

# GENOME-WIDE GENOMIC AND FUNCTIONAL ASSOCIATION STUDY FOR DIRECT CALVING EASE IN HOLSTEIN CATTLE

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# DATA

- 30,603 bulls from Holstein – Friesian dairy cattle
- Genotypic platform: Illumina BovineSNP50 BeadChip version 2
- Filtration criteria:  $MAF > 0.01$ , call rate  $> 99\%$
- 46,216 SNP after quality control



# METHODS

## 1. GWAS with SNPs from the panel:

$$\mathbf{y} = \mathbf{X}\boldsymbol{\beta} + \mathbf{Z}_1\mathbf{g} + \mathbf{Z}_2\mathbf{a} + \boldsymbol{\varepsilon},$$

- $\mathbf{y}$  - vector of DRP;
- $\boldsymbol{\beta}$  - vector of fixed effects representing a general mean;
- $\mathbf{X}$  - design matrix;
- $\mathbf{Z}_1$  - design matrix for SNP genotypes;
- $\mathbf{g}$  - vector of random additive SNP effects;
- $\mathbf{Z}_2$  - design matrix for polygenic effect;
- $\mathbf{a}$  - vector of random bulls' additive polygenic effects
- $\boldsymbol{\varepsilon}$  - vector of residuals.

## 2. Remapping to newest genome (ARS\_UCD1.2) - Remap



# METHODS II

3. Annotation to genes - Variant Effect Predictor (VEP)
4. Annotation to KEGG pathways and GO:terms - DAVID
5. Second-stage genome association study using pathway analysis:

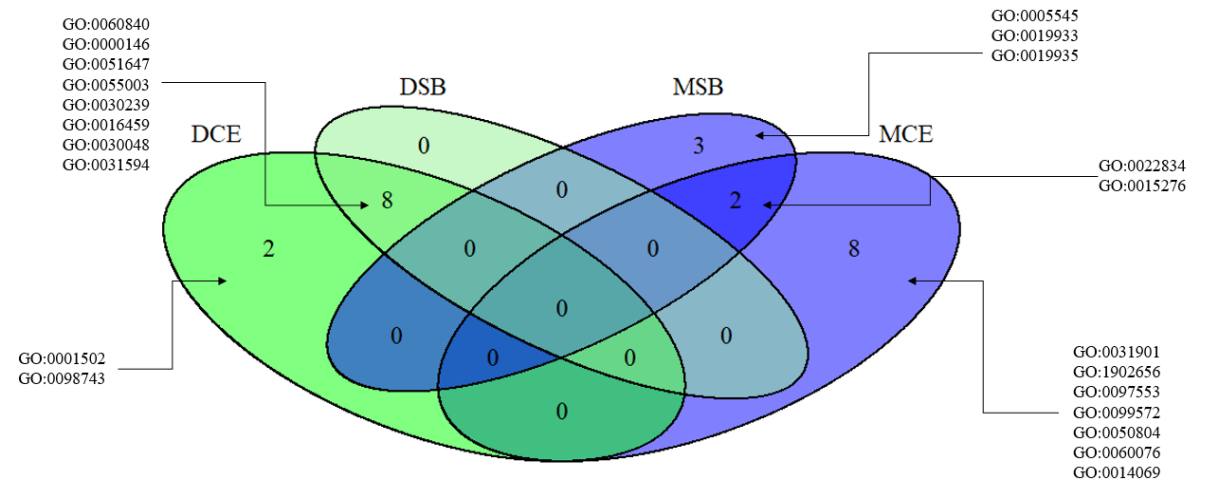
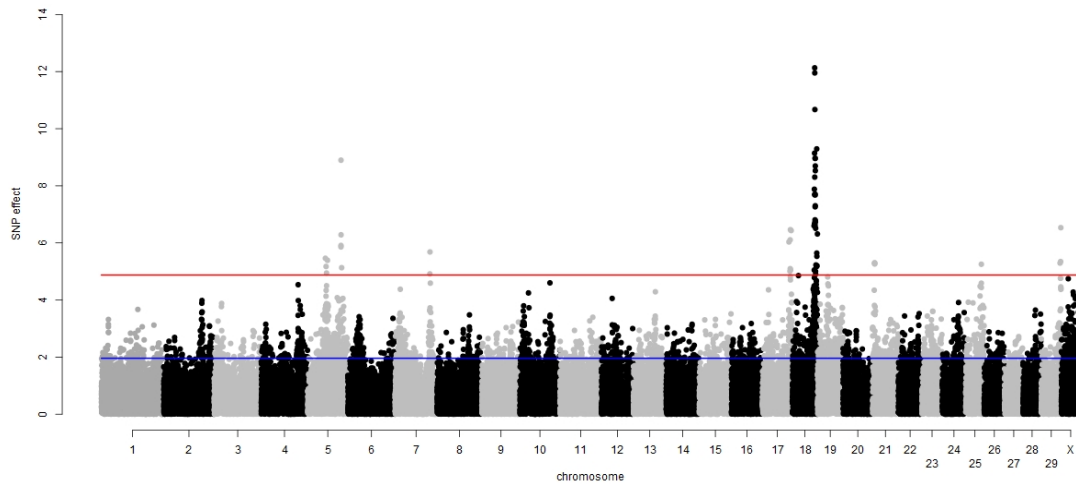
$$\mathbf{y}^* = \mu + \mathbf{Z}^* \mathbf{p} + \boldsymbol{\varepsilon}^*$$

- $\mathbf{y}^*$  - absolute value of the SNP effect for a given trait;
- $\mathbf{p}$  - random KEGG or GO:TERM pathway effect
- $\mathbf{Z}^*$  - the incidence matrix for pathway effect  $\mathbf{p}$ .
- $\boldsymbol{\varepsilon}^*$  - residual term.



# RESULTS

- 59 significant SNPs
- 32/59 highly significant of BTA18
- BTA5 – 9, BTA7 – 2, BTA17 – 7, BTA19 – 1, BTA21 – 4, BTA52 – 1, BTA29 – 3
- 41 from all, mapped to genes
- 10 significant GO: terms
- No significant KEEGs



# CONCLUSIONS

We identified seven significant biological processes.

One significant molecular function.

Two significant cell components.

The majority of significant genes are located on BTA18.



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