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Genomic insights on the history and selection trajectories of the local goat breed Comune di Sicilia

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The "Capra Comune di Sicilia" (CCS), also known as “Mascaruna” due to its typical black "badgerface" markings, is a Sicilian local goat population described since 1870 and currently subjected to a recovery project for its characterization and ethnic recognition. This study details the ancestral genetic components and selection trajectories of CCS. We analysed the genotypes of 78 CCS goats using the Goat 60k SNP BeadChip along with data from 1,920 individuals of 66 goat breeds of Mediterranean and African origin. The CCS recorded relatively high levels of heterozygosity (0.408) and moderate inbreeding (0.04), with an effective population size ranging from 96 to 185. Genetic ancestry analysis identified gene flow signals with Maltese, Girgentana, Rossa Mediterranea and Saanen populations, along with a putative Greek ancestry shared with most of the Mediterranean breeds in our dataset, reflecting Sicily’s deep historical and cultural links with Greece. In order to understand the evolutionary trajectories of this population and exploit the potential ancestry sourcing from Greek populations, we investigated selection signatures within CCS through iHS and ROH analysis, identifying 76 and 31 SNPs falling within 38 and 12 genes, respectively, under putative selection. Then, we applied XP-nSL and ROH using Greek populations as ancestors, and identified 21 and 431 SNPs, associated with four and 157 genes, respectively, under putative selection. Overall, all selection signature analyses highlighted genes under positive selection linked to milk (*MGFGE8, ANPEP, POLG, AXIN2*) and meat (*MRPL46*) production, body size and growth (*FAM184B, LCORL*), fertility (*PFOXic, FANCA, NTRK3, CPEB1, BLM, SPP1, MGFGE8*), coat colour (*AP3B2*), fat deposition (*KLF5, COPS8*), and ear and horn development *(PFOXic, ORC6, LCORL, MEPE, FAM184B, ISBP*). Our results shed light on the history of CCS, emphasizing its genetic uniqueness and providing essential information about its background, which is crucial for informed efforts to recognize it as a distinct and valuable breed.
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