



NOVEL RUNS OF HOMOZYGOSITY ISLANDS IN THE FINNISH AYRSHIRE POPULATION

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INTRODUCTION



FINNISH AYRSHIRE (FAY)

- One of the Nordic Red dairy breeds (RDC) (NAV 2023B)
- Many good qualities, e.g., good fertility and high milk fat (Faba 2023, ProAgria 2023)
- 40 % of the total number of cows in national milk recording are FAY (ProAgria 2023)
- Breeding programme based on genomic selection (GS)
 - Breeding values estimated jointly for RDC in Finland, Sweden, and Denmark (NAV 2023B)
 - Selection based on the Nordic Total Merit Index (NTM)



Figure 1. Finnish Ayrshire. (I. Hennola)



RUNS OF HOMOZYGOSITY ISLANDS

- Runs of homozygosity emerge from inbreeding and selection (Broman & Weber 1999, Forutan et al. 2018)
 - Split by recombination (Howrigan et al. 2011)
 - ROHs due to inbreeding are randomly distributed along the genome (Zhang et al. 2015)
- Runs of homozygosity islands (ROH-islands)
 - Increased frequency of ROHs at a certain genomic site (Nothnagel et al. 2010)
 - Selection signatures (Purfield et al. 2017, Gorssen et al. 2019)
 - Expected near quantitative trait loci (QTL) (Sonesson et al. 2010)
- Selection in one population compared to the other: difference in ROH-patterns between populations (Δ ROH-islands) (Cortellari et al. 2021)



AIM

The aim of the present study was to identify differences in ROH-island patterns, Δ ROH-islands, in the Finnish Ayrshire, that have emerged after genomic selection was introduced.

In addition, we aimed to identify quantitative trait loci and genes within the selected regions affecting traits relevant in the breeding programme of the FAY.



MATERIAL & METHODS



MATERIAL

- Genotypes of 53,469 FAY cows from Nordic Cattle Genetic Evaluation (NAV)
 - 45,834 autosomal SNPs (positions updated to ARS-UCD1.2 assembly)
- Pedigree data including birth years from the Finnish Animal Breeding Association (Faba)
 - Animals born in Finland and registered as RDC were considered FAY
- Assumed transition from traditional BLUP-based selection to GS between years 2012–2014 → animals divided into two groups:
 - Those born between years 1980–2011 (6,108 cows) and selected with traditional EBV
 - Those born between years 2015–2020 (47,361 cows) selected with GEBVs



DETECTION OF Δ ROH-ISLANDS

- ROHs identified using PLINK version 1.90b6.20 (Chang et al. 2015, Purcell & Chang 2021)
 - Parameters to detect ROHs set following Meyermans et al. (2020)
- Δ ROH-islands identified from ROH distribution
 - Difference in the occurrence of each SNP in a ROH (H-score) between the two groups ($\rightarrow \Delta$ H-score) (Cortellari et al. 2021)
 - The top 1 % of SNPs with the largest Δ H-scores were considered significant
 - Significant SNPs separated by a maximum of two non-significant SNPs formed a Δ ROH-island



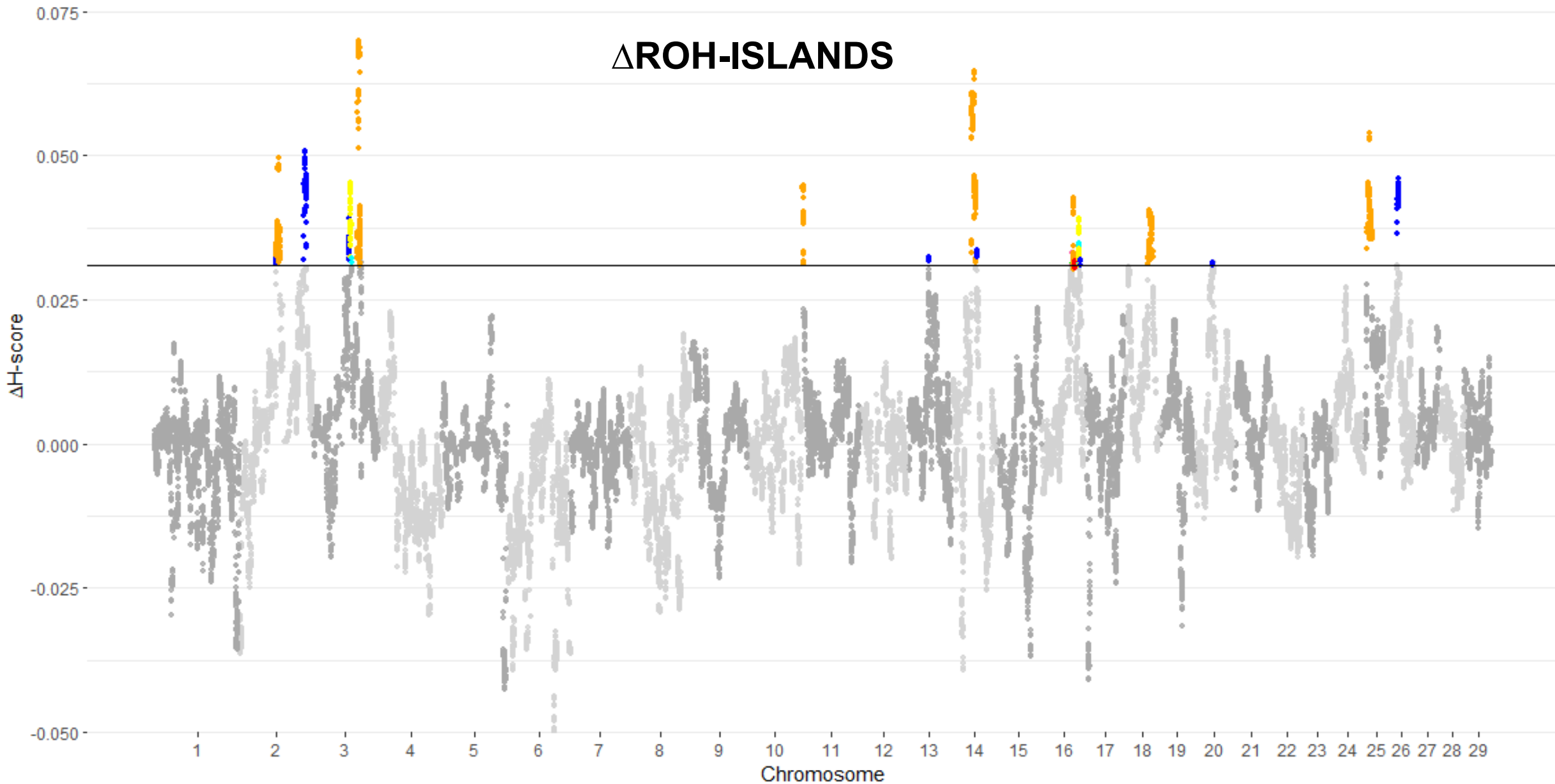
QTL AND GENES WITHIN Δ ROH-ISLANDS

- QTL within the identified Δ ROH-islands
 - CattleQTLdb version 14 release 47 (2022-04-25) (Hu et al. 2013)
 - Limited to QTL identified in Nordic Red breeds
- Genes within the identified Δ ROH-islands
 - UCSC Table browser (ARS-UCD1.2/bosTau9 assembly, April 2018)

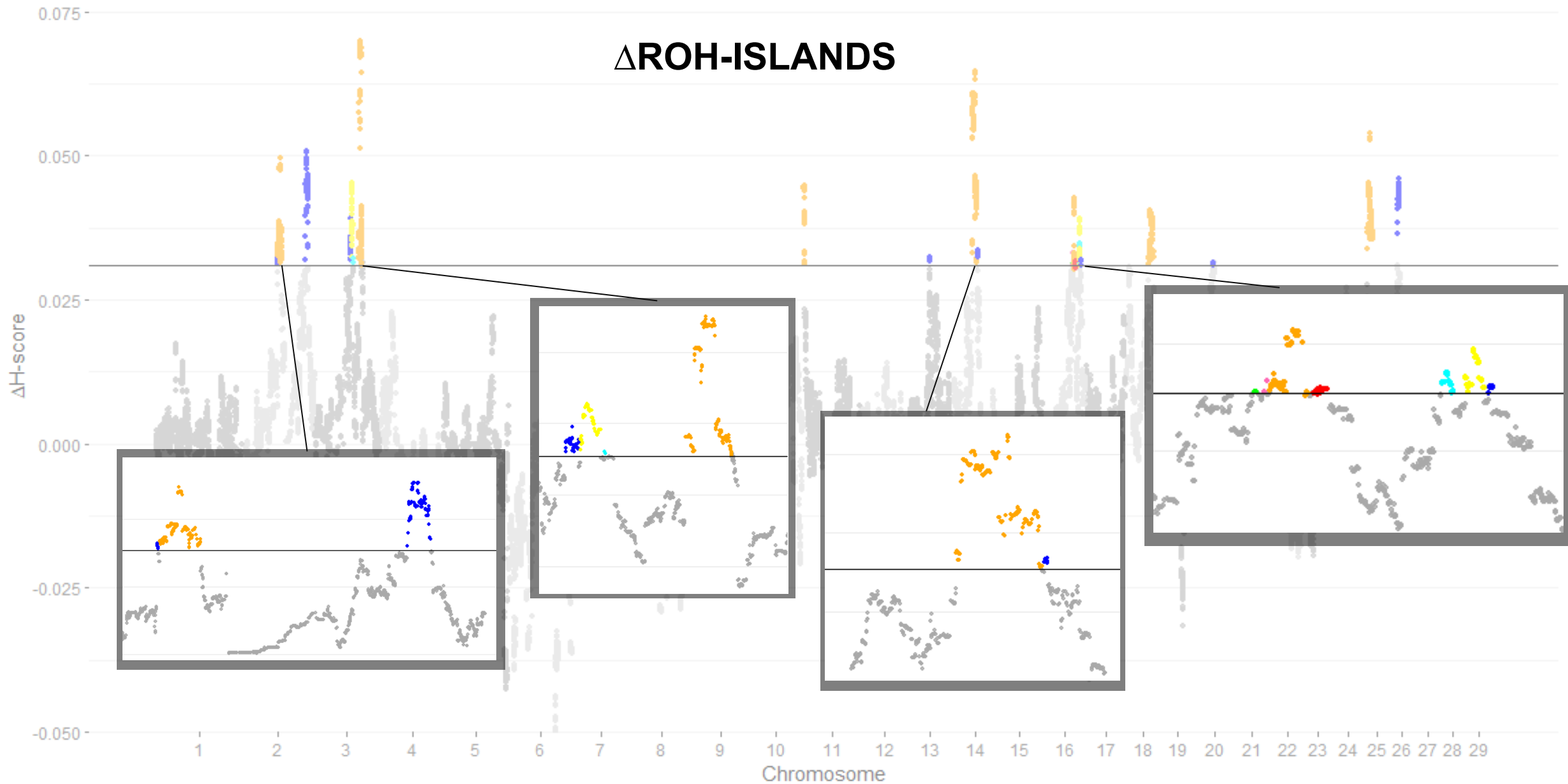


RESULTS & DISCUSSION

Δ ROH-ISLANDS



Δ ROH-ISLANDS





Δ ROH-ISLAND DEFINITION

- ROH-patterns vary between breeds (Szmatoła et al. 2016, Mastrangelo et al. 2018)
 - Depend on e.g., parameters set to detect ROHs
- No consensus in definition of ROH- or Δ ROH-islands between previous studies
 - From top 0.1 % (e.g., Gorssen et al. 2021) to top 1 % (Szmatoła et al. 2019) of SNPs generally form the ROH-islands



QTL WITHIN Δ ROH-ISLANDS

- Several Δ ROH-islands overlapped QTL regions related to
 - Stature, interval from first estrus to calving, fertility index, and milk fat yield (Höglund et al. 2014, Iso-Touru et al. 2016A, Bouwman et al. 2018, Fang et al. 2018)
- A short Δ ROH-island (46 Kbp) in chromosome 20 was located near several QTL regions related to stature and milk yield (Iso-Touru et al. 2016, Fang et al. 2018, Bouwman et al. 2018)
- Some overlapped or were near ROH-islands identified in other dairy breeds (Szmatoła et al. 2016, Mastrangelo et al. 2018, Szmatoła et al. 2019) yet did not overlap any QTL region identified in the RDC breed
 - May indicate genomic regions potential for selection or are under hitchhiking effect



GENES WITHIN Δ ROH-ISLANDS



- 424 genes within Δ ROH-islands identified
- Genes overlapped were related to e.g., milk composition and lactation:
 - *ATF3, MECR, CHUK, COX15, SOAT1*
 - Milk yield and milk composition in Holstein (Han et al. 2017)
 - Cellular metabolism in mammary gland in lactating cows (Invernizzi et al. 2012)
 - Milk fatty acid traits in Holstein (Ibeagha-Awemu et al. 2016, Palombo et al. 2018, Shi et al. 2019)
 - Milk cholesterol content in Holstein (Do et al. 2020)



GENES WITHIN Δ ROH-ISLANDS



- ...fertility, health, and metabolic and feed efficiency:
 - *RICTOR*, *OSMR*, *SEMA4G*
 - *RICTOR* is a regulator for highly expressed genes in fertile cows (Matsuyama et al. 2022)
 - Mammary epithelium regeneration after clinical mastitis in Holstein (Tiezzi et al. 2015)
 - Metabolic efficiency in dairy cattle (Wærp et al. 2019)
 - Feed efficiency in beef cattle (Abo-Ismael et al. 2018)
 - Amount of dietary energy in non-lactating Holstein cows (Moisá et al. 2017)



GENES WITHIN Δ ROH-ISLANDS



- ...growth and beef production traits:
 - *SLC44A5*, *SOAT1*, *KCNK2*, *SHISA9*, *PROX1*, *HSPB6*
 - Birth weight in Holstein (Sugimoto et al. 2012)
 - Intramuscular fat content and meat and carcass traits in beef and dairy crossbreds and dual-purpose cattle (Komolka et al. 2016, Picard et al. 2023)
 - Average daily gain, meat quality, and maturity rate (growth) in beef cattle (Sorbolini et al. 2016, Wang et al. 2019, Duan et al. 2021)



CONCLUSIONS



CONCLUSIONS

Signatures of selection emerged during the introduction of genomic selection were identified in several chromosomes in the Finnish Ayrshire.

Homozygosity had increased due to selection at the QTL regions and genes related to traits included in the Nordic Total Merit Index (stature, udder health, fertility, milk yield and milk composition, metabolic and feed efficiency, growth, and beef production traits) after the introduction of genomic selection in the Finnish Ayrshire.

References



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