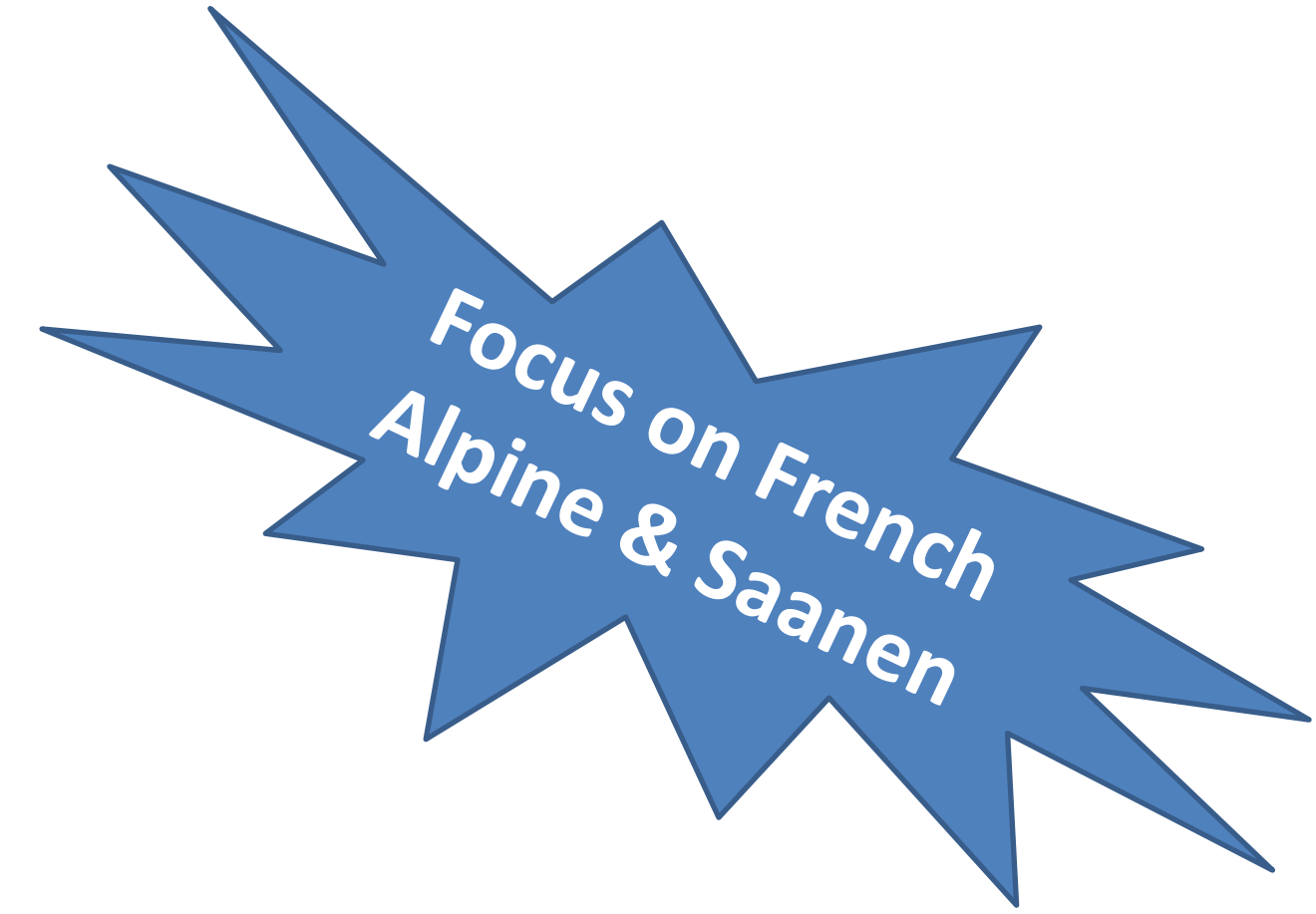




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CONTEXT

- ❖ VarGoats project is a re-sequencing program which aims at covering at best genetic diversity
 → to date : 829 sequenced individuals including 44 French Alpine and 37 French Saanen
- ❖ Expectations of accessing whole-genome variability
 - ✓ identification of causal mutations
 - ✓ improvement of genomic evaluations accuracy



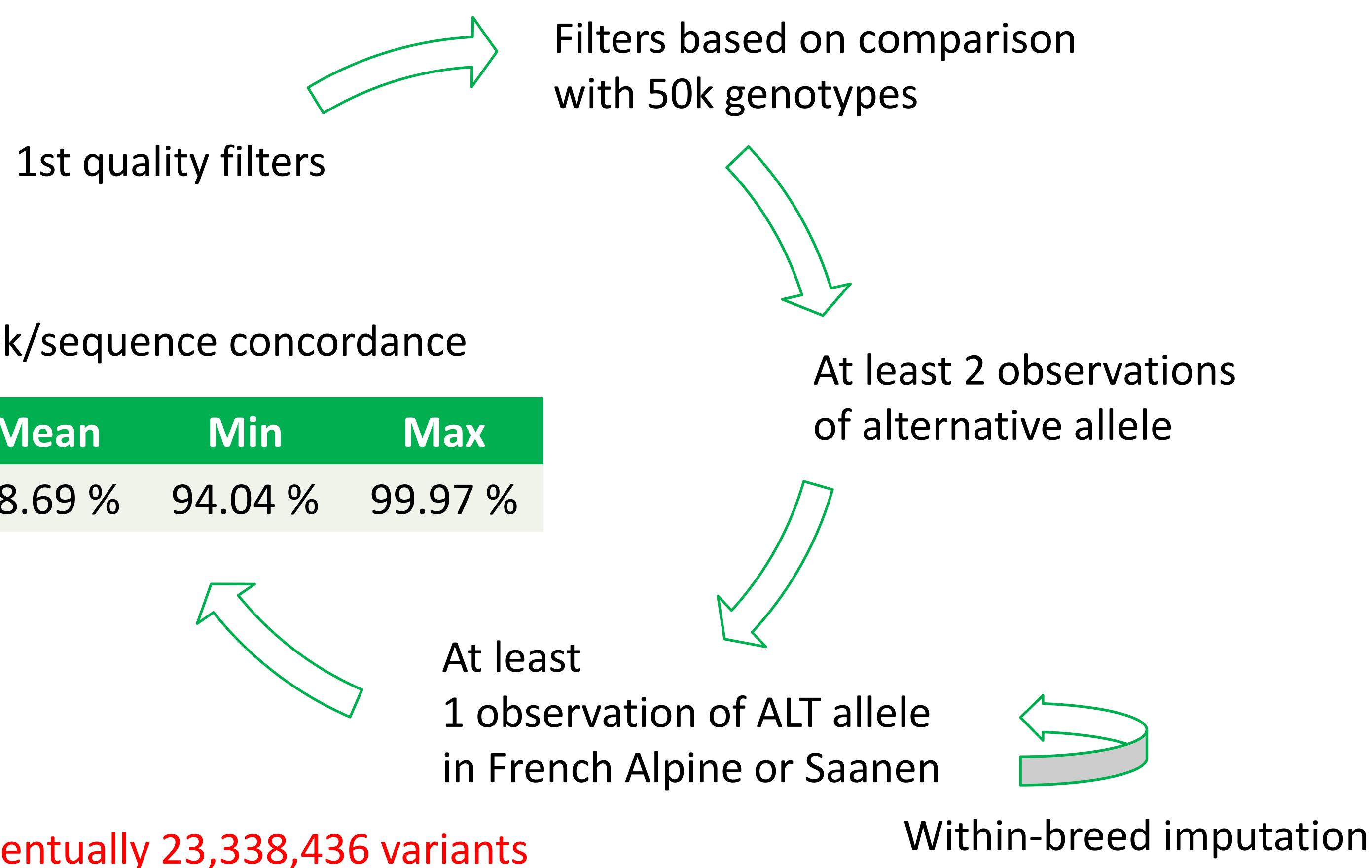
OBJECTIVE

Assess utility of sequence data in French dairy goats

Dilemma : filter enough to impute correctly while keeping enough variants for GWAS analysis

1. Data filtering

Initially 110,193,942 variants



2. Imputation

23,338,436 variants including 40,491 50k-chip SNPs

Leave-one-out scenario

Comparison

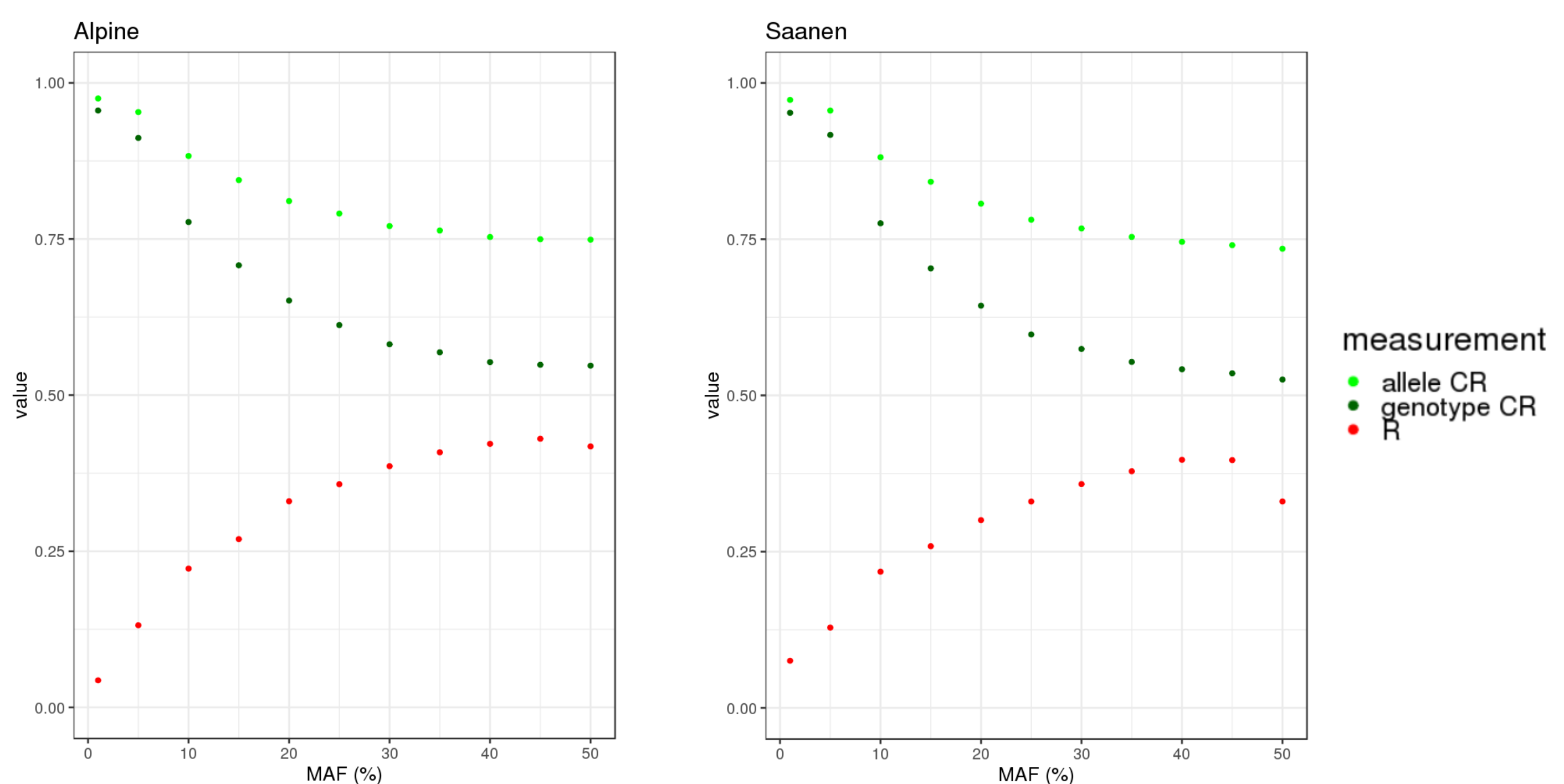
Fimpute including pedigree

Sequence

Masked sequence

Imputed sequence

Alpine			Saanen		
R	genotype CR	allele CR	R	genotype CR	allele CR
0,28	0,73	0,86	0,23	0,76	0,87



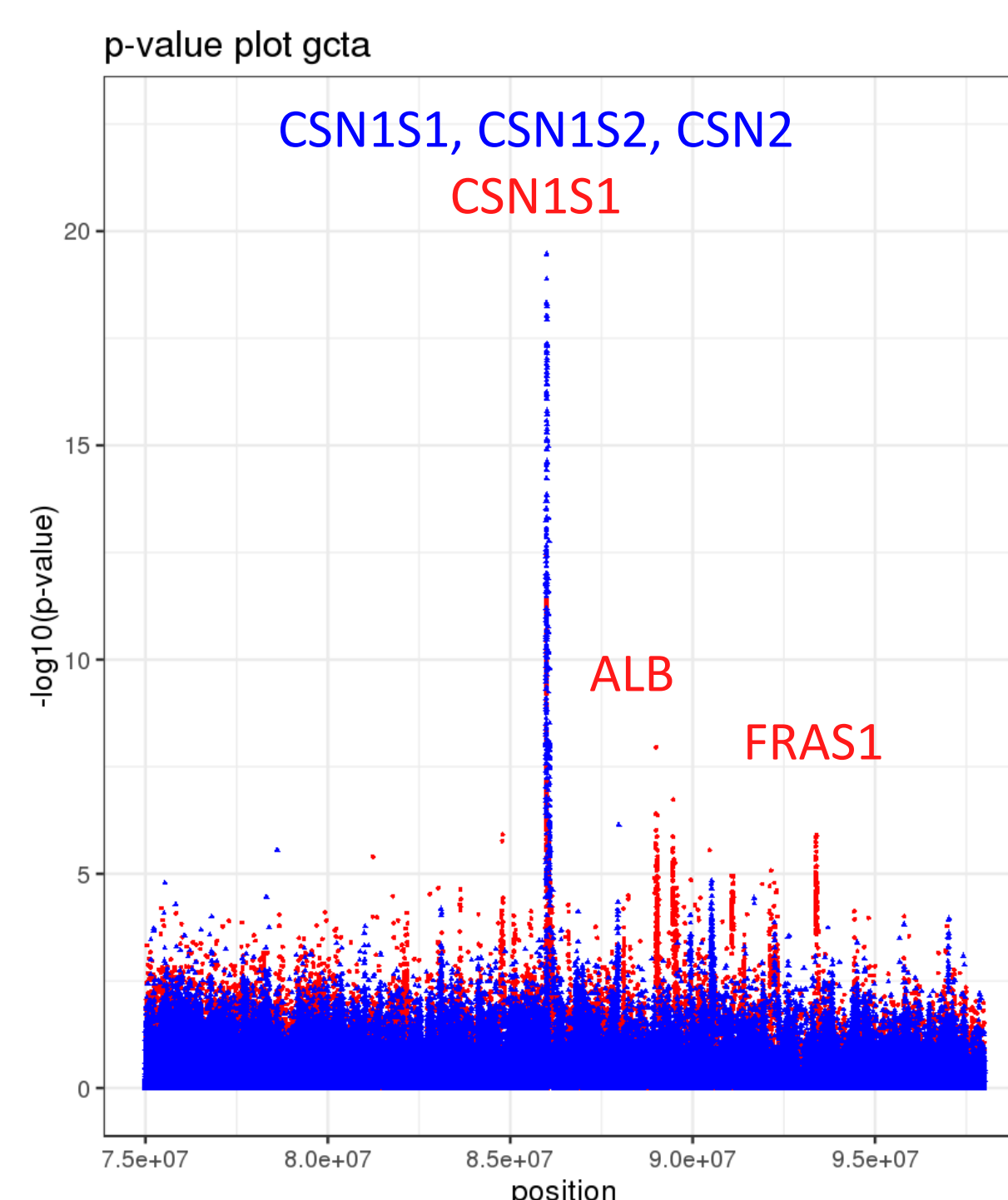
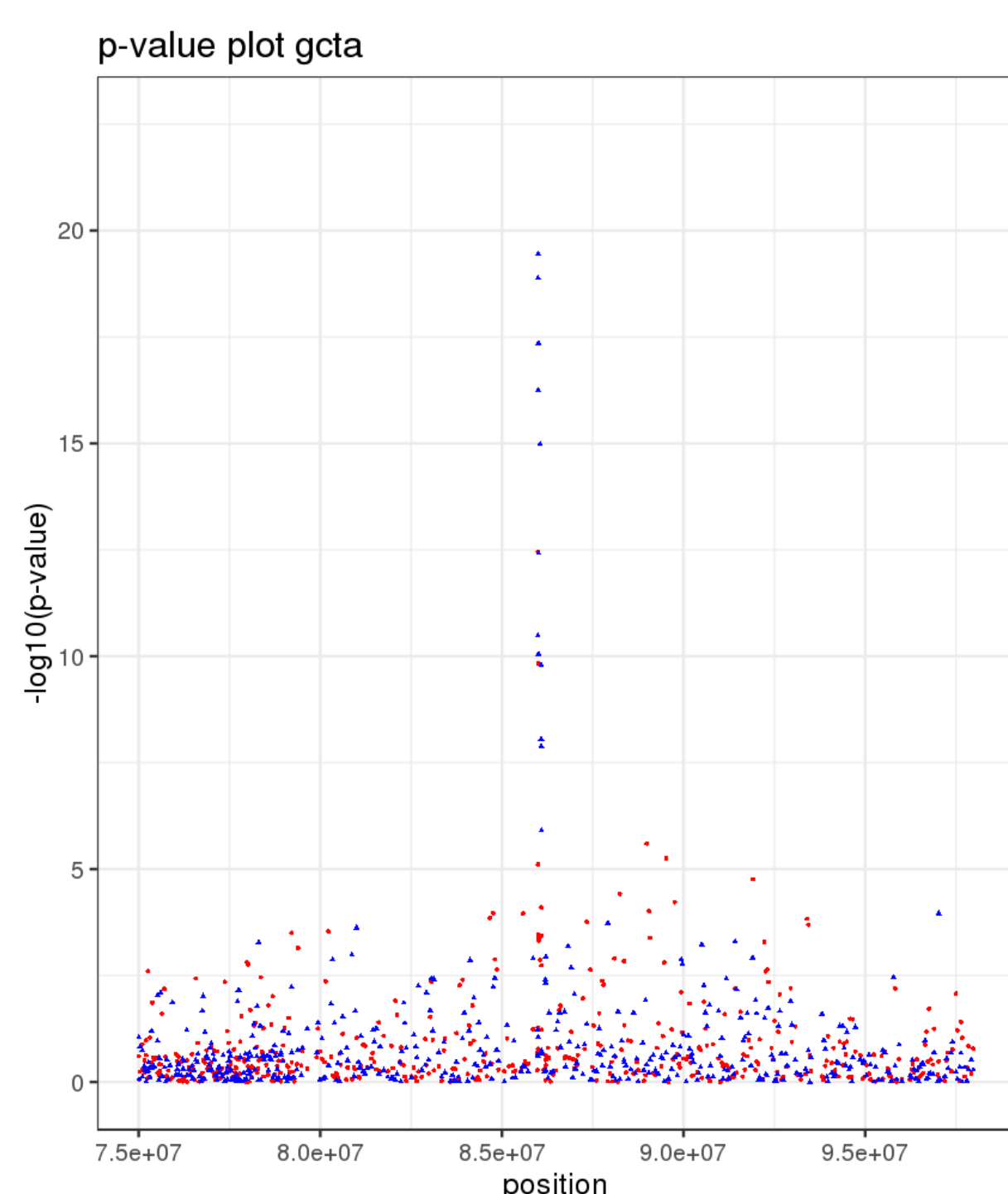
- On CHI 6 in both Alpine and Saanen breeds:
 - ✓ Caseins region confirmed
 - ✓ New regions detected → need an improved annotation
- On CHI 19: 460 variants above chromosome threshold
 - ✓ 117 fixed in Alpine (MAF < 5%) with 115 with MAF >25% in Saanen → 58 annotated genes
 - ✓ Among annotated genes → 13 genes highly expressed in testis
 - ✓ Other genes need to be investigated → no explicit link with trait

3. GWAS analysis on imputed sequence

50k chip genotypes (40,491 SNP)

649 sequenced Alpine
503 sequenced Saanen

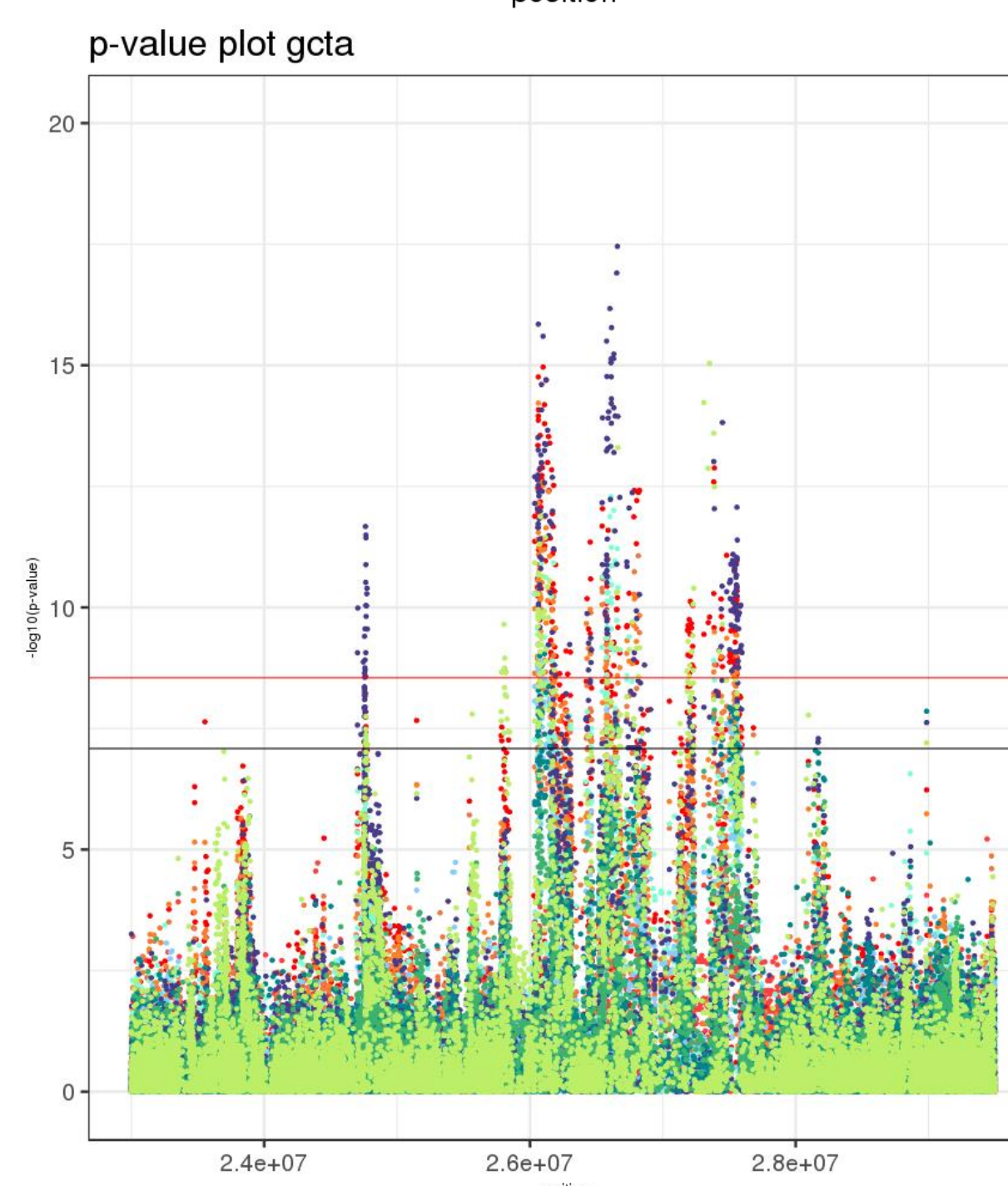
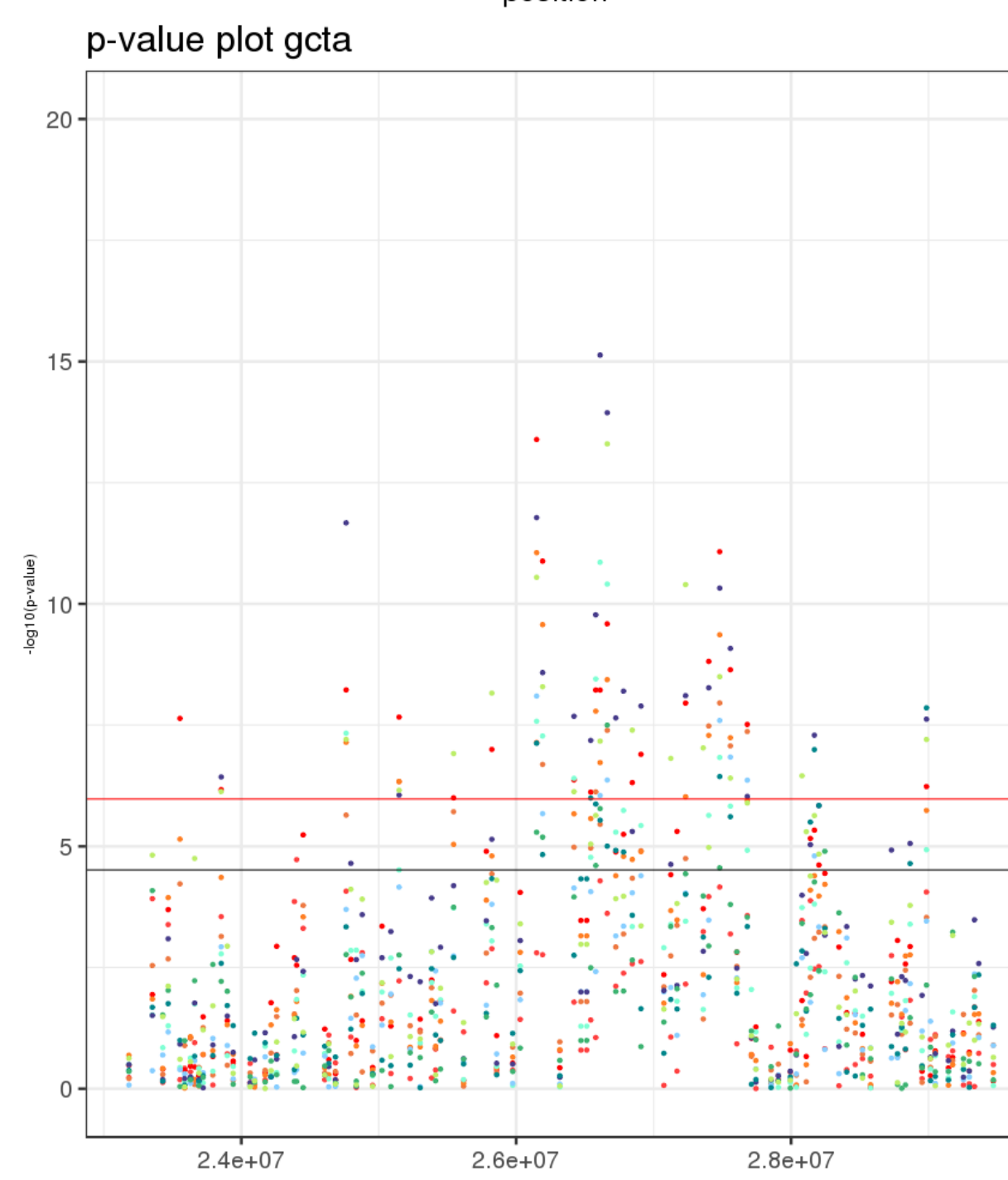
CHI 6 (protein content)



breed

- Alpine
- Saanen

CHI 19 (Saanen)



trait

- CD
- FU
- FY
- LSCS
- MY
- PY
- RUA
- SN
- SV
- UFP

Type traits : fore udder (FU), chest depth (CD), Rear Udder Attachment (RUA), Udder Floor Position (UFP)
 Production traits : fat and protein yield (FY, PY), somatic cells score (LSCS), milk yield (MY)
 Semen production traits : spermatozoa number (SN), semen volume (SV)

CONCLUSIONS

- ❖ Sequence data
 - ✓ Refining known QTL regions
 - ✓ Identifying candidate mutations
 BUT need an improved annotation of *Capra hircus* genome
- ❖ Future perspectives
 - ✓ Evaluation of impact on genomic selection
 - ✓ Functional analysis to confirm candidate mutations