

Unravelling the Genetic Background of Clinical Mastitis in Cattle Using Whole Genome Sequence

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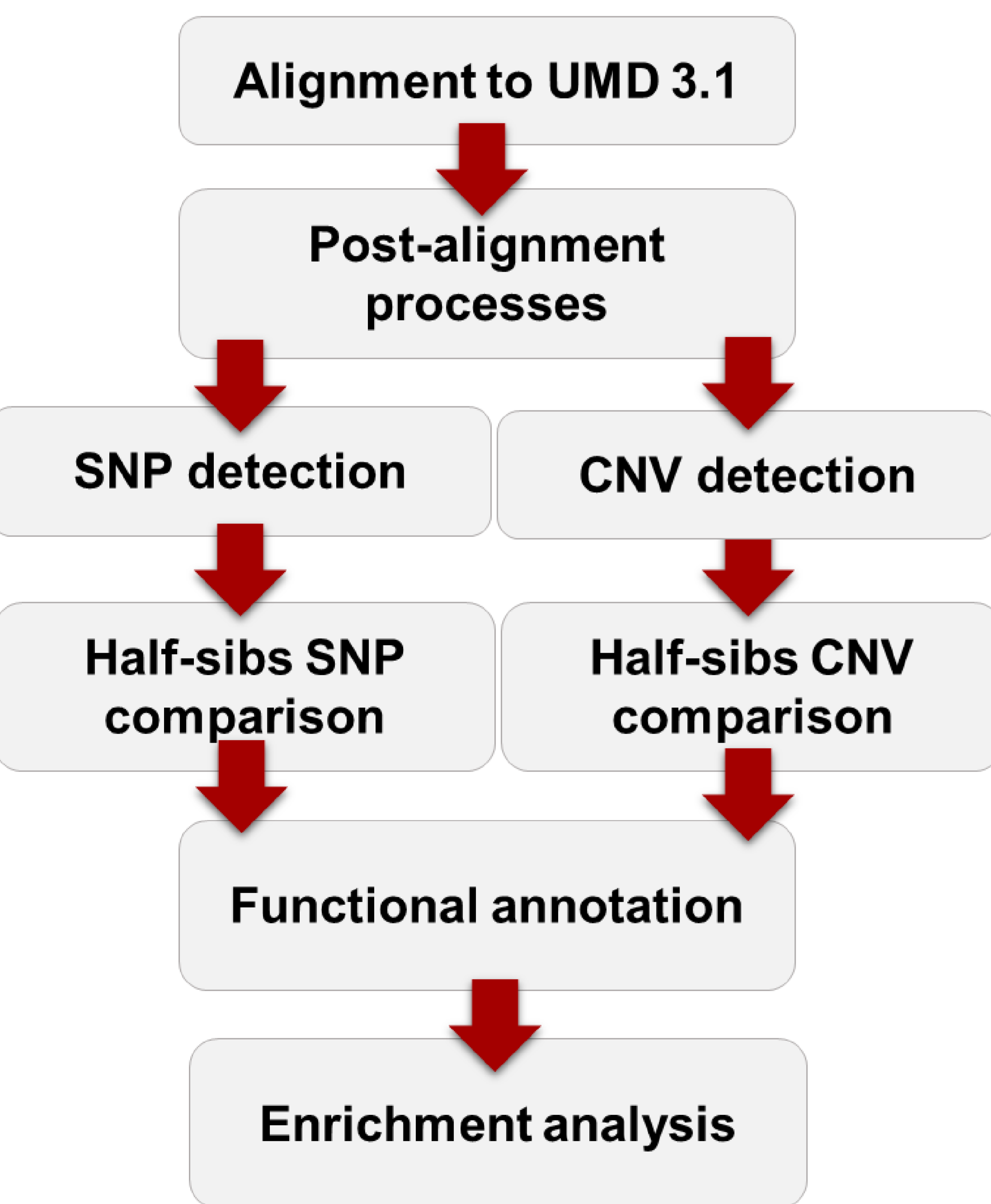
Objectives

Characterization of links between single nucleotide polymorphisms (SNPs), copy number variants (CNVs) and the incidence of clinical mastitis (CM)

Conclusions

- CNVs play an important role in the susceptibility to CM
- Identified genes are involved in immune response
- Deletions – more severe consequences on reducing resistance against clinical mastitis, than...
- Duplications – on increasing resistance to clinical mastitis

Methods

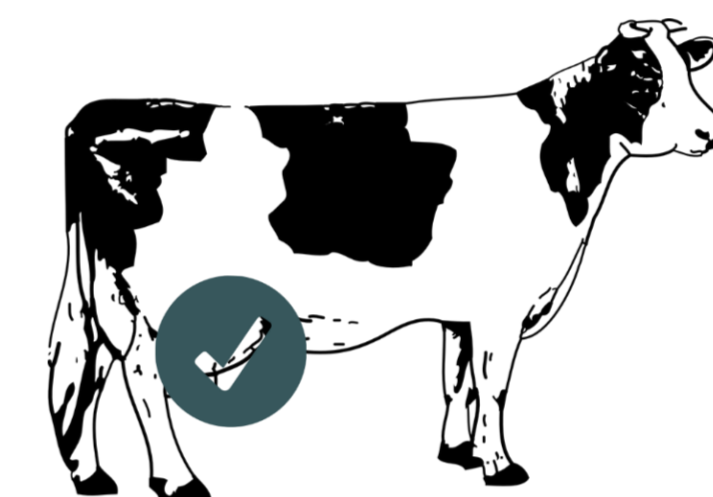


Acknowledgements

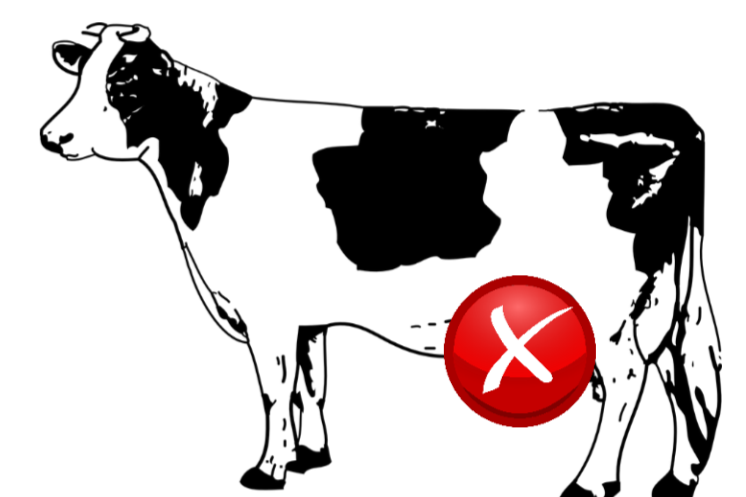


Data set

32 Polish Holstein Friesian cows
 16 paternal half-sibs
 Illumina HiSeq 2000 (WGS)
 Coverage 5x – 17x
 NCBI BioProject PRJNA359667



Healthy (H)



Sick (S)

Results

SNPs

Introns of genes with differences in SNP genotypes between sick and healthy cows

RefSeq coding ring finger protein 122 (10/16 half-sibs)

- cell viability and immune response

MET proto-oncogene receptor tyrosine kinase (9/16 half-sibs)

- processes related to inflammation, cancerogenesis

WRN gene (8/16 half-sibs)

- premature aging in humans, increased susceptibility to infection

APP: amyloid beta precursor

- bactericidal and antifungal activities in human
- molecular markers for SCM in ruminants

FOXL2: forkhead box L2

- role in inflammation

SSFA2: sperm specific antigen 2

- associated with SCS = indicator of CM
- overlaps with QTL for bovine immunoglobulin G

CNV DELETIONS

Exons deleted in a sick cow, but present in its healthy half-sib exhibits potential causal influence CM (at least 7 half-sibs pairs)

NDUFS6: NADH:ubiquinone oxidoreductase subunit S6

- QTL for SCS = CM indicator
 DGVa: Boussaha *et al.* 2015

ADORA2A: adenosine A2a receptor

- modulating tissue response to inflammation
- in mice highly expressed in mammary gland
 DGVa: Hou *et al.* 2011

TXNRD2: thioredoxin reductase 2

- candidate for influencing susceptibility to *S.aureus*
 DGVa: Hou *et al.* 2011

OTUD3: deubiquitinase 3

- associated with inflammatory bowel disease in humans
 DGVa: Keel *et al.* 2017