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Identification of Five Novel Mutations Responsible for Prenatal Mortality in Dairy Cattle

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Sébastien Fritz UNCEIA Genetics Team, Aurelien Capitan UNCEIA Genetics Team, Anis Djari INRA SIGENAE Bioinformatics Team, Sabrina C Rodriguez INRA SIGENAE Bioinformatics Team, Sabrina C Rodriguez INRA UMR1313 Animal Genetics and Integrative Biology, Anne Barbat INRA UMR1313 Animal Genetics and Integrative Biology, Aurélia Baur UNCEIA Genetics Team, Cécile Grohs INRA UMR1313 Animal Genetics and Integrative Biology, Bernard Weiss INRA UMR1313 Animal Genetics and Integrative Biology, Mekki Boussaha INRA UMR1313 Animal Genetics and Integrative Biology, Mekki Boussaha INRA UMR1313 Animal Genetics and Integrative Biology, Mekki Boussaha INRA UMR1313 Animal Genetics and Integrative Biology, Mekki Boussaha INRA UMR1313 Animal Genetics and Integrative Biology, Mekki Boussaha INRA UMR1313 Animal Genetics and Integrative Biology, Diane Esquerré INRA GeT Genomics Facility, Christophe Klopp INRA Toulouse, Dominique Rocha INRA UMR1313 Animal Genetics and Integrative Biology, Didier A. Boichard INRA UMR1313 Animal Genetics and Integrative Biology

P0568 - Identification of Five Novel Mutations Responsible for Prenatal Mortality in Dairy Cattle

Genomic regions harboring recessive deleterious mutations responsible for mortality during gestation were detected in three dairy cattle breeds by identifying frequent haplotypes (>1%) with a deficit of homozygotes. Material consisted in Illumina 50k Beadchip genotypes of 76,177 animals (47,878 Holstein, 16,833 Montbeliarde, and 11,466 Normande). Forteen to 25 candidate regions (57 in total) were detected in each breed, including Brachyspina, CVM, HH1, and HH3 in Holstein breed. The two most frequent haplotypes (9 and 7%) were observed in Montbeliarde. A negative effect on calving rate, consistent in heifers and in lactating cows, was observed for 13 regions in matings between carrier bulls and daughters of carrier sires. Among these 13 regions, five new deleterious mutations were identified after sequencing the genome of heterozygous bull carriers and control animals (35 animals in total). In addition, deleterious mutations for 3 other regions without any confirmed effect on fertility were also found. All of them were located in coding sequences, were non synonymous and had very damaging predicted effects. Previously reported causative mutations for Brachyspina and CVM were found with this approach. Finally we showed that some causative mutations were in the neighborhood of - but outside - these non homozygous regions, due to incomplete linkage disequilibrium.