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Evaluating the Genetic Diversity and Autozygosity of Indigenous South African Sheep Breeds Characterized by Small Population Sizes

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Abstract

Indigenous South African (SA) sheep populations are characterised by relatively small population sizes, with some at risk of becoming endangered. These unique genetic resources are utilised in the communal and smallholder systems, and generally disregarded in the commercial sector. The small population sizes could possibly increase the likelihood of bottleneck events and thus the loss of genetic variation. This could lead to an increased risk of adaptive traits as well as increased inbreeding levels. The aim of this study was to investigate genetic diversity parameters and marker- and runs of homozygosity-based inbreeding levels of SA indigenous sheep breeds. Single nucleotide polymorphism (SNP) genotypes were available for 158 animals representing five indigenous SA sheep populations namely the Black Headed Persian (BHP), Damara (DAM), Namakwa Afrikaner (NAM; endangered), Pedi (PED; endangered) and non-descript Fat-tailed sheep (FTT). The available SNP genotypes, generated using the Illumina Ovine 50K SNP BeadChip, were subjected to standard call rate (0.05), MAF (0.02) and Hardy-Weinberg Equilibrium ($p < 0.001$) quality control procedures and a subset of 48,753 autosomal SNPs were retained for diversity analysis. The study showed moderate levels of heterozygosity, with HO (HE) values ranging from 0.326 (0.335) for FTT sheep to 0.356 (0.374) for PED. The number of ROH ranged from 30 for DAM and 93 for PED and the majority of ROH segments were short in length (1 to 3.99 Mb). Inbreeding coefficients were all positive but low, with FROH ranging from 0.002 (DAM and NAM) to 0.007 (FTT). Lastly, the predicted effective population sizes (N_e) were extremely small for all populations; ranging from $N_e=35$ (BHP) to $N_e=90$ (PED). The moderate heterozygosity levels, relatively large within- and between breed variation, together with the low levels of overall inbreeding, indicate that despite the small population sizes, sufficient genetic variation exists to allow effective conservation and sustainable management of these populations.

Keywords: Conservation, diversity, inbreeding, indigenous