S., San, S., Davun, H., & Magnusson, U. (2017). Urban and peri-urban family-based pig-keeping in Cambodia: Characteristics, management and perceived benefits and constraints. *PLOS ONE*, 12(8), e0182247. <https://doi.org/10.1371/JOURNAL.PONE.0182247>
- Takele, A., Melesse, A., Taye, M., Takele, A., Melesse, A., & Taye, M. (2021). APPLICATION OF MULTIVARIATE ANALYSIS TO DIFFERENTIATE HARARGHE HIGHLAND GOAT POPULATIONS REARED IN THE WEST HARARGHE ZONE, ETHIOPIA. *Chilean Journal of Agricultural & Animal Sciences*, 37(3), 209–220. <https://doi.org/10.29393/CHJAAS37-23AMAM30023>
- Tolenkhomba, T., Saikia, P., Hmar, L., Lalthazuali, Prava, M., & Singh, N. S. (2013). Principal component analysis of body measurements in Mizo local pigs. *Indian Journal of Veterinary Research*.
- Traoré, A., Tamboura, H. H., Kabore, A., Royo, L. J., Fernández, I., Álvarez, I., Sangare, M., Bouchel, D., Poivey, J. P., Francois, D., Sawadogo, L., & Goyache, F. (2008). Multivariate analyses on morphological traits of goats in Burkina Faso. *Aab.Copernicus.Org* A Traoré, HH Tamboura, A Kabore, LJ Royo, I Fernandez, I Álvarez, M Sangare, D BouchelArchives Animal Breeding, 2008•aab.Copernicus.Org, 51, 588–600. <https://aab.copernicus.org/articles/51/588/2008/>
- Wickham, H. (2016). Programming with ggplot2. 241–253. https://doi.org/10.1007/978-3-319-24277-4_12
- Yunusov, K., Djambilov, B., Xolmirzayev, D., Ibragimov, B., & Daniyerov, R. (2024). The period of gestation of rabbits and its fertility connection. *BIO Web of Conferences*, 95, 01029. <https://doi.org/10.1051/BIOCONF/20249501029>

4 GENETIC DIVERSITY, ADMIXTURE LEVELS, AND EXTINCTION RISK ASSESSMENT OF SOUTH AFRICAN INDIGENOUS AND VILLAGE PIG POPULATIONS RESULTING FROM CROSSBREEDING WITH EXOTIC BREEDS

4.1 Abstract

Genetic diversity among populations is crucial for adaptability, including adaptability to the harsh conditions associated with climate change. The present study examined genetic diversity, admixture levels, and potential risk to extinction of South African indigenous and village pig populations, potentially affected by crossbreeding, uncontrolled mating, and breeding practices. We genotyped 384 pig ear tissue samples from South African villages across six provinces using the Illumina PorcineSNP80K Bead Chip. Additionally, 100 pig genotypes from previous studies were included to compare the clustering and relationships between populations. Genetic variability was assessed using polymorphic information content (PIC), percentage of polymorphic loci ($%P$), total (N_a), effective allele numbers ($N_e^{alleles}$), P-value for the Global Hardy Weinberg test (P), observed (H_o), and expected heterozygosity (H_E). The effective population size (N_e) results indicate a rapid decline in N_e across generations for all populations, leading to increased inbreeding and loss of genetic diversity, and thereby escalating the risk of population collapse and eventual extinction. Low F_{ST} values (0.016 – 0.111) were observed among the populations, indicating relatively low to low genetic differentiation among the pig populations. Principal component analysis produced five distinct clusters, with PC1 and PC2 explaining 18.17% and 8.92% of total genetic variability, respectively, forming five distinct clusters. Optimal ADMIXTURE clustering ($K = 14$) revealed admixture in all village pig populations. Treemix analysis provided insights into relationships among breeds and populations. The results indicated a decline in population size alongside high levels of admixture, suggestive of extensive interbreeding among the investigated populations. Further studies on mating systems and their effects on population diversity and structure are recommended.

Keywords: *Genetic diversity, population structure, SNPs, pig, PorcineSNP80 BeadChip*

4.2 Introduction

Pigs are among the most significant livestock species worldwide, playing a crucial role in livelihood, food security, and economic growth, particularly in developing and underdeveloped countries, where they often survive under unfavourable environments (i.e., limited or poor-quality feed, irregular access to clean water, suboptimal housing or overcrowding, high disease

pressure with minimal veterinary support, and harsh climatic conditions) (Mutua *et al.*, 2010; Niemi *et al.*, 2010). The domesticated pig (*Sus scrofa*), a member of the family Suidae, evolved about 20 to 30 million years ago, independently from Asian and European wild boar subspecies (Giuffra *et al.*, 2000). Other members of the family Suidae included wild hog, warthog (*Phacochoreus africanus*), pig deer (*Babyrusa babyrussa*), pygmy hog (*Porcula salvania*), as well as wild boar (*Sus scrofa*) that has been domesticated and distributed across several continents, showcasing its adaptability to multiple environments and climates (Ruvinsky *et al.*, 2011; Frantz *et al.*, 2016; Groenen, 2016).

In South Africa, pig populations have been shaped by complex evolutionary forces including domestication, selection, gene flow, genetic drift, and historical introductions that reflect ongoing interactions among wild, domestic, and indigenous populations (D'Alessandro *et al.*, 2022). While indigenous breeds likely trace their origins to domestic pigs that migrated to South Africa from sub-Saharan Africa through the Nile Corridor, European settlers introduced commercial pig breeds from America and Europe in the 1600s for commercial farming (Penrith and Vosloo, 2011; Blench and MacDonald, 2000). South African domesticated pigs are reared under various production systems, including commercial intensive systems, small-scale farming, and free backyard management systems characterized by the absence of adequate housing or shelter (Matabane *et al.*, 2015; Mokoele *et al.*, 2014; Molotsi *et al.*, 2021). Notably, the South African pig production sector is highly saturated by commercial breeds favoured for their fast growth rate and superior production quality (Ai *et al.*, 2013). Conversely, the two breeds considered indigenous to South Africa, Kolbroek and Windsnyer (Hlongwane *et al.*, 2020; Madzimure *et al.*, 2012), are rarely used in the formal production sector due to their slow growth and lower meat yield, rendering them unsuitable for intensive commercial breeding (Mendelsohn, 2003; Prolit, 2004).

Despite being less productive than commercial breeds, South African indigenous and undefined village pigs are well-adapted to harsh climates and exhibit disease resistance (Madzimure *et al.*, 2012). Their resilience is because these pigs have evolved and adapted over centuries to survive in harsh climatic conditions (*e.g.*, extreme temperatures, limited water availability, and poor-quality feed) that are common in most rural areas of South Africa as well as traditional farming practices where the pigs are often reared in environments with inadequate access to veterinary care, supplemental feeding, and modern housing (Medrano, 2013). Their adaptability to thrive on minimal resources, resistance to diseases, and hardiness makes the indigenous and village pigs particularly well-suited to low-input production systems that are prevalent among small-scale and village farmers. Despite the constraints, indigenous and

undefined village pigs continue to perform well, providing a reliable source of meat, income, and cultural value to the communities that rear them.

Although the South African indigenous and undefined village pigs are generally less productive in terms of growth rate and meat yield compared to commercial breeds, this resilience makes them invaluable for small-scale and village farmers (Madzimore *et al.*, 2012). Moreover, these indigenous breeds represent a critical reservoir of genetic diversity, crucial for coping with environmental changes and climate change and ensuring food security and the sustainability of pig farming in the face of future uncertainties. However, the introduction of commercial breeds, favoured for their rapid growth and high productivity, threatens these indigenous populations (Medrano, 2013). This is because the selection for traits such as rapid growth and high productivity may result in the loss of genetic diversity, increase the risk of inbreeding, genetic drift, and the potential fixation of deleterious alleles. These consequences could severely compromise adaptation potential, viability, and elevate the risk of extinction (Frankham, 1996; Palstra & Ruzzante, 2008). Despite the increased extinction risk, the risk status of these breeds is unknown due to missing or outdated population data (FAO, 2016). It is therefore important to understand and mitigate these risks.

To understand and mitigate these risks, accurate methods are needed to investigate the loss of genetic diversity, increased risk of inbreeding, genetic drift, and the potential fixation of deleterious alleles, of assessing genetic diversity and population structure are essential. Although microsatellites have long been a reliable tool for investigating genetic variability due to their high polymorphism, codominant inheritance, and even distribution throughout the genome, Single Nucleotide Polymorphisms (SNP) marker analysis has become the standard method for diversity analysis and genome-wide research (Vignal *et al.*, 2002). This is because SNP markers are abundant, genetically stable, and suitable for high-throughput automatic analysis, despite being bi-allelic. Advances in genome sequencing, although expensive, can identify SNPs and are used in various studies to characterize genetic diversity. However, SNPs generated from the SNP BeadChip present a cheaper alternative to sequencing and have successfully been used in population studies of pigs. For instance, Diao *et al.* (2019) observed a rapid decline in genetic diversity in indigenous pigs in South China using SNPs generated from the SNP BeadChip. In addition, technological advancements allow us to work with the big datasets produced with the SNP BeadChip. This includes softwares such as Golden Helix SNP Variation Suite (SVS) and Plink, which are useful to estimate effective population size (N_e), a theoretical measure of an idealized population size expected to experience the same rate of genetic diversity loss due to genetic drift (Corbin *et al.*, 2012). Additionally, tools like

Principal Component Analysis (PCA) (Patterson *et al.*, 2006), ADMIXTURE (Alexander *et al.*, 2009; Alexander & Lange, 2011), and Treemix (Pickrell & Pritchard, 2012) are crucial for assessing genetic diversity and population structure. The integration of these methods is vital for developing informed strategies to manage, improve, and conserve indigenous and village pig breeds.

As commercial breeds continue to dominate the market due to their superior productivity, the conservation of indigenous pig populations becomes even more urgent, as their genetic traits might be diluted or lost through crossbreeding and genetic erosion. This study aimed to evaluate the risk of inbreeding and crossbreeding with exotic commercial breeds within South African indigenous and village pig populations. This entailed genotyping indigenous and village pigs sampled from villages across South Africa using SNP BeadChip technology to determine genetic diversity, the level of admixture, and the risk of extinction of these pig populations due to crossbreeding with prevailing exotic breeds. The results of this study will assist in making decisions regarding the improvement, management, and conservation of indigenous and village pig breeds, ensuring their continued contribution to agricultural sustainability in South Africa. Ultimately, the results of this study will help protect indigenous and village breeds, which could serve as a safeguard against the unforeseen challenges that the agricultural sector may face in the years to come. Preserving these breeds is therefore not just about maintaining cultural heritage but also about securing genetic resources that could be crucial for future agricultural innovations.

4.3 Materials and Methods

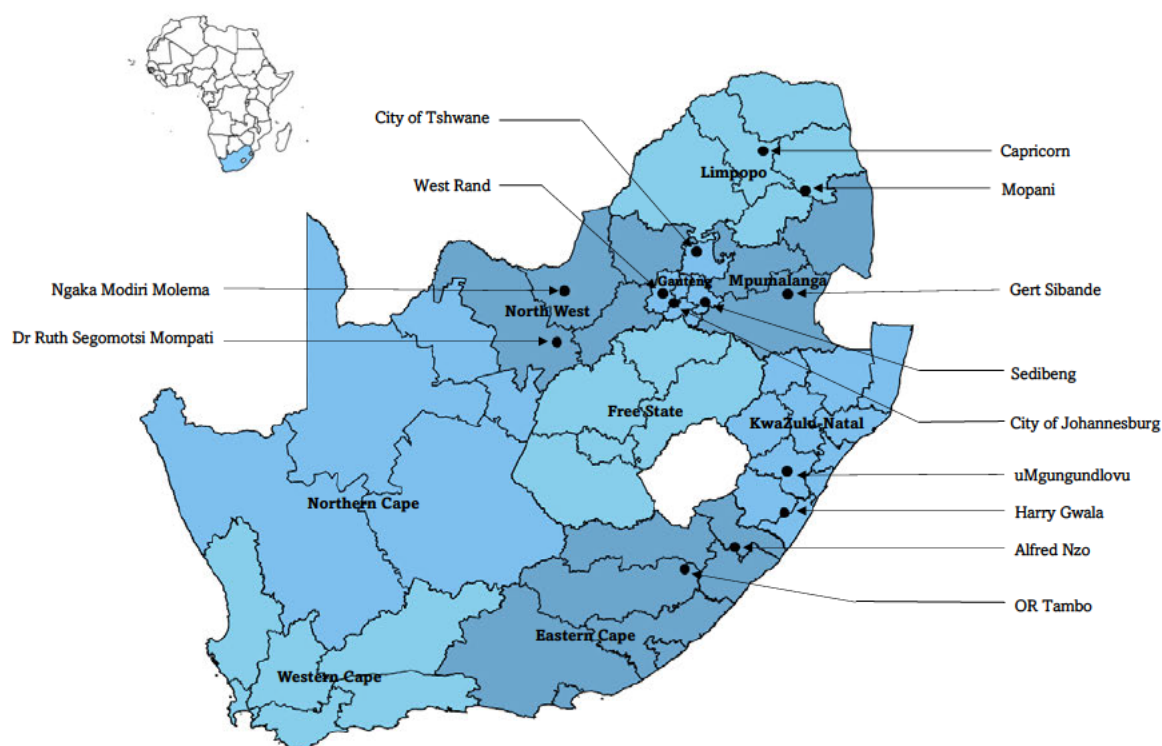


Figure 4.12 South African map showing the locations of the population sample sites.

4.3.1 Site Identification and Sample Collection

A total of 384 ear tissue samples were collected from villages across South Africa to represent the national status of the indigenous and village pig populations. Samples were collected from three pigs per household, specifically targeting a piglet, a sow, and a boar. This collection captured familial relationships within households, enabling assessment of both within- and between-population genetic variation. Pigs were selected based on their phenotypic traits (coat colour and body frame) and availability. The indigenous and village pig populations were sampled from the following regions: Eastern Cape in Alfred Nzo (EC-ALN, $n = 28$) and Oliver Reginald Tambo (EC-ORT, $n = 32$) districts; Gauteng in City of Johannesburg (GP-JHB, $n = 08$), Sedibeng (GP-SBD, $n = 34$), Tshwane (GP-TSW, $n = 30$) and West Rand (GP-WRD, $n = 12$) districts; KwaZulu-Natal in Harry Gwala (ZN-HGL, $n = 38$) and uMgungundlovu (ZN-UMG, $n = 25$) districts; Limpopo in Capricorn (LP-CAP, $n = 35$) and Mopani (LP-MOP, $n = 49$) districts; Mpumalanga in Gert Sibande (MP-GSD, $n = 37$) district; and North West in Ngaka Modiri Molema (NW-NMM, $n = 21$) and Dr. Ruth Segomotsi Mompati (NW-RSM, $n = 35$) districts (**Figure 4.1**).

4.3.2 DNA Extraction, SNP Genotyping, and Quality Control

Genomic DNA was extracted from ear tissue samples at the Agricultural Research Council – Biotechnology Platform (ARC-BTP) using Qiagen DNA Extraction Kit (Qiagen, Valencia,

CA), following the manufacturer's instructions. The quality and quantity of DNA were assessed using a Qubit® 4.0 fluorometer (Life Technologies, Thermo Fisher Scientific, USA) and gel electrophoresis. The extracted DNA was genotyped using the Porcine SNP80k BeadChip (Illumina, San Diego, CA, USA) with 68,528 SNPs and an average gap length of 38kb. SNPs were scanned using the Illumina iScan® system, and genotype calling, and SNP clustering were performed using GenomeStudio software (Illumina Inc., San Diego, CA, USA). Plink files (map file for SNP panel and ped file for pedigree) were generated for analysis.

SNP quality control and data combination were performed using Golden Helix Variation Suite (SVS) version 8.9.1 (Golden Helix Inc., Bozeman, MT, USA; www.goldenhelix.com). SNPs with call rate >85%, Minor Allele Frequency (MAF) <0.02, and Hardy Weinberg (HWE) < 10⁻⁴ were removed to generate a data set for F_{ST} analysis and SNeP. SNPs located on the sex chromosomes (X, XY, and Y) and 0 were also removed. For downstream analysis, additional QC filtering was applied to the combined dataset, removing SNPs with high LD ($r^2 = 0.5$) and estimating Identity-by-descent ($IBD \geq 0.65$) to prune closely related and duplicate samples. SVS was used to generate a dataset for analysing population structure with Principal Component Analysis (PCA) and ADMIXTURE.

4.3.3 Data Analyses

4.3.3.1 Genetic Diversity Indices

To observe the genetic diversity among the village pig populations, various genetic variability parameters including polymorphic information content (PIC), percentage of polymorphic loci ($\%P$), mean allele number per locus (N_a), and effective allele number ($N_e^{alleles}$). The calculations for N_a , $N_e^{alleles}$, PIC , and $\%P$ were done in Rstudio using data.table and dplyr packages. Observed and Expected heterozygosity (H_O and H_E) were estimated at the base of SNP information using PLINK 1.09 (Purcell *et al.*, 2007). Heterozygote deficiency within breed (F_{IS}) was estimated using formula $(H_E - H_O)/H_E$ from heterozygosity.

4.3.3.2 Effective Population Size

Effective population size (N_e) was estimated using Corbin *et al.* (2012) formula embedded in SNeP software (Barbato *et al.*, 2015), a software developed using the linkage disequilibrium method for analysing effective population size.

The formula:

$$N_{T(t)} = \left(1/(4f(Ct))\right) * \left(1/E[r^2_{adj} | Ct]\right) - a$$

where:

$NT(t)$ = effective population size t generations ago

Ct = recombination rate of specific distance between markers (formula by (Sved, 1971))

α = correction for mutation occurrence

$r2adj$ = adjusted LD for sample size and gametic phase

4.3.3.3 Population Genetic Differentiation, Structure and Ancestry Estimation

The formula of Weir and Cockerham (1984) embedded in the SNP & Variation Suite was used to estimate the pairwise F_{ST} values of the populations. For the analysis of population structure, admixture, and ancestry estimation, all autosomal SNPs that successfully passed the initial quality control check were merged with genotypes from indigenous (Kolbroek and Windsnyer) and commercial (Duroc, Large White, and South African Landrace) breeds, extracted from the Hlongwane *et al.* (2020) dataset. These additional 100 genotypes were included as reference populations representing pure breeds, providing a baseline for comparison to accurately assess admixture, crossbreeding effects, and genetic integrity in South African indigenous and village pig populations. PCA (Patterson *et al.*, 2006) was used to explore the population structure and patterns of admixture among the village pig populations in relation to the reference indigenous and commercial pig breeds (Hlongwane *et al.*, 2020). The PCA was done using SNP & Variant Suite version 8.9.1 (Golden Helix Inc., Bozeman, MT, USA; www.goldenhelix.com) and eigenvector methodology for population clustering. To unravel historical admixture and ancestry estimation, we ran admixture analysis using ADMIXTURE version 1.3.0 (Alexander *et al.*, 2009; Alexander and Lange, 2011). The number of ancestral clusters was set from $K = 1$ to $K = 25$ to reassign each sample to its population of origin using the block-relaxation method algorithm for point estimation and terminating the objective when the estimates increased by < 0.0001 . The optimal K -value was the one with the lowest cross validation (CV) error. The ancestry proportions at the optimal K -value were tabulated. Genesis version 0.2.5 (Buchmann & Hazelhurst, 2014) was subsequently used to construct ADMIXTURE boxplots.

To complement the PCA and admixture analysis, TreeMix (Pickrell & Pritchard, 2012) was used to assess the genetic relationships and divergence of the South African village pig populations. This technique draws a maximum likelihood-based phylogram that illustrates ancestral admixture and divergence between populations. Migrations were tested from $m = 0$ to $m = 10$ using five replicates per $-m$ to assess consistency. The most predictive number of migration edges ($m = 6$) was selected using the *optM* function in the R package *OptM* (Fitak, 2021). For this analysis, the Duroc breed was set as the root of the tree due to their clear separation from other breeds in the previously described analyses. Blocks of 1000 SNPs and

the -se option were used to calculate standard errors of migration proportions. To visualize the graph, the R script plotting_funcs.R was used on ggplot2 package.

4.4 Results

4.4.1 Genotyping and Quality Control

The number of SNPs and individuals remaining after initial QC and further QC are presented in **Table 4.1**. The 384 pig genotypes and the merged dataset with 484 individuals included 32,743 SNPs distributed over the autosomes, which remained after QC to remove SNPs (<85% call rate, MAF <0.2, HWE <10⁻⁶). Among the populations included, the ZN-HGL population retained the greatest number of SNPs (37,871) and EC-ALN retained the least number of SNPs (33,862) (**Supplementary Figure 4.1**). After LD and IBD pruning, a total of 20,761 SNPs and 448 individuals were retained and used to analyse population structure.

Table 4.1 Summary Results of SNP Quality Control

Quality Control Standard	SNPs/Individuals	
	Initial Quality	Further Quality
	Control	Control
Number of Individuals	384	484
Total number of SNPs	58 690	58 690
Total number of Autosomal SNPs	41 752	41 752
SNP with MAF < 0,2	2 867	2 858
SNP not in Hardy-Weinberg Equilibrium $p < 10^{-6}$	5 236	10 387
With call rate < 0.85	1 565	1 572
LD pruning	-	10 839
Identity-by-descent	-	28
SNPs on chromosome X	1 431	1 431
SNPs on chromosome Y	22	22
SNPs on chromosome 0	15 503	15 503
Individuals after quality control	380	448
SNPs used after quality control	32 743	20 761

4.4.2 Genetic Diversity Indices

Overall, the population-level indices of genetic diversity varied across populations (**Table 4.2**) according to the following: for all the populations, the set of SNPs analysed had PIC values below 0.35, with a mean PIC of 0.330. The ZN-HGL population recorded the highest PIC (0.342) and polymorphic percentage of 94%. However, GP-JHB population recorded the lowest PIC (0.311) and polymorphic percentage of 84%. The mean N_a varied from 1.837 ± 0.370 to 1.929 ± 0.257 with a mean of 1.908 ± 0.289 , whereas $N_e^{alleles}$ varied from 1.534 ± 0.352 to 1.590 ± 0.331 with a mean of 1.575 ± 0.337 . Heterozygosities for each population studied are shown in **Figure 4.2**, values in **Supplementary Table 4.1**. The observed heterozygosity values ranged from 0.295 ± 0.054 (EC-ALN) to 0.400 ± 0.187 (GP-JHB). H_o values ranged from 0.362 ± 0.132 (ZN-UMG) to 0.372 ± 0.126 (EC-ALN). H_E was lower than observed in some populations (i.e. GP-WRD, $H_o = 0.381 \pm 0.177$ and $H_E = 0.364 \pm 0.128$). The EC-ALN, GP-JHB and GP-WRD exhibited negative F_{IS} values. The average H_o , H_E and heterozygote deficiency within breed are 0.347 ± 0.139 , 0.368 ± 0.126 and 0.038 ± 0.254 , respectively.

Table 4.2 Population genetic variability parameters

Population	N	PIC	%P	N_a	$N_e^{alleles}$
EC-ALN	28	0.309	88	1.876 ± 0.330	1.534 ± 0.352
EC-ORT	32	0.333	91	1.911 ± 0.284	1.581 ± 0.340
GP-JHB	08	0.311	84	1.837 ± 0.370	1.540 ± 0.354
GP-SBD	34	0.332	92	1.920 ± 0.271	1.577 ± 0.336
GP-TSW	30	0.336	92	1.925 ± 0.263	1.584 ± 0.331
GP-WRD	12	0.324	89	1.891 ± 0.312	1.562 ± 0.340
LP-CAP	35	0.340	93	1.929 ± 0.257	1.590 ± 0.328
LP-MOP	49	0.337	93	1.927 ± 0.260	1.589 ± 0.334
MP-GSD	37	0.339	92	1.924 ± 0.265	1.590 ± 0.331
NW-NMM	21	0.327	90	1.900 ± 0.300	1.568 ± 0.339
NW-RSM	35	0.336	92	1.923 ± 0.267	1.585 ± 0.333
ZN-HGL	38	0.342	94	1.929 ± 0.296	1.596 ± 0.327
ZN-UMG	25	0.330	91	1.910 ± 0.286	1.573 ± 0.338
Mean		0.330 ± 0.010	91	1.908 ± 0.289	1.575 ± 0.337

N , sample size; PIC, polymorphic information content; %P, percentage of polymorphic loci; N_a , mean allele number per locus; $N_e^{alleles}$, effective alleles number ($1 / (\sum p_i^2)$). Mean $\pm$ standard deviation.

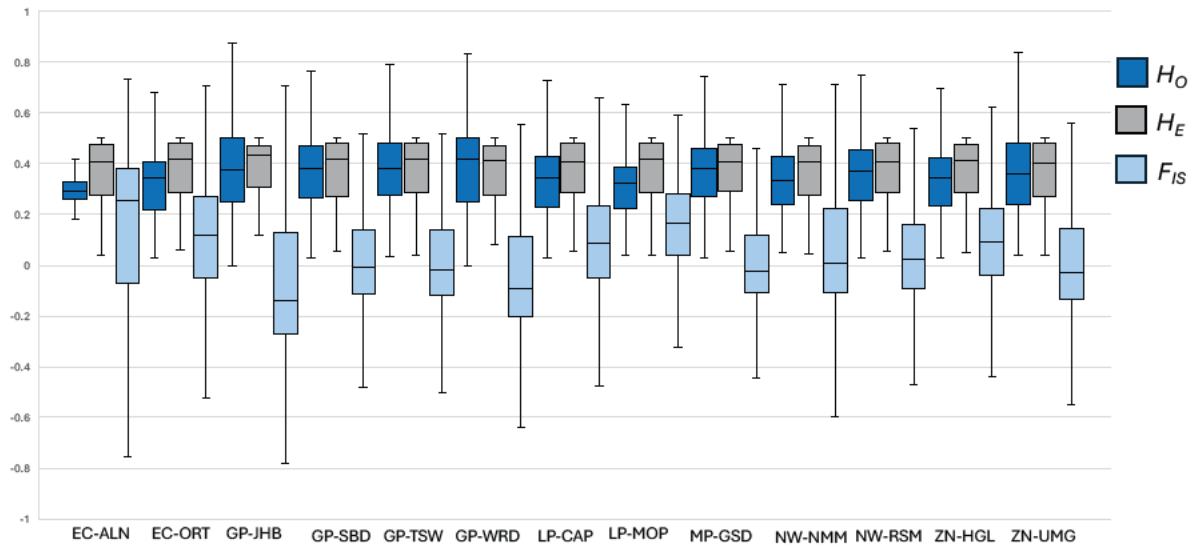


Figure 13 Heterozygosity analysis (observed heterozygosity (H_O), expected heterozygosity (H_E), and heterozygote deficiency within population (F_{IS}))

4.4.3 Effective Population Size

The estimated population size over generations is presented in **Figure 4.3**, illustrating the historical and recent demographic trends of the studied pig populations. The population size kept declining over the years across all populations. However, $N_e > 80$ for all the populations (**Supplementary Table 4.2**). The variation in N_e across generations was lowest for GP-JHB ($N_e = 81$) and highest for LP-MOP ($N_e = 252$). A sharp decline was observed for GP-JHB population between 292 and 12 generations ago. Although the rate of decline varied slightly among populations, all showed the same downward trajectory.

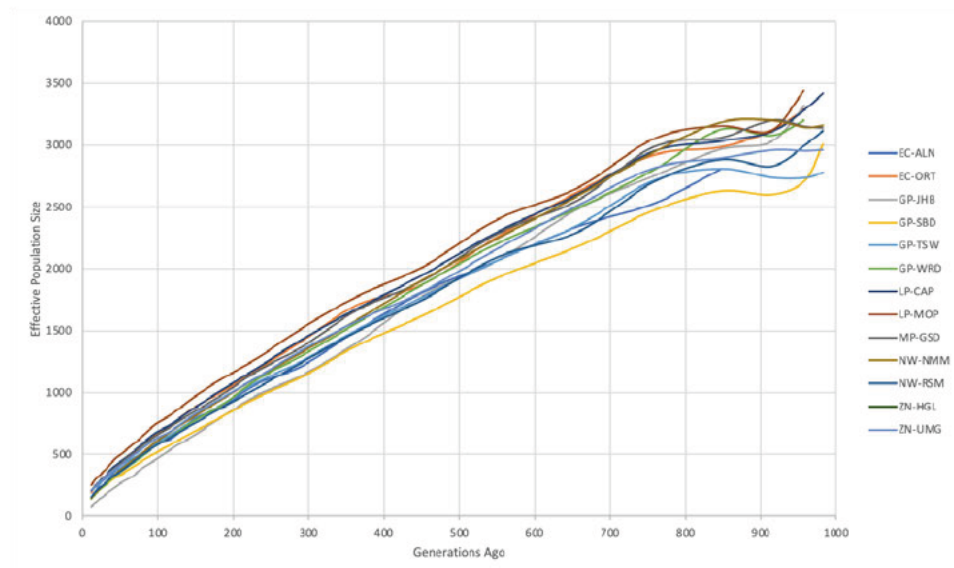


Figure 14 The estimated effective population size of 11 South African village pig populations over time with GONE (y-axis denotes the estimated N_e , while the x-axis represents generations ago. The different coloured lines indicate each population)

4.4.4 Population Genetic Differentiation, Structure, and Ancestry Estimation

F_{ST} values range from zero (complete sharing) to one (no sharing) (Bird *et al.*, 2017). The values from population pairwise F_{ST} are presented in **Supplementary Table 4.3** and graphically illustrated in **Figure 4.4**, where the colour gradient displayed in the scale at the top of the figure indicates the magnitude of genetic distances between populations, where similar colours reflect closer genetic relationships and increasingly contrasting colours represent greater divergence. Considerably low F_{ST} values were observed between the village populations, ranging from 0.016 to 0.111. The lowest differentiation was shown by GP-TSW compared to GP-SBD and GP-WRD ($F_{ST}= 0.016$). The highest observed F_{ST} was exhibited by EC-ALN when compared to GP-JHB ($F_{ST}= 0.111$).

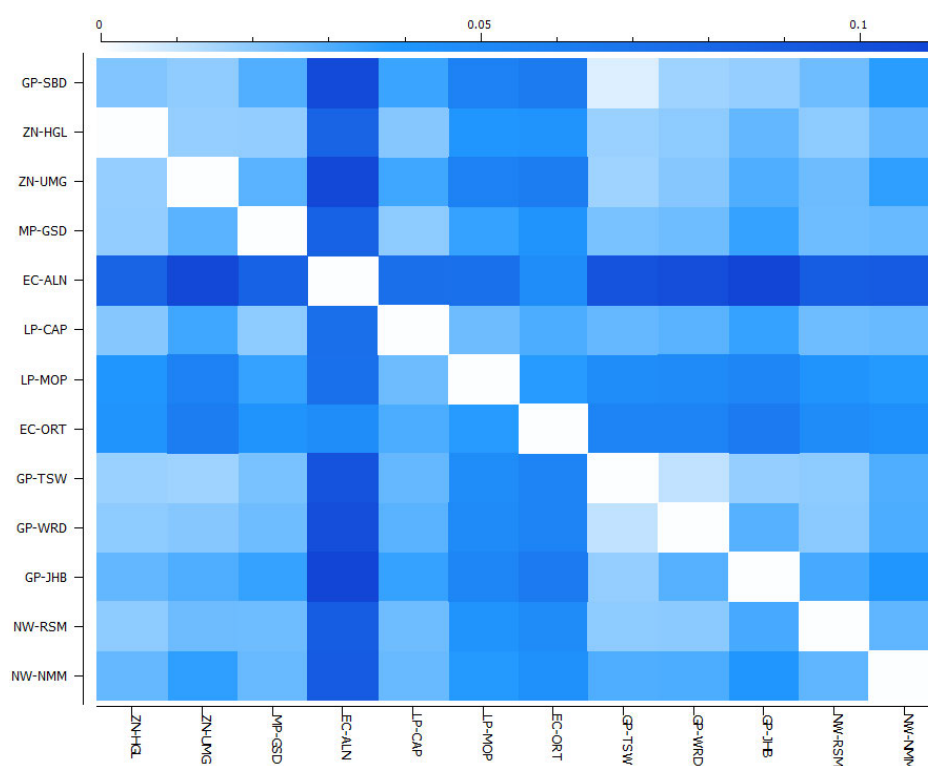


Figure 15 Graphically representation of the pairwise F_{ST} values among the 13 village pig populations studied. Colours representing populations distances are defined on the scale at the top of the figure.

Principal Component Analysis was used to identify the genetic structure of the South African village pig populations. The PCA clustered the populations based on their genetic composition, which resulted in five distinct clusters (**Figure 4.5**). Principal component one (PC1) and two (PC2) accounted for 18.17% and 8.92% of the total genetic variability, respectively. The PB-DUR, PB-LWT, and PB-SAL distinctly clustered in Clusters 1, 4, and 5, respectively. The indigenous breeds (PB-KOL and PB-WIN) clustered together with the village pig populations

in Cluster 3. Cluster 2 was formed by GP-SBD population which some of which was found in Cluster 3 with other village populations. Overall, the PCA demonstrated tight clustering of commercial breeds, and separately, due to strong artificial selection and controlled breeding while the indigenous and village populations formed a broad, overlapping cluster. This reflects their history of uncontrolled mating, interbreeding, and shared ancestry, leading to high levels of genetic admixture across communities and regions.

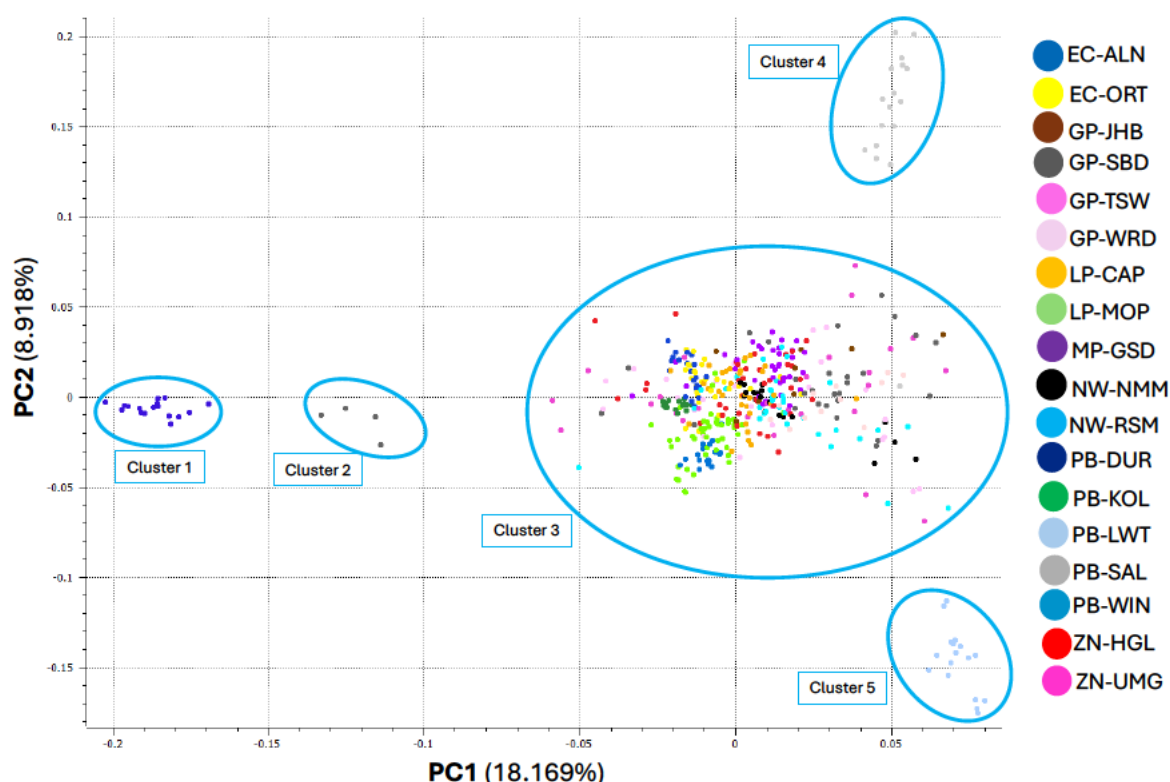


Figure 16 Principal Component Analysis Results of the eighteen pig populations including Hlongwane *et al.*, (2020) SNP genotypes (21 084 SNPs)

The genetic structure amongst the village pig populations was also evidenced using ADMIXTURE analysis, and the optimum K -value was observed at $K = 14$ (Figure 4.6). The structure results based on ADMIXTURE are shown in Figure 4.7 for values ranging between $K = 2$ to $K = 14$. Based on the visual observation of the admixture plot, at $K = 2$, commercial, indigenous, and village populations share common ancestry. At $K = 3$, PB-DUR and PB-KOL pigs separated from the rest, and by $K = 6$, PB-LWT, PB-WIN, and PB-SAL pigs also formed distinct clusters. As K increased, commercial and indigenous populations remained distinct, while village populations showed high levels of admixture, indicating they are not homogenous. Ancestry proportions of each pre-defined pig population in the fourteen genetic clusters identified from analysis are shown in Supplementary Table 4.4. The pig populations

PB-DUR and PB-KOL resulted in the highest ancestry proportions (100%; 1.000 ± 0.000) and (99%; 0.993 ± 0.022) at Clusters 12 and 5, respectively. Cluster 1 was dominated by PB-SAL (81%; 0.815 ± 0.200) and had slight contributions from all the village populations. In Cluster 2, PB-SAL dominated with (94%; 0.938 ± 0.075) and had significant contributions from GP-TSW (11%; 0.106 ± 0.112), NW-RSM (12%; 0.119 ± 0.121), and ZN-UMG (12%; 0.123 ± 0.119). The village population EC-ORT was dominant in Cluster 3 at 52% (0.516 ± 0.282), followed by EC-ALN (26%; 0.259 ± 0.239) and LP-CAP (10%; 0.103 ± 0.100). The Eastern Cape village population, EC-ALN dominated Cluster 8 with 64% (0.640 ± 0.328) followed by EC-ORT with (15 %; 0.152 ± 0.096). Cluster 6 was dominated by Gauteng populations, GP-JHB (21%; 0.214 ± 0.332), GP-SBD (22%; 0.219 ± 0.211), GP-TSW (22%; 0.221 ± 0.284), GP-WRD (18%; 0.184 ± 0.228), and substantial contribution from KZN population, ZN-UMG (11%; 0.113 ± 0.170). Clusters 4, 9, and 13 constituted proportions from pig populations that had the most variations, except PB-DUR and PB-KOL. PB-WIN population were represented in cluster 10 at (92%; 0.916 ± 0.126). Clusters 7, 11, and 14 were dominated by MP-GSD (39%; 0.387 ± 0.353), NW-RSM (24%; 0.244 ± 0.418), and LP-MOP (22%; 0.224 ± 0.327), respectively. The proportions obtained and the clustering structure did indicate that the individuals within the different populations share similar genetic makeup.

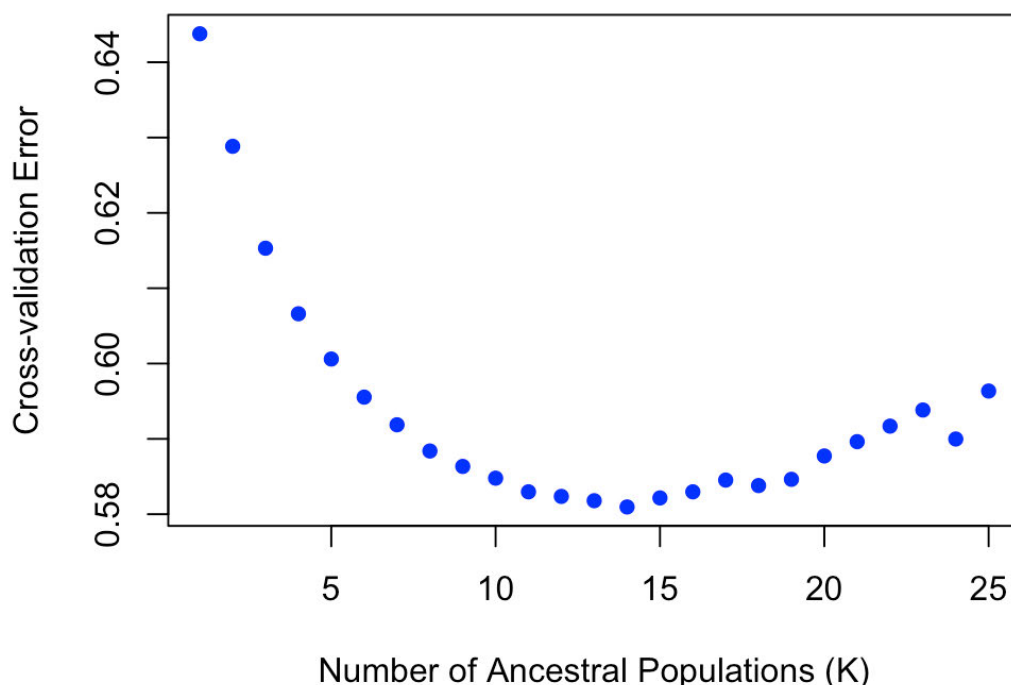


Figure 17 Cross-validation plot of the admixture analysis

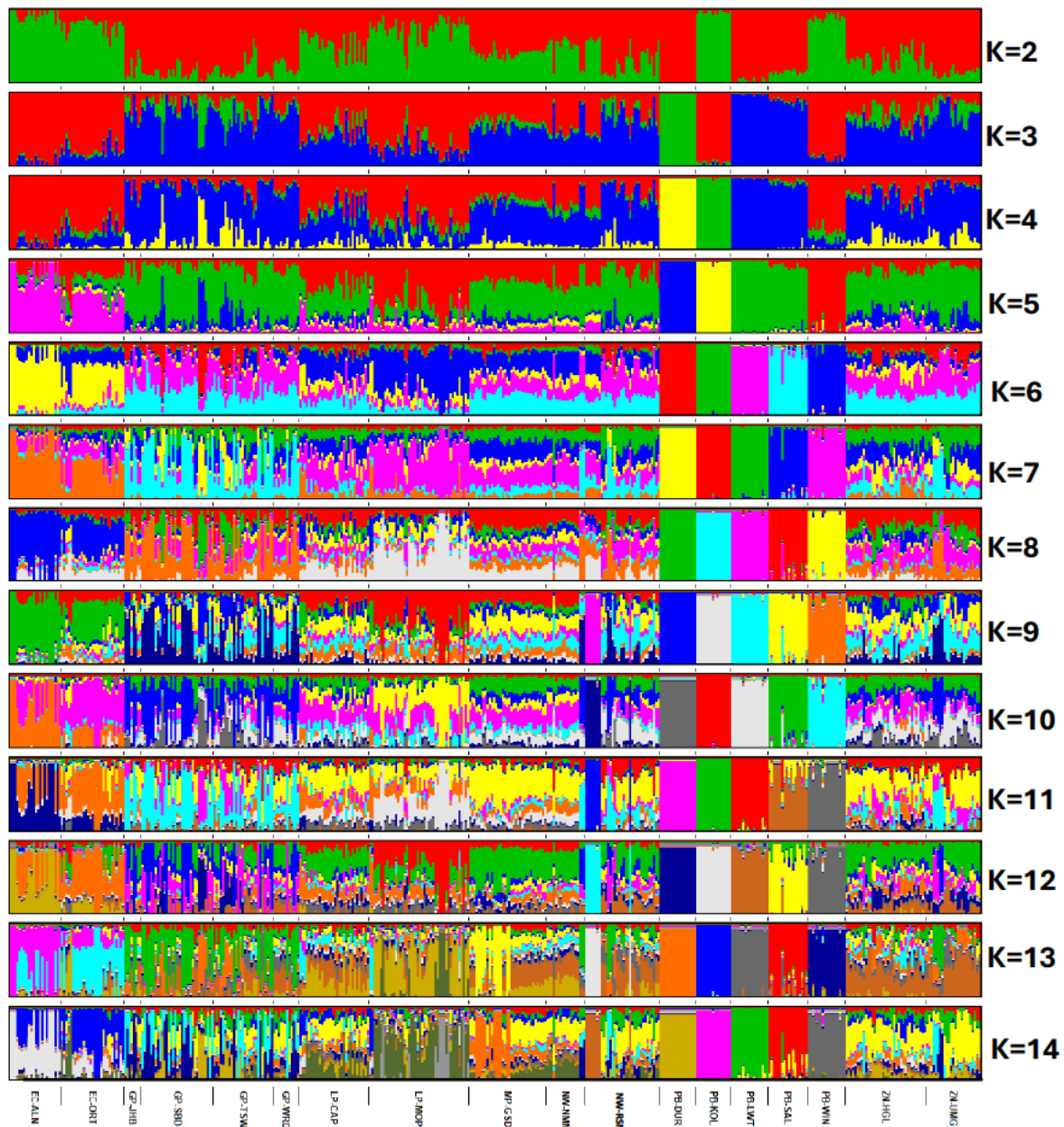


Figure 18 Admixture plot showing population clustering at $K = 14$. Each animal is represented by a single vertical line divided into K colours, where K is the number of assumed ancestral clusters that ranged from $K = 2$ to $K = 14$; the coloured segment shows the individual's estimated membership proportions in each cluster. Populations are grouped by type: the village/local populations (EC-ALN, EC-ORT, GP-JHB, GP-SBD, GP-TSW, GP-WRD, LP-CAP, LP-MOP, MP-GSD, NW-NMM, NW-RSM, ZN-HGL, ZN-UMG), commercial exotic breeds (PB-DUR, PB-LWT, PB-SAL), and indigenous breeds (PB-WIN, PB-KOL).

The maximum likelihood phylogram constructed by Treemix analysis (Figure 4.9) provided additional evidence supporting the findings of PCA and ADMIXTURE analyses. The linear method implemented in the *optM* function indicated a major changing point in the log likelihood at six migration events explaining marginally higher 99.8% (Figure 4.8) variance for the relatedness between the populations. High migration gene flow was observed between

$m = 1$ (PB-WIN to PB-SAL), $m = 3$ (GP-TSW to MP-GSD). Two migration events with a weaker signal of gene flow were highlighted between $m = 2$ (PB-DUR to EC-ORT) and $m = 4$ (GP-TSW to EC-ALN). There was gene flow at $m = 5$ (PB-LWT to PB-KOL) and $m = 6$ (PB-DUR TO EC-ALN).

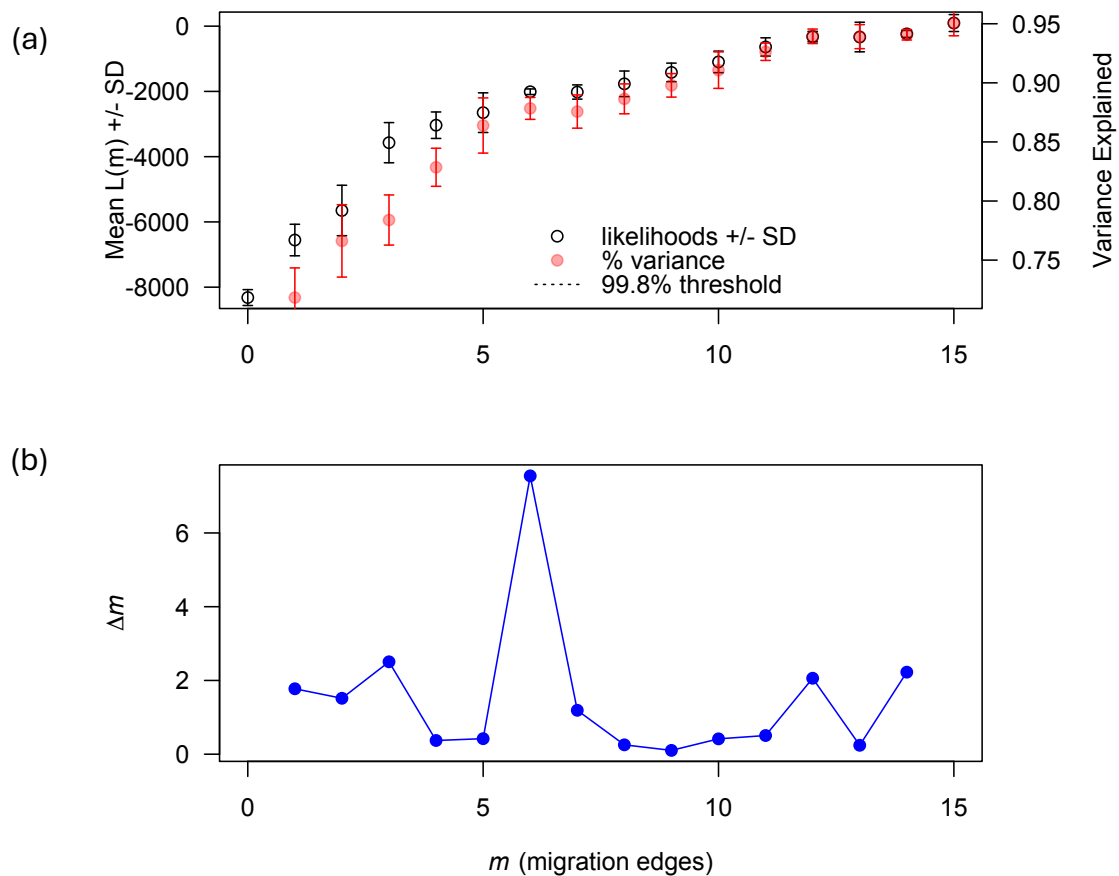


Figure 19 The output generated by OptM for the simulated dataset with $m = 6$ migration edges. (a) The mean and standard deviation (SD) across 10 iterations for the composite likelihood $L(m)$ (left axis, black circles) and the proportion of variance explained (right axis, red "x"s). The 99.8% threshold is shown as a horizontal dotted line. (b) The second-order rate of change (Δm) across different values of m .

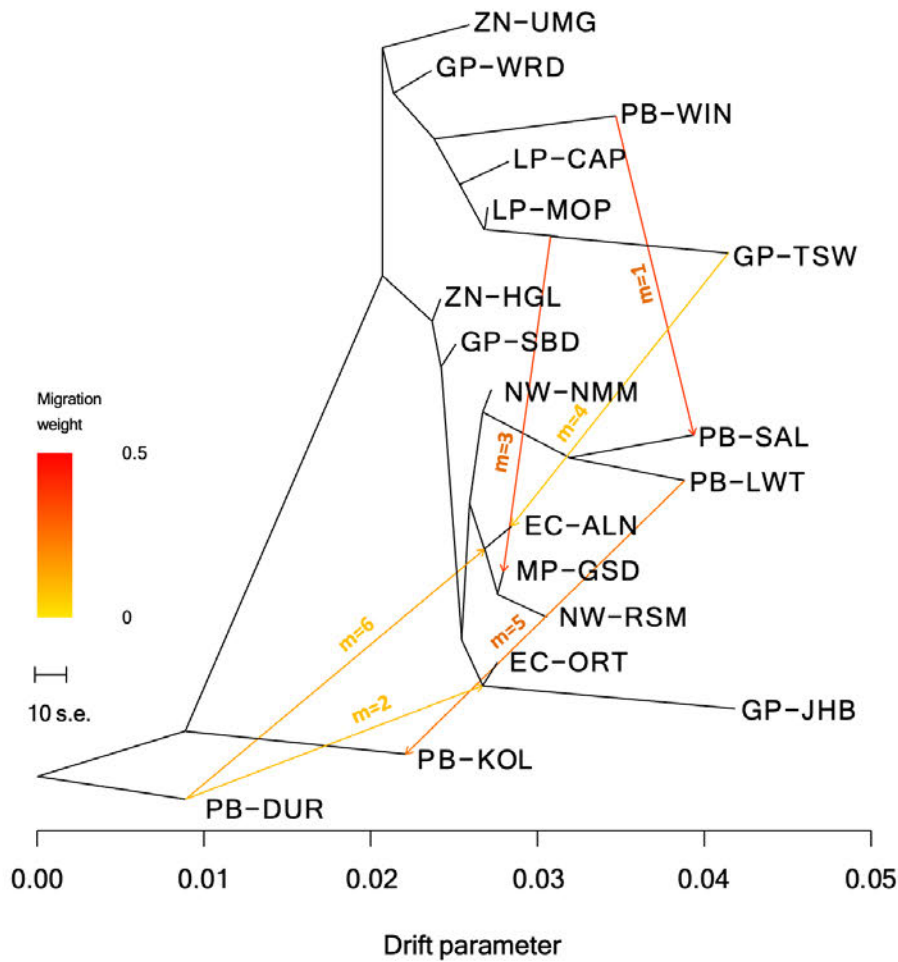


Figure 20 Population splits and mixture of the tested commercial, indigenous and village pig populations inferred using TreeMix. Six migrations were allowed as inferred by OptM. The migration edges are coloured according to their weight. The scale bar indicates ten times the average standard error of the values in the covariance matrix.

4.5 Discussion

The Illumina PorcineSNP80 was used to study the genetic diversity of indigenous and village pig populations in South Africa. Even though several studies successfully employed this array for analysis (Herrero-Medrano *et al.*, 2013), our results indicated that the SNP markers were moderately informative. This is because our *PIC* values (0.330 ± 0.010) fell within the range of 0.25 to 0.50, and *PIC* values between 0.25 and 0.50 are considered moderately informative (Botstein *et al.*, 1980). This might be due to SNP markers being bi-allelic by nature, which limits *PIC* values to 0.5, as well as the relatively low mutation rates of SNPs (Coates *et al.*, 2009; Eltaher *et al.*, 2018). Additionally, fewer SNPs were informative for the indigenous and village populations, likely due to their lower representation in the SNP array design. This is similar to what was found in studies of genetic diversity of Asian goats (Lin *et al.*, 2013) and

Chinese cattle (Wang *et al.*, 2018). This emphasized the need to develop a SNP array that included South African indigenous and village populations.

In spite of these limitations, our results clearly demonstrated that the introduction of commercial breeds has impacted the diversity of the indigenous and village breeds. The mean N_a was 1.908 ± 0.289 and $N_e^{alleles}$ was 0.575 ± 0.337 , indicating relatively low genetic diversity likely influenced by small population size, genetic drift, and inbreeding. However, it is important to note that high N_a values alone do not provide a complete picture of genetic diversity. Other factors, such as heterozygosity, the effective number of alleles, and the presence of private alleles, also contribute to the overall genetic diversity of a population (Pham *et al.*, 2013). In this study, low heterozygosity values ($H_o = 0.347 \pm 0.139$ and $H_E = 0.368 \pm 0.126$) were observed, indicating poor genetic variability. Similar values were reported by (Muñoz *et al.*, 2019) for European local pig breeds, which across-breeds averaged values for H_o and H_E were $0.297 (\pm 0.053)$ and $0.303 (\pm 0.054)$, respectively. High mean observed heterozygosity levels were reported by (Swart *et al.*, 2010) in the South African (0.634), the Mozambican (0.692), and the Namibian local pigs (0.531). However, these studies used microsatellites and not SNPs; thus, the high genetic diversity is expected. Also, the observed heterozygosity in the village pig populations did not differ from the expected value by more than 0.030. The results indicate that South African village pigs are genetically heterogeneous, reflecting high levels of admixture among individuals across populations, even though morphometric traits presented them as relatively homogeneous in **Chapter 3**. Mating and crossbreeding practices were largely unsupervised, making these populations susceptible to inbreeding and uncontrolled genetic mixing with other breeds (Halimani *et al.*, 2010). This continuous gene flow among the populations likely explains the low levels of genetic differentiation observed.

The F_{IS} provides valuable insights into the levels of heterozygosity and relatedness within populations, clearly showing high inbreeding occurrence within the 10 village populations. The Alfred-Nzo, Johannesburg, and West Rand populations showed an excess of heterozygotes, as indicated by the negative F_{IS} values (-0.015 ± 0.738 , -0.078 ± 0.318 , and -0.046 ± 0.281) obtained. The negative F_{IS} values suggest that mates are not as closely related on average as expected, as indicated by negative F_{IS} values, which signify excess heterozygotes. This suggests that individuals are less related than expected in random-mated subpopulations (Kardos *et al.*, 2016). Across the other 10 village pig populations, a significant heterozygote deficit was observed relative to Hardy–Weinberg proportions, indicating that high inbreeding occurred within these populations. Additionally, the low N_e levels in recent generations of

South African village pig populations are concerning. This is because N_e values below 65 suggest that the pig populations may be at risk (Simon, 1999). In this study, $N_e < 65$, even though there is a decline in population size over the years, they cannot currently be classified as being at risk. The ongoing decline in population size may persist as these populations are anticipated to endure fragmented sizes due to inadequate breeding and management practices. Populations with narrow genetic backgrounds, frequently the case for locally adapted pig breeds, are mostly vulnerable to issues such as loss of genetic diversity and inbreeding depression (Biscarini *et al.*, 2015). The long-term viability of populations may be threatened by their small size, which reduces fitness traits and increases the effects of genetic drift (Frankham *et al.*, 1995).

The results of this study clearly demonstrate that geographical proximity, environmental similarity, and gene flow have shaped the evolution of these pigs. Previous studies have shown that habitat isolation and local environmental conditions significantly influence genetic differentiation among pig breeds across different regions (Wang *et al.*, 2024; Zhong *et al.*, 2023). Results obtained from the PCA were in congruence with those observed from ADMIXTURE, indicating low genetic differentiation within and between populations. The PCA analyses identify five genetic clusters, which present individuals that share a significant proportion of their ancestry and were graphically close to each other in cluster 3. The separate clustering of the Duroc from other populations on PCA clustering could be attributed to their distinct genetic profiles and evolutionary origins (Hlongwane *et al.*, 2020). The Duroc breed is thought to have been imported into America by Christopher Columbus from Portugal or Spain and then undergone further breed development in the late 1800s in the US (Rothschild and Ruvinsky, 2011). Moreover, the differentiation between Duroc and other populations may be due to different selection pressures in its genome, which caused genetic mutation in some genes (Amaral *et al.*, 2011; Yu *et al.*, 2020). The values in this study contrast with those reported by Diao *et al.* (2019), who observed F_{ST} values ranging from 0.010 to 0.852 among indigenous pigs in South China. In comparison, the low pairwise F_{ST} values obtained in this study (0.016 – 0.111) indicate weak genetic differentiation, suggesting higher gene flow or admixture among South African village pig populations (Frankham *et al.*, 2002). A value of 0 indicates no genetic differentiation, while a value of 1 indicates complete genetic separation. This suggests high genetic similarity among the groups, substantial historical or ongoing gene flow, and overall weak population structure. These results coincided with the PCA results, where all the village populations clustered together in cluster 3, accounting for 27.09% of the total variation. This showed that the presented individuals share a significant proportion of their ancestry. By using PCA together with the admixture analysis, we were able to better understand

the genetic relationship, population structure, and genetic proportions of the eleven village pig populations.

The TreeMix analysis revealed significant introgressive hybridization between sampled populations and pure breeds, influencing their genetic diversity. Notably, Duroc and Kolbroek breeds shared a common ancestry, likely due to the historical relationship between Iberian pigs and American breeds (Lee *et al.*, 2020). This shared ancestry suggests that genomic tools developed for Duroc could also be applied to Kolbroek pigs, as Misztal *et al.* (2022) proposed for closely related breeds. Gene flow was detected at $m = 1$ between Windsnyer and South African Landrace and at $m = 5$ between Large White and Kolbroek, indicating potential introgression of genetic material from shared ancestral sources or earlier migration events (Ciani *et al.*, 2014). Additionally, gene flow was observed at $m = 3$ and $m = 4$ among Gauteng (Tshwane), Eastern Cape (Alfred-Nzo), and Mpumalanga (Gert Sibande), supporting previous findings of low genetic differentiation and significant admixture among village pigs (Hlongwane *et al.*, 2020; Mujibi *et al.*, 2018). This gene flow may be driven by the exchange and trade of pigs between farmers across regions (Madzimure *et al.*, 2012; Matabane *et al.*, 2015; Gcumisa *et al.*, 2016). Furthermore, gene flow at $m = 2$ and $m = 6$ from Duroc to Eastern Cape populations (Alfred Nzo and OR Tambo) is likely a result of the introduction of commercial breeds like Duroc to improve traits such as growth rate and meat quality (Banerjee *et al.*, 2020). These findings highlight the complex genetic interactions shaping the diversity and production potential of South African pig populations, driven by both historical migrations and modern crossbreeding practices.

4.6 Conclusion

These findings highlight the complex genetic landscape of South African pig populations, emphasizing the ongoing impact of crossbreeding on their genetic diversity. While studies on commercial pig breeds are relatively common and help understand the broader genetic landscape of domestic pigs, research on indigenous and village pig populations remains limited, despite their importance for conservation and sustainable management. The findings obtained from this study can inform preservation strategies and the management of genetic diversity within South African indigenous and village pig populations. In addition, these results can guide decisions regarding the extent of crossbreeding with exotic breeds, helping to mitigate risks of extinction and maintain the unique characteristics of these local pig populations. It is advised that further studies investigating the mating systems and their effects on population diversity and structure should be undertaken to ensure the prevention of the risk of extinction and that careful management strategies are encouraged.

4.7 References

- Ai, H., Huang, L., & Ren, J. (2013). Genetic Diversity, Linkage Disequilibrium and Selection Signatures in Chinese and Western Pigs Revealed by Genome-Wide SNP Markers. *PLoS ONE*, 8(2), 56001. <https://doi.org/10.1371/journal.pone.0056001>
- Alexander, D. H., & Lange, K. (2011). Enhancements to the ADMIXTURE algorithm for individual ancestry estimation. *BMC Bioinformatics*, 12. <https://doi.org/10.1186/1471-2105-12-246>
- Alexander, D. H., Novembre, J., & Lange, K. (2009). Fast model-based estimation of ancestry in unrelated individuals. *Genome Research*, 19(9), 1655–1664. <https://doi.org/10.1101/gr.094052.109>
- Amaral, A. J., Ferretti, L., Megens, H. J., Crooijmans, R. P. M. A., Nie, H., Ramos-Onsins, S. E., Perez-Enciso, M., Schook, L. B., & Groenen, M. A. M. (2011). Genome-Wide Footprints of Pig Domestication and Selection Revealed through Massive Parallel Sequencing of Pooled DNA. *PLOS ONE*, 6(4), e14782. <https://doi.org/10.1371/JOURNAL.PONE.0014782>
- Banerjee, P., Carmelo, V. A. O., & Kadarmideen, H. N. (2020). Genome-Wide Epistatic Interaction Networks Affecting Feed Efficiency in Duroc and Landrace Pigs. *Frontiers in Genetics*, 11. <https://doi.org/10.3389/FGENE.2020.00121/FULL>
- Barbato, M., Orozco-terWengel, P., Tapio, M., & Bruford, M. W. (2015). SNeP: A tool to estimate trends in recent effective population size trajectories using genome-wide SNP data. *Frontiers in Genetics*, 6(MAR), 1–6. <https://doi.org/10.3389/fgene.2015.00109>
- Bird, K. A., An, H., Gazave, E., Gore, M. A., Pires, J. C., Robertson, L. D., & Labate, J. A. (2017). Population structure and phylogenetic relationships in a diverse panel of Brassica rapa L. *Frontiers in Plant Science*, 8. <https://doi.org/10.3389/fpls.2017.00321>
- Biscarini, F., Nicolazzi, E., Alessandra, S., Boettcher, P., & Gandini, G. (2015). Challenges and opportunities in genetic improvement of local livestock breeds. *Frontiers in Genetics*, 5(JAN), 1–16. <https://doi.org/10.3389/FGENE.2015.00033/BIBTEX>
- Blench, R.M.; MacDonald, K.C. *The Origins and Development of African Livestock: Archaeology, Genetics, Linguistics and Ethnography*; UCL Press: London, UK, 2000.
- Botstein, D., White, R. L., Skolnick, M., & Davis, R. W. (1980). Construction of a Genetic Linkage Map in Man Using Restriction Fragment Length Polymorphisms. In *Am JHum Genet* (Vol. 32).
- Charlesworth, B. (2009). Fundamental concepts in genetics: Effective population size and patterns of molecular evolution and variation. *Nature Reviews Genetics*, 10(3), 195–205. <https://doi.org/10.1038/nrg2526>
- Ciani, E., Crepaldi, P., Nicoloso, L., Lasagna, E., Sarti, F. M., Moiola, B., Napolitano, F., Carta, A., Usai, G., D'Andrea, M., Marletta, D., Ciampolini, R., Riggio, V., Occidente, M., Matassino, D., Kompan, D., Modesto, P., Macciotta, N., Ajmone-Marsan, P., & Pilla, F. (2014). Genome-wide analysis of Italian sheep diversity reveals a strong geographic pattern and cryptic relationships between breeds. *Animal Genetics*, 45(2), 256–266. <https://doi.org/10.1111/AGE.12106>
- Coates, B. S., Sumerford, D. V., Miller, N. J., Kim, K. S., Sappington, T. W., Siegfried, B. D., & Lewis, L. C. (2009). Comparative Performance of Single Nucleotide Polymorphism and Microsatellite Markers for Population Genetic Analysis. *Journal of Heredity*, 100(5), 556–564. <https://doi.org/10.1093/jhered/esp028>
- Corbin, L. J., Liu, A. Y. H., Bishop, S. C., & Woolliams, J. A. (2012). Estimation of historical effective population size using linkage disequilibria with marker data. *Journal of Animal Breeding and Genetics*, 129(4), 257–270. <https://doi.org/10.1111/j.1439-0388.2012.01003.x>
- D'Alessandro, E., Arfuso, F., Floridia, V., Tardiolo, G., Fazio, F., Giannetto, C., Piccione, G., & Zumbo, A. (2022). Different genotype and a liquid whey-supplemented diet influence the resilience of pigs through immune-modulation and anti-inflammatory response. *Frontiers in Veterinary Science*, 9. <https://doi.org/10.3389/FVETS.2022.1046101>
- Diao, S., Huang, S., Xu, Z., Ye, S., Yuan, X., Chen, Z., Zhang, H., Zhang, Z., & Li, J. (2019). Genetic diversity of indigenous pigs from South China area revealed by SNP array. *Animals*, 9(6), 1–17. <https://doi.org/10.3390/ani9060361>
- Eltaher, S., Sallam, A., Belamkar, V., Emara, H. A., Nower, A. A., Salem, K. F. M., Poland, J., & Baenziger, P. S. (2018). Genetic diversity and population structure of F3:6 Nebraska Winter wheat genotypes using genotyping-by-sequencing. *Frontiers in Genetics*, 9(MAR), 330669. <https://doi.org/10.3389/FGENE.2018.00076/BIBTEX>

- FAO. (2016). Status of Animal Genetic Resources. Food and Agriculture Organization of the United Nations. Rome, 6 - 8 July 2016. June.
- Fitak, R. R. (2021). OptM: estimating the optimal number of migration edges on population trees using Treemix. *Biology Methods and Protocols*, 6(1). <https://doi.org/10.1093/BIOMETHODS/BPAB017>
- Frankham, R. (1996). Relationship of Genetic Variation to Population Size in Wildlife. *La Relación Entre la Variación Genética y el Tamaño Poblacional en Vida Silvestre. Conservation Biology*, 10(6), 1500–1508.
- Frantz, L., Meijaard, E., Gongora, J., Haile, J., Groenen, M. A. M., & Larson, G. (2016). The evolution of suidae. *Annual Review of Animal Biosciences*, 4, 61–85. <https://doi.org/10.1146/annurev-animal-021815-111155>
- Gcumisa, S. T., Oguttu, J. W., & Masafu, M. M. (2016). Pig farming in rural South Africa: A case study of uthukela district in KwaZulu-Natal. *Indian Journal of Animal Research*, 50(4), 614–620. <https://doi.org/10.18805/IJAR.11175>
- Giuffra, E., Kijas, J. M. H., Amarger, V., Carlborg, O. ", Jeon, J.-T., & Andersson, L. (2000). The Origin of the Domestic Pig: Independent Domestication and Subsequent Introgression. <https://academic.oup.com/genetics/article/154/4/1785/6047989>
- Groenen, M. A. M. (2016). A decade of pig genome sequencing: A window on pig domestication and evolution. *Genetics Selection Evolution*, 48(1), 1–9. <https://doi.org/10.1186/s12711-016-0204-2>
- Halimani, T. E., Muchadeyi, F. C., Chimonyo, M., & Dzama, K. (2010). Pig genetic resource conservation: The Southern African perspective. In *Ecological Economics* (Vol. 69, Issue 5, pp. 944–951). <https://doi.org/10.1016/j.ecolecon.2010.01.005>
- Herrero-Medrano, J. M., Megens, H. J., Groenen, M. A. M., Ramis, G., Bosse, M., Pérez-Enciso, M., & Crooijmans, R. P. M. A. (2013). Conservation genomic analysis of domestic and wild pig populations from the Iberian Peninsula. *BMC Genetics*, 14, 1–13. <https://doi.org/10.1186/1471-2156-14-106>
- Hlongwane, N. L., Hadebe, K., Soma, P., Dzomba, E. F., & Muchadeyi, F. C. (2020). Genome Wide Assessment of Genetic Variation and Population Distinctiveness of the Pig Family in South Africa. *Frontiers in Genetics*, 11(May), 1–19. <https://doi.org/10.3389/fgene.2020.00344>
- Kardos, M., Taylor, H. R., Ellegren, H., Luikart, G., & Allendorf, F. W. (2016). Genomics advances the study of inbreeding depression in the wild. In *Evolutionary Applications* (Vol. 9, Issue 10, pp. 1205–1218). Wiley-Blackwell. <https://doi.org/10.1111/eva.12414>
- Lin, B. Z., Kato, T., Kaneda, M., Matsumoto, H., Sasazaki, S., & Mannen, H. (2013). Genetic diversity and structure in Asian native goat analyzed by newly developed SNP markers. *Animal Science Journal = Nihon Chikusan Gakkaiho*, 84(8), 579–584. <https://doi.org/10.1111/asj.12039>
- Madzimure, J., Chimonyo, M., Zander, K. K., & Dzama, K. (2012). Diurnal heat-related physiological and behavioural responses in South African indigenous gilts. *Journal of Arid Environments*, 87, 29–34. <https://doi.org/10.1016/J.JARIDENV.2012.05.010>
- Matabane, M. B., Nethenzheni, P., Thomas, R., Netshirovha, T. R., Norris, D., Nephawe, K. A., & Nedambale, T. L. (2015). *Applied Animal Husbandry & Rural Development. Appl. Anim. Husb. Rural Develop*, 8, 19–25.
- Medrano, J. H. (2013). Conservation genetics of local and wild pig populations : insight in genetic diversity and demographic history.
- Mendelsohn, R. (2003). The challenge of conserving indigenous domesticated animals. *Ecological Economics*, 45(3), 501–510. [https://doi.org/10.1016/S0921-8009\(03\)00100-9](https://doi.org/10.1016/S0921-8009(03)00100-9)
- Mokoele, J. M., Spencer, B. T., van Leengoed, L. A. M. G., & Fasina, F. O. (2014). Efficiency indices and indicators of poor performance among emerging small-scale pig farmers in the Limpopo Province, South Africa. *The Onderstepoort Journal of Veterinary Research*, 81(1). <https://doi.org/10.4102/OJVR.V81I1.774>
- Molotsi, A. H., Cupido, M., & Hoffman, L. C. H. (2021). Characterization of smallholder pig production systems in the Western Cape, South Africa. *Tropical Animal Health and Production*, 53(2). <https://doi.org/10.1007/S11250-021-02649-8>
- Mujibi, F. D., Okoth, E., Cheruiyot, E. K., Onzere, C., Bishop, R. P., Fèvre, E. M., Thomas, L., Masembe, C., Plastow, G., & Rothschild, M. (2018). Genetic diversity, breed composition and admixture of Kenyan domestic pigs. *PLOS ONE*, 13(1), e0190080. <https://doi.org/10.1371/JOURNAL.PONE.0190080>

- Muñoz, M., Bozzi, R., García-Casco, J., Núñez, Y., Ribani, A., Franci, O., García, F., Škrlep, M., Schiavo, G., Bovo, S., Utzeri, V. J., Charneca, R., Martins, J. M., Quintanilla, R., Tibau, J., Margeta, V., Djurkin-Kušec, I., Mercat, M. J., Riquet, J., ... Óvilo, C. (2019). Genomic diversity, linkage disequilibrium and selection signatures in European local pig breeds assessed with a high density SNP chip. *Scientific Reports*, 9(1), 1–14. <https://doi.org/10.1038/s41598-019-49830-6>
- Mutua, F., Arimi, S., Ogara, W., Dewey, C., & Schelling, E. (2010). Farmer Perceptions on Indigenous Pig Farming in Kakamega District, Western Kenya. *Nordic Journal of African Studies*, 19(1), 15–15. <https://doi.org/10.53228/NJAS.V19I1.213>
- Niemi, J. K., Sevón-Aimonen, M. L., Pietola, K., & Stalder, K. J. (2010). The value of precision feeding technologies for grow–finish swine. *Livestock Science*, 129(1–3), 13–23. <https://doi.org/10.1016/J.LIVSCI.2009.12.006>
- Palstra, F. P., & Ruzzante, D. E. (2008). Genetic estimates of contemporary effective population size: What can they tell us about the importance of genetic stochasticity for wild population persistence? *Molecular Ecology*, 17(15), 3428–3447. <https://doi.org/10.1111/j.1365-294X.2008.03842.x>
- Patterson, N., Price, A. L., & Reich, D. (2006). Population structure and eigenanalysis. *PLoS Genet*, 2(12), 190. <https://doi.org/10.1371/journal.pgen.0020190>
- Penrith, M. L., Vosloo, W., & Mather, C. (2011). Classical swine fever (hog cholera): review of aspects relevant to control. *Transboundary and Emerging Diseases*, 58(3), 187–196. <https://doi.org/10.1111/J.1865-1682.2011.01205.X>
- Pham, L. D., Do, D. N., Nam, L. Q., van Ba, N., Minh, L. T. A., Hoan, T. X., Cuong, V. C., & Kadarmideen, H. N. (2014). Molecular genetic diversity and genetic structure of Vietnamese indigenous pig populations. *Journal of Animal Breeding and Genetics*, 131(5), 379–386. <https://doi.org/10.1111/JBG.12068>
- Pickrell, J. K., & Pritchard, J. K. (2012). Inference of population splits and mixtures from genome-wide allele frequency data. *Nature Precedings* 2012, 1–1. <https://doi.org/10.1038/npre.2012.6956.1>
- Purcell, S., Neale, B., Todd-Brown, K., Thomas, L., Ferreira, M. A. R., Bender, D., Maller, J., Sklar, P., De Bakker, P. I. W., Daly, M. J., & Sham, P. C. (2007). PLINK: A tool set for whole-genome association and population-based linkage analyses. *American Journal of Human Genetics*, 81(3), 559–575. <https://doi.org/10.1086/519795>
- Rothschild M. F., Ruvinsky A. (eds). (2011). *The genetics of the pig*. Wallingford: Cabi Publishing.
- Sha, Y., Gao, C., Liu, M., & Zhao, S. (2020). Evaluation of the genetic diversity of six Chinese indigenous chickens. *Asian-Australasian Journal of Animal Sciences*, 33(10), 1566. <https://doi.org/10.5713/AJAS.19.0606>
- Simon. (1999). European approaches to conservation of farm animal genetic resources. *Animal Genetic Resources Information*, 25, 77–97.
- Sved, J. A. (1971). Linkage disequilibrium and homozygosity of chromosome segments in finite populations. *Theoretical Population Biology*, 2(2), 125–141. [https://doi.org/10.1016/0040-5809\(71\)90011-6](https://doi.org/10.1016/0040-5809(71)90011-6)
- Swart, H., Kotze, A., Olivier, P. A. S., & Grobler, J. P. (2010). Microsatellite-based characterization of Southern African domestic pigs (*Sus scrofa domestica*). In *South African Journal of Animal Science* (Issue 2). <http://www.sasas.co.za/sajas.asp>
- Vignal, A., Milan, D., SanCristobal, M., & Eggen, A. (2002). A review on SNP and other types of molecular markers and their use in animal genetics. *Genetics Selection Evolution*, 34(3), 275–305. <https://doi.org/10.1051/gse:2002009>
- Wang, W., Gan, J., Fang, D., Tang, H., Wang, H., Yi, J., & Fu, M. (2018). Genome-wide SNP discovery and evaluation of genetic diversity among six Chinese indigenous cattle breeds in Sichuan. *PLoS ONE*, 13(8). <https://doi.org/10.1371/journal.pone.0201534>
- Wang, Z., Song, B., Yao, J., Li, X., Zhang, Y., Tang, Z., & Yi, G. (2024). Whole-genome analysis reveals distinct adaptation signatures to diverse environments in Chinese domestic pigs. *Journal of Animal Science and Biotechnology*, 15(1), 97-. <https://doi.org/10.1186/S40104-024-01053-0/FIGURES/5>
- Weir, B. S., & Cockerham, C. C. (1984). Estimating F-Statistics for the Analysis of Population Structure. *Evolution*, 38(6), 1358. <https://doi.org/10.2307/2408641>
- Wright, S. (1990). Evolution in mendelian populations. *Bulletin of Mathematical Biology*, 52(1–2), 241–295. <https://doi.org/10.1007/BF02459575>

- Yu, J., Zhao, P., Zheng, X., Zhou, L., Wang, C., & Liu, J. F. (2020). Genome-Wide Detection of Selection Signatures in Duroc Revealed Candidate Genes Relating to Growth and Meat Quality. *G3* (Bethesda, Md.), 10(10), 3765–3773. <https://doi.org/10.1534/G3.120.401628>
- Zhong, Z., Wang, Z., Xie, X., Tian, S., Wang, F., Wang, Q., Ni, S., Pan, Y., & Xiao, Q. (2023). Evaluation of the Genetic Diversity, Population Structure and Selection Signatures of Three Native Chinese Pig Populations. *Animals: An Open Access Journal from MDPI*, 13(12). <https://doi.org/10.3390/ANI13122010>

5 GENOME-WIDE IDENTIFICATION OF RUNS OF HOMOZYGOSITY AND ASSOCIATED GENES AND ESTIMATES OF GENOMIC INBREEDING IN SOUTH AFRICAN INDIGENOUS AND VILLAGE PIG POPULATIONS

5.1 Abstract

South African pig populations are experiencing a systematic decline in functional genetic diversity accompanied by the accumulation of deleterious alleles. Inbreeding, stemming from the mating of closely related individuals, exacerbates this decline and increases the frequency of harmful recessive alleles. Similarly, genetic drift, driven by fluctuations in allele frequencies within small populations, intensified the decline in genetic diversity. A total of 384 pig ear tissue samples were collected from villages across the provinces of the Eastern Cape, Gauteng, KwaZulu-Natal, Limpopo, Mpumalanga, and North West in South Africa. These samples underwent genotyping using Illumina PorcineSNP80K BeadChip. Only autosomal SNPs were analysed. Identity-by-Descent analyses were carried out in Plink. Runs of homozygosity (ROH) were computed using the R package *detectRUNS*. Mean number, length, and total number of ROH per animal were determined across various size categories. Genomic inbreeding coefficient (F_{ROH}) was calculated using the formula $F_{ROH} = L_{ROH} / L_{AUTO}$. The *tableRuns* function with 50% was used for the identification of common runs and Ensembl database was used to identify genes located within the detected genomic regions. From IBD analysis, a notable genetic similarity among these animals was observed. A total of 8,981 homozygous segments with mean ROH per population ranging from 17.500 (Johannesburg) to 32.061 (Mopani) were detected. Only two individuals did not yield any ROH. Most of the ROHs identified have short segments (0 – 5 Mb). Of all the investigated populations, genomic inbreeding coefficient based on ROH (F_{ROH}), the Harry Gwala ($F_{ROH} = 0.118$) population exhibited the highest value, and inbreeding based on excess homozygosity (F_{HOM}) was the highest in Mopani ($F_{HOM} = 0.211$). Common runs per SNP across populations were only detected on chromosome 7 (SSC7). The detected genes in ROH islands were associated with numerous quantitative trait loci (QTLs) linked to economically important traits such as growth, reproduction, and meat quality in pigs. These findings offer valuable insights into genetic inbreeding background and highlight key breed-specific traits that can contribute to selective breeding and trait improvement. Further studies investigating the causes of inbreeding and the identification of candidate genes in pigs from South Africa are encouraged.

Keywords: *Runs of homozygosity, ROH island, inbreeding, PorcineSNP80 BeadChip*

5.2 Introduction

Genetic diversity plays a crucial role in the adaptability and long-term survival of animal populations (Birader, 2023). It enables populations to adapt to changing environments, resist diseases, and maintain overall population health. Pigs are a valuable livestock species that were domesticated thousands of years ago. However, in recent years, they have encountered challenges such as uncontrolled crossbreeding, inbreeding, and reduced population sizes, which may impact their productivity and genetic diversity. Loss of genetic diversity in a population can reduce fitness and adaptability, increasing the risk of extinction (Boyce, 1992).

Populations with low genetic variation have a higher probability of becoming genetically inbred. Additionally, small populations are more susceptible to inbreeding, which can harm their health and reproductive capabilities (Lynch *et al.*, 1995). Inbreeding, which results from the mating of closely related individuals, exacerbates this decline and increases the frequency of harmful recessive alleles, leading to inbreeding depression characterized by reduced fertility, increased susceptibility to diseases, and lower survival rates (Hedrick & Kalinowski, 1870). Genetic drift and inbreeding events affect the gene pool by increasing homozygosity and, consequently, revealing the phenotypic effects of deleterious recessive alleles usually present in low frequencies. Moreover, inbreeding reduces genetic diversity, leading to increased homozygosity, which can be fatal under conditions where deleterious alleles become more prevalent and fertility declines (Keller & Waller 2002; Hedrick 2004). The fitness reduction seen in inbred progeny, compared to outbred progeny, is referred to as “inbreeding depression” (Keller & Waller, 2002). This phenomenon is primarily attributed to the accumulation of recessive deleterious mutations in the genome, as inbreeding raises the likelihood of these mutations becoming homozygous (Agrawal & Whitlock, 2012; Charlesworth & Willis, 2009; Ohta, 1973). The possible detrimental effects of inbreeding on reproduction and health in comparison to an outbred population are known as genetic load (Crow, 2017; 1970), which is primarily due to the expression of recessive homozygous harmful mutations (García-Dorado, 2003; Lynch *et al.*, 1995).

With the availability of high-density single nucleotide polymorphisms (SNPs), several genomic estimators have been proposed to assess inbreeding (Keller *et al.*, 2011; Peripolli *et al.*, 2018). These genomic estimators measure the realized autozygosity and are independent of the depth and completeness of the pedigree using genomic data. Several studies showed superiority of using genomic data to estimate true inbreeding compared to using pedigree information (Forutan *et al.*, 2018; Saura *et al.*, 2013). Methods for estimating genomic inbreeding coefficients from genomic data include using a genomic relationship matrix (GRM)

to examine IBD information marker by marker (Powell *et al.*, 2010) and using runs of homozygosity (ROH), which was first proposed for humans by McQuillan *et al.* (2008) and then for cattle by Ferencakovic *et al.* (2011).

ROHs can be used for the inbreeding coefficient estimation when pedigree information is absent. Inbreeding estimated from continuous stretches of homozygous SNPs, known as ROH, is considered one of the most accurate. These ROH segments occur when two identical haplotypes are inherited from a shared ancestor (Broman & Weber, 1999). If an individual's parents have a relatively recent shared ancestor, they are likely to share large portions of their genomes that are identical-by-descent. When both parents pass down the same segment to the progeny, it becomes homozygous in that segment, leading to ROH formation (Curik *et al.*, 2014; Gibson *et al.*, 2006; Kirin *et al.*, 2010; Purfield *et al.*, 2012). Since ROH results from both haplotypes being identical by descent, the likelihood of a low-frequency recessive deleterious allele becoming homozygous is higher within these regions than regions outside IBD (Szpiech *et al.*, 2013). Research has identified ROHs in numerous livestock, including chickens (Fedorova *et al.*, 2022), cattle (Mastrangelo *et al.*, 2016; Szmatoła *et al.*, 2019), goats (Chokoe *et al.*, 2022), pigs (Shi *et al.*, 2020), and sheep (Dzomba *et al.*, 2021). However, the available literature shows a lack of information on ROHs in South African village pigs.

Beyond genomic inbreeding estimation, ROH analysis can also assist in identifying common genomic regions found in homozygous segments across individuals within a population through the detection of ROH islands or hotspots, and subsequently reveal selection signatures (Peripolli *et al.*, 2018; Purfield *et al.*, 2012). These genomic regions may harbour genes associated with key functional and economically significant traits such as growth, reproduction, adaptability, and resistance (Gorssen *et al.*, 2021; Mastrangelo *et al.*, 2017, 2018). Assessing genes found within ROH islands provides important information on the genetic processes that underlie the functional characteristics and adaptability of pig populations. Furthermore, these regions might also include deleterious alleles that accumulate due to small or fragmented population sizes, highlighting the necessity of cautious genetic management (Hoffmann *et al.*, 2020; Sun *et al.*, 2025). Each individual carries deleterious mutations in its genome that can lower their fitness and health depending on their dominance and selection coefficient.

The present study utilised the latest technologies to investigate inbreeding and identify candidate genes in the genomes of South African indigenous and village pig populations using Identity by Descent (IBD) and Runs of Homozygosity (ROH) analysis.

5.3 Materials and Methods

5.3.1 Sample Site and Data Collection

Total of 384 individuals from eleven pig populations from South African villages were included in this study: 32 Alfred Nzo (ALN), 86 Capricorn (CAP), 20 Gert Sibande (GSD), 10 Harry Gwala (HGL), 11 City of Johannesburg (JHB), 12 Mopani (MOP), 13 OR Tambo (ORT), 14 Sedibeng (SBD), 15 Tshwane (TSW), 16 uMgungundlovu (UMG), and 17 West Rand (WRD) **Table 5.1**.

Table 5.1 Sampled South African village pig populations

Province	District	No. of Samples	Total
Eastern Cape	Alfred Nzo (EC-ALN)	28	384
	OR Tambo (EC-ORT)	32	
Gauteng	Johannesburg (GP-JHB)	08	
	Sedibeng (GP-SBD)	34	
	Tshwane (GP-TSW)	30	
	West Rand (GP-WRD)	12	
KwaZulu-Natal	Harry Gwala (ZN-HGL)	38	
	uMgungundlovu (ZN-UMG)	25	
Limpopo	Capricorn (LP-CAP)	35	
	Mopani (LP-MOP)	49	
Mpumalanga	Gert Sibande (LP-GSD)	37	
North West	Ngaka Modiri Molema (NW-NMM)	21	
	Dr. Ruth Segomotsi Mompati (NW-RSM)	35	

5.3.2 DNA Extraction, SNP Genotyping, and Quality Control

Genomic DNA extractions from the sampled pig ear tissues were conducted at the Agricultural Research Council – Biotechnology Platform using a commercially available using Qiagen DNA Extraction Kit (Qiagen, Valencia, CA), as per the manufacturer’s protocol. The quality and quantity of DNA were assessed using a Qubit® 4.0 fluorometer (Life Technologies, Thermo Fisher Scientific, USA) and gel electrophoresis (2%). The 384 pigs were genotyped using the Illumina porcine 80K SNP BeadChip (Illumina, San Diego, CA, USA) containing 68,528 SNPs and an average gap length of 38 kilobases. The calling of SNP genotypes was performed using the genotyping module integrated into GenomeStudio software (Illumina, Inc.). Plink Ped (Pedigree file) and Map (SNP panel file) were generated for use in analysis.

SNP genome coordinates were obtained from the *Sus scrofa* 11.2 porcine genome reference assembly. The Golden Helix Variation Suite (SVS) version 8.9.1 (Golden Helix Inc., Bozeman, MT, USA; www.goldenhelix.com) was used for quality control of the SNP data and the following standards were set; discarding of SNPs with a call rate of less than 0.85, minor allele frequency (MAF) of less than 0.02 (a minor allele frequency higher than 0.98) and Hardy Weinberg (HWE) of less than 0.0001. Only autosomal SNPs were retained for analyses.

5.3.4 Identity by Descent

Genome-wide IBD between pairs of samples was estimated using the PI-HAT coefficient, which represents the probability of sharing 0, 1, or 2 alleles IBD between individuals from the same homogeneous random-mating population (Purcell *et al.*, 2007). A heatmap plot was generated in R software using the ggplot2 package (Wickham, 2016).

5.3.5 Genomic Inbreeding Coefficient Estimation

In this study, two methods were used to estimate genomic inbreeding coefficients, F_{ROH} (based on ROH) and F_{HOM} (based on excess homozygosity). The F_{ROH} coefficient was estimated for each individual as the ratio between continuous homozygous segments to the autosomal genome length (McQuillan *et al.*, 2008) using *detectRUNs* (Biscarini *et al.*, 2018) as follows:

$$F_{ROH} = \frac{\sum L_{ROH}}{L_{auto}}$$

where L_{ROH} is the total length of ROHs on autosomes, and L_{auto} is the total length of the genome covered by SNPs. Genomic inbreeding coefficient (F_{HOM}) was calculated on PLINK (Purcell *et al.*, 2007) using the `-het` function as follows:

$$F_{HOM} = \frac{(E_{HOM} - O_{HOM})}{(L - E_{HOM})},$$

where E_{HOM} is the number of expected homozygous genotypes, O_{HOM} is the number of observed homozygous genotypes, and L is the total number of genotyped autosomal SNPs. Furthermore, Pearson's correlation was used to compare F_{ROH} and F_{HOM} using *cor.test* function in R version 4.3.3 for the thirteen pig populations.

5.3.6 Runs of Homozygosity Detection

The ROHs were detected with the *detectRUNs* package (Biscarini *et al.*, 2018) of R version 0.9.6 software using a sliding-window-based and consecutive-SNP-based detection method. No LD pruning was performed and ROH was defined according to the criteria: (i) 0.05 threshold to call a SNP in a run; (ii) The minimum number of SNPs in a sliding window was set to 50; (iii) One SNP with a missing genotype and up to one possible missing genotypes in

the run (iv) The minimum ROH length was 1000 kb. In this study, we divided ROH into five different classes based on their physical length: > 40 Mb, 20 - 40 Mb, 10 - 20 Mb, 5 - 10 Mb, and 1 - 5 Mb. For each length category, we calculated the average length of an ROH and the frequency of ROH numbers for each population.

5.3.7 Detection of Common Runs of Homozygosity and Gene Annotation

To identify within-population top runs, the detectRUNS R-package version 0.96 (Biscarini *et al.*, 2018) was used to detect genomic regions most commonly associated with ROH. The “topRuns” function from the detectRUNS package was used to extract the most common runs using a 0.50 threshold. This criterion required that an ROH be present in at least 50% of individuals within a given population for the region to be classified as part of an ROH island. No ROH islands were identified across populations when a 70% threshold was applied; therefore, 50% threshold was used, which has been previously used by Cesarani *et al.* (2018) and Martínez-Rocha *et al.* (2022) in their studies. The genes located directly in the detected regions (ROH islands) were identified using the latest available porcine reference genome obtained at the ENSEMBL BIOMART database (<http://www.ensembl.org/biomart/martview/>) and the NCBI file SSCROFA11.1 GFF. The detected gene associations with known QTLs were searched using previously published protocols implemented in BEDTools (Quinlan & Hall, 2010).

5.4 Results

After quality control, the imputed and cleaned genotypic data comprised 384 individuals with 32,743 SNPs from 13 village pig populations.

5.4.1 Identity by Descent

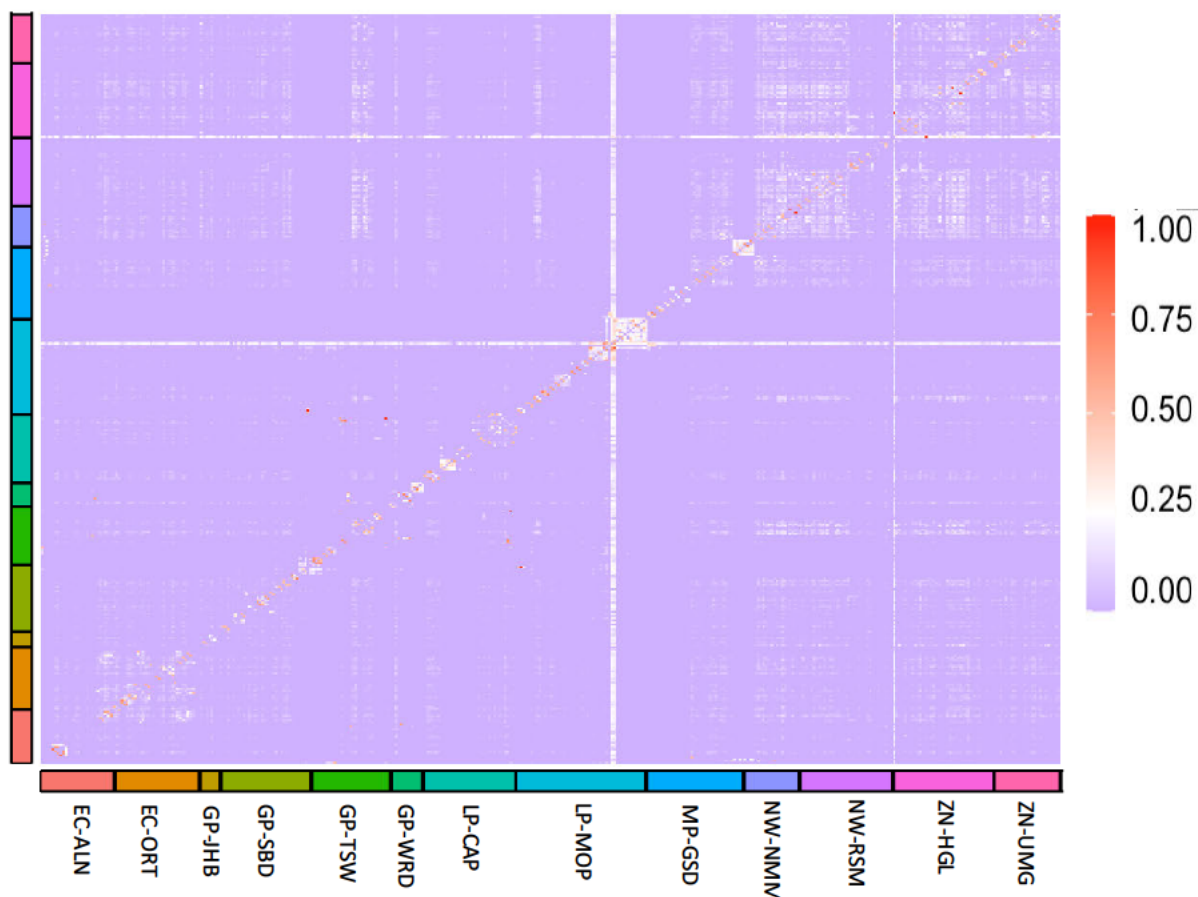


Figure 5.21 Heatmap representing genome-wide identity by descent (IBD) between pairs of South African village pigs estimated with the PI-HAT coefficient

The degree of genome-wide IBD among South African village pigs was inferred (**Figure 5.1**). Out of 73,536 pairwise comparisons from IBD analysis, 23.39% values were equal to zero, indicating no detectable genetic relatedness. Remarkably, only 0.16% of the pairwise comparisons exhibited values exceeding 0.5, indicating that a small subset of individuals had a high degree of genetic similarity between them. Moreover, one comparison between animals from GP-TSW had an exceptionally high value of 0.991, suggesting that these individuals are nearly genetically identical and likely share the majority of their genetic material.

5.4.2 Genomic Inbreeding Coefficients

The F_{ROH} and F_{HOM} , their range of variation across populations, and their distribution are illustrated in **Figure 2(a)** and **2(b)** respectively. The F_{ROH} values ranged between 0.001 to 0.218, and F_{HOM} values ranged between -0.384 to 0.353. Mean F_{ROH} was the highest at ZN-HGL ($F_{ROH}=0.118$) and lowest at GP-WRD ($F_{ROH}=0.049$). Mean F_{HOM} was the highest at LP-MOP ($F_{HOM}=0.211$) and lowest at GP-WRD ($F_{HOM}=0.016$). The estimated mean Pearson's

squared correlation coefficient (r) value between F_{ROH} and F_{HOM} was ($r = 0.121$, $P < 1.298 \times 10^{-42}$).

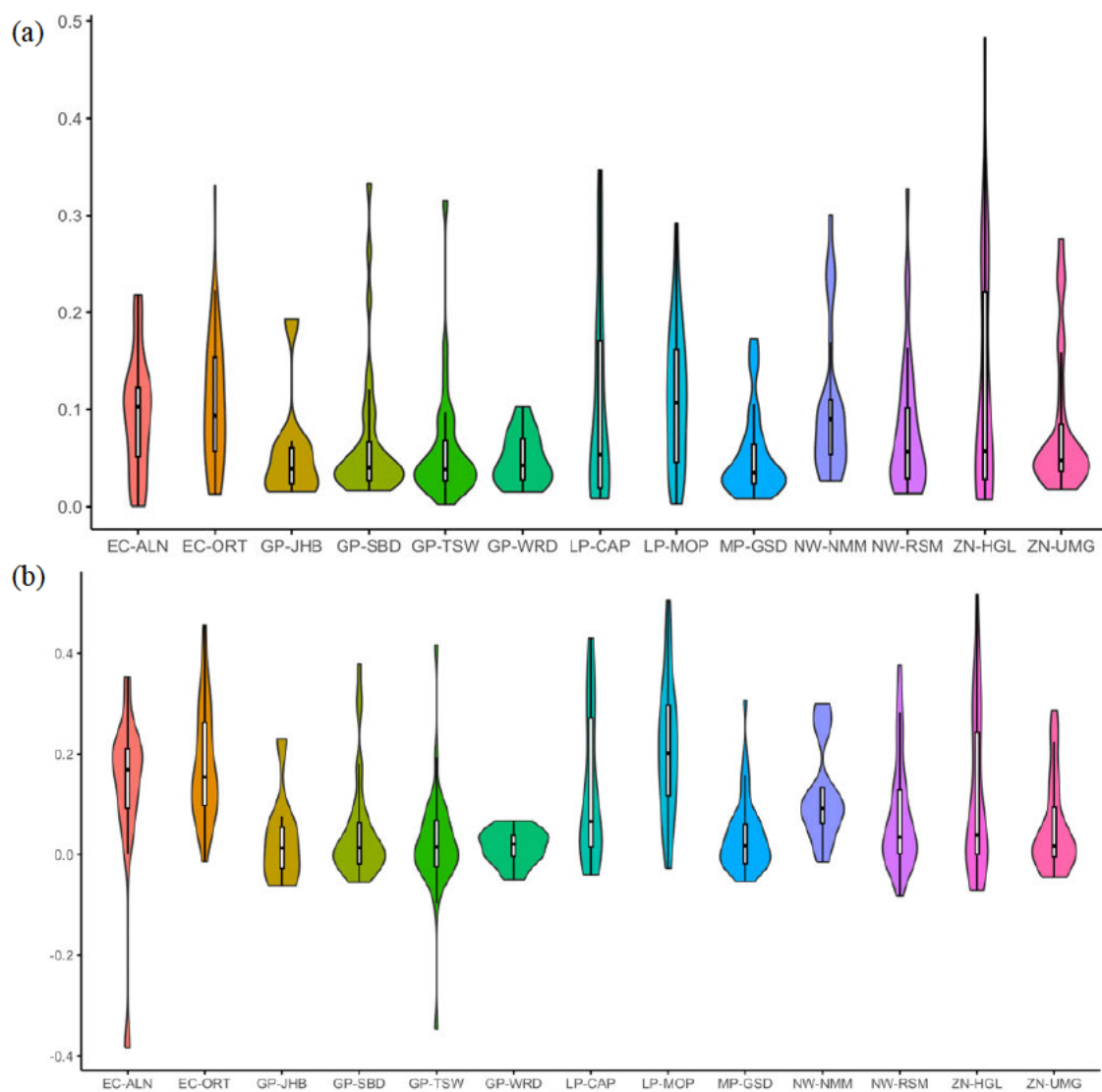


Figure 5.22 Genomic inbreeding coefficients within each population (a) F_{ROH} distribution in the thirteen studied populations (b) F_{HOM} distribution in the thirteen studied populations

Table 5.2 Mean and standard deviation of ROH based on inbreeding (F_{ROH}) and excess of homozygosity-based inbreeding (F_{HOM}) of South African village pig populations

Population	Mean F_{ROH}	SD	Mean F_{HOM}	SD
EC-ALN	0.094	0.061	0.129	0.167
EC-ORT	0.111	0.072	0.179	0.113
GP-JHB	0.057	0.058	0.031	0.093
GP-SBD	0.068	0.072	0.047	0.104
GP-TSW	0.059	0.060	0.022	0.115
GP-WRD	0.049	0.027	0.016	0.035
LP-CAP	0.104	0.104	0.128	0.149
LP-MOP	0.113	0.071	0.211	0.133
MP-GSD	0.052	0.044	0.038	0.073
NW-NMM	0.108	0.081	0.121	0.096
NW-RSM	0.079	0.071	0.077	0.110
ZN-HGL	0.118	0.119	0.120	0.158
ZN-UMG	0.082	0.007	0.054	0.092
Overall	0.084	0.025	0.090	0.063

5.4.3 Distributions of Runs of Homozygosity

A total of 8,981 homozygous segments were detected across populations; however, both the length and frequency of ROH varied with each population. Of the 384 individuals, only two belonging to the EC-ALN population lacked ROH. ROH ranged from 119 in GP-JHB to 1.571 in LP-MOP population. The mean number of ROH per population, arranged in ascending order, was as follows: MP-GSD (15.784), followed by GP-WRD (17.417), GP-JHB (17.500), GP-TSW (18.467), ZN-UMG (21.800), GP-SBD (21.971), NW-RSM (22.000), ZN-HGL (23.605), LP-CAP (23.686), EC-ALN (25.071), EC-ORT (26.375), NW-NMM (28.048), and highest in LP-MOP (32.061). The relationship between the number and total length of ROH per individual is illustrated in **Figure 5.3**. Complementary to the preceding figures, the plotting of ROH per individual allowed the visualization of possible trends and patterns in the size and distribution of ROH in the genome for the populations.

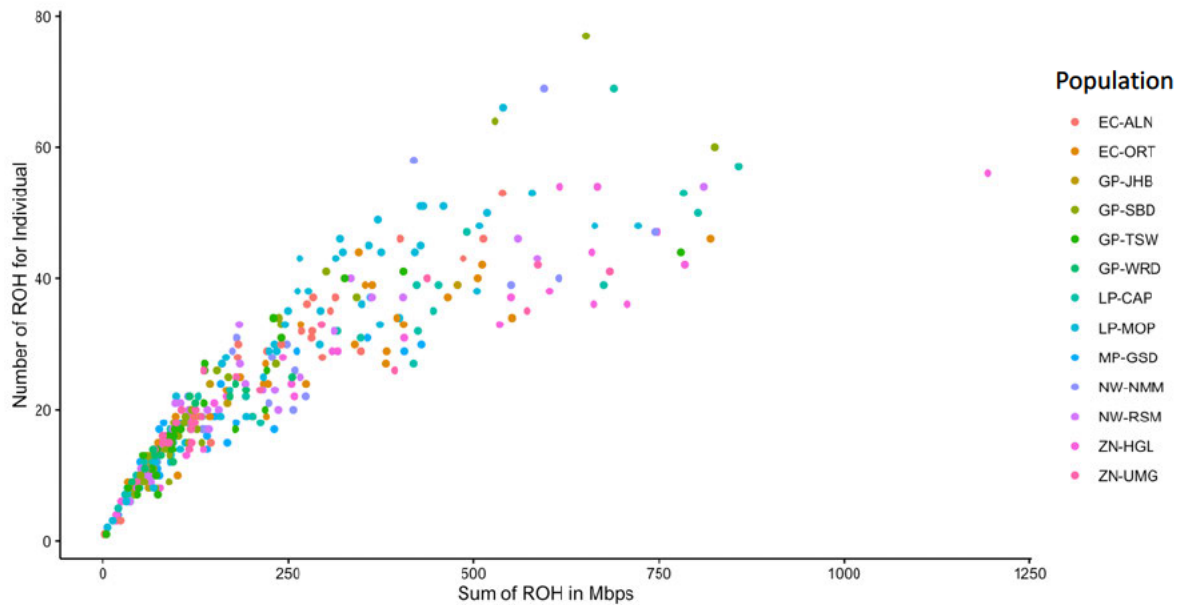


Figure 5.23 Individual patterns of ROH (the number of ROH compared to the total length of ROH for individuals). Each dot represents an individual.

To study the ROH patterns, we classified them into five classes: > 40 Mb, 20 - 40 Mb, 10 - 20 Mb, 5 - 10 Mb, and 1 - 5 Mb. Distribution of ROHs by total length and number for each population is depicted in **Figure 5.4**. Long and short ROHs were detected at different proportions across all the populations. The ZN-HGL population presented the highest percentage of long ROHs (> 40 Mb), of 23.55%, which indicates recent inbreeding. The LP-MOP population exhibited higher frequencies of short ROHs (0 - 5 Mb), of about 17.05%. Overall, LP-MOP exhibited the highest ROHs in total (17.49%).

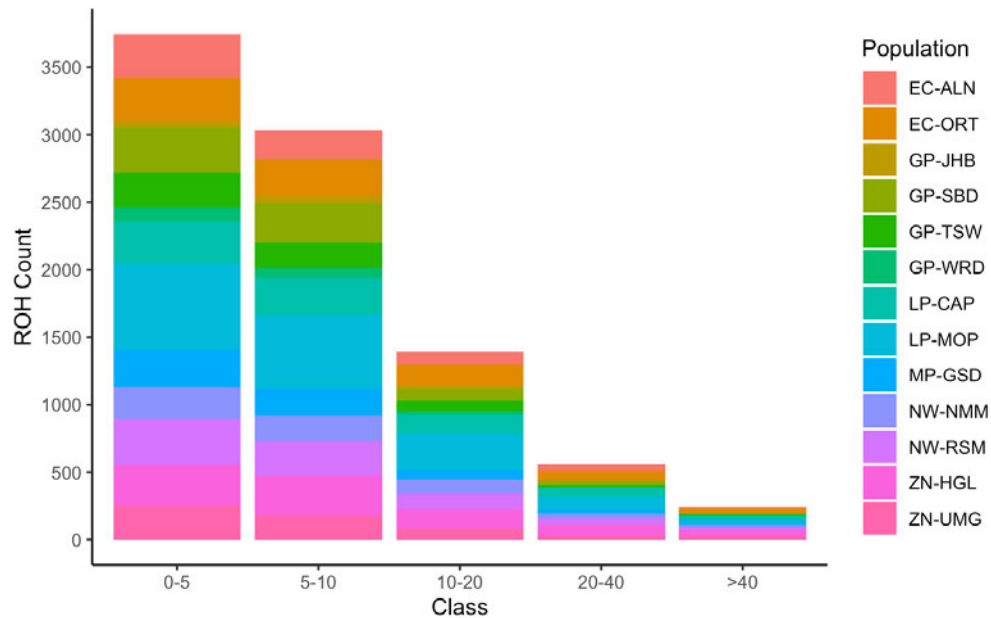


Figure 5.24 ROH count per population in different classes

The proportion and distribution of ROH coverage were further investigated at a chromosomal level for all groups. The chromosomes SSC3, SSC6, SSC8, and SSC15 had the longest ROH mean length. However, SSC1, SSC7, SSC13, and SSC14 showed the highest number of ROH. The scaffolds were zoomed in to highlight clear patterns of distributions (**Figure 5.5**). SSC1 had the highest number across all populations with 1,145, followed by SSC14, SSC7, and SSC13 with 845, 736, and 682, respectively. Long and short ROHs were detected at different proportions across all the populations.

Table 5.3 Mean length of ROH in Mb across 18 autosomes in South African village populations

Chr	Population												
	EC- ALN	EC- ORT	GP- JHB	GP- SBD	GP- TSW	GP- WRD	LP- CAP	LP- MOP	MP- GSD	NW- NMM	NW- RSM	ZN- HGL	ZN- UMG
1	5.705603	7.461148	5.342923	7.637943	7.454087	5.911578	8.026885	8.861358	8.309891	6.764593	8.668416	9.760473	9.912376
2	9.580909	12.02779	5.565755	9.685297	8.504821	7.627940	11.31095	10.35820	7.626264	12.05336	7.618143	13.84776	9.028444
3	8.591119	13.07097	8.250805	6.440759	12.20319	12.93386	19.07517	8.917913	8.730892	13.91691	10.46365	13.47133	17.97415
4	8.542096	9.282855	8.889473	7.199020	10.27553	7.607808	8.123915	7.662420	8.100227	9.208883	6.620819	9.521398	8.522155
5	8.604549	10.85081	8.161830	4.179065	6.934397	5.195546	11.20061	10.14895	7.751940	7.311506	11.45309	15.53917	6.968202
6	14.07746	10.76182	11.95143	9.893822	8.128770	7.148786	12.88671	11.31775	9.023390	10.29300	10.78220	16.70533	12.14052
7	8.151218	9.581418	6.549358	6.262020	7.221614	5.228607	10.83284	8.143450	7.398543	6.868595	10.30460	11.33578	5.576642
8	10.16046	12.03857	6.779816	8.607095	10.07925	14.16032	12.51742	10.32472	9.486902	10.33045	9.864492	15.03682	10.02610
9	9.252490	11.01246	5.372767	9.006625	6.862961	7.374056	8.069679	7.365747	8.281463	8.146291	7.518004	11.54132	7.070462
10	8.207228	9.719841	4.790418	6.422136	6.637031	5.199699	8.551898	7.853450	7.524052	6.460751	7.325267	9.611840	12.03815
11	10.34077	10.60566	7.448185	6.398234	5.591140	5.671754	10.93064	9.007261	8.441677	8.979742	8.684743	12.23889	8.576201
12	6.421064	8.975785	14.72926	6.010378	6.167639	12.20485	10.19077	6.992550	7.243511	8.667367	9.120203	12.95001	9.495168
13	8.350832	11.64138	8.431609	8.731400	7.546468	6.540491	12.72449	8.717529	10.58070	10.02948	7.047363	16.72306	13.10416
14	7.863238	10.84689	8.319101	7.288992	7.487429	4.569349	8.119196	7.367648	6.461181	9.277685	7.954070	9.918258	6.644391
15	10.81514	11.28181	60.18575	6.814428	10.60070	12.46616	17.75473	9.207507	7.984670	13.02115	13.69281	15.23204	7.162890
16	6.980948	8.984768	4.913534	7.350754	9.569195	5.603294	12.69654	7.707010	8.810840	12.15382	9.051784	10.52173	11.36122
17	4.519400	9.732672	4.297972	4.996592	6.005644	3.516078	6.605202	7.870416	8.711997	6.436894	5.509171	8.683785	6.941287
18	11.25675	8.720031	11.29740	10.02380	5.814411	3.911134	7.325121	8.634778	7.072027	6.518402	7.884332	13.61384	6.386004

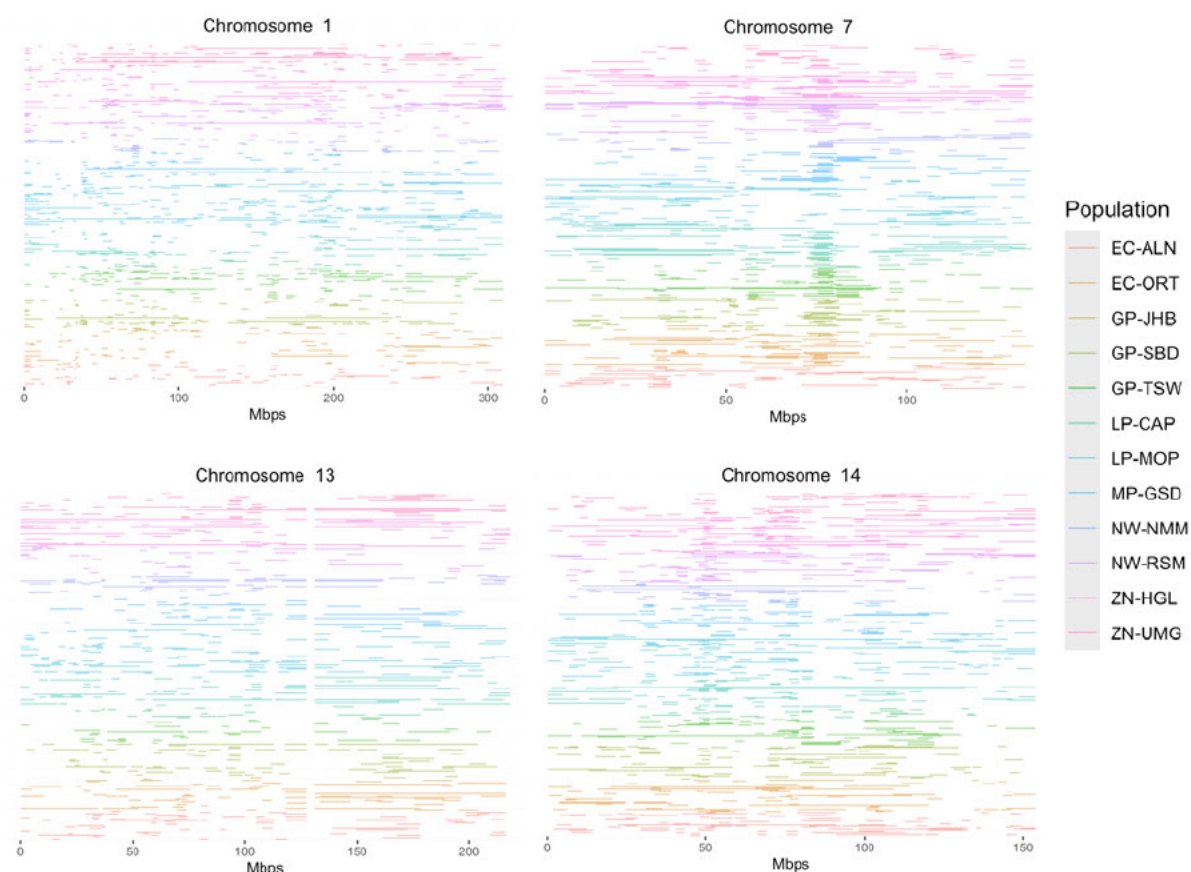


Figure 5.25 Distribution across chromosomes with the highest number of detected ROH (SSC1, SSC7, SSC13, and SSC14)

5.4.4 Common Runs Detected

Analysis of SNP frequency within ROH regions across the studied populations enabled for the assessment of the genomic distribution of ROHs (**Figure 5.6**) and the identification of ROH islands; the genomic regions that contain the most frequently occurring SNPs. The ROH islands were detected exclusively in Chr 7 (SSC7) on only three populations (GP-TSW, GP-WRD, and NW-RSM), with the total length of these regions covering 3.935 Mb, 2.930 Mb, and 1.145 Mb, respectively (**Table 5.4**). A total of 864 genes were identified inside the detected regions, including economically important genes such as *ABCD4*, *RPGRIP1*, *PSMC1*, and *BDKRB2* for teat number, dressing percentage, lean meat percentage, and intramuscular fat content, respectively. Moreover, candidate genes for reproductive traits such as *PPP1R3E* and *PABPN1* were also determined. The supplementary file shows the genes detected in common ROHs and their associated QTLs (**Supplementary Table 5.1**).

Table 5.4 Top 50 percentile of runs of homozygosity

Pop	Chr	Start SNP	End SNP	SNPs	Start (bp)	End (bp)	Length (Mb)
GP-TSW	7	DRGA0007785	DRGA0007846	48	75,490,772	79,426,745	3.935
GP-WRD	7	DRGA0007785	DRGA0007831	31	75,490,772	78,420,342	2.930
NW-RSM	7	MARC0001935	DRGA0007831	15	77,275,077	78,420,342	1.145

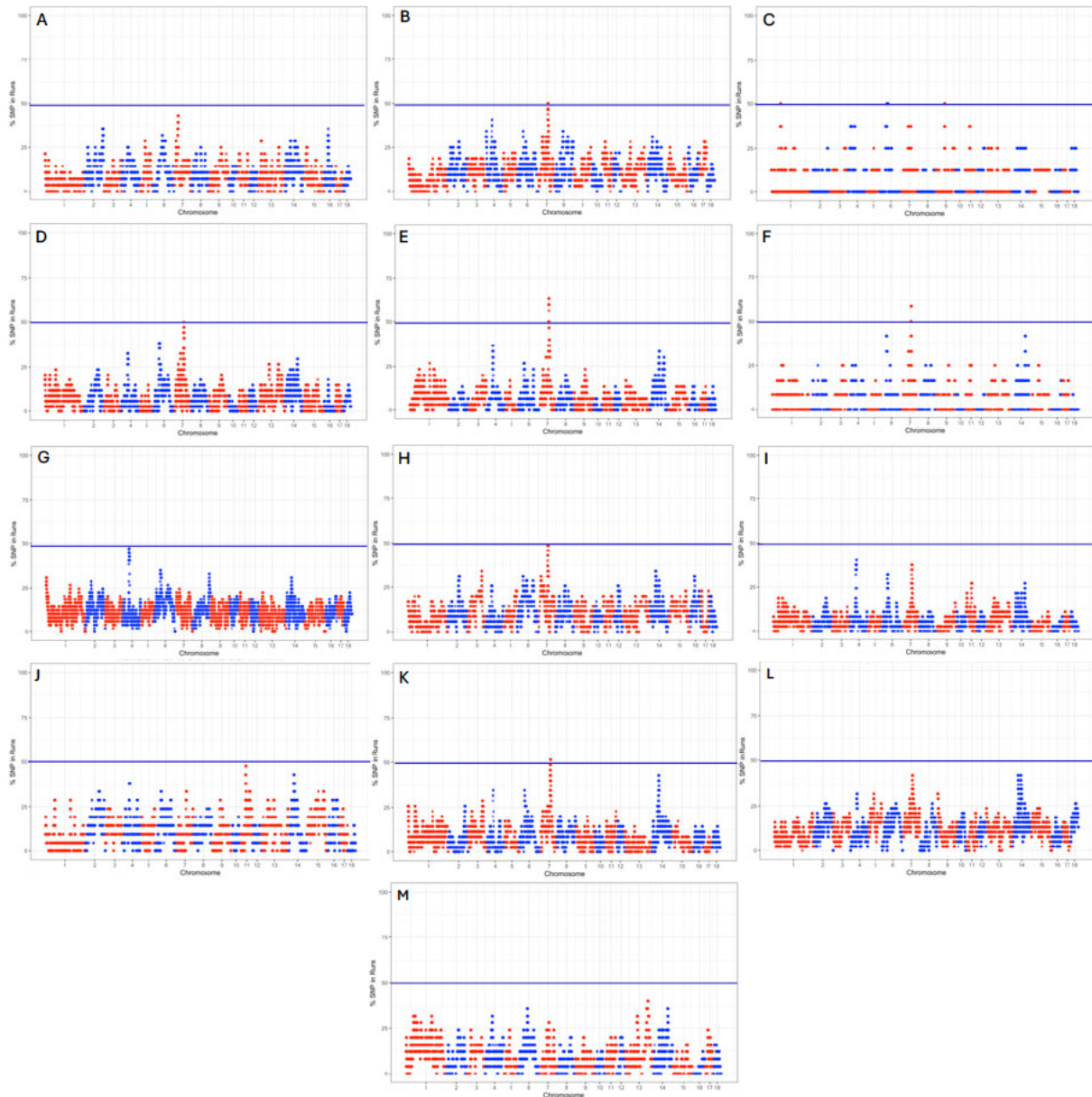


Figure 5.26 Manhattan plot displaying the distribution of runs of ROH hotspots in the studied South African village pigs. The x-axis represents the SNP genomic coordinate, while the y-axis shows the percentage (%) of overlapping ROH shared among individuals. (a) EC-ALN; (b) EC-ORT; (c) GP-JHB; (d) GP-SBD; (e) GP-TSW; (f) GP-WRD; (g) LP-CAP; (h) LP-MOP; (i) MP-GSD; (j) NW-NMM; (k) NW-RSM; (l) ZN-HGL; (m) ZN-UMG. The line parallel to the X-axis marks the top 50% of SNP frequency in a run of homozygosity (ROH). SNPs above that line correspond to ROH islands.

5.5 Discussion

Pigs are valuable genetic resources that are crucial to the local economy. Numerous studies have explored the ROH pattern and its associated inbreeding depression related to traits at the genomic level in multiple pig populations. However, the ROH distributions and patterns in South African village populations remain unexplored. To the best of our knowledge, this study

is the first of its kind to use high-density SNP arrays to examine the distribution and occurrence of ROH in local pigs from South Africa.

Analysing the length, quantity, and distribution of ROHs can reveal the level of inbreeding within the populations' genomes (Schiavo *et al.*, 2022). The distribution and length of ROH segments provide a reliable alternative for inferring inbreeding, haplotype age, and genetic diversity (Xu *et al.*, 2019). Homozygosity distributions can be interpreted by clustering ROHs into different length groups. Short ROH segments are linked to distant inbreeding, whereas long ROH segments are associated with recent inbreeding events (De Cara *et al.*, 2013; Gomez-Raya *et al.*, 2019; Marras *et al.*, 2015). ROHs resulting from recent inbreeding tend to be longer, due to limited opportunity of recombination to break up segments that are identical by descent (Curik *et al.*, 2014), and ancient ROHs are typically shorter due to chromosomal segments being broken down by recurrent meiosis (Curik *et al.*, 2014; Kirin *et al.*, 2010). In this study, the South African village pigs exhibited higher frequencies of short ROH fragments (0-5 Mb), encompassing over 41.72% of the total ROH length, indicating that inbreeding levels within these populations are relatively low and are rarely affected by recent inbreeding events. Nonetheless, it should be noted that the ROH length depends not only on inbreeding events but also on the stochastic nature of gamete formation and recombination rate (Liu *et al.*, 2010).

In our study, the distribution of ROH reveals chromosome-specific patterns, with the physical length of the chromosome influencing the quantity of ROHs. The ROH distribution on chromosomes varied, with SSC1 recording the highest number of ROHs (1,145), aligning with previous findings that reported a positive correlation between chromosomal length and ROH (Li *et al.*, 2022). Notably, the genomic regions with the lowest and highest ROH coverage in our study were on *Sus scrofa* chromosome 1 (SSC1) and *Sus scrofa* chromosome 15 (SSC15), respectively. These ROH distribution variations are attributed to the effects of artificial or natural selection.

Inbreeding can create homozygous populations, but it may also result in inbreeding depression, which can impair growth rates, reproductive capacity, and other aspects of production performance (Lozada-Soto *et al.*, 2021). Historically, inbreeding coefficients were estimated using pedigree data; however, pedigree information might be incomplete and incorrect due to misidentified or missing parents (Sell-Kubiak *et al.*, 2018). Estimating inbreeding coefficients is even more challenging for crossbred individuals since genealogical relationships are poorly established (Xu *et al.*, 2021). Moreover, the pedigree-based estimates do not account for the stochastic recombination events that may have taken place during meiosis (Marras *et al.*, 2015).

Thus, genetic markers offer more accurate estimates of genetic relationships compared to traditional pedigree-based methods (Goddard, 2009). Hence, we used genomic data to estimate inbreeding coefficients using two different methods, including F_{ROH} and F_{HOM} . The genomic inbreeding coefficients (F_{ROH} and F_{HOM}) calculated from the genotypes exhibited low to moderate values, < 0.300 . The average for the F_{ROH} was 0.084 ± 0.025 and was 0.090 ± 0.063 for the F_{HOM} . The very low correlation ($r = 0.121$, $P < 1.298 \times 10^{-42}$) observed between the two indicates that F_{ROH} and F_{HOM} capture different aspects of inbreeding, with F_{ROH} primarily reflecting recent inbreeding through long runs of homozygosity while F_{HOM} is more influenced by genome-wide allele frequencies and heterozygosity levels. These findings contrast with earlier studies conducted in other pig populations, which reported higher correlations between F_{ROH} and F_{HOM} , suggesting that the genetic structure and recent breeding practices (Shi et al., 2020; Xu et al., 2019). Also, we noted several individuals with negative F_{HOM} values, likely due to the sensitivity of the parameter to allele frequency in populations with higher heterozygosity levels compared to F_{ROH} estimators (Zhang *et al.*, 2015). We suspected that the ROH type, sample populations, and SNP array platform might cause these discrepancies. These results suggest that F_{HOM} may not be a reliable proxy for recent inbreeding in populations with high genetic diversity. Moreover, the genome-wide IBD revealed pairwise comparisons equal to zero, showing no relatedness, which suggests a diverse genetic structure likely due to traditional breeding practices. Only 0.159% of comparisons had IBD values above 0.5, suggesting limited instances of close genetic relationships. However, an exceptionally high IBD value (0.991) was observed among GP-TSW individuals, indicating extensive genetic similarity, likely due to inbreeding or small effective population size.

ROH islands in pigs have been utilized to detect genomic regions under selection that may contribute to the definition of breed-specific characteristics or environmental or production system adaptation (Szmatola *et al.*, 2020). From the analysis of ROH islands, we identified three high-frequency genomic regions from the South African village populations. ROH islands were detected on Chromosome 7 (SSC7) and are mostly associated with several candidate genes that play crucial roles in various biological functions.

The ROH islands harboured genes such as *PSMCI*, *CALMI*, *EFCAB11*, *TDPI*, *KCNK13*, *NRDE2*, *FOXN3* and *TTC7B* which are linked to muscle moisture percentage, lean meat percentage, loin muscle area, polyunsaturated fatty acid content, shoulder subcutaneous fat thickness, longissimus muscle length, body weight, linoleic acid content, average backfat thickness and backfat weight QTLs. To further investigate the biological functions of the

candidate genes identified through ROH analysis, a literature review was conducted to determine whether any of these genes have been previously linked to economically important traits. Among them, the *PSMCI* (Proteasome 26S Subunit, ATPase 1) gene was identified, which has been reported to influence growth and carcass traits in beef cattle (Liang *et al.*, 2014). *CALMI* (Calmodulin 1) and *TDPI* (Tyrosyl-DNA Phosphodiesterase 1) genes were recently identified as candidate genes associated with ham weight loss during the first salting process (Zappaterra *et al.*, 2021). The *CALMI* gene regulates the movement of glucose and other carbohydrates within hepatic cells (Chen *et al.*, 2017) and has been reported as a downregulated gene in the Longissimus muscle of pigs with elevated intramuscular fat, suggesting this gene's potential to influence pork quality from the metabolic pathways (Hamill *et al.*, 2012). The *EFCAB11* (EF-Hand Calcium Binding Domain 11) gene was recently identified as a candidate gene for ham weight at end of salting (Zappaterra *et al.*, 2021) and the *FOXN3* (Forkhead Box N3) gene was proposed as a candidate gene affecting intramuscular fat content in swine (Sato *et al.*, 2011). Morton *et al.* (2011) have identified *TTC7B* (Tetratricopeptide Repeat Domain 7B) as a promising candidate gene in pigs because it has been previously detected as a candidate gene for obesity in mice (Rothammer *et al.*, 2014). The *KCNK13* (Potassium Two Pore Domain Channel Subfamily K Member 13) gene is associated with calcium concentration in the first 24 hours postpartum, encodes a potassium channel containing two pore-forming domains in cows (Novo *et al.*, 2023).

Additionally, we detected some genes reported to be related to reproduction in pigs or other livestock (Protein phosphatase 1 regulatory subunit 3E (*PPP1R3E*) and *PABPN1*). The presence of a number of stillborn QTLs within ROH regions suggests that these loci may have undergone inbreeding or selection pressures. *PPP1R3E* is associated with glycogen metabolism, which is critical for energy storage and regulation during development (Blumer *et al.*, 2022). Its presence in the stillborn QTL suggests that variations in energy storage or metabolism could influence foetal viability. *PABPN1* is involved in mRNA processing and the regulation of muscle function. It is possible that this gene influences muscle development during gestation, and disruptions in its function could lead to developmental issues that result in stillbirth (Zhu *et al.*, 2022).

5.6 Conclusion

The findings from this genomic study reported the frequency and distribution of ROHs of South African village pig populations and highlighted their inbreeding levels. Despite the low average inbreeding levels, there were individuals with high inbreeding coefficients, highlighting the

necessity of considering inbreeding during the designation of mating strategies. The pattern of distribution of ROH and levels of inbreeding corresponded to the breed history and production system under which they are raised. Genes detected within ROH islands are mainly associated with important breeding traits such as reproduction and meat quality. Understanding the genes linked to reproductive and meat quality traits is essential for breeding and can act as valuable genetic markers for molecular breeding designs. In summary, the combined analysis of IBD and ROH offered a comprehensive view of the genetic landscape, inbreeding levels, and historical events shaping South African indigenous and village pig populations. These insights are invaluable for guiding effective conservation and breeding strategies and propose necessary theoretical support for the conservation and utilisation of these populations.

5.7 References

- Birader, K. (2023). Journal of Biodiversity & Endangered Species Perspective Genetic Diversity and the Adaptation of Species to Changing Environments. <https://doi.org/10.37421/2332-2543.2023.11.474>
- Biscarini, F., Cozzi, P., Gaspa, G., & Marras, G. (2018). detectRUNS: Detect runs of homozygosity and runs of heterozygosity in diploid genomes. <https://cran.r-project.org/web/packages/detectRUNS/index.html>
- Blumer, M., Brown, T., Freitas, M. B., Destro, A. L., Oliveira, J. A., Morales, A. E., Schell, T., Greve, C., Pippel, M., Jebb, D., Hecker, N., Ahmed, A. W., Kirilenko, B. M., Foote, M., Janke, A., Lim, B. K., & Hiller, M. (2022). Gene losses in the common vampire bat illuminate molecular adaptations to blood feeding. *Science Advances*, 8(12), 6494. https://doi.org/10.1126/SCIADV.ABM6494/SUPPL_FILE/SCIADV.ABM6494_TABLES_S1_TO_S5.ZIP
- Boyce, M. S. (1992). POPULATION VIABILITY ANALYSIS. In *Annu. Rev. Ecol. Syst* (Vol. 23). <https://about.jstor.org/terms>
- Broman, K. W., & Weber, J. L. (1999). Long homozygous chromosomal segments in reference families from the Centre d'Etude du Polymorphisme Humain. *American Journal of Human Genetics*, 65(6), 1493–1500. <https://doi.org/10.1086/302661>
- Cesarani, A., Sorbolini, S., Criscione, A., Bordonaro, S., Pulina, G., Battacone, G., Marletta, D., Gaspa, G., & Macciotta, N. P. P. (2018). Genome-wide variability and selection signatures in Italian island cattle breeds. *Animal Genetics*, 49(5), 371–383. <https://doi.org/10.1111/AGE.12697>
- Chen, Z., Ding, L., Yang, W., Wang, J., Chen, L., Chang, Y., Geng, B., Cui, Q., Guan, Y., & Yang, J. (2017). Hepatic Activation of the FAM3C-HSF1-CaM Pathway Attenuates Hyperglycemia of Obese Diabetic Mice. *Diabetes*, 66(5), 1185–1197. <https://doi.org/10.2337/DB16-0993>
- Chokoe, T. C., Hadebe, K., Muchadeyi, F. C., Nephawe, K. A., Dzomba, E. F., Mphahlele, T. D., Matelele, T. C., & Mtileni, B. J. (2022). Conservation status and historical relatedness of South African communal indigenous goat populations using a genome-wide single-nucleotide polymorphism marker. *Frontiers in Genetics*, 13, 909472. <https://doi.org/10.3389/FGENE.2022.909472/BIBTEX>
- Crow, J. (2017). An introduction to population genetics theory. https://books.google.com/books?hl=en&lr=&id=mpVEEAAAQBAJ&oi=fnd&pg=PP1&ots=KnfnVZe-w6&sig=J_S5bD5ZUNLJMnOEWUCxBYWRJqY
- Crow, J. F. (1970). Genetic Loads and the Cost of Natural Selection. *Mathematical Topics in Population Genetics*, 128–177. https://doi.org/10.1007/978-3-642-46244-3_5
- Curik, I., Ferencaković, M., & Sölkner, J. (2014a). Inbreeding and runs of homozygosity: A possible solution to an old problem. *Livestock Science*, 166(1), 26–34. <https://doi.org/10.1016/J.LIVSCI.2014.05.034>

- Curik, I., Ferencaković, M., & Sölkner, J. (2014b). Inbreeding and runs of homozygosity: A possible solution to an old problem. *Livestock Science*, 166(1), 26–34. <https://doi.org/10.1016/J.LIVSCI.2014.05.034>
- De Cara, M. Á. R., Villanueva, B., Toro, M. Á., & Fernández, J. (2013). Using genomic tools to maintain diversity and fitness in conservation programmes. *Molecular Ecology*, 22(24), 6091–6099. <https://doi.org/10.1111/MEC.12560>
- Dzomba, E. F., Chimonyo, M., Pierneef, R., & Muchadeyi, F. C. (2021). Runs of homozygosity analysis of South African sheep breeds from various production systems investigated using OvineSNP50k data. *BMC Genomics*, 22(1), 1–14. <https://doi.org/10.1186/S12864-020-07314-2/TABLES/2>
- Fedorova, E. S., Dementieva, N. V., Shcherbakov, Y. S., & Stanishevskaya, O. I. (2022). Identification of Key Candidate Genes in Runs of Homozygosity of the Genome of Two Chicken Breeds, Associated with Cold Adaptation. *Biology* 2022, Vol. 11, Page 547, 11(4), 547. <https://doi.org/10.3390/BIOLOGY11040547>
- Ferencakovic, M., Hamzic, E., Gredler, B., Curik, I., & Sölkner, J. (2011). Runs of Homozygosity Reveal Genome-wide Autozygosity in the Austrian Fleckvieh Cattle. *Agriculturae Conspectus Scientificus*, 76(4), 325–329.
- Forutan, M., Ansari Mahyari, S., Baes, C., Melzer, N., Schenkel, F. S., & Sargolzaei, M. (2018). Inbreeding and runs of homozygosity before and after genomic selection in North American Holstein cattle. *BMC Genomics*, 19(1), 1–12. <https://doi.org/10.1186/S12864-018-4453-Z/FIGURES/6>
- García-Dorado, A. (2003). Tolerant versus sensitive genomes: The impact of deleterious mutation on fitness and conservation. *Conservation Genetics*, 4(3), 311–324. <https://doi.org/10.1023/A:1024029432658/METRICS>
- Gibson, J., Morton, N., genetics, A. C.-H. molecular, & 2006, undefined. (2006). Extended tracts of homozygosity in outbred human populations. *Academic.Oup.ComJ Gibson, NE Morton, A CollinsHuman Molecular Genetics*, 2006•academic.Oup.Com. <https://doi.org/10.1093/hmg/ddi493>
- Goddard, M. (2009). Genomic selection: Prediction of accuracy and maximisation of long term response. *Genetica*, 136(2), 245–257. <https://doi.org/10.1007/S10709-008-9308-0/FIGURES/4>
- Gomez-Raya, L., Rauw, W. M., Dunkelberger, J. R., & Dekkers, J. C. M. (2019). Autozygosity and genetic differentiation of landrace and large white pigs as revealed by the genetic analyses of crossbreds. *Frontiers in Genetics*, 10(JUL), 454561. <https://doi.org/10.3389/FGENE.2019.00739/BIBTEX>
- Gorssen, W., Meyermans, R., Janssens, S., & Buys, N. (2021). A publicly available repository of ROH islands reveals signatures of selection in different livestock and pet species. *Genetics, Selection, Evolution : GSE*, 53(1). <https://doi.org/10.1186/S12711-020-00599-7>
- Hamill, R. M., McBryan, J., McGee, C., Mullen, A. M., Sweeney, T., Talbot, A., Cairns, M. T., & Davey, G. C. (2012). Functional analysis of muscle gene expression profiles associated with tenderness and intramuscular fat content in pork. *Meat Science*, 92(4), 440–450. <https://doi.org/10.1016/J.MEATSCI.2012.05.007>
- Hedrick, P. W., & Kalinowski, S. T. (n.d.). INBREEDING DEPRESSION IN CONSERVATION BIOLOGY.
- Hoffmann, A. A., Miller, A. D., & Weeks, A. R. (2020). Genetic mixing for population management: From genetic rescue to provenancing. *Evolutionary Applications*, 14(3), 634. <https://doi.org/10.1111/EVA.13154>
- Keller, L. F., & Waller, D. M. (2002). Inbreeding effects in wild populations. *Trends in Ecology & Evolution*, 17(5), 230–241. [https://doi.org/10.1016/S0169-5347\(02\)02489-8](https://doi.org/10.1016/S0169-5347(02)02489-8)
- Keller, M. C., Visscher, P. M., & Goddard, M. E. (2011). Quantification of Inbreeding Due to Distant Ancestors and Its Detection Using Dense Single Nucleotide Polymorphism Data. *Genetics*, 189(1), 237–249. <https://doi.org/10.1534/GENETICS.111.130922>
- Kirin, M., McQuillan, R., Franklin, C. S., Campbell, H., Mckeigue, P. M., & Wilson, J. F. (2010). Genomic runs of homozygosity record population history and consanguinity. *PLoS ONE*, 5(11). <https://doi.org/10.1371/JOURNAL.PONE.0013996>

- Li, Y., Chen, Z., Fang, Y., Cao, C., Zhang, Z., Pan, Y., & Wang, Q. (2022). Runs of Homozygosity Revealed Reproductive Traits of Hu Sheep. *Genes*, 13(10), 1848. <https://doi.org/10.3390/GENES13101848/S1>
- Liang, W., Zhang, H., Liu, Y., Lu, B., Liu, X., ... Q. L.-Genet. M., & 2014, undefined. (2014). Investigation of the association of two candidate genes (H-FABP and PSMC1) with growth and carcass traits in Qinchuan beef cattle from China. *Funpecrp.Com.BrW Liang, HL Zhang, Y Liu, BC Lu, X Liu, Q Li, Y CaoGenet. Mol. Res*, 2014•funpecrp.Com.Br, 13(1), 1876–1884. <https://doi.org/10.4238/2014.March.17.15>
- Liu, G. E., Hou, Y., Zhu, B., Cardone, M. F., Jiang, L., Cellamare, A., Mitra, A., Alexander, L. J., Coutinho, L. L., Dell'Aquila, M. E., Gasbarre, L. C., Lacalandra, G., Li, R. W., Matukumalli, L. K., Nonneman, D., Regitano, L. C. D. A., Smith, T. P. L., Song, J., Sonstegard, T. S., ... Keele, J. W. (2010). Analysis of copy number variations among diverse cattle breeds. *Genome Research*, 20(5), 693–703. <https://doi.org/10.1101/GR.105403.110>
- Lozada-Soto, E. A., Maltecca, C., Lu, D., Miller, S., Cole, J. B., & Tiezzi, F. (2021). Trends in genetic diversity and the effect of inbreeding in American Angus cattle under genomic selection. *Genetics Selection Evolution*, 53(1), 1–15. <https://doi.org/10.1186/S12711-021-00644-Z/FIGURES/6>
- Lynch, M., Conery, J., & Bu7rger3, R. (1995). MUTATION ACCUMULATION AND THE EXTINCTION OF SMALL POPULATIONS (Vol. 146, Issue 4). <http://www.journals.uchicago.edu/t-and-c>
- Lynch, M., Conery, J., & Burger, R. (1995). Mutation Accumulation and the Extinction of Small Populations. <https://doi.org/10.1086/285812>, 146(4), 489–518.
- Marras, G., Gaspa, G., Sorbolini, S., Dimauro, C., Ajmone-Marsan, P., Valentini, A., Williams, J. L., & MacCiotto, N. P. P. (2015). Analysis of runs of homozygosity and their relationship with inbreeding in five cattle breeds farmed in Italy. *Animal Genetics*, 46(2), 110–121. <https://doi.org/10.1111/AGE.12259>
- Martínez-Rocha, R., Hidalgo, J., Cesarani, A., Ramírez-Valverde, R., Núñez-Domínguez, R., García-Muñiz, J. G., & Domínguez-Viveros, J. (2022). Assessment of Genetic Diversity, Runs of Homozygosity, and Signatures of Selection in Tropical Milking Criollo Cattle Using Pedigree and Genomic Data. *Genes* 2022, Vol. 13, Page 1896, 13(10), 1896. <https://doi.org/10.3390/GENES13101896>
- Mastrangelo, S., Sardina, M., Tolone, M., Animal, R. D. G.-, & 2018, undefined. (2018). Genome-wide identification of runs of homozygosity islands and associated genes in local dairy cattle breeds. *Cambridge.OrgS Mastrangelo, MT Sardina, M Tolone, R Di Gerlando, AM Sutera, L Fontanesi, B PortolanoAnimal*, 2018•cambridge.Org, 12(12), 2480–2488. <https://doi.org/https://doi.org/10.1017/S1751731118000629>
- Mastrangelo, S., Tolone, M., Di Gerlando, R., Fontanesi, L., Sardina, M. T., & Portolano, B. (2016). Genomic inbreeding estimation in small populations: evaluation of runs of homozygosity in three local dairy cattle breeds. *Animal*, 10(5), 746–754. <https://doi.org/10.1017/S1751731115002943>
- Mastrangelo, S., Tolone, M., Sardina, M. T., Sottile, G., Sutera, A. M., Di Gerlando, R., & Portolano, B. (2017). Genome-wide scan for runs of homozygosity identifies potential candidate genes associated with local adaptation in Valle del Belice sheep. *Genetics, Selection, Evolution : GSE*, 49(1). <https://doi.org/10.1186/S12711-017-0360-Z>
- McQuillan, R., Leutenegger, A. L., Abdel-Rahman, R., Franklin, C. S., Pericic, M., Barac-Lauc, L., Smolej-Narancic, N., Janicijevic, B., Polasek, O., Tenesa, A., MacLeod, A. K., Farrington, S. M., Rudan, P., Hayward, C., Vitart, V., Rudan, I., Wild, S. H., Dunlop, M. G., Wright, A. F., ... Wilson, J. F. (2008). Runs of homozygosity in European populations. *American Journal of Human Genetics*, 83(3), 359–372. <https://doi.org/10.1016/J.AJHG.2008.08.007>
- Morton, N. M., Nelson, Y. B., Michailidou, Z., Rollo, E. M., Ramage, L., Hadoke, P. W. F., Seckl, J. R., Bunker, L., Horvat, S., Kenyon, C. J., & Dunbar, D. R. (2011). A stratified transcriptomics analysis of polygenic fat and lean mouse adipose tissues identifies novel candidate obesity genes. *PloS One*, 6(9). <https://doi.org/10.1371/JOURNAL.PONE.0023944>
- Novo, L. C., Poindexter, M. B., Rezende, F. M., Santos, J. E. P., Nelson, C. D., Hernandez, L. L., Kirkpatrick, B. W., & Peñagaricano, F. (2023). Identification of genetic variants and individual

- genes associated with postpartum hypocalcemia in Holstein cows. *Scientific Reports* 2023 13:1, 13(1), 1–9. <https://doi.org/10.1038/s41598-023-49496-1>
- Peripolli, E., Stafuzza, N. B., Munari, D. P., Lima, A. L. F., Irgang, R., Machado, M. A., Panetto, J. C. do C., Ventura, R. V., Baldi, F., & da Silva, M. V. G. B. (2018). Assessment of runs of homozygosity islands and estimates of genomic inbreeding in Gyr (*Bos indicus*) dairy cattle. *BMC Genomics*, 19(1), 1–13. <https://doi.org/10.1186/S12864-017-4365-3/FIGURES/5>
- Powell, J. E., Visscher, P. M., & Goddard, M. E. (2010). Reconciling the analysis of IBD and IBS in complex trait studies. *Nature Reviews Genetics* 2010 11:11, 11(11), 800–805. <https://doi.org/10.1038/nrg2865>
- Purcell, S., Neale, B., Todd-Brown, K., Thomas, L., Ferreira, M. A. R., Bender, D., Maller, J., Sklar, P., De Bakker, P. I. W., Daly, M. J., & Sham, P. C. (2007). PLINK: A tool set for whole-genome association and population-based linkage analyses. *American Journal of Human Genetics*, 81(3), 559–575. <https://doi.org/10.1086/519795>
- Purfield, D., Berry, D., McParland, S., genetics, D. B.-B., & 2012, undefined. (2012). Runs of homozygosity and population history in cattle. SpringerDC Purfield, DP Berry, S McParland, DG BradleyBMC Genetics, 2012•Springer, 13. <https://doi.org/10.1186/1471-2156-13-70>
- Quinlan, A. R., & Hall, I. M. (2010). BEDTools: a flexible suite of utilities for comparing genomic features. *Bioinformatics* (Oxford, England), 26(6), 841–842. <https://doi.org/10.1093/BIOINFORMATICS/BTQ033>
- Rothhammer, S., Kremer, P. V., Bernau, M., Fernandez-Figares, I., Pfister-Schär, J., Medugorac, I., & Scholz, A. M. (2014). Genome-wide QTL mapping of nine body composition and bone mineral density traits in pigs. *Genetics Selection Evolution*, 46(1), 1–11. <https://doi.org/10.1186/S12711-014-0068-2/TABLES/2>
- Sato, S., Ohnishi, C., Uemoto, Y., & Kobayashi, E. (2011). Haplotype analysis within quantitative trait locus affecting intramuscular fat content on porcine chromosome 7. *Czech J. Anim. Sci*, 56(12), 521–528.
- Saura, M., Fernandez, A., Rodriguez, M. C., Toro, M. A., Barragan, C., Fernandez, A. I., & Villanueva, B. (2013). Genome-Wide Estimates of Coancestry and Inbreeding in a Closed Herd of Ancient Iberian Pigs. *PLOS ONE*, 8(10), e78314. <https://doi.org/10.1371/JOURNAL.PONE.0078314>
- Schiavo, G., Bovo, S., Ribani, A., Moscatelli, G., Bonacini, M., Prandi, M., Mancin, E., Mantovani, R., Dall’Olio, S., & Fontanesi, L. (2022). Comparative analysis of inbreeding parameters and runs of homozygosity islands in 2 Italian autochthonous cattle breeds mainly raised in the Parmigiano-Reggiano cheese production region. *Journal of Dairy Science*, 105(3), 2408–2425. <https://doi.org/10.3168/JDS.2021-20915>
- Sell-Kubiak, E., Czarniecki, Ł., & Strabel, T. (2018). Challenges in inbreeding estimation of large populations based on Polish Holstein-Friesian cattle pedigree. *Journal of Applied Genetics*, 59(3), 313–323. <https://doi.org/10.1007/S13353-018-0441-X>
- Shi, L., Wang, L., Liu, J., Deng, T., Yan, H., Zhang, L., Liu, X., Gao, H., Hou, X., Wang, L., & Zhao, F. (2020). Estimation of inbreeding and identification of regions under heavy selection based on runs of homozygosity in a Large White pig population. *Journal of Animal Science and Biotechnology*, 11(1), 1–10. <https://doi.org/10.1186/S40104-020-00447-0/TABLES/2>
- Sun, L., Yuan, C., Guo, T., Bai, Y., Lu, Z., & Liu, J. (2025). The accumulation of harmful genes within the ROH hotspot regions of the Tibetan sheep genome does not lead to genetic load. *BMC Genomics*, 26(1), 60. <https://doi.org/10.1186/S12864-025-11207-7/TABLES/2>
- Szmatola, T., Gurgul, A., Jasielczuk, I., Ząbek, T., Ropka-Molik, K., Litwińczuk, Z., & Bugno-Poniewierska, M. (2019). A Comprehensive Analysis of Runs of Homozygosity of Eleven Cattle Breeds Representing Different Production Types. *Animals* 2019, Vol. 9, Page 1024, 9(12), 1024. <https://doi.org/10.3390/ANI9121024>
- Szmatola, T., Jasielczuk, I., Semik-Gurgul, E., Szyndler-Nędza, M., Blicharski, T., Szulc, K., Skrzypczak, E., & Gurgul, A. (2020). Detection of runs of homozygosity in conserved and commercial pig breeds in Poland. *Journal of Animal Breeding and Genetics*, 137(6), 571–580. <https://doi.org/10.1111/JBG.12482>
- Szpiech, Z. A., Xu, J., Pemberton, T. J., Peng, W., Zöllner, S., Rosenberg, N. A., & Li, J. Z. (2013). Long runs of homozygosity are enriched for deleterious variation. *American Journal of Human Genetics*, 93(1), 90–102. <https://doi.org/10.1016/j.ajhg.2013.05.003>

- Wickham, H. (2016). Programming with ggplot2. 241–253. https://doi.org/10.1007/978-3-319-24277-4_12
- Xu, L., Zhao, G., Yang, L., Zhu, B., Chen, Y., Zhang, L., Gao, X., Gao, H., Liu, G. E., & Li, J. (2019). Genomic Patterns of Homozygosity in Chinese Local Cattle. *Scientific Reports* 2019 9:1, 9(1), 1–11. <https://doi.org/10.1038/s41598-019-53274-3>
- Xu, Z., Mei, S., Zhou, J., Zhang, Y., Qiao, M., Sun, H., Li, Z., Li, L., Dong, B., Oyelami, F. O., Wu, J., & Peng, X. (2021). Genome-Wide Assessment of Runs of Homozygosity and Estimates of Genomic Inbreeding in a Chinese Composite Pig Breed. *Frontiers in Genetics*, 12, 720081. <https://doi.org/10.3389/FGENE.2021.720081/BIBTEX>
- Xu, Z., Sun, H., Zhang, Z., Zhao, Q., Olasege, B. S., Li, Q., Yue, Y., Ma, P., Zhang, X., Wang, Q., & Pan, Y. (2019). Assessment of autozygosity derived from runs of homozygosity in Jinhua pigs disclosed by sequencing data. *Frontiers in Genetics*, 10(MAR), 409192. <https://doi.org/10.3389/FGENE.2019.00274/BIBTEX>
- Zappaterra, M., Zambonelli, P., Schivazappa, C., Simoncini, N., Virgili, R., Stefanon, B., & Davoli, R. (2021). Investigating the features of PDO green hams during salting: Insights for new markers and genomic regions in commercial hybrid pigs. *Animals*, 11(1), 1–19. <https://doi.org/10.3390/ANI11010068>
- Zhang, Q., Calus, M. P. L., Guldbrandtsen, B., Lund, M. S., & Sahana, G. (2015). Estimation of inbreeding using pedigree, 50k SNP chip genotypes and full sequence data in three cattle breeds. *BMC Genetics*, 16(1), 1–11. <https://doi.org/10.1186/S12863-015-0227-7/FIGURES/6>
- Zhu, J., Yang, Z., Hao, W., Li, J., Wang, L., Xia, J., Zhang, D., Liu, D., & Yang, X. (2022). Characterization of a Read-through Fusion Transcript, BCL2L2-PABPN1, Involved in Porcine Adipogenesis. *Genes*, 13(3), 445. <https://doi.org/10.3390/GENES13030445/S1>

6 GENERAL DISCUSSION, CONCLUSIONS, CHALLENGES, AND RECOMMENDATIONS FOR FURTHER RESEARCH

6.1 Introduction

The importance of livestock species such as pigs, sheep, and goats to the socio-economic well-being of people in developing and underdeveloped countries in the tropics in terms of nutrition, income, and intangible benefits (i.e., savings, insurance against emergencies, cultural and ceremonial purposes) cannot be overemphasised. South African pigs are a significant source of protein, a major contributor to household income, and a key factor in food security and poverty alleviation, also playing a crucial role in the nation's economic well-being. Pork is easily digestible, rich in essential amino acids, has high biological value, and shares the nutritional qualities of both white and red meat; thus, it is regarded as “pink meat” (Adeschinwa *et al.*, 2024). Lately, there has been a heightened demand for pork consumption (Yu & Jensen, 2022). The rising demand for pork triggers an increase in swine production, thus necessitating pig rearing improvement. This includes adopting better sustainable farming methods and improving standards of animal welfare. Investing in better production systems and breeding techniques can ensure healthier pigs and more efficient production processes.

The South African pig population is complex and consists of breeds kept and thriving under various production systems and is a host to numerous non-descript local ecotypes. These local ecotypes remain largely uncharacterized and are primarily raised under minimal-input production systems, where they are subjected to a range of natural selection pressures, including climatic variability, nutritional limitations, and disease challenges. The non-descript or undefined ecotype gene pool is hypothesised to harbour distinct genes from the known commercial and indigenous breeds. Over the years, the genetic makeup of the national pig population has evolved into a blend of commercial and indigenous breeds, with many populations having been present in South Africa long enough to adapt to local environments. This genetic admixture presents an opportunity for increased genetic diversity and heterosis.

6.2 Aims and Objectives

The present study aimed to investigate the genomic diversity of South African indigenous and village pig populations to provide information useful for assessing their potential risk of extinction and informing conservation strategies. Initially, we investigated mating, breeding, and production systems through surveys (Chapter 3). Furthermore, the commercially available

Porcine 80K SNP BeadChip was used to analyse the population structure and genetic diversity in the population (Chapter 4) and to investigate inbreeding and identify candidate genes in the genomes of South African indigenous and village pig populations (Chapter 5). The Porcine 80K SNP BeadChip is a high-throughput genotyping array developed by Illumina for the detection and analysis of genetic variations in pigs. Several South African studies have indicated the Porcine 80K SNP BeadChip beneficial applicability (Feng *et al.*, 2022; Lin *et al.*, 2024; Xu *et al.*, 2023). The porcine 80K BeadChip has recently been upgraded from porcine SNP60K. A high number of SNPs enables broader genomic region coverage and is advantageous for research examining associations between genetic markers and traits (Herrero-Medrano *et al.*, 2013).

6.3 General Discussion

Upon analysing mating, breeding, and production systems, it was found that pig farming is mainly dominated by males between the ages of 40 and 60 years; however, women play a significant role in pig rearing and daily management. Women do light work, such as cleaning the stalls and providing feed, while males do jobs that require physical energy, such as constructing stalls (Rindia Wati *et al.*, 2022). Pigs were mostly kept under extensive or semi-extensive systems, provided with housing constructed from available resources (i.e., iron, untreated wood, wire, mud, etc). In most parts of Africa, pigs are primarily raised under extensive production systems where they scavenge for food with little human care provided (Dione *et al.*, 2014). A significant portion (38%) of uncontrolled breeding was observed. It was also noted that the number of boars kept for breeding was relatively low, with most households not keeping any. This emphasises the need for farmers to prioritize preserving breeding males as they play a significant role in attaining high genetic diversity and lowering levels of inbreeding. Feed and water were provided. The feeding practices include commercial feed, supplemented with swill in some systems.

Additionally, the study examined the morphological traits of pig populations present in these villages. Phenotypic traits are important distinguishers used by farmers to identify and classify breeds. Using the PCA based on the qualitative and quantitative morphometric traits also revealed significant morphological variation among the pig populations. High numbers of pigs that are admixed in these populations lacking differentiation were shown by the PCA through the overlapping of clusters. This admixture across provinces is thought to be due to geographical proximity. Therefore, variations in management practices, production limitations, and geographical factors are likely to influence genetic diversity and adaptive potential of pigs

from different production systems and agro-ecological environments. Results from this chapter highlight the importance of local adaptation for pig populations to survive and perform well under the challenging conditions of smallholder, low-input production systems, including limited and seasonal feed availability, climate variability, poor housing and management, limited veterinary care, uncontrolled breeding, and lack of formal recordkeeping.

As a follow-up to Chapter 3, Chapter 4 genotyped 384 village pigs raised under different locations and production systems from six provinces of South Africa (Eastern Cape, Gauteng, KwaZulu-Natal, Limpopo, Mpumalanga, and North West) to investigate their genetic diversity, population genetic structure, admixture levels, and potential risk to extinction. The populations reported low inbreeding levels; Alfred-Nzo, Johannesburg, and West Rand had the highest inbreeding levels. Low diversity levels were also reported in the overall population drawn from polymorphic information content (PIC), percentage of polymorphic loci ($\%P$), total (N_a), effective allele numbers ($N_e^{alleles}$), observed (H_o), and expected heterozygosity (H_E). The low levels of genetic differentiation, as observed in principal component analysis and pairwise F_{ST} comparisons among village populations, suggest a shared genetic background with fewer unique alleles. Results from the ADMIXTURE analysis were congruent with those observed in the PCA and pairwise F_{ST} , showing high levels of admixture. These findings were further supported by Treemix, which revealed significant introgressive hybridization. This congruence supported the impacts of geographic origin on population structuring assumptions. This pattern implies that, due to high admixture and low differentiation, the populations may possess fewer unique functional alleles, which could limit their adaptive potential to specific agro-ecological zones and low-input production systems. Effective population size estimates also indicated a decline over time. Observations have also shown that farmers and even research flocks often have different pig breeds raised on the same farm.

Chapter 5 estimated genomic levels of inbreeding, detected runs of homozygosity, and examined associated genes. In this chapter, the pairwise comparisons from genome-wide IBD revealed some individuals with IBD values exceeding 0.5, which showed the higher population relatedness, suggesting a less diverse genetic structure likely influenced by traditional breeding practices and inbreeding. Therefore, we proceeded to investigate genomic regions for ROHs as they correspond to IBD segments within a single individual that descends from shared parental ancestry, with longer ROH indicating more recent ancestry and shorter ROH indicative of ancient shared ancestry (Curik *et al.*, 2014). A higher frequency of shorter ROHs was detected

for all populations in this study. This may be due to poor breeding management and small population sizes observed in Chapters 3 and 4, or to historical recombination events. Low inbreeding coefficients were observed based on both F_{ROH} and F_{HOM} . These results are supported by the low and, in some cases, negative F_{IS} values, indicating little to no deficit of heterozygotes within populations, as reported in Chapter 4. There was no clear correlation between ROH and the inbreeding coefficient. In this study, ROH islands were only determined in SSC7 from three populations. The identified ROH islands, which are highly homozygous genomic regions due to selection pressures, were found to be associated with various QTLs. The most commonly shared QTLs in these populations were linked to economically important traits (teat number, dressing percentage, lean meat percentage, intramuscular fat content). Genes linked to reproductive traits such as *PPP1R3E* and *PABPN1* were identified (number of stillborn). The identification of these genes will help improve breeding programs, productivity, and genetic selection, leading to more efficient and sustainable livestock production.

6.4 Conclusion

Overall, this study provides comprehensive information on production systems and the genomic makeup of South African pig populations. The village pig populations are characterized by inadequate production systems, inbreeding, and small effective population sizes. These factors contribute to higher ROH frequencies, low genetic diversity, and an increased risk of population extinction. The findings also highlight genome fragmentation and alterations to unique adaptation patterns caused by both historical migrations and modern crossbreeding practices.

6.5 Study Challenges & Recommendations

6.5.1 This study relied on data collected through surveys with pig farmers, and the lack of records and essential information, such as breeding line records, posed a significant challenge. Additionally, the small sample size from some populations due to a lack of availability further limited the effectiveness of the study. Smaller sample size compared to other populations reduces statistical power, hinders the representation of genetic diversity, and limits the generalizability of the findings. These limitations may lead to biased conclusions, increased error margins, and the potential omission of rare genetic variants, ultimately affecting the accuracy and applicability of the results.

6.5.2 The following recommendations are based on the study's findings, taking into account its stated shortcomings:

- (i) Pig production enhancement in South Africa is recommended to meet the needs of the increasing population and transform the industry from the traditional method of scavenging pigs to a more productive system. This requires holding workshops and educational seminars for pig keepers and smallholder farmers about management practices and implementation of policies that support proper management of indigenous breeds and undefined village pigs, thereby ensuring their conservation and maintenance of genetic diversity.
- (ii) There are many complementary approaches for analysing genomic regions such as CNVs, which assist in assessing the genetic diversity of the genomes, their makeup, and differences. Numerous studies have used CNVs to evaluate genetic diversity in genomes, the genetic composition of various species, and individual variations. However, no study has been conducted in South African indigenous and village pig breeds using CNVs; thus, it is recommended.

6.6 References

- Adeshinwa, A. O. K., Boladuro, B. A., Dunmade, A. S., Idowu, A. B., Moreki, J. C., & Wachira, A. M. (2024). Pig production in Africa: current status, challenges, prospects and opportunities. *Animal Bioscience*, 37(4), 730. <https://doi.org/10.5713/AB.23.0342>
- Curik, I., Ferencaković, M., & Sölkner, J. (2014). Inbreeding and runs of homozygosity: A possible solution to an old problem. *Livestock Science*, 166(1), 26–34. <https://doi.org/10.1016/J.LIVSCI.2014.05.034>
- Dione, M. M., Ouma, E. A., Roesel, K., Kungu, J., Lule, P., & Pezo, D. (2014). Participatory assessment of animal health and husbandry practices in smallholder pig production systems in three high poverty districts in Uganda. *Preventive Veterinary Medicine*, 117(3–4), 565–576. <https://doi.org/10.1016/J.PREVETMED.2014.10.012>
- Feng, X., Diao, S., Liu, Y., Xu, Z., Wei, C., Yuan, X., & Li, J. (2022). Analysis of selection signatures for Hainan pigs across the whole genome based on SNP beadchips. <https://www.cabidigitallibrary.org/doi/full/10.5555/20220153870>
- Herrero-Medrano, J. M., Megens, H. J., Groenen, M. A. M., Ramis, G., Bosse, M., Pérez-Enciso, M., & Crooijmans, R. P. M. A. (2013). Conservation genomic analysis of domestic and wild pig populations from the Iberian Peninsula. *BMC Genetics*, 14, 1–13. <https://doi.org/10.1186/1471-2156-14-106>
- Lin, D., Qiu, Y., Zhou, F., Li, X., Deng, S., Yang, J., Chen, Q., Cai, G., Yang, J., Wu, Z., & Zheng, E. (2024). Genome-wide detection of multiple variants associated with teat number in French Yorkshire pigs. *BMC Genomics*, 25(1), 1–12. <https://doi.org/10.1186/S12864-024-10611-9/TABLES/4>
- Rindia Wati, N. K., Amrawaty, A. A., & Sri Lestari, V. (2022). Gender roles at pig farming. *Advances in Environmental Biology*, 16(11), 1–5. <https://doi.org/10.22587/AEB.2022.16.11.1>
- Xu, C., Zhang, W., Jiang, Y., Zhou, M., Liu, L., Su, S., Li, X., & Wang, C. (2023). Genome-Wide Detection and Analysis of Copy Number Variation in Anhui Indigenous and Western Commercial Pig Breeds Using Porcine 80K SNP BeadChip. *Genes*, 14(3). <https://doi.org/10.3390/genes14030654>
- Yu, W., & Jensen, J. D. (2022). Sustainability implications of rising global pork demand. *Animal Frontiers*, 12(6), 56–60. <https://doi.org/10.1093/AF/VFAC070>

7 APPENDIX: SUPPLEMENTARY FILES

Supplementary files can be found on the following link:

https://drive.google.com/drive/folders/1vY_YdpTlfKPDdcGtCG0OPyWHbv89HTQg?usp=s hare_link

Chapter 4: Supplementary files

Supplementary Table 4.1 Summary of expected, observed heterozygosity, and inbreeding coefficient across populations

Supplementary Table 4.2 Effective population size over the past generations.

Supplementary Table 4.3 Pairwise genetic differentiation between 13 pig populations.

Supplementary Table 4.4 Ancestry proportions of each pre-defined pig population in each of fourteen genetic clusters identified from admixture analysis.

Supplementary Figure 4.1 SNPs before and after quality control per village population.

Chapter 5: Supplementary files

Supplementary Table 5.1 ROH islands with identified genes and associated QTLs.