



**HAL**  
open science

## Search for new mutations in cattle by systematic whole genome resequencing

Mekki Boussaha, C Eché, C Iampietro, C Grohs, A Capitan, Denis Milan, S Fritz, C Donnadieu, D Boichard

► **To cite this version:**

Mekki Boussaha, C Eché, C Iampietro, C Grohs, A Capitan, et al.. Search for new mutations in cattle by systematic whole genome resequencing. 74th Annual Meeting of the European Federation of Animal Science, INRAE, Aug 2023, Lyon, France. 10.3920/978-90-8686-936-7 . hal-04195854

**HAL Id: hal-04195854**

**<https://hal.inrae.fr/hal-04195854v1>**

Submitted on 4 Sep 2023

**HAL** is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

**Search for new mutations in cattle by systematic whole genome resequencing**

M. Boussaha<sup>1</sup>, C. Eché<sup>2</sup>, C. Escoufflaire<sup>3</sup>, C. Grohs<sup>1</sup>, C. Iampietro<sup>2</sup>, A. Capitan<sup>1</sup>, M. Denis<sup>2,4</sup>, S. Fritz<sup>3</sup>, C. Donnadiou<sup>2</sup> and D. Boichard<sup>1</sup>

<sup>1</sup>Université Paris-Saclay, INRAE, AgroParisTech, GABI, Jouy-en-Josas, 78350, France, <sup>2</sup>INRAE, US1426, GeT-PlaGe, Genotoul, France Genomique, Université Fédérale de Toulouse, Castanet-Tolosan, 31326, France, <sup>3</sup>Eliance, Paris, 75012, France, <sup>4</sup>GenPhySE, Université de Toulouse, INRAE, INPT, ENVT, Castanet-Tolosan, 31326, France; mekki.boussaha@inrae.fr

Systematic whole genome sequencing provides a rapid and powerful method to identify recent novel mutations on a cattle population scale. It helps farmers to detect early carriers of new genetic anomalies and more generally to boost genomic selection. Regarding genetic defects, the aim is to identify new candidate mutations that may have deleterious potential before they are widely spread in the population to inform the breeders. A major drawback is the high rate of false positives, ie of variants with a strong annotation but without any effect. Therefore the annotation is the most critical step. We focussed only on still unknown variants (ie de novo or likely very recent mutations) in highly conserved sequences in other species and/or with effects predicted to be similar to those described in OMIM or MGI databases. We applied this strategy on whole genome sequences on 571 artificial insemination bulls from 14 dairy and beef breeds. In large breeds, recently marketed bulls were selected. In smaller breeds, influential ancestors not yet sequenced were chosen. In total, we identified 1,548 novel genomic variants with a potential link to certain quantitative traits of interest or possible genetic abnormalities. These variants were investigated by searching for carrier or homozygous descendants, and by adding these variants on the custom part of the EuroGenomics SNP chip for an easy screening of the population. This information is also returned back to bull's owners, with a specific alert only for the few most critical variants. Although the annotation still lacks accuracy, we believe that whole genome sequencing of all artificial insemination bulls is an early, rapid and always cheaper method for identifying genetic defects before they disseminate in the population. The SeqOccIn project was funded by the Occitanie region, FEDER, and Apis-Gene.

**Estimation of non-additive genetic effects for semen production traits in beef and dairy bulls**

R. Nagai<sup>1</sup>, M. Kinukawa<sup>2</sup>, T. Watanabe<sup>2</sup>, A. Ogino<sup>2</sup>, K. Kurogi<sup>3</sup>, K. Adachi<sup>3</sup>, M. Satoh<sup>1</sup> and Y. Uemoto<sup>1</sup>

<sup>1</sup>Graduate School of Agricultural Science, Tohoku University, Laboratory of Animal Breeding and Genetics, Sendai, Miyagi, 980-8572, Japan, <sup>2</sup>Livestock Improvement Association of Japan, Inc., Maebashi Institute of Animal Science, Maebashi, Gunma, 371-0121, Japan, <sup>3</sup>Livestock Improvement Association of Japan, Inc., Cattle Breeding Department, Koto-ku, Tokyo, 135-0041, Japan; rintaro.nagai.q7@dc.tohoku.ac.jp

Non-additive genetic effects are considered as a key factor to understand reproductive traits, such as semen production traits, in cattle. In order to evaluate the relevance of non-additive genetic effects for semen production traits in beef and dairy bulls using the Illumina BovineSNP50 BeadChip, we performed genome-wide association studies (GWAS) to detect non-additive quantitative trait loci (QTLs). We also evaluated non-additive polygenic effects by estimating non-additive genetic (i.e. dominance and epistatic) variance components. In total, 65,463 records for 615 genotyped Japanese Blacks (JB) as beef bulls and 50,734 records for 873 genotyped Holstein (HOL) as dairy bulls were used to detect QTLs and to estimate non-additive genetic variance components for five semen production traits: semen volume (VOL), sperm number (NUM), sperm motility (MOT), MOT after freeze-thawing (aMOT) and sperm concentration (CON). Additive QTL was detected on *Bos taurus* autosome (BTA) 24 for MOT in JB and on BTA17 for MOT in HOL. Non-additive QTLs were detected on BTA2 for MOT and on BTA26 for CON in JB and BTA6 for MOT, BTA11 for aMOT and BTA17 for NUM, MOT, aMOT and CON in HOL. In HOL, a non-additive QTL on BTA17 has pleiotropic effects on these traits. Assuming non-additive genetic variance components, the broad-sense heritability (0.17 to 0.43) was more than twice as high as the narrow-sense heritability (0.04 to 0.11) for all traits and breeds. In addition, the difference between the broad-sense heritability and repeatability were very small for VOL, NUM and CON, because a large proportion of permanent environmental variance was explained by epistatic variance. In conclusion, our present study suggest that non-additive QTLs and polygenic effects play important roles in semen production traits in beef and dairy bulls.