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Mutational load and deleterious mutations in goat genome from Kenya

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Abstract

Different domestication processes such as selection and effective population size are among other factors responsible for the accumulation of mutations in an animal. This study investigated the mutation load and accumulation of deleterious mutations in Kenyan goat populations using Single Nucleotide Polymorphism (SNP) data obtained from local (Galla = 12) and exotic (Saanen = 24, Alpine = 28, and Toggenburg = 30) goat genotypes. The SNP annotation was done on the ENSEMBLE goat (*Capra hircus*) using the Variant Effect Predictor (VEP). Sorting Intolerant from Tolerant (SIFT) was done on the annotated file using a score of > 0.01 to describe highly deleterious mutations. Biomart tool in the ENSEMBL goat *C. hircus* was used for Gene Ontology (GO) for the genes identified from highly deleterious mutations. The analysis disclosed no differences in mutation load among the studied genotypes. Overall, the synonymous mutations were in abundance compared to missense mutations, totaling 693 and 258 representing 1.01% and 0.37%, respectively. The calculated mutation load was similar (0.37) suggesting exposure of goats to similar domestication processes that are creating similar effects on the animal's genome. Further analysis of the missense variants revealed 126 deleterious mutations and 132 tolerated mutations. A total of 111 genes were identified from the deleterious mutations and only 5 were among the highly deleterious mutations (SIFT score > 0.01) which include PROS1, EHBP1, LTN1, LRRN4, and FNDC3A. Gene Ontology describes these genes as responsible for different functions associated with animal reproduction, diseases, and memory. This study's results have confirmed the levels of mutation load and the presence of deleterious mutations in Kenyan goats. This can be used as a base to predict the future rate of mutation load to ensure better implementation of conservation programs to avoid an accumulation of deleterious mutations.

Keywords: Annotation, deleterious mutations, goats, genes, mutation load