

# Genome-Wide Association Study for Milking Speed and Temperament in Holstein-Friesian bulls

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## Objectives

Identify SNPs and biological pathways significant such as milking speed (MSP) and temperament (TEM).

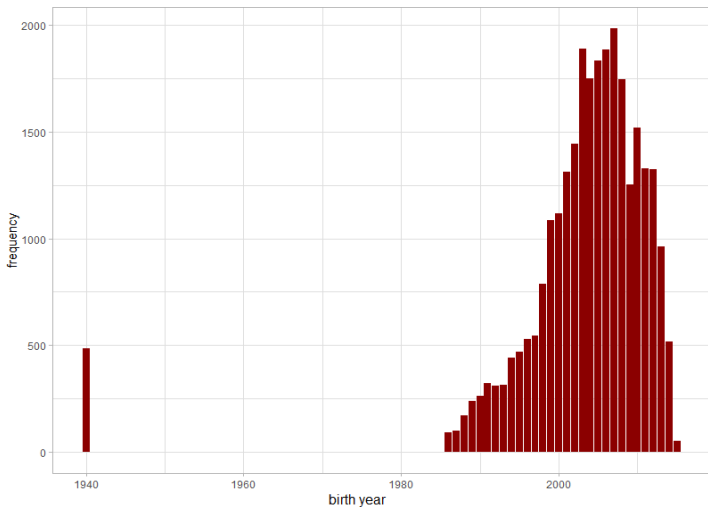


## Material/Animals

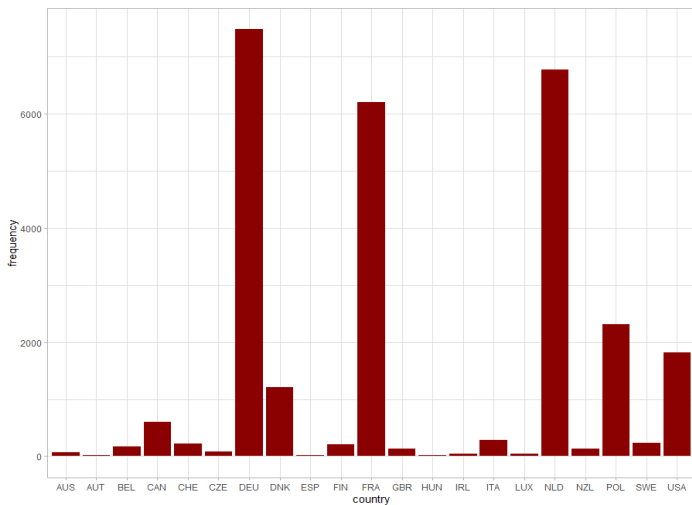
- ❑ EuroGenomics consortium (December 2019 evaluation)
- ❑ 28 315 Holstein-Friesian bulls
- ❑ Illumina Bovine 50K BeadChip → 54 609 SNPs
- ❑ MAF 0.01; call rate 99 % → 46 216 SNPs



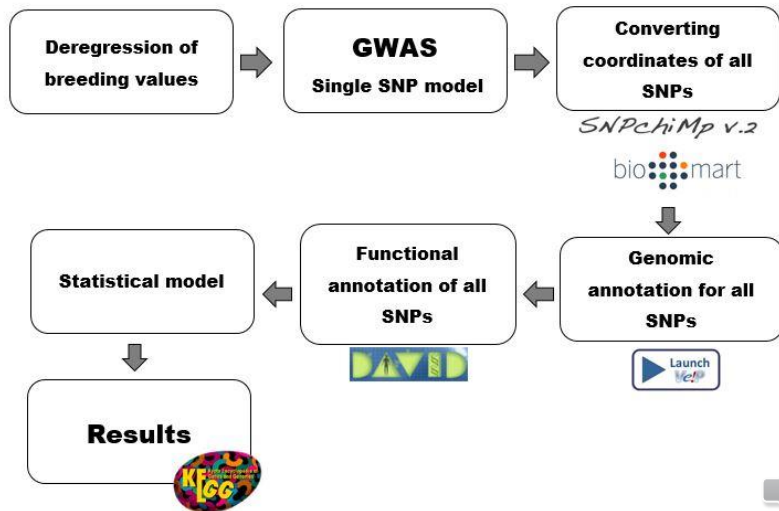
# Material/Animals



# Material/Animals



## Methods/Workflow



## Methods/GWAS model

$$\mathbf{y} = \mu + \mathbf{Z}_1\mathbf{g} + \mathbf{Z}_2\mathbf{a} + \varepsilon \quad (1)$$

$\mathbf{y}$  - vector of deregressed breeding values

$\mu$  - general mean

$\mathbf{Z}_1$  - design matrix for SNP genotypes (-1, 0, 1)

$\mathbf{g}$  - vector of random additive SNP effect

$\mathbf{Z}_2$  - design matrix for polygenic effect

$\mathbf{a}$  - vector of random additive polygenic effect of bulls

$\varepsilon$  - vector of residuals



## Methods/Functional annotation model

$$\hat{\mathbf{g}} = \mathbf{X}\mathbf{b} + \mathbf{e} \quad (2)$$

$\hat{\mathbf{g}}$  - vector of estimated SNP effects from model (1)

$\mathbf{X}$  – design matrix

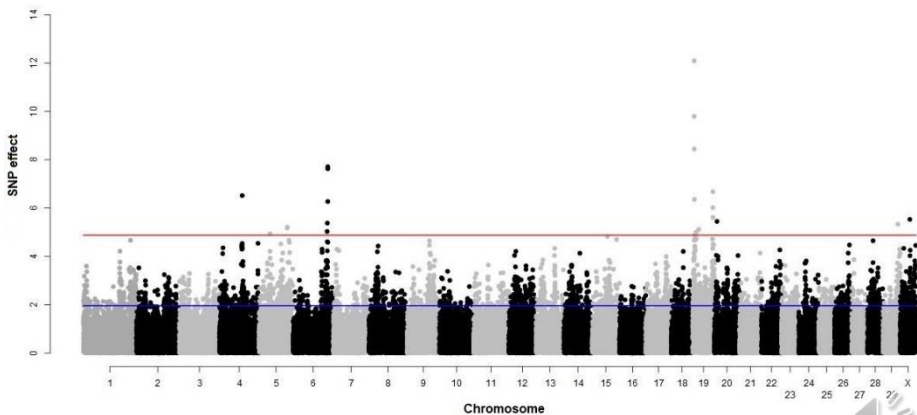
$\mathbf{b}$  - is a vector of KEGG pathway effects

$\mathbf{e}$  - vector of residuals





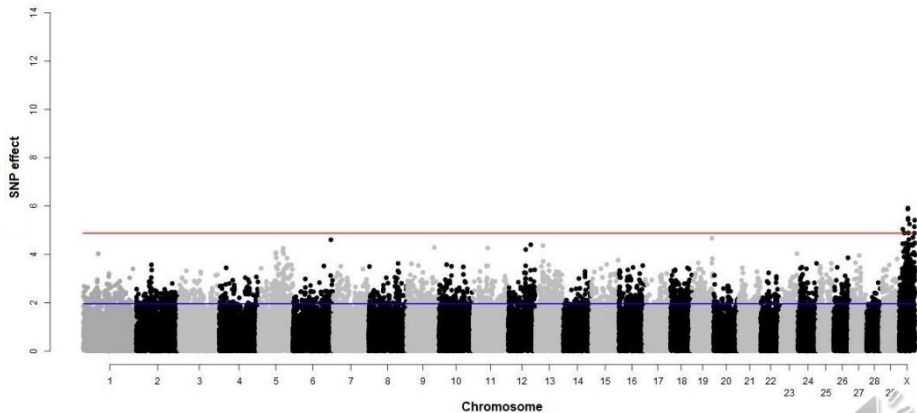
## Results/GWAS/milking speed



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rs	p-value	Location on ARS-UCD1.2	Genomic variant	Gene name
rs41620153	<0.0001	4: 70201858	intergenic	-
rs41599885	0.0375	5: 37711059	intergenic	-
rs41655452	0.0087	5: 105350319	intron of potassium voltage-gated channel subfamily A member 6	ENSBTAG000000047191
rs41257416	0.0107	5:105474132	3 prime UTR variant of NADH:ubiquinone oxidoreductase subunit A9	ENSBTAG000000005465
rs109422338	0.0226	6: 102771937	intron of protein phosphatase 2 regulatory subunit Bgamma	ENSBTAG000000020598
rs42326910	0.0035	6: 103186091	intron of lncRNA	ENSBTAG000000053771
rs109188808	0.0002	6: 104310525	intergenic	-
rs41655065	<0.0001	6: 104350026	intergenic	-
rs41584906	0.0002	19: 7250802	intergenic	-
rs41641989	0.0035	19: 7300658	located in intron of a novel gene	ENSBTAG000000038823
rs110015884	0.0108	19: 7333959	intergenic	ENSBTAG000000021292
rs110036994	<0.0001	19: 7717717	exon of tripartite motif containing 25	ENSBTAG000000009948
rs41579796	0.0259	19: 13808381	3 prime UTR variant of dual specificity phosphatase 14	ENSBTAG000000010279
rs29019325	0.0144	19: 19814327	3 prime UTR variant of transmembrane protein 97	ENSBTAG000000008109
rs29020026	0.0001	19: 59364966	intergenic	-
rs41653204	<0.0001	19: 59437500	intergenic	-
rs109682344	0.0009	19: 59547890	intron of lncRNA	ENSBTAG000000048685
rs43461171	0.0024	20: 6586964	intergenic	-
rs41586565	0.0046	29: 45629483	intron of choline kinase alpha	ENSBTAG000000110304
rs41626488	0.0014	X: 86996906	intergenic	-

## Results/GWAS/temperament



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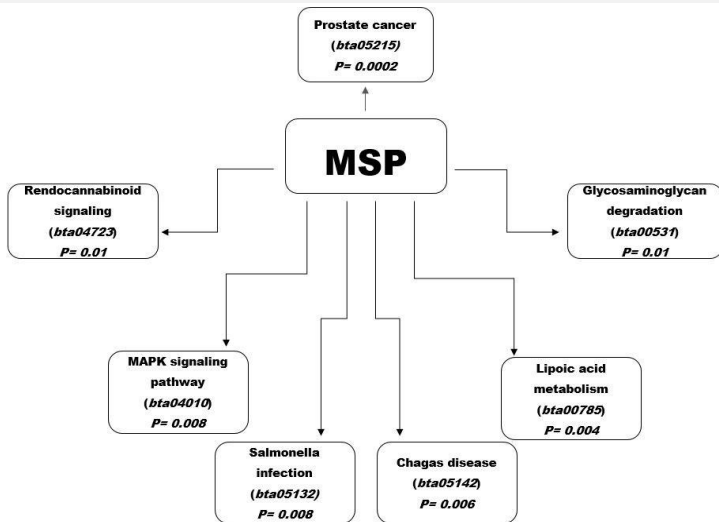


## Results/GWAS/temperament

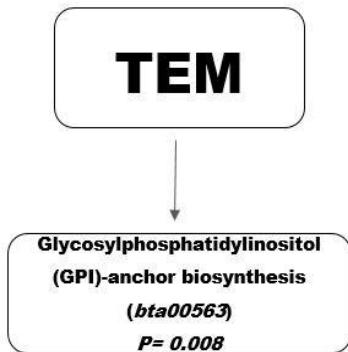
rs	p-value	Location on ARS-UCD1.2	Genomic variant	Gene name
rs110689641	0.0122	X: 863652	intergenic	-
rs29025415	0.0492	X: 11576369	intergenic	-
rs41662793	0.0002	X: 62929712	intergenic	-
rs41631246	0.0018	X: 74200611	intron of fibronectin type III domain containing protein 3C1	ENSBTAG00000026025
rs41621075	0.0025	X: 74239322	3 prime UTR variant of TATA-box binding protein associated factor 9b	ENSBTAG00000000895
rs41623769	0.0002	X: 74434775	intron of ATPase copper transporting alpha	ENSBTAG00000010018
rs109814431	0.0068	X: 99134084	intergenic	-
rs41628806	0.0028	X: 131311943	intergenic	-



## Results/Functional annotation model



## Results/Functional annotation model



## Conclusions

- ❑ Most of significant polymorphisms for MSP are located on chromosome 19.
- ❑ Including the potential causal mutation of tripartite motif containing 25 (TRIM25).
- ❑ All of most of the significant polymorphisms for TEM are located on the X chromosome.
- ❑ Based on the results from functional annotation model seven KEGG pathway were found to associated with MSP and only one with TEM.
- ❑ None of the significant pathways were found to be associated with MSP and TEM.



## Acknowledgements

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