Abstracts

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(*CALD1*, $P = 1.2E{\times}10^{-7}$). Our findings will be the basis for the development of the genomic evaluation concept for udder health and milk quality traits for the Russian Holstein and Black-and-White cattle population. Supported by the Russian Scientific Foundation, project number 15-16-00020.

Key Words: GWAS, Holstein cattle, somatic cell score
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168 The distribution for LoF mutations in the FANCI, APAF1, SMC2, GART, and APOB genes of the Russian Holstein cattle population.

O. S. Romanenkova, V. V. Volkova, O. V. Kostyunina, E. A. Gladyr', E. N. Naryshkina, A. A. Sermyagin*, and N. A. Zinovieva, *L.K. Ernst Institute of Animal Husbandry, Moscow, Russian Federation.*

LoF (loss-of-function) mutations causing hereditary abnormalities and embryonic mortality are one of the reasons for a decline in fertility of cows. Currently, more than fifteen LoF mutations are known in Holsteins. In our work, we studied the frequency of LoF mutations in *FANCI*, *APAF1*, *SMC2*, *GART*, and *APOB* genes that are associated with fertility haplotypes HH0, HH1, HH3, HH4, and HCD in the Russian Holstein cattle population. We analyzed the estimated breeding values (EBVs) for daughters' milk production and reproduction traits in sires carrying LoF mutations compared to non-carrier sires using the BLUE approach. Genotyping LoF mutations was performed by PCR, PCR-RFLP or allele-specific PCR. In total, 1,521, 636, 880, 690, and 574 bulls and 896, 630, 773, 482, and 727 cows were genotyped for LoF mutations in *FANCI*, *APAF1*, *SMC2*, *GART*, and *APOB* genes, respectively. The ratio of carriers among bulls and cows was, respectively, 2.89 and 4.13% for *FANCI*, 2.04 and 1.83% for *APAF1*, 1.14 and 2.98% for *SMC2*, 1.30 and 1.04% for *GART*, and 5.57 and 2.06% for *APOB*. Generally, the sires carrying LoF mutations had higher EBVs for milk production traits. The greatest effect was observed for *SMC2* genotype: +236 kg for 305-days milk yield, 9.0 kg for fat yield, and 7.6 kg for protein yield. The bulls carrying LoF mutations in the *SMC2* and *GART* genes were characterized by the higher number semen doses per insemination (0.02 to 0.24 units) compared with the non-carrier bulls. The higher EBVs for the number of days open (from 12.6 ($P < 0.001$) to 7.2 days ($P < 0.05$)) and calving interval (from 9.0 ($P < 0.001$) to 10.5 days ($P < 0.01$)) were observed in bulls, which were carriers of LoF mutations in *SMC2* and *GART* genes. The group of sires with the mutation in the *FANCI* gene had the smallest length of calving interval (−6.0 days, $P < 0,001$). Analysis of the best linear unbiased estimates showed that for insemination of offspring from bull carriers of HH3 haplotype spent +0.026 units semen than for bulls with other mutations. The longest interval from calving to the first insemination and days open by +1.9 to 3.9 days were daughters of bulls with the *GART* gene (HH4). Given the dissemination of LoF mutations among the breeding sires and cows, with the aim of reducing genetically caused embryonic losses, matings of carriers bulls with mutations to cows whose fathers are hidden carriers of LoF mutations in the *FANCI*, *APAF1*, *SMC2*, *GART*, and *APOB* gene must be excluded.

Key Words: fertility, haplotype, LoF mutations
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169 A dairy calf DNA biobank for the discovery of new recessive genetic disorders.

J. B. Cole*, *Animal Genomics and Improvement Laboratory, Agricultural Research Service, United States Department of Agriculture, Beltsville, MD.*

This abstract describes the establishment of a new DNA biobank to support the discovery of new recessive genetic disorders in the U.S. dairy cattle population. High-density single-nucleotide polymorphism genotypes have recently been used to identify a number of novel recessive mutations that adversely affect fertility in dairy cattle, but the lack of consistent procedures for collecting DNA samples and supporting information for those projects underscores the need for a standardized process. The new Dairy Calf DNA BioBank in Beltsville, MD, is a repository for the collection and storage of samples. It complements the automated process of searching for new haplotypes that is part of the national dairy genetic evaluation system. The goal is to collect whole blood from calves that are born dead or that die shortly after birth, particularly if the calf appears to suffer from a congenital defect, as well as DNA from its dam and a sibling in the herd. Kits that include all sampling materials, a pre-paid return shipping label, a material transfer agreement, and a protocol for sample collection are shipped upon receipt of a request through the website. DNA providers are also able to provide substantial descriptive information about the calves for which they're providing blood, including digital photographs. When the samples are received in Beltsville, the DNA is extracted and stored for future analysis. The material transfer agreement ensures that a clear chain of permissions is available for every sample. If a pattern emerges to suggest that there is a new recessive genetic disorder in the population, the DNA is available for sequencing and causal variant discovery. Standardized protocols for DNA extraction and whole-genome sequencing will help ensure that data are of high quality. Information on carrier status for new recessives will be distributed through the Council on Dairy Cattle Breeding (Bowie, MD). The URL for the BioBank website is: http://aipl.arsusda.gov/BioBank/.

Key Words: dairy cattle, DNA bank, recessive disorders
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