

analysis, to shed light on the molecular pathways or mechanisms underlining the variability of the trait. Firstly, we inspected the position of each SNP via UCSC Genome Browser and we determined if it was located within or close to a gene or another functional DNA element. Then, we examined the role of the element in any molecular pathway via databases such as Gene Ontology and KEGG. Finally, we evaluated how each specific pathway or molecular element could shape the phenotype of interest.

We detected some significant associations between SNPs and key fitness and productive trait. As an example, we found that SNPs situated within the MCM9 gene was linked to udder physiology. Polymorphisms in this gene have been found to be linked to estrous behaviour in cattle and several ovarian pathologies in humans. Our results underscore the power of GWAS and give insight into the genetic architecture of key traits in local breeds.

O119

Genome-wide search of distinctiveness selection signatures in Maremmana cattle compared to Italian Podolian-derived breeds

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Several Italian breeds such as Marchigiana, Romagnola, Chianina, Calvana, Podolica, Cinisara, Modicana and Maremmana belong to the so-called 'Podolian' group. The Maremmana is one of the most distinctive cattle breeds because it is raised in semi-feral conditions and it still displays many ancestral features such as horn shape and size, sexual dimorphism and longevity. The aim of this study was to map genomic regions that might explain the phenotypic distinctiveness observed in Maremmana cattle compared to the other Podolian-derived Italian breeds. Putative selection signatures in Maremmana were investigated using genotypes generated for 146 Maremmana and 174 individuals belonging to Podolian-derived Italian breeds using the Bovine SNP50K v2 BeadChip and combining three statistical approaches designed to quantify the excess of haplotype homozygosity either within (iHS) or among pairs of populations (Rsb and XP-EHH).

We used $-\log_{10}(p\text{-value}) = 2.5$ as a threshold to define significant iHS, Rsb and XP-EHH candidate regions. Multidimensional scaling and neighbor-joining tree revealed a clear separation between Maremmana and the other Podolian-derived breeds. Overall, the three haplotype-based analyses revealed selection signatures distributed over 19 genomic regions. Of these, six regions spanning 87 known genes, were identified by at least two approaches. Within these regions, we found genomic signatures of selective sweeps spanning genes related to muscle development, growth and meat traits (SCIN, THSD7A, ETV1, UCHL1 and MYOD1) which reflects the different breeding schemes between Maremmana and the other Podolian-derived breeds. We also identified several genes possibly linked to Maremmana adaptation to the environment of the western central part of Italy, known to be hyperendemic for malaria and other tick-borne diseases. These include several chemokine (C-C motif) ligand genes crucially involved in both innate and adaptive immune responses to intracellular parasite infections and other genes playing key roles in pulmonary disease (HEATR9, MMP28, ASIC2) or strongly associated with malaria resistance/susceptibility (AP2B1). Information about the location of these regions can be used to detect variants that may underlie important traits of practical interest for the beef industry as well as to enhance our understanding of the genomic basis of the millenarian environmental adaptation of Maremmana to the semi-wild conditions of its rearing cradle.

O120

Whole-genome resequencing reveals specific genomic variants in Italian insular sheep breeds

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The development of high-throughput sequencing technologies revolutionized the way genetic variation studies are assessed. In livestock, whole-genome sequencing approaches with pooled DNA have recently developed into a powerful tool to identify genetic divergences among breeds. In this study, we applied pooled whole-genome sequencing to reveal differentiation and specific genomic variants in five Italian insular sheep breeds: Barbaresca (BAR), Comisana (COM), Pinzirita (PIN), Sarda (SAR) and Valle del Belice (VDB). Each pool consisted of 30 animals. These breeds