Allele specific expression analysis in bovine muscle tissue

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Abstract

Imprinted genes have been target of many studies, mainly in human and mouse, and lately in bovines due to the interest of understanding the epigenetic mechanisms underlying important meat quality phenotypes and the possibility of applying it in animal breeding programs in the future. Genomic DNA from 146 steers was genotyped using the Illumina BovineHD BeadChip in order to identify heterozygous individuals with known allele origin. Total mRNA from muscle was extracted and sequenced by Illumina HiScanSQ. The software ALEA was used to create a diploid reference genome for each individual, in which haplotype regions were reconstructed from the individual haplotypes. ALEA also invokes BWA to map short sequencing reads to the in silico genome constructed and detects reads that are uniquely aligned only to one of the two haploid genomes. In-house software was developed to compute the frequency of reads mapped to each allele and to perform binomial statistical tests in order to identify allele specific expression. From 742,906 SNPs contained in the Illumina BovineHD BeadChip, 419 were assigned to be imprinted based on the following criteria: heterozygous in the individual, homozygous in its sires, at least 20x RNA-Seq coverage, and $p<0.05$ for the binomial statistical test. The Ensembl software VeP (Variant Effect Predictor) was used to determine the effect of these SNPs on genes, transcripts, and protein sequence, as well as regulatory regions. The VeP report together with phenotype information of the steers will be carefully examined in order to elucidate the molecular mechanisms involved with beef quality.