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

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

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)

METHODS

16 HEALTHY cows*

• no mastitis evidence
• coverage: 5 – 17x
• #SNPs: 2 063 811 – 6 117 976
• #CNV deletions: 4 012 – 6 218
• #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
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