

Comparative approach of missing homozygosity and GWAS in Brown Swiss

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Introduction and Dataset

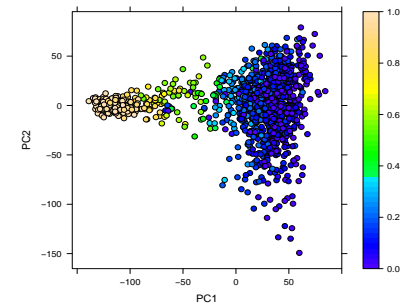
- 2 BSW populations

	Characterization	No. of genotyped animals (08/19)	No. of trios genotyped (12/18)	No. of paternal halfsib groups genotyped (12/18)
BS	Dairy focus (LO)	60,000	5'600	38,000
OB	Dual purpose (HI)	6,900	1'200	2,200

- No. of evaluated traits: 60 (+3)
 - 2-step genomic evaluation system

- Software:

- Imputation: FImpute (Sargolzei et al. , Seefried et al. EAAP 2019)
- Haplotype analysis: snp1101 (Sargolzei et al. 2014)

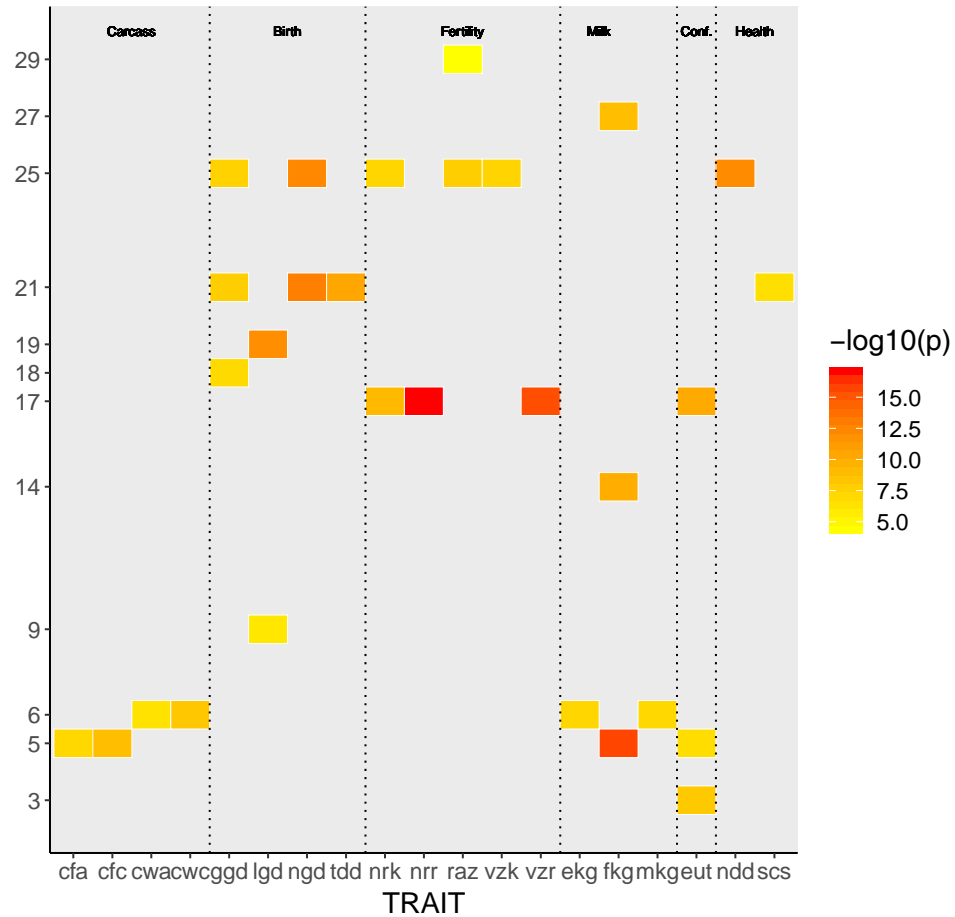


GWAS and phenotypic analyses

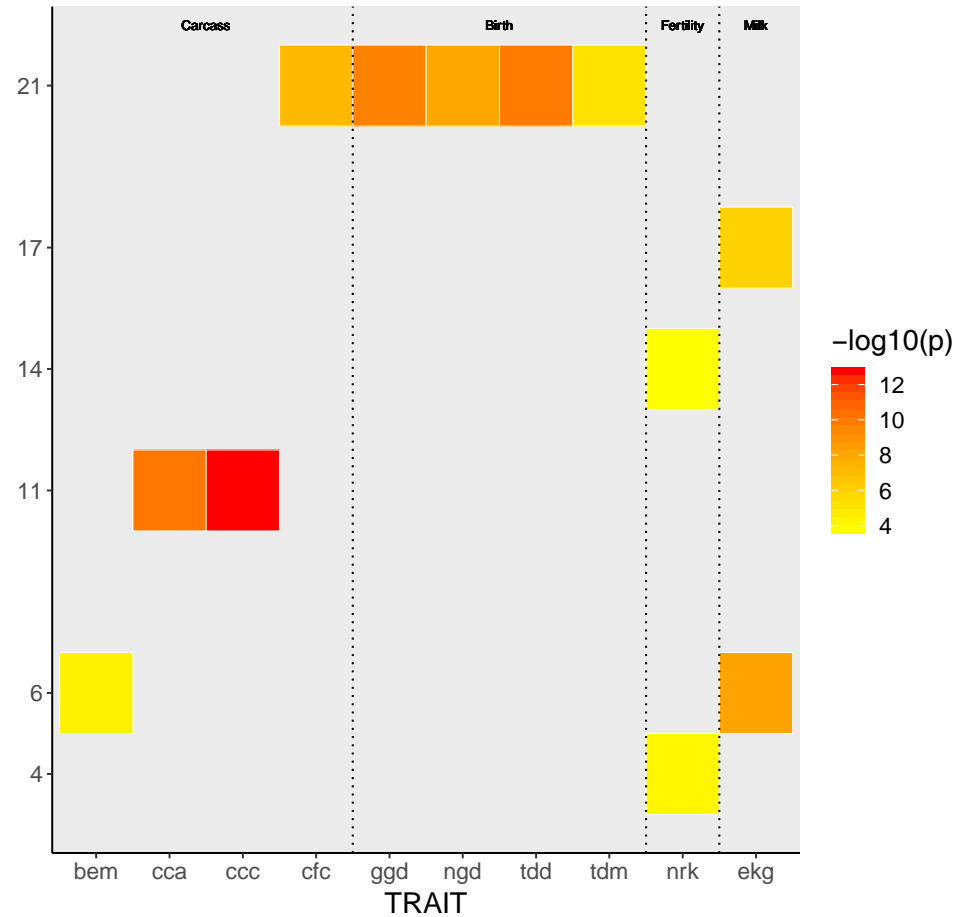
- Assembly ARS-UCD1.2
- GWAS:
 - Single SNP regression model
 - Stratification: G
 - SNP-Density: 110K
- Trait blocks related to missing homozygosity
 - Fertility
 - Birth
 - Carcass
 - Milk production traits (pleiotropy)

GWAS results

GWAS Heatmap BS



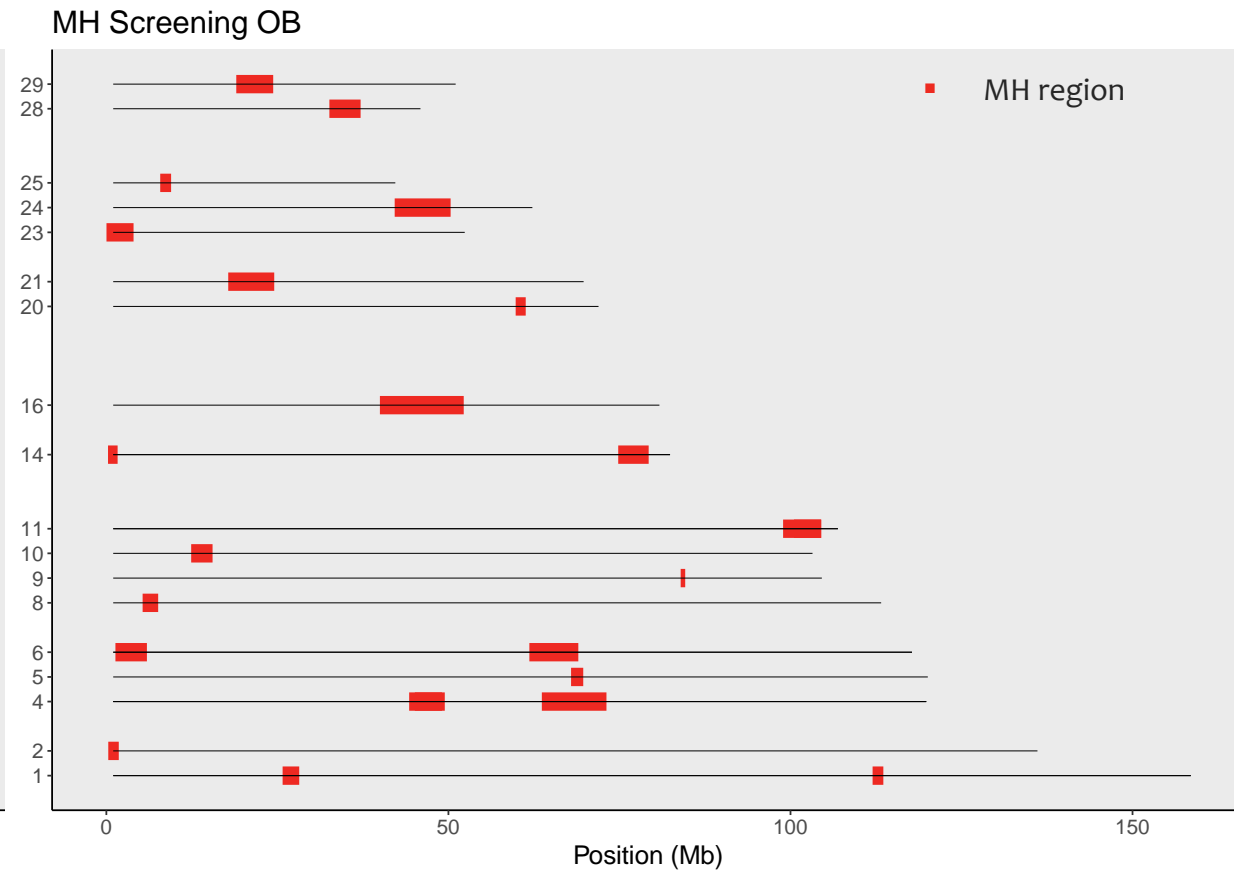
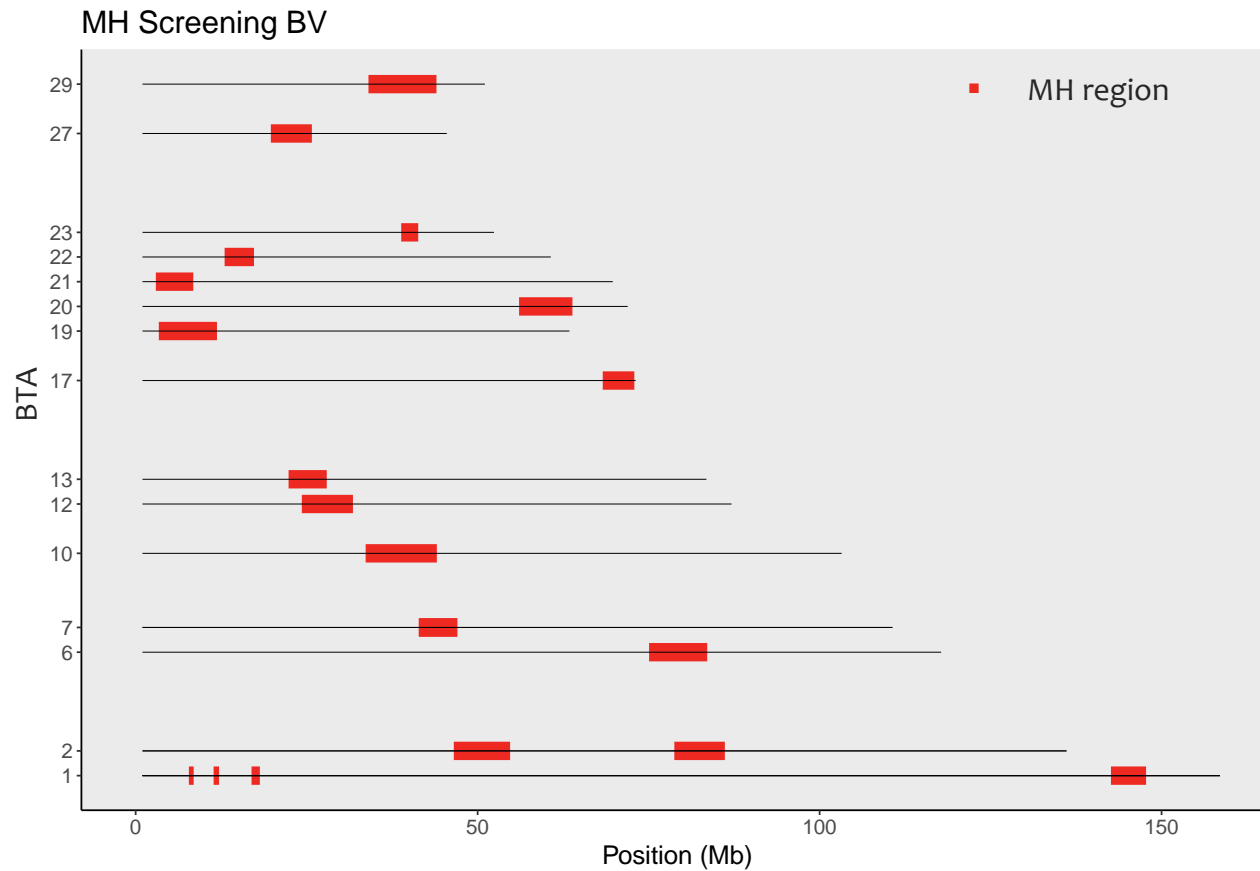
GWAS Heatmap OB



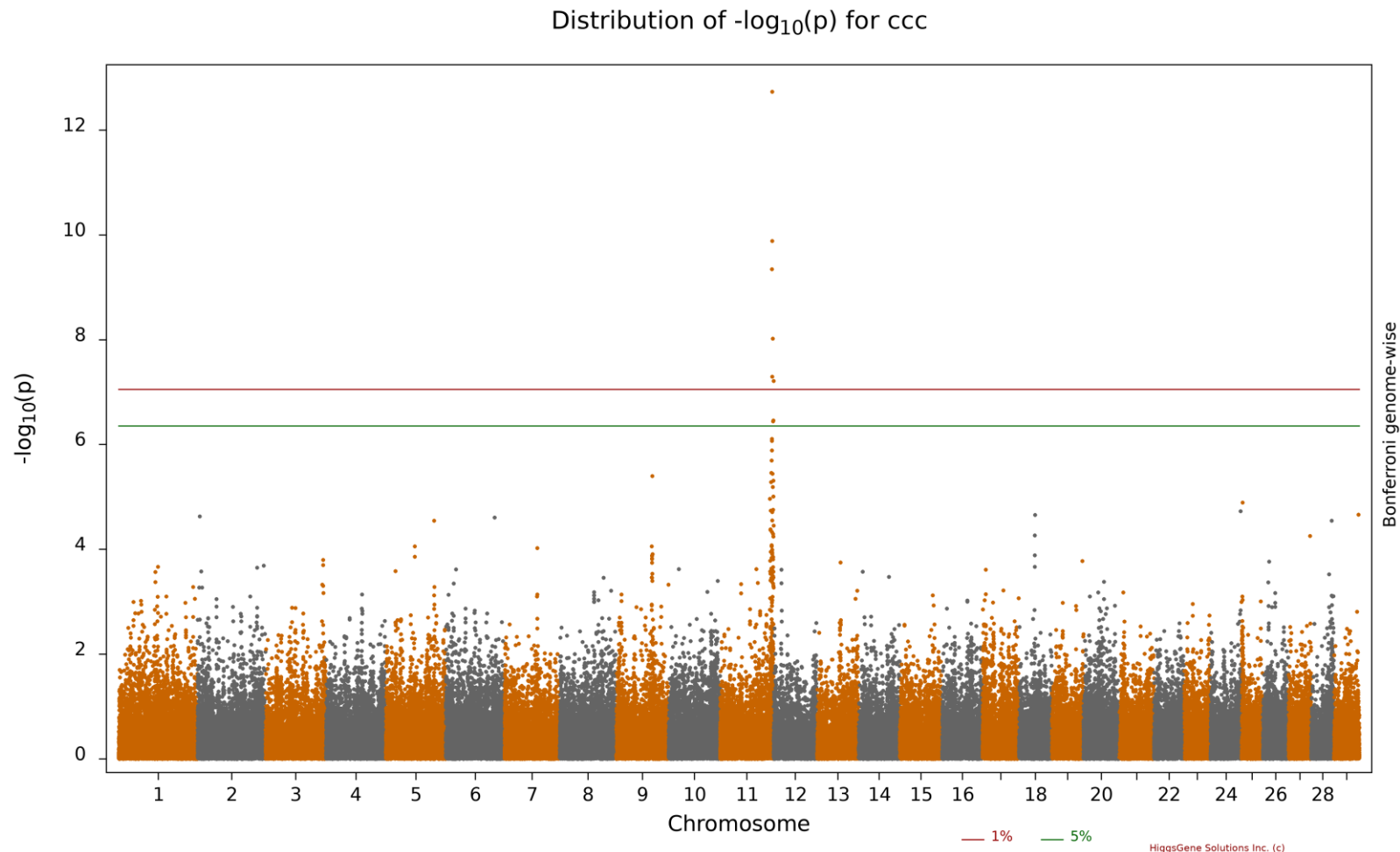
Screening for missing homozygosity (MH)

- Genome wide haplotype screening for reduced or missing homozygosity
- Sliding window approach: different window sizes (50 – 200 SNPs)
- Datasets: Paternal grand parent groups / Trios
- Validation:
 - De-regressed EBVs
 - Traits: fertility -, birth -, growth – and milk production traits
 - Haplotype selection: window's longest haplotype with significant MH
 - $y_{EBV} = \mu + G + \beta H + e$
 - GCTA software (Yang et al. 2011)

Haplotype Screening Summary

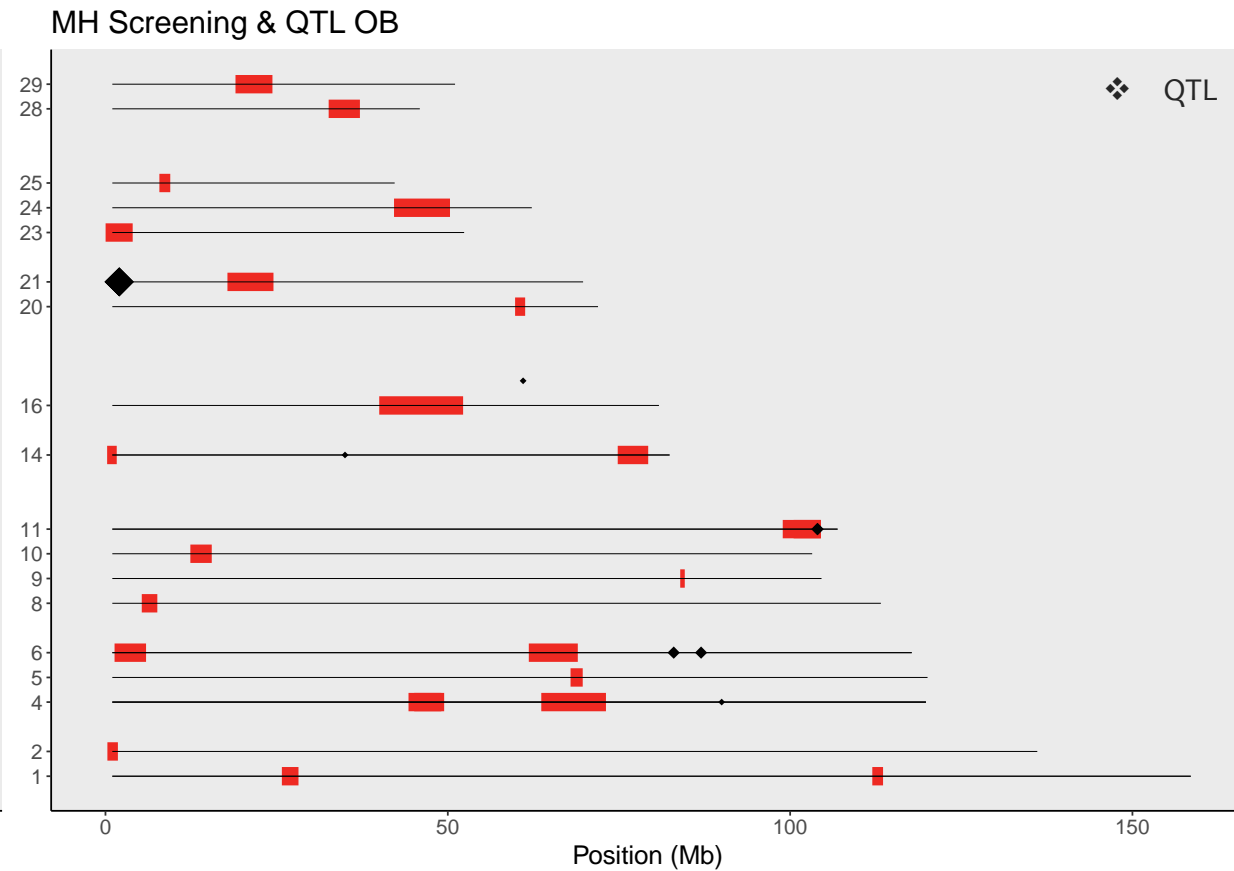
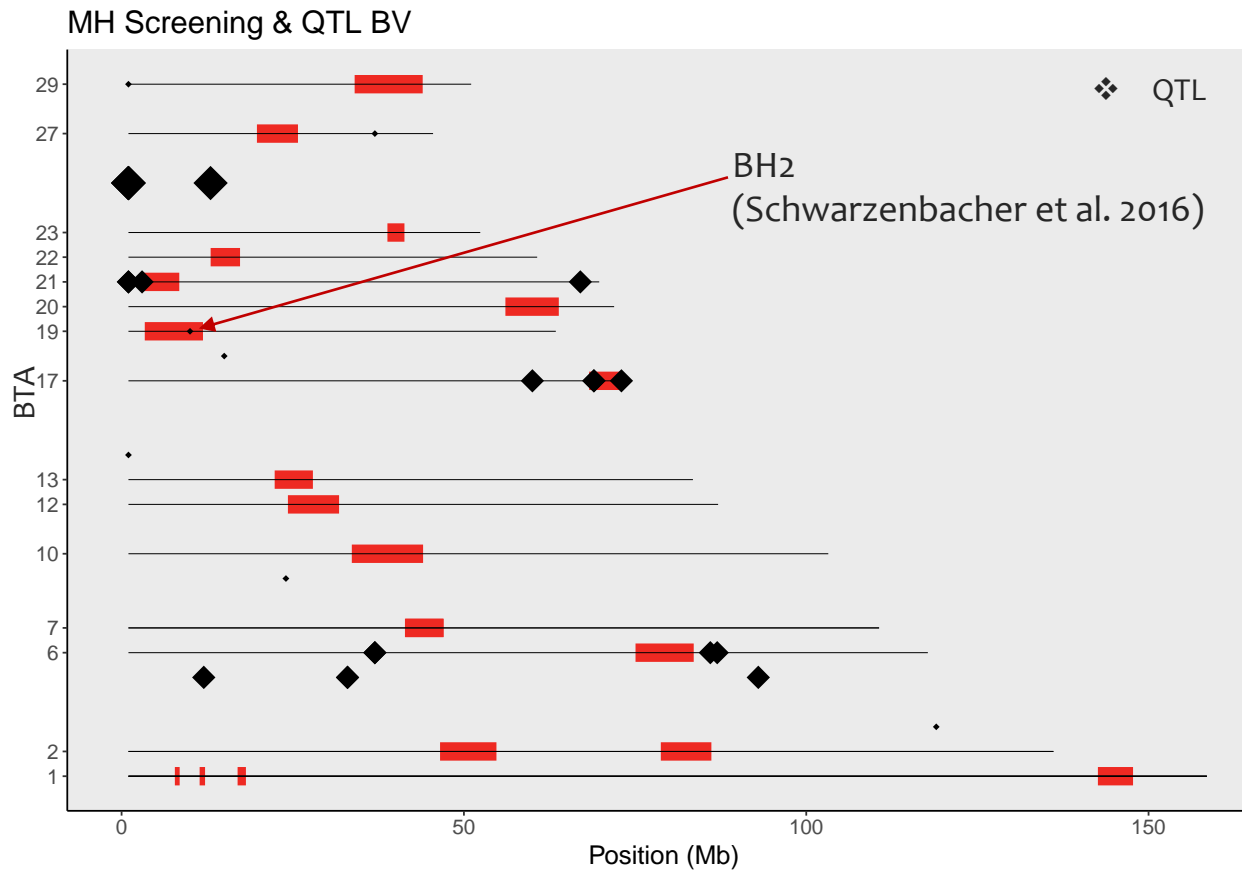


GWAS example OB: carcass conformation calf

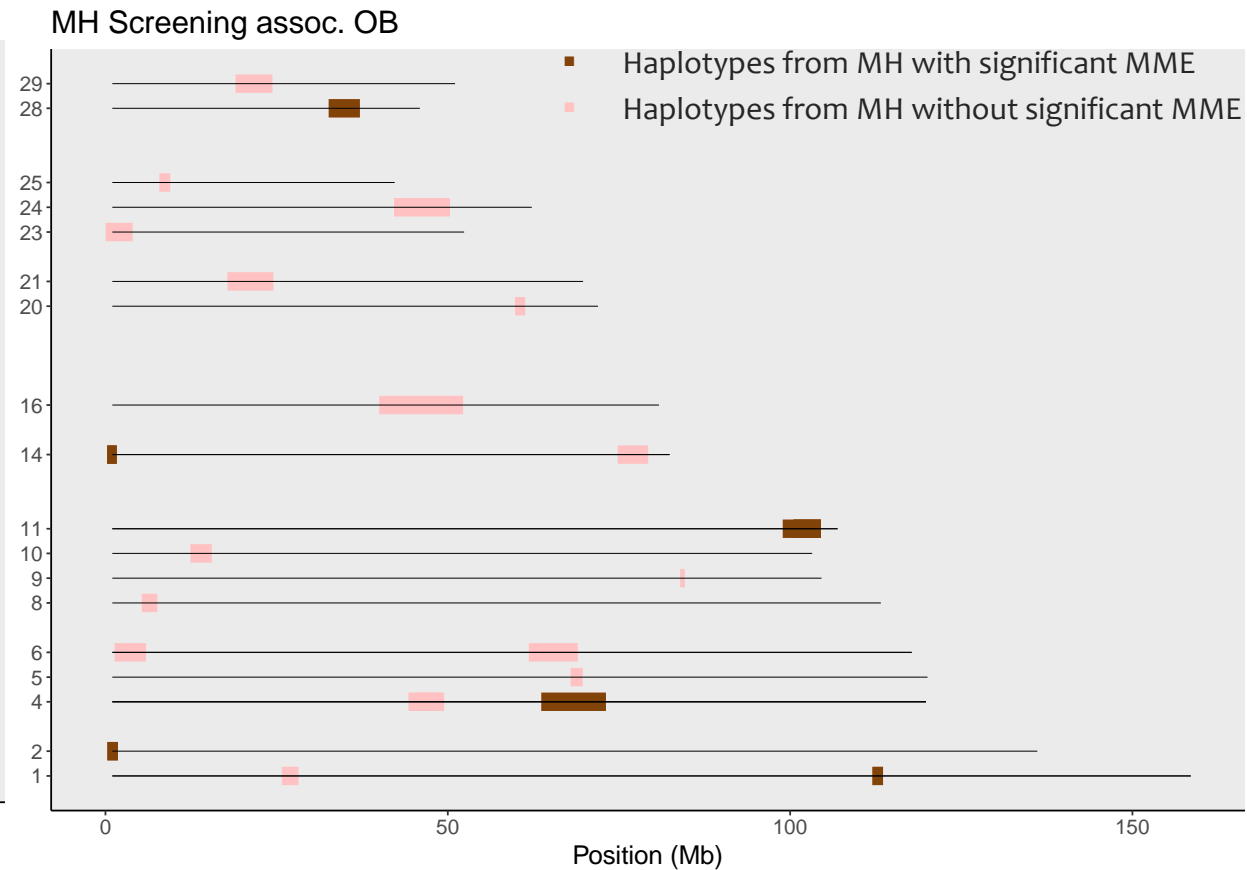
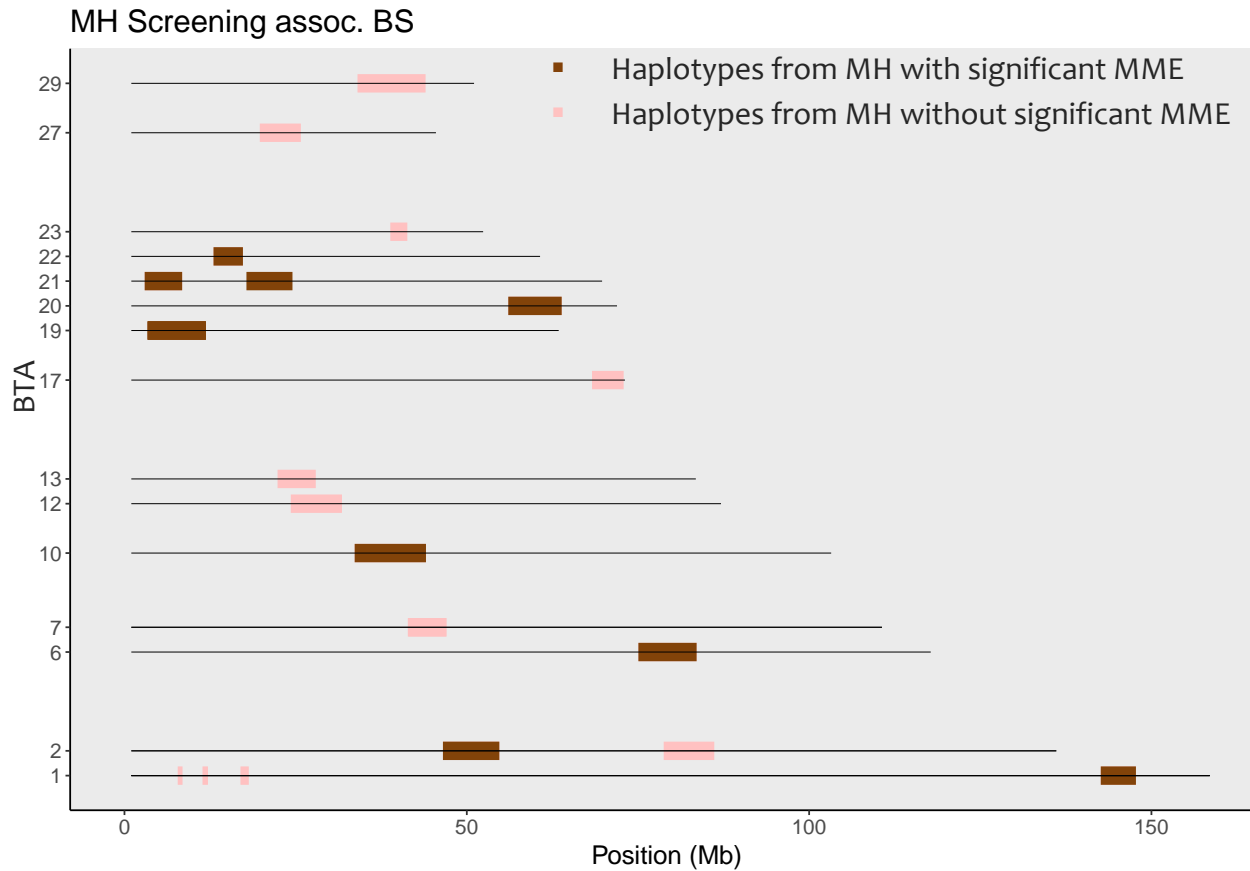


HiggsGene Solutions Inc. (c)

Overlap – MH Screening & QTL

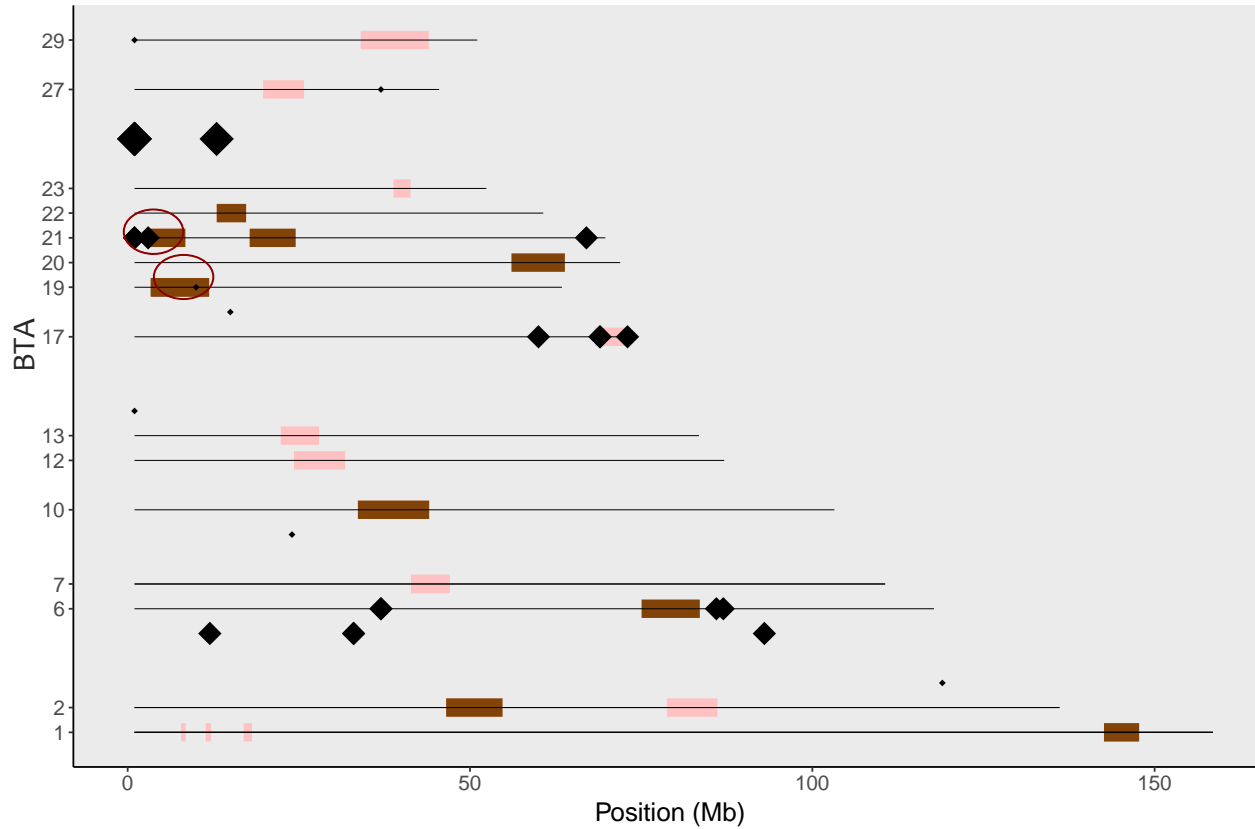


Phenotypic analyses using haplotype information

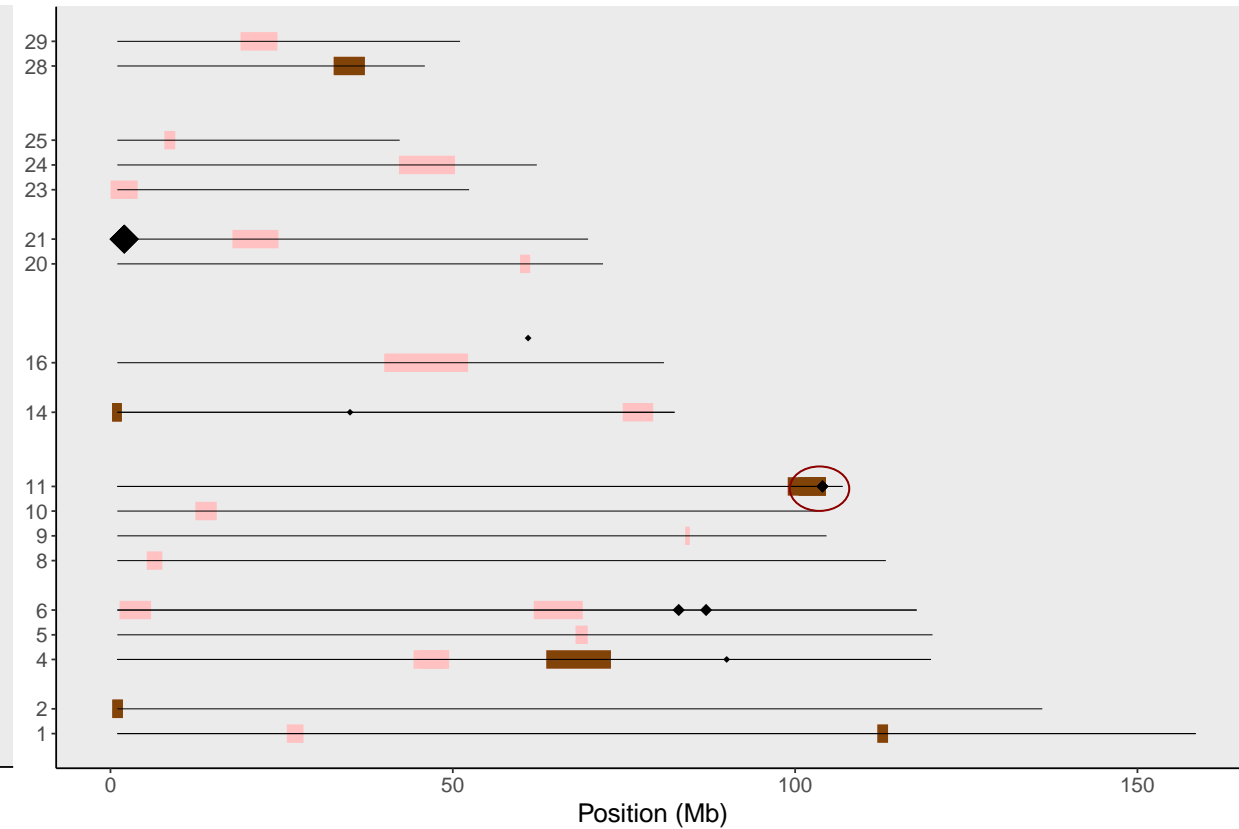


Associated Haplotypes & QTL

MH Screening assoc. + QTL BS



MH Screening assoc. + QTL OB



Summary

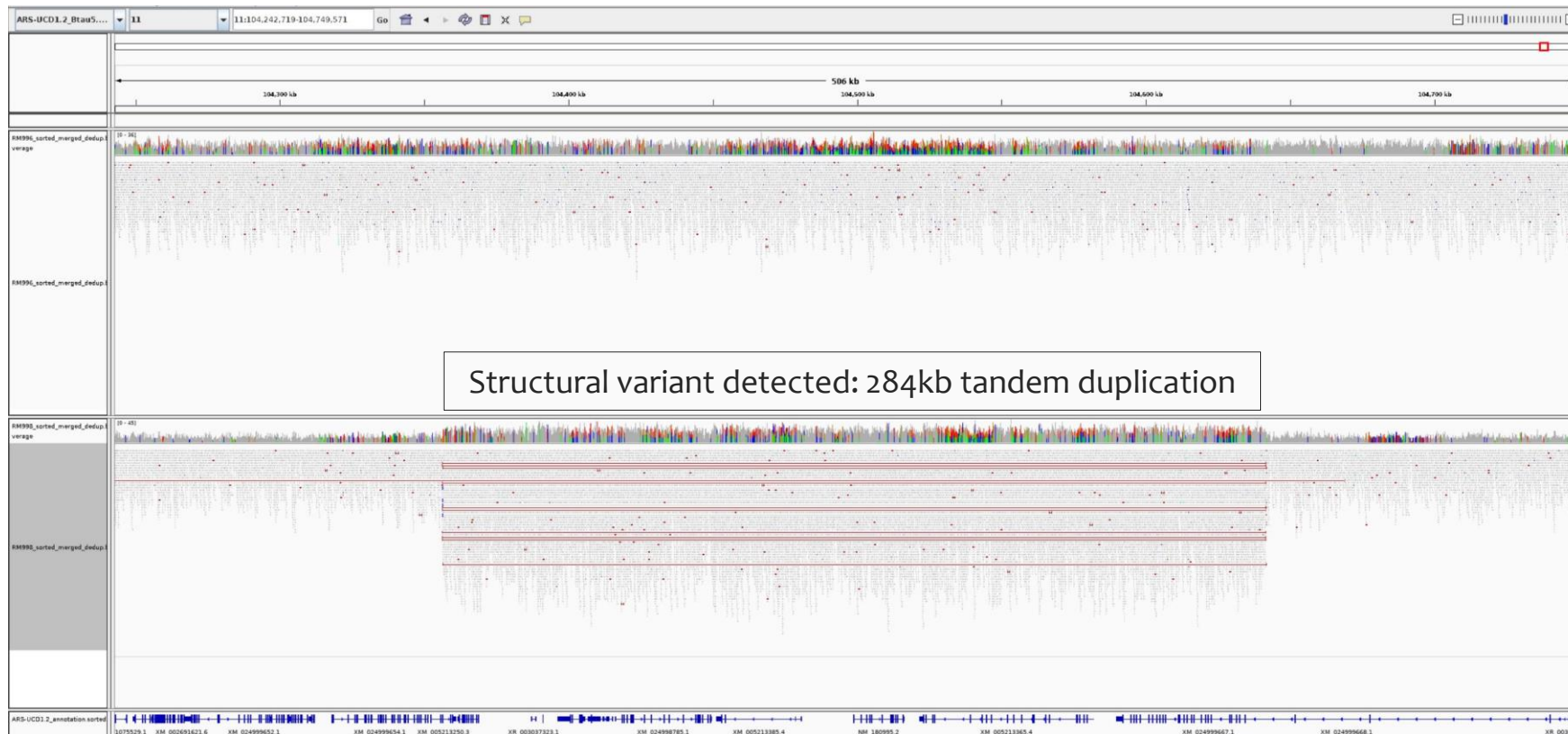
MH Screening	Haplotype associated traits	QTL traits	Population	Validation
19-3385283-11898822	Birth weight still birth rearing success period 1	Still birth	BV	BH2
21-17825892-24537688	Muscularity	Calving ease	BV	
17-68292838-72912770		Non return rate heifer Non return rate cow Interval first - last insemination heifer	BV	
11-100519730-104493671	Carcass conformation score calf Carcass conformation score adults	Carcass conformation score calf Carcass conformation score adults	OB	

Outlook

- Genome Segments with significant reduced homozygosity detected in both populations
- Some regions show associations with related traits
- Some regions overlap with previous QTL / GWAS studies
 - BS: $n = 2$ (BTA19 / BTA21)
 - OB: $n = 1$ (BTA11)

OB 11-100519730-104493671 - Variants

HIGH impact variants	MODERATE impact variants	LOW impact variants	MODIFIER variants	TOTAL
20	345	527	14,469	14,949



- Thank you for your attention!
- Project partners:



Schweizerische Eidgenossenschaft
Confédération suisse
Confederazione Svizzera
Confederaziun svizra



FONDS NATIONAL SUISSE
DE LA RECHERCHE SCIENTIFIQUE

- Third parties:



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