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Runs of homozygosity detects inbreeding coefficients in South African beef cattle

B. Mtileni[1*], K. Nephawe[1], and M. Modiba[1]

[1]*Tshwane University of Technology, Pretoria, South Africa;* [*]*Corresponding author:* mtilenib@tut.ac.za

Introduction

Fitness is a fundamental term in population biology, influencing both a species' ecological stability and evolutionary trajectory and it is often quantified by proxies such as survival or reproductive success (Wadgymar *et al*., 2024). In small populations, inbreeding can lead to a drop-in fitness, refer to as inbreeding depression, which may result in population decreases and eventual extinction (Keller, 2002; Hedrick and Garcia-Dorado, 2016). So genome-wide runs of homozygosity (ROH) are excellent in understanding population history, estimating genomic inbreeding, and deciphering genetic architecture of complex traits and diseases, as well as identifying genes of agro-economic traits (Modiba *et al*., 2022). Runs of homozygosity (ROH) are contiguous stretches of DNA where both chromosomes in a pair are identical because they were inherited from a common ancestor. These patterns result from both recent and historical inbreeding, with segment length acting as an indicator of the timing of genetic relatedness; longer segments are more indicative of recent inbreeding events. This study aimed to assess the level of inbreeding in South African beef cattle using Runs of Homozygosity (ROH) to estimate the inbreeding coefficient (FROH).

Materials & Methods

A total of 96 semen samples were collected from South African cattle breeds: Nguni (n = 28), Bonsmara (n = 21), Angus (n = 22), and Simmental (n = 25) and genotyped using the Illumina BovineSNP 150K BeadChip array. The LEA package in R version 3.18.0 was used for population structure analysis. Using selective non-negative matrix factorization (sNMF) models with the cross-entropy criterion, as used in LEA, the most likely number of ancestral populations (K) was calculated. Triangle plots were then used to classify individuals in the population in the StructuRly database. DetectRUNS package (version 0.9.6) in R was used to categorizing ROH into five length classes: 0–6 Mb, 6–12 Mb, 12–24 Mb, 24–48 Mb, and >48 Mb across all four breeds. ROH were calculated per breed, and chromosomal ROH coverage was defined as the overall length of each chromosome by ROH across all individuals in the population. Furthermore, the proportion of SNPs in ROH was determined using the frequency of each SNP present in ROH across individuals. Individual homozygosity was quantified following the approach described by (McQuillan *et al*., 2008). The genomic inbreeding coefficient based on runs of homozygosity (Froh) was calculated as:

$$Froh = {\sum \text{Lroh}}/{\text{Lauto}}$$

where ∑Lroh represents the total length of ROH in an individual and Lauto represents the total length of the autosomal genome.

Results

The analysis, using K = 4, revealed significant genetic distinctions between Nguni and Bonsmara cattle, and between Angus and Simmental cattle (Figure 1). The Nguni and Bonsmara populations showed some genetic mixing. These observed clustering patterns are consistent with the known breed histories.

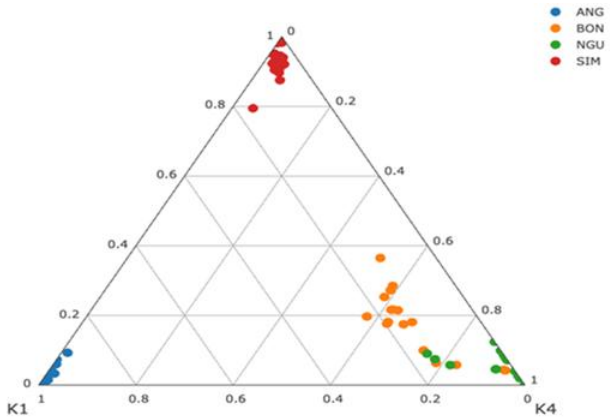

Figure 1: Population structure of individual animals inferred using ADMIXTURE analysis at K = 4, with colours indicating genetic ancestry corresponding to Nguni (NGU), Bonsmara (BON), Simmental (SIM), and Angus (ANG) breeds.

The distribution of runs of homozygosity (ROH) length across the genome revealed marked differences among breeds (Figure 2). Nguni cattle exhibited the highest number of ROH segments within the shortest length class (0–6 Mb), followed by Simmental, Angus, and Bonsmara. While in contrast, within the intermediate ROH length classes (6–12 Mb and 12–24 Mb), Angus cattle displayed the highest number of ROH segments, reflecting more recent inbreeding and intensive selection. Overall, Bonsmara cattle consistently showed the lowest number of ROH segments across all length classes, suggesting a relatively lower level of genomic autozygosity.

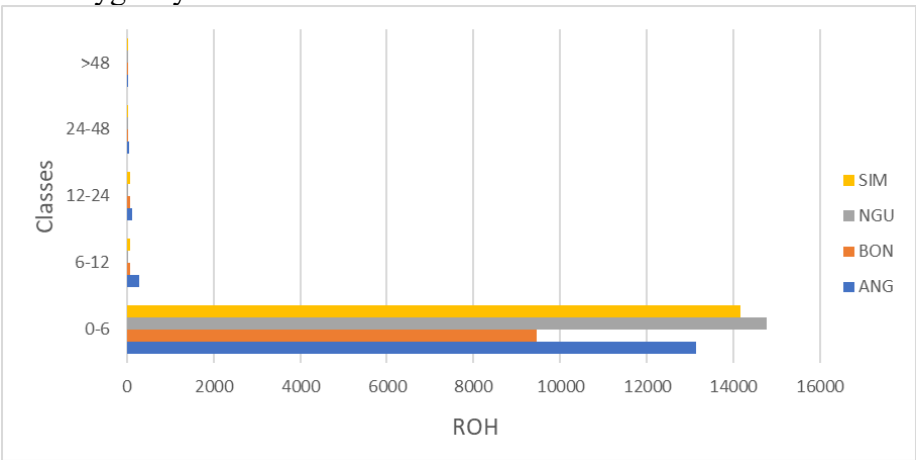

Figure 2: Distribution of ROH in class >24Mb across the genome with color representing the number of ROH per breed.

Manhattan plots illustrating the proportion of SNPs within ROH (Figure 3) revealed distinct breed-specific patterns of ROH hotspots. Bonsmara cattle revealed comparatively lower ROH signals, with most peaks ranging between 50–60%. In contrast, Nguni cattle showed several pronounced ROH hotspots, with peaks exceeding 75% on chromosomes 1 and 6. Simmental cattle displayed multiple high-intensity ROH peaks (>75%) across chromosomes 2, 6, 7, 11, 14, 21, and 22. Angus cattle revealed ROH peaks on 1, 4, 6, 9, 11, 13, 14, 16, 19 and 21, reflecting a greater degree of shared presented the most extensive in a genome.

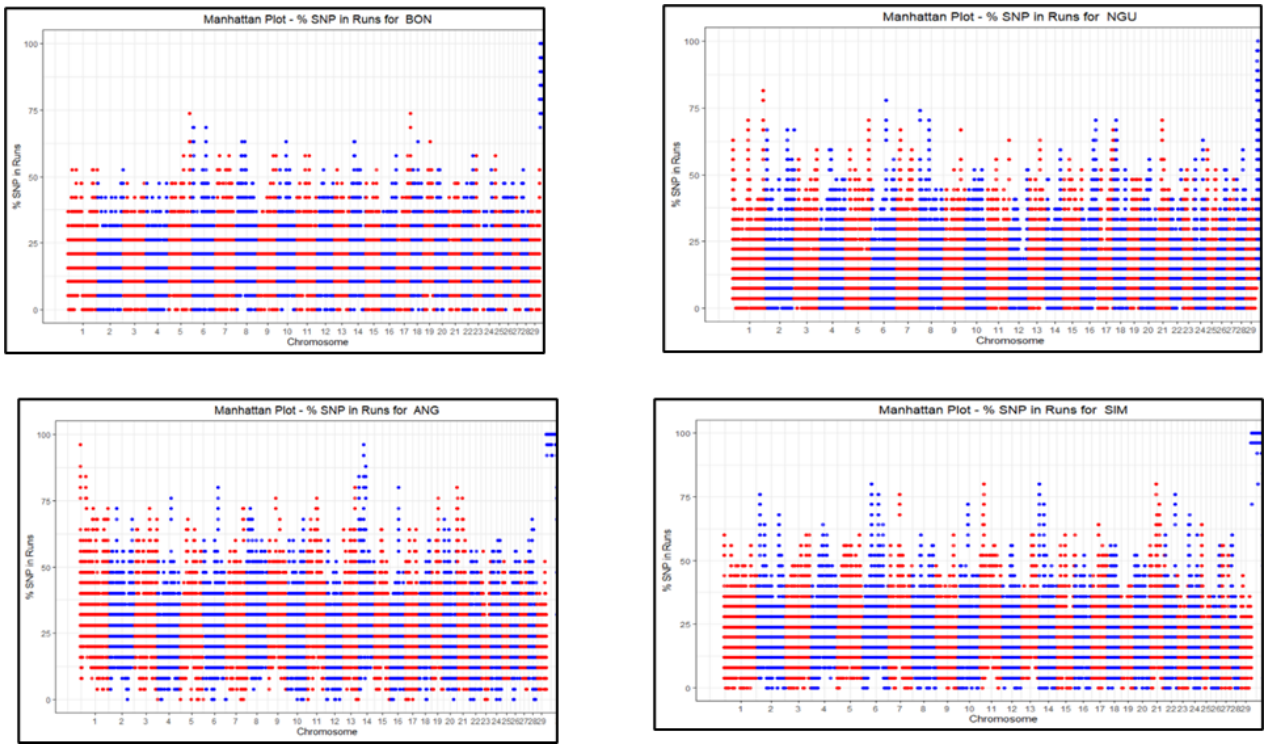

Figure 3: Manhattan plots of the ROH hotspots in South African Bonsmara (BON), Nguni (NGU), Angus (ANG) and Simental (SIM) (A), with each dotted colour representing SNP and high peaks regions of the genome that have the greatest ROH. X-axis (Chromosome): Autosomes 1–29 and Y-axis (% SNP in Runs).

The distribution of genomic inbreeding coefficients (FROH) differed among the studied breeds (Figure 4). Angus exhibited the highest FROH values around ~0.30, indicating a higher level of genomic inbreeding, followed by Simental cattle FROH values around ~0.25, suggesting moderate levels of autozygosity with greater individual variation. Then Bonsmara displayed lower FROH values (~0.22), while Nguni cattle showed the lowest FROH values (~0.15) indicating higher genetic diversity and lower recent inbreeding. Analysis of IBD, PI_HAT values were highest (~0.5) when Z0 was near 0.0, reflecting close relatives highlighting shared genome. While PI_HAT values Z0 near 1.0, indicating unrelated individuals with nearly all segments non-shared.

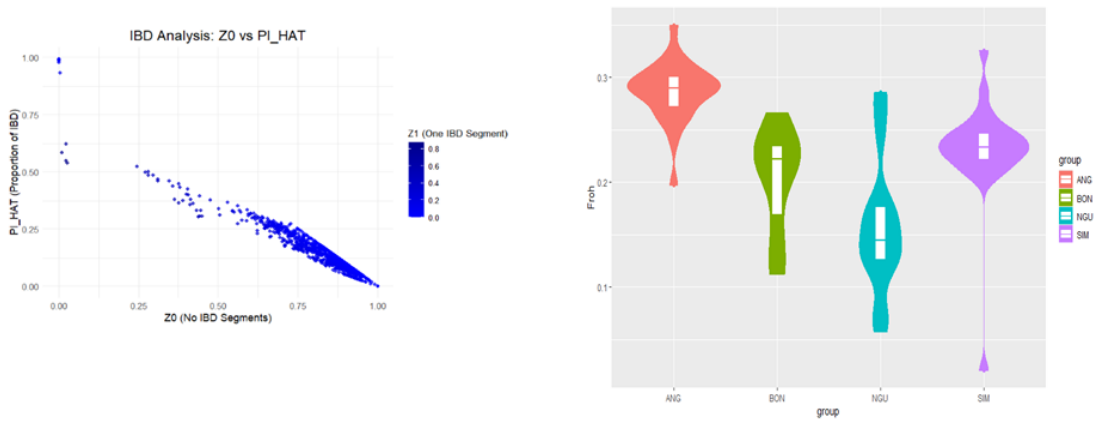

Figure 4: Box plot for Inbreeding coefficient (FROH) and PI_Hat explaining the estimated proportion of alleles shared identical-by-descent (IBD) between two individuals.

Discussion

Population admixture confirmed a clear genetic differentiation among the four breeds with a mix individual between the Bonsmara and Nguni. These results are consistent with previous studies reporting higher genetic diversity and admixture in indigenous and composite breeds than in commercial breeds due to historical gene flow, adaptation to diverse environments, and less intensive artificial selection (Makina *et al*., 2015). The distribution of runs of homozygosity (ROH) length across the genome further highlighted marked differences in demographic history and selection intensity among breeds. Nguni cattle exhibited the highest number of short ROH segments (0–6 Mb), followed by Simmental, Angus, and Bonsmara. Short ROH are generally associated with ancient inbreeding and long-term population history rather than recent mating among close relatives (Howrigan *et al*., 2011; Zavarez *et al*., 2015). Angus cattle exhibited the highest number of ROH segments in the intermediate length classes (6–12 Mb and 12–24 Mb), extensive ROH across many chromosomes and the highest FROH values (0.28–0.30) indicating more recent inbreeding. For Angus, the combination of intermediate ROH, genome-wide hotspots, and high FROH shows that the breed has undergone recent, intensive selection, leading to higher genomic inbreeding and less genetic diversity compared to more diverse or composite breeds like Nguni or Bonsmara. A genome-wide study on ROH for Aberdeen Angus cattle identified multiple ROH islands consistent with regions of high homozygosity shared across individuals (Kolpakov *et al*., 2024).

Conclusions

Overall, these findings highlight the contrasting genetic architectures between indigenous and composite breeds, which maintain greater diversity and lower recent inbreeding. In contrast, the selected Angus population revealed extensive ROH hotspots and high FROH values, reflecting more recent inbreeding driven by intensive selection.

Acknowledgements

Tshwane University of Technology

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