SELECTION SIGNATURE ANALYSES REVEALED MULTIPLE GENOMIC REGIONS ASSOCIATED WITH MILK PRODUCTION TRAITS IN THE GIRGENTANA BREED

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Introduction

Both natural and artificial selection are among the main forces shaping genetic variation across the genome of livestock species.

The high-throughput techniques have facilitated the detection of selection signatures.

The Girgentana is an ancient goat breed with distinctive morphological, adaptive, and productive traits. Its long breeding history makes it an excellent model for detecting footprints of selection, which would help identify genes related to biological processes and traits of interest.
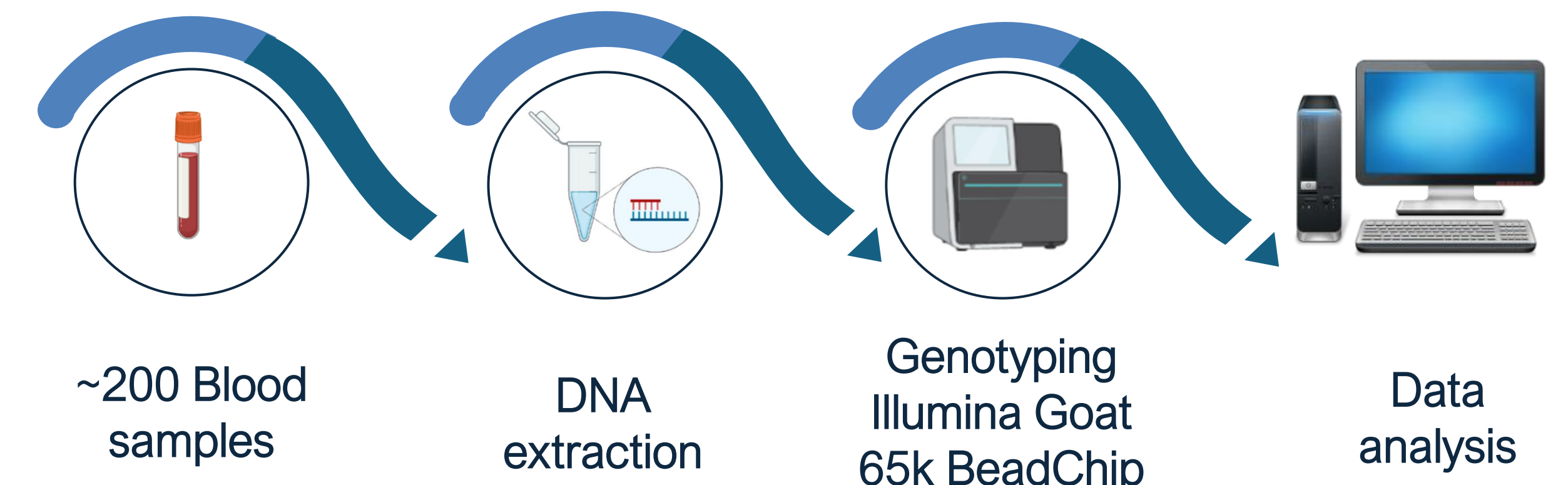

Figure 1. Specimens of Girgentana goats.

Aim

In this study, we investigated genomic regions under selection in the Girgentana goat breed using two Extended haplotype homozygosity (EHH) tests.

Methods

~200 Blood samples   DNA extraction   Genotyping Illumina Goat 65k BeadChip   Data analysis

The raw data were merged with the genotypes of other 59 Girgentana samples and those of 13 Italian breeds (5 from Southern Italy and 8 from Northern Italy).

The final dataset consisted of 536 individuals, including 210 Girgentana animals, genotyped for 48,744 SNPs distributed over the 29 autosomal chromosomes.

We used two EHH-based metrics implemented in the *rehh* package in R (Gautier et al., 2012): the within-population iHS (Voight et al., 2006) and the between-population Rsb test (Tang et al., 2007) where the EHH profile of Girgentana was compared to EHH profiles of Northern and Southern Italian goats.

Results

A total of 14 genomic regions were detected: three within the Girgentana (iHS) and eleven from the comparisons Girgentana vs Northern and Girgentana vs Southern breeds (Rsb).

The largest overlap between the two approaches was on chromosomes 1 (110 - 112 Mb) and 5 (38 – 40 Mb).

Among the several selection sweeps, it is worth noting the region on chromosome 6, which harbours casein genes (*CSN2*, *CSN1S1*, *CSN3*) known to be involved in milk quality.

Other selection signatures related to genes associated with body size, reproduction, and immune resistance have been identified.

Conclusion

Our results suggest that both natural and artificial selection shaped the genome of Girgentana.

This study provides a foundation for detecting mutations that underlie genetic variation of economically important traits for the Girgentana breed.

Maintaining these traits is fundamental for preserving this reservoir of genetic diversity and maximizing the adapting ability to the changing environment of this traditional livestock system.

Figure 2. Manhattan plots of the: a) genome-wide integrated haplotype score (iHS) analysis for Girgentana breed; Rsb tests in the comparison between b) Girgentana *vs* Northern Italy and c) Girgentana *vs* Southern Italy. Horizontal dashed lines mark the significance threshold applied to detect the outlier SNPs - $\log_{10}$ (p-value) = 3.