## Identification of ROH and HRR islands across the genome of European sheep breeds

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## Abstract

Runs of Homozygosity (ROH) are continuous homozygous segments in an individual's genome, typically resulting from inbreeding or population bottlenecks (Ceballos et al., 2018). ROH can be categorized by length, with long ROH indicating recent inbreeding and shorter ROH reflecting historical demographic events. Within ROH, certain genomic regions may show recurrent homozygosity across multiple individuals, forming ROH-enriched islands (Bosse et al., 2014). That may arise from selection, genetic drift or evolutionary processes, aiding studies on local adaptation and deleterious mutation accumulation. Heterozygosity Rich Regions (HRR) are genomic regions with consistently high heterozygosity across a population, indicating loci under balancing selection, recombination hotspots or introgressed regions from hybridization events. Studies on ROH and HRR islands provide insights into demographic history, inbreeding, and selection (Szpiech et al., 2019). In the present study, 11 sheep breeds originating from northern (Babolna Tetra), central (Rouge du Rusillon, Hortobagy Racka), southern Europe (including Greek native populations) (Turcana, Rusty Tsigai, Pelagonia, Frizarta, Boutsiko, Mytlini, Ojalada) and South America (Creole) were analyzed to explore ROH and HRR patterns from different geographic clusters. Medium-density (Ovine SNP50) genotypes from SMARTER database (Cozzi et al., 2024) were analyzed using the detectRUNS R package with a "sliding-window" approach for ROH and "consecutive runs" for HRR. Total ROH/HRR count and length were estimated for each breed and SNPs with >1% occurrence in runs were selected to define ROH and HRR islands. Creole breed from Latin America showed the highest ROH number and length compared to the European breeds, likely due to intense historical selective pressures, while also sharing genomic components with European breeds as a descendant of those introduced by Spanish and Portuguese explorers during the colonial period (Revelo et al., 2022). On the other hand, Romanian Turcana and Rusty Tsigai sheep had the lowest ROH number and length, reflecting high genetic diversity. Similar HRR patterns were observed across the sheep breeds. Common ROH islands were detected on chromosomes 2, 3, 8, 13 and 22, while common HRR islands were found on chromosomes 2, 3, 7 and 9, indicating potential functional importance of chromosomes 2 and 3. Greek Mytilini sheep demonstrated the highest number of

ROH islands possibly linked to its geographical isolation. Hungarian Hortobagy Racka and Romanian Rusty Tsigai sheep had one ROH island, suggesting higher gene flow and crossbreeding events, while they share HRR hotspots with the Greek Pelagonia and Frizarta sheep. The Spanish breed Ojalada shared the highest number of ROH islands with other breeds, in consistence with domestic sheep migration from eastern Europe to Spain during the Roman period (Ciani et al., 2020). Ongoing identification and functional annotation of the genes on ROH and HRR islands aim to provide further insights into the relationship among the studied sheep breeds.

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