

# Mining the genetic background of clinical mastitis in dairy cattle using whole genome DNA sequences of 32 cows

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## CONCLUSIONS

- genes with putative predisposition to mastitis resistance identified by CNV regions comparison: KBTBD3, TPT1, GLG1 (part of "Cell adhesion molecules,, pathway involved in inflammation)
- genes identified by SNP allele freq. comparison: CA10, IGLV548 (immunoglobulin lambda variable)

## MATERIAL

whole genome NGS with IlluminaHiSeq2000, Polish Holstein-Friesian breed, 16 paternal halfsib pairs

### 16 HEALTHY cows\*

- no mastitis evidence
- coverage 5 – 17x
- #SNPs: 2 063 811 – 6 117 976
- #CNV deletions: 4 012 – 18 413
- #CNV duplications: 1 849 – 6 218
- CNV deletion length: 4 233 – 10 531
- CNV duplication length: 7 254 – 20 13

### 16 SICK cows\*

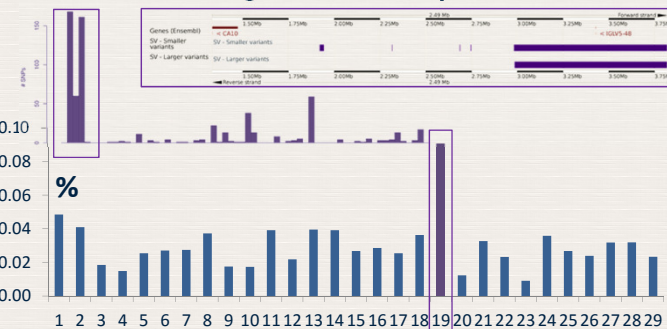
- multiple mastitis case
- coverage 6 – 17x
- #SNPs: 5 041 702 – 5 956 889
- #CNV deletion: 14 518 – 22 496
- #CNV duplication: 1 641 – 2 347
- CNV deletion length: 4 275 – 14 917
- CNV duplication length: 7 591 – 9 124

\* Coverage = average genome coverage per cow; #SNPs = total number of SNP per cow; #CNV deletion (duplication) = total number of deleted (duplicated) regions per cow; CNV deletion (duplication) length = average length of deleted (duplicated) regions per cow

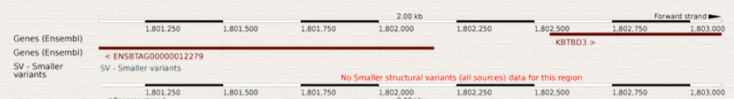
## RESULTS

### SNP allele freq. differences SICK vs HEALTHY

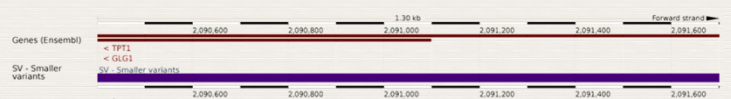
- no significance with Bonferroni correction
- SNPs with large allele freq. differences



### CNV regions deleted in SICK cows



- BTA15 → 6 deletions of 2000 bp, KBTBD3



- BTA18 → 6 deletions of 1300 bp, TPT1, GLG1

## METHODS

### NGS DATA PROCESSING

Raw read filtering → Trimmomatic

Alignment to UMD3.1 → BWA-MEM

Post alignment filtering → Picard

SNP calling → FreeBayes

CNV calling → CNVnator

### VARIANT ANALYSIS

SNP allele count estimation → Jackknife of halfsibs

SNP allele frequency difference → Odds Ratio

CNV regions detection specific to only sick halfsibs