







120

# Genomic patterns of homozygosity around casein gene cluster in Italian Holstein cattle

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## INTRODUCTION

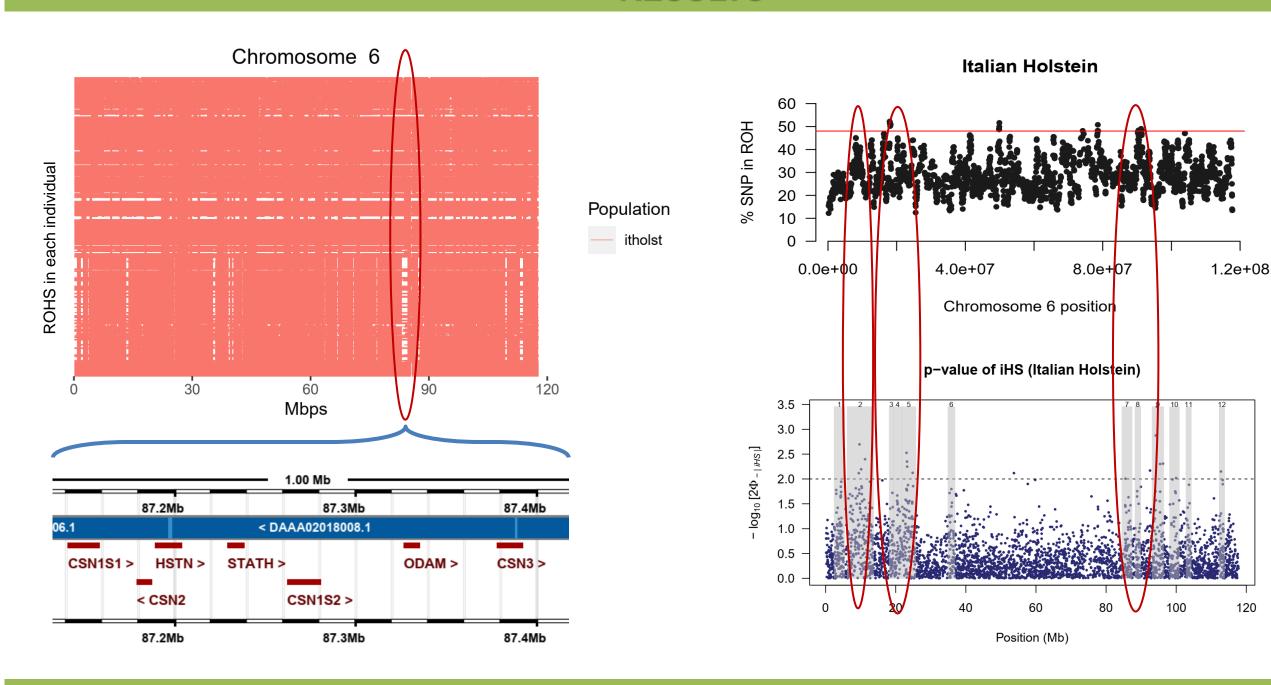
Most of the milk produced in Italy is processed into various types of cheese. Therefore, the insight into the milk processing properties is highly important. Caseins are milk proteins that are considered the most important in the context of cheese production. 54 SNP polymorphisms have been detected in the casein gene cluster on chromosome 6 in Italian Holsteins (Chessa et. al 2021).

The aim of this study was to analyze Italian Holstein population using two approaches, Runs of Homozygosity (ROH) and Extended Haplotype Homozygosity (EHH) in order to provide insights into the past and ongoing selection events around casein cluster on Chromosome 6.

### MATERIAL AND METHODS

In this study, 160 000 Italian Holstein cows routinely genotyped with 50k SNP arrays were used in the analyses. Only autosomal SNPs with known positions were analysed. SNPs where more than 5% of genotypes were missing were excluded from the analysis while animals for which > 10% of the genotype was missing were also excluded. Thresholds to filter out all SNPs with minor allele frequency was <1% and due to departures from Hardy-Weinberg equilibrium (HWE) <0.01%, respectively. After QC, 2170 markers and 71079 animals remained for further analysis. ROH analysis was performed by detectRUNS using the consecutive method with parameters as recommended by Ferencakovic et al (2013). Analyses of iHS were calculated in R software using rehh package (Gautier et al. 2017) and if 3 or more SNPs with extreme values in a defined window exceeded the significance threshold of  $-\log 10$  p value = 4, it was marked as a selection signature.

#### **RESULTS**



### CONCLUSION

The results showed that a certain number of animals have high levels of autozygosity on chromosome 6 and clear separation of population in that regard, which is also confirmed for homozygosity at the haplotype level based on high iHS scores. These analyses confirm the effects of selection on certain casein gene variants, although they were not included in the selection scheme. Since the casein gene cluster is important for the Italian Holstein population, the results of the selection signature analysis should be deeply evaluated and hopefully contribute to the selection plans.