



Genome-wide scan for Runs of Homozygosity in Valle del Belice sheep

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Abstract:	The current availability of very large numbers of single nucleotide polymorphisms (SNPs) throughout the genome makes these markers particularly suitable for the detection of genomic regions where a reduction in heterozygosity occurred and offers new opportunities to improve the accuracy of inbreeding (F) estimates. Runs of homozygosity (ROH) are contiguous lengths of homozygous segments of the genome where the two haplotypes inherited from the parents are identical. Here, we investigated the occurrence and the distribution of ROH in medium-density SNP genotypes (~ 50 000) in order to characterize autozygosity in 512

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4 individuals of Valle del Belice sheep and identify the regions of the genome
5 with high ROH frequencies. A total of 11 629 ROH were identified. All
6 individuals displayed at least one ROH > 1 Mb. The mean value of
7 FROH>1Mb was 0.084±0.061. ROH that were shorter than 10 Mb
8 predominated. The highest coverage of chromosome (OAR) by ROH was
9 observed on OAR24, whereas the lowest one was observed on OAR1. A
10 typical pattern was observed for the number of ROH per OAR with higher
11 values in the first three chromosomes. There was a considerable difference
12 among animals for the number of ROH segments and the length of the
13 genome covered by ROH. The genomic regions most commonly associated
14 with ROH were identified by selecting the top 1% of the SNPs most
15 commonly observed in ROH within breed. A total of 239 SNPs were
16 considered as candidate SNPs and we identified 107 potential candidate
17 genes that may be under directional selection. Six genomic regions located
18 on six chromosomes (OAR2, OAR3, OAR4, OAR10, OAR11 and OAR23),
19 corresponding to ROH island, presented hotspot of autozygosity. According
20 to KEGG database, a majority of the genes were involved in multiple
21 signaling and signal transduction pathways in a wide variety of cellular and
22 biochemical processes. The ROH islands spanned several candidate genes
23 which influence traits that are associated with adaptability and with the
24 regulation of immune responses (NPAS2, PDCL3, SERPINF1 and SERPINF2)
25 and we did not identified candidate genes with important influence on milk
26 production traits in sheep. The Valle del Belice breed is subjected to limited
27 breeding selection programs for milk production traits, but shows excellent
28 adaptability to the local environments. Therefore, these results suggest at
29 least a partial role of natural selection in shaping the genome of Valle del
30 Belice sheep breed.
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Genome-wide scan for Runs of Homozygosity in Valle del Belice sheep

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The current availability of very large numbers of single nucleotide polymorphisms (SNPs) throughout the genome makes these markers particularly suitable for the detection of genomic regions where a reduction in heterozygosity occurred and offers new opportunities to improve the accuracy of inbreeding (F) estimates. Runs of homozygosity (ROH) are contiguous lengths of homozygous segments of the genome where the two haplotypes inherited from the parents are identical. Here, we investigated the occurrence and the distribution of ROH in medium-density SNP genotypes (~ 50 000) in order to characterize autozygosity in 512 individuals of Valle del Belice sheep and identify the regions of the genome with high ROH frequencies. A total of 11 629 ROH were identified. All individuals displayed at least one ROH > 1 Mb. The mean value of FROH>1Mb was 0.084±0.061. ROH that were shorter than 10 Mb predominated. The highest coverage of chromosome (OAR) by ROH was observed on OAR24, whereas the lowest one was observed on OAR1. A typical pattern was observed for the number of ROH per OAR with higher values in the first three chromosomes. There was a considerable difference among animals for the number of ROH segments and the length of the genome covered by ROH. **The genomic regions most commonly associated with ROH were identified by selecting the top 1% of the SNPs most commonly observed in ROH within breed.** A total of 239 SNPs were considered as candidate SNPs and we identified 107 potential candidate genes that may be under directional selection. Six genomic regions located on six chromosomes (OAR2, OAR3, OAR4, OAR10, OAR11 and OAR23), corresponding to ROH island, presented hotspot of autozygosity. According to KEGG database, a majority of the genes were involved in multiple signaling and signal transduction pathways in a wide variety of cellular and biochemical processes. The ROH islands spanned several candidate genes which influence traits that are associated with adaptability and with the regulation of immune responses (*NPAS2*, *PDCL3*, *SERPINF1* and *SERPINF2*) and we did not identified candidate genes with important influence on milk production traits in sheep. The Valle del Belice breed is subjected to limited breeding selection programs for milk production traits, but shows excellent adaptability to the local environments. Therefore, these results suggest at least a partial role of natural selection in shaping the genome of Valle del Belice sheep breed.