

# Strategy to imputation of large genomic deletions and utilizing them in mapping and genomic prediction in cattle

Md Mesbah-Uddin<sup>1,2</sup>, Bernt Guldbrandtsen<sup>1</sup>, Aurélien Capitan<sup>2</sup>, Mogens Sandø Lund<sup>1</sup>, Didier Boichard<sup>2</sup>, Goutam Sahana<sup>1\*</sup>

<sup>1</sup> Centre for Quantitative Genetics and Genomics, Aarhus University, Denmark

<sup>2</sup> Animal Genetics and Integrative Biology, UMR 1313 GABI, INRAE, AgroParisTech, Université Paris-Saclay, France

\*Contact: [goutam.sahana@qgg.au.dk](mailto:goutam.sahana@qgg.au.dk)

- Large genomic deletions can be detected from whole-genome sequencing (WGS), and imputed to SNP array-typed cattle population with high accuracy
- Use of deletions, beside SNPs and indels, provide better mapping signal, and gains in genomic prediction accuracy

## Hypothesis

- Large deletions can be identified from WGS and imputed with high accuracy
- Addition of deletions will improve mapping and genomic prediction

## Objectives

- To identify large deletions, and impute to existing SNP-array genotyped animals
- To use large deletions in GWAS and genomic prediction

## Methods

- Large deletions were identified from WGS using GenomeSTRIP-2 software
- Genotype phasing was performed using Beagle-4 and SHAPIT-2, and imputation was done using Minimac-3 software
- Single-marker GWAS and conditional association, and genomic prediction was performed using GCTA software



Fig1. A 525-KB deletion on chromosome 23 in Nordic Red Dairy Cattle.

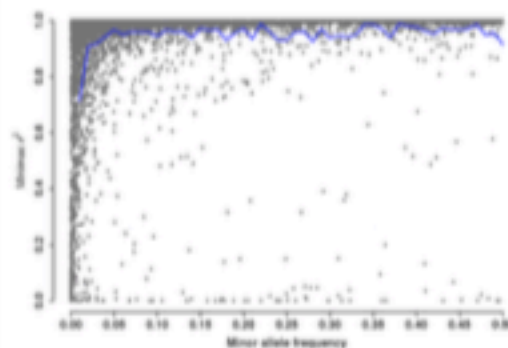


Fig2. Scatter plot of imputation accuracy of 5,719 large deletions.

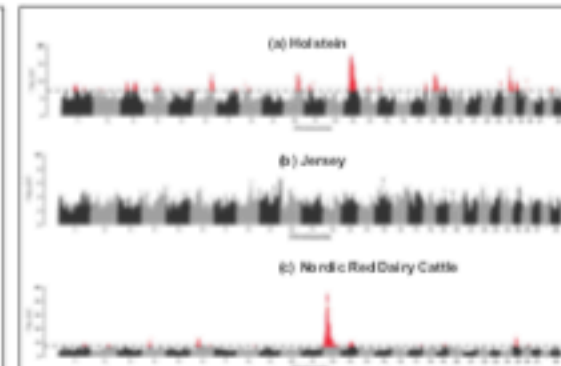


Fig3. Manhattan plot of single-marker GWAS for Fertility Index in (a) Holstein, (b) Jersey, and (c) Nordic Red Dairy Cattle.

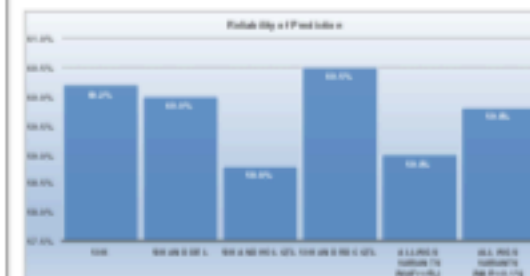


Fig4. Reliability of genomic prediction (Fertility) in Jersey cattle.

## Results

- Provided a high-resolution map of large deletions (Fig 1, Ref 1)
- Deletions can be imputed with high accuracy (Fig 2, Ref 2)
- Besides SNPs and indels, some deletions showed strong association with female fertility in dairy cattle (Fig 3, Ref 3)
- Addition of GWAS markers provided higher gains in prediction accuracy (Fig 4, Ref 3)

## Reference

1. Mesbah-Uddin, M., et al., Genome-wide mapping of large deletions and their population-genetic properties in dairy cattle. *DNA Res*, 2018, 25(1): p. 49-69.
2. Mesbah-Uddin, M., et al., Joint imputation of whole-genome sequence variants and large chromosomal deletions in cattle. *J Dairy Sci*, 2019, 102(12): p. 11193-11206.
3. Mesbah-Uddin, M. (2019). Identification of causal factors for recessive lethals in dairy cattle with special focus on large chromosomal deletions. Retrieved from <http://www.theses.fr/2019/AN/0018/document>. Available from <http://www.theses.fr/2019/AN/0018>