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Genome-wide homozygosity in Maremmana Cattle

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The current availability of large numbers of single nucleotide polymorphisms (SNPs) throughout the genome makes these markers particularly suitable for the detection of patterns of genetic diversity and of genome-wide homozygosity in animal populations. The aim of this work was to estimate genetic diversity and homozygosity in the Maremmana cattle breed. We used a sample of 149 animals (males and females) genotyped with the BovineSNP50 v2 (54K) Illumina BeadChip. After editing for call-rate >0.9 and removing SNP unassigned or on the sex chromosomes, 128 animals and 50,814 SNPs were left. We estimated the following genetic parameters: observed and expected heterozygosity (Ho and He), minor allele frequency (MAF), and the FIS statistic. We also scanned the genome for runs of homozygosity (ROH). In the present study, ROH were detected based on 20-SNP-long sliding-windows, and allowing for a maximum of 1 missing and 1 heterozygote genotype, and a maximum gap between consecutive SNP of 105 bp. ROH contained minimum 10 SNPs, and had a minimum length of 1 Mb and a minimum density of 1 SNP every 50 kbps. The average Ho and He were 0.374 ± 0.132 and 0.365 ± 0.120, respectively, and the average MAF was 0.274 ± 0.130. These values are consistent with the range observed in other cattle breeds. We obtained some negative values for FIS (-0.162 to 0.180) which corresponded to animals with lower than average homozygosity. In total, 10,465 ROH were detected (81.75 per animal), with an average length of 2.69 Mb. Most ROH (74%) had length 2 Mb. ROH are contiguous lengths of homozygous genomic segments where the two inherited haplotypes are identical. ROH indicate genomic regions where a reduction in heterozygosity occurred, and offer new opportunities to estimate inbreeding (F). The inbreeding coefficient based on ROH (FROH) was estimated by the ratio between the total ROH length and the size of the genome in each animal. Average FROH was 0.0869 ± 0.032. Unlike inbreeding estimated based on Ho, FROH is not influenced by allele frequencies (sampling) and can distinguish recent from ancient inbreeding. However,

FROH requires SNP positions to be known (unlike Ho). Two genomic regions with ROH in over 60% of the animals were found: one on BTA6 (38.6-39.7 Mbps), one on BTA13 (54.3-54.8 Mbps). These may highlight regions where selective pressures have shaped the genome of the Maremmana breed.

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