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Identification of six Mutations Responsible for Prenatal Mortality in Dairy Cattle

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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, six new deleterious mutations were identified in GART, SHBG, NOA1, SLC37A2, UPF1, and SMC2 (responsible for HH3), after sequencing the genome of heterozygous bull carriers and control animals (36 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. This study was funded by the ANR-10-GENM-0018 ANR-Apisgene Cartoseq project.