



Inbreeding Estimation and Effective Population size of four South African Dairy Breeds

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Background

- Studies indicate that inbreeding due to intense selection is increasing in dairy cattle populations across the world.
- The development of reproductive technologies such as artificial insemination (AI) and multiple ovulation and embryo transfer (MOET) is a contributing factor.
- Traditionally inbreeding and genetic variability were assessed for a population from its pedigree information but these pedigree-based estimates (F_{ped}) can be unreliable.
- Inbreeding estimates is expected to be more accurate when it is based on genomic estimates such as runs of homozygosity (ROH) using genome-wide DNA markers, usually SNP.

Motivation and aim

This study aims to estimate genomic inbreeding and effective population sizes for four SA dairy breeds.

Materials and Methods

- A total of 1 006 animals (Holstein = 416, Jersey = 416, Ayrshire = 112 and SA Dairy Swiss = 62) were included in the study.
- Animals were genotyped using the 50K Illumina SNP bead chip.
- Quality control was performed using PLINK v1.07 (Purcell *et al.*, 2007) based on missing genotypic data (call rate < 90%), marker call rate (< 95%) and minor allele frequency (MAF < 0.02).
- ROH was estimated using PLINK v1.07 (Purcell *et al.*, 2007). ROH was calculated at different lengths: ROH1 = 1Mb, ROH2 = 2Mb, ROH4 = 4Mb, ROH8 = 8Mb, ROH16 = 16Mb.
- Effective population size (N_e) was estimated using SNeP tool (Barbato *et al.*, 2015)

Results & Discussion

- Limited loss of heterozygosity was observed across all four breeds.
- The Jersey had the lowest mean heterozygosity of all four breeds.
- The Holstein and the SA Dairy Swiss breeds show the highest mean expected heterozygosity (H_e) levels.
- Negative F_{IS} values for all four breeds may be due to effective management against inbreeding and use of both local and international bulls.

Table 1. Summary statistics before and after LDP (linkage disequilibrium pruning) for the four Dairy cattle breeds.

	n	H_o^*	H_o^{**}	H_e^*	H_e^{**}	F_{IS}^*	F_{IS}^{**}
Holstein	406	0.35882	0.35731	0.35646	0.35497	-0.00664	-0.00657
Jersey	414	0.33991	0.33769	0.33644	0.33413	-0.01033	-0.01059
Ayrshire	112	0.35478	0.34906	0.34148	0.34129	-0.0389	-0.02281
SA Dairy Swiss	62	0.34446	0.36505	0.33801	0.35813	-0.01903	-0.02067

*Before LDP
**After LDP

- Inbreeding is present in all four breeds, with higher amount of inbreeding in recent years (F_{ROH16})
- The Holstein and Jersey show the highest amount of inbreeding in recent years.

Table 2. F_{ROH} distribution for the four Dairy cattle breeds.

	F_{ROH1}	F_{ROH2}	F_{ROH4}	F_{ROH8}	F_{ROH16}
Holstein	0.0904	0.0989	0.1060	0.1538	0.2522
Jersey	0.0770	0.0862	0.0981	0.1596	0.2439
Ayrshire	0.0899	0.0913	0.0981	0.1381	0.1921
SA Dairy Swiss	0.0991	0.0995	0.1082	0.1413	0.1848

$F_{ROH1} = 1Mb$, $F_{ROH2} = 2Mb$, $F_{ROH4} = 4Mb$, $F_{ROH8} = 8Mb$, $F_{ROH16} = 16Mb$

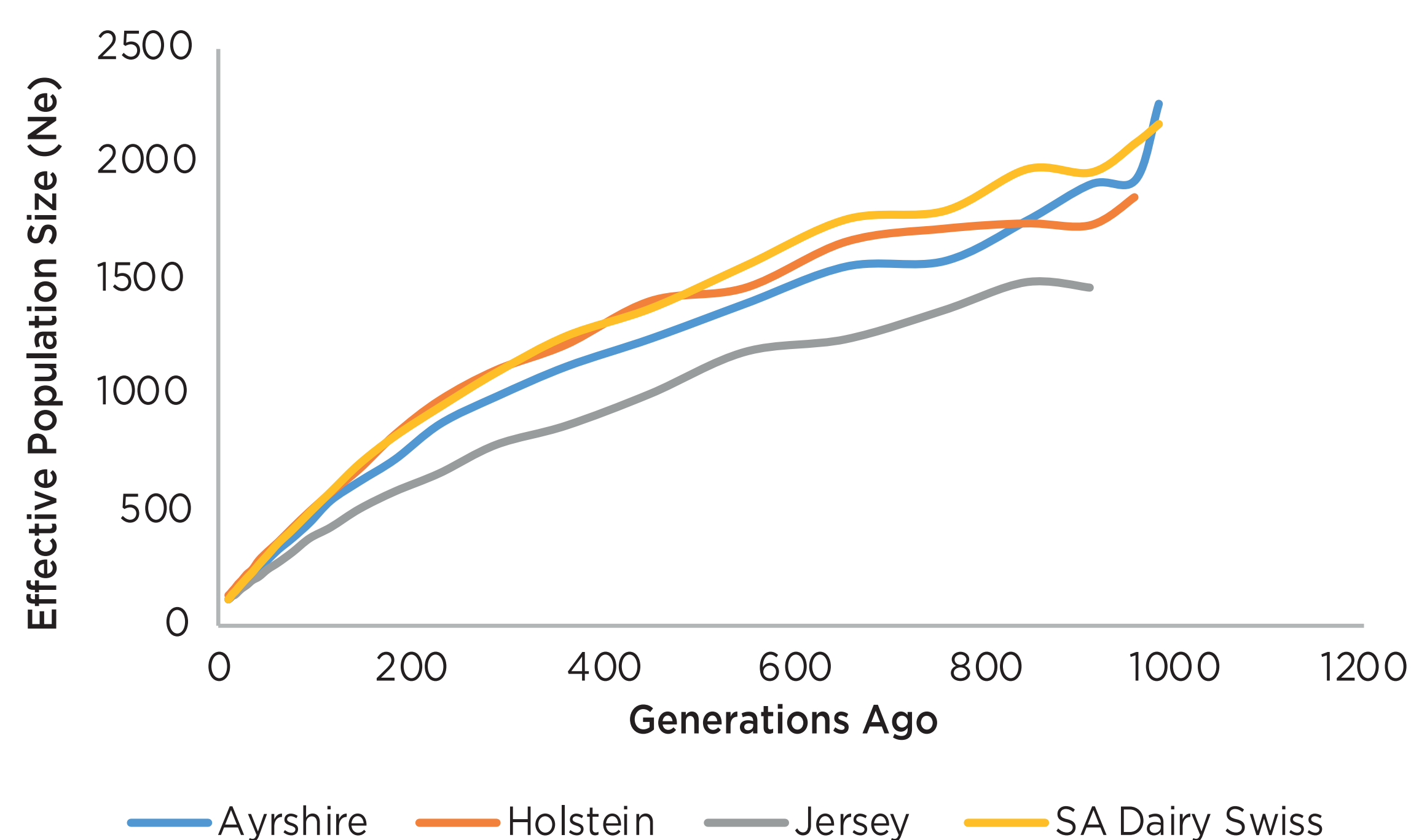


Fig 1. Trends in historic effective population size (N_e) for the four Dairy cattle breeds.

- A general decrease in N_e can be seen for all four breeds, indicating a loss of genetic diversity.
- The Holstein shows the largest decrease in N_e from 1853 animals 995 generations ago, to 133 animals 13 generations ago.



SA Holstein (www.saholstein.co.za)



SA Jersey (www.jerseysa.co.za)



SA Ayrshire (www.ayrshire.co.za)



SA Dairy Swiss (www.dairyswiss.co.za)

Conclusion

The loss of genetic diversity as well as the increase in inbreeding is a cause for major concern in the dairy industry. Mating strategies, with the use of genomic data, should be implemented to minimize inbreeding.