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P0490: Cattle

Copy Number Variation in Dairy Cattle using Next-Generation Sequencing

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Gene copy number variants (CNV) have been shown to be associated with several production traits in dairy cattle; however, the detection and validation of CNVs in crossbred cattle is currently lacking. In order to provide a basis for future association studies, we sought to identify CNV regions (CNVRs) within the Girolando composite breed resulting from a mating of the Holstein (taurine) and Gir (indicine) breeds. A read depth method was performed using CNVnator software on NGS data from two Girolando, two Gir and ten Holstein bulls. The individual CNVs were merged into CNVRs based on genomic regions overlapping by at least 1 bp. In total, we identified a composite of 1,286 CNVRs (520 deletions, 255 duplications, 511 mixed) on the genomes of all samples. We observed 34 CNVRs (nine deletions, 25 mixed) in common (overlapping > 50%) only between Girolando and Holstein and 181 CNVRs (20 deletions, 21 duplications, 140 mixed) only in Girolando and Gir, suggesting parent-of-origin inheritance from Holstein and Gir cattle, respectively. One of these Holstein-specific CNVRs intersected with the interleukin 6 family cytokine (*LIF*) gene which is linked to fat production and fertility traits in Holstein. Genes related to disease resistance (e.g. the *CD4* gene) also coincided with CNVRs present only in Girolando cattle suggesting an indicine origin for the CNV. These results showed evidence of specific CNVRs shared by Girolando and purebred breeds which may be targeted for future selective breeding.

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P0491: Cattle

Genome-Wide Scan of Copy Number Variants (CNVs) in Valdostana Red Pied and Comparison with Italian Brown Swiss and Mexican Holstein

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Copy Number Variants (CNVs) are an important source of genomic structural variation. Many studies have focused on identified CNVs within and between populations, in livestock as well as in human, but only a few studies have explored population-genetic proprieties in cattle based on CNVs derived from the high-density SNP array. Here we report a high resolution CNV scan from Illumina's 777k BovineHD Beadchip data for the Valdostana Red Pied (VRP), an autochthonous Italian dual-purpose cattle population reared in the Alps that did not undergo strong selection for production traits. We perform a genetic comparison among the VPR, the Italian Brown Swiss (IBS 164 sires) and the Mexican Holstein (HOL 124 males and females) based on CNVs. In the VRP we identified a total of 6,784 CNVs that were summarized to 1,723 CNV regions (CNVR) on 29 autosomes covering a total of ~59 Mb of the UMD3.1 autosome. A total of 171 CNVRs were shared by all breeds, 474 were common to VRP and IBS, while 313 overlapped between VRP and HOL, indicating a more similar genetic background for the populations originating from the Alps. The PCA and the NJ tree showed a clear separation of the two Alpine breeds from the HOL. Vst statistics was calculated for CNV common regions among breeds identified in at least 5 individuals. Genes annotated in the CNVRs were compared to disclose diverging paths of selection among breeds.