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179 Identification of Regulatory Mutations Affecting Complex Traits.

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Abstract: Understanding genetic bases controlling complex traits of economic importance is fundamental to improve breeding programs. Recent efforts from FAANG consortium resulted in significant progress in the identification of regulatory regions in farm animals. However, most studies in cattle have focused on *Bos taurus*, and limited information is available for *Bos indicus*. Considering that *Bos indicus* is the predominant subspecies in tropical regions, the objective of this work was to identify regulatory variants associated with meat quality by combining information from whole genome sequence (WGS), gene expression (RNA-seq), and open chromatin regions (ATAC-seq). For genome wide association study (GWAS), an imputed panel of SNPs was obtained from WGS of 26 sires, a Bovine HD 770K panel of 374 progenies, and SNPs called from RNA-seq data of Longissimus thoracis muscle (LT) of 192 progenies. The SNPs were identified using GATK4, and the dataset included almost 4.5 million variants that were pruned to 553,581 tag-SNPs (PLINK, r2 = 0.8). ATAC-seq data was obtained from duplicated samples of LT collected from two animals. Phenotype data included back fat thickness (BFT), intramuscular fat (IMF), ribeye area (REA) and shear force (SF). Association of genotypes (553,581 SNPs) and gene expression (12,991 genes), performed in Matrix eQTL resulted in 36,916 cis-eQTL and 14,408 trans-eQTL (FDR < 0.05). For GWAS analysis of the eQTLs with phenotypic traits, we used a linear model in PLINK program (considering population stratification, contemporary group and hot carcass weight). GWAS resulted in 24 eQTLs associated with IMF, three with BFT, 12 with REA, and eight with SF (FDR < 0.10). ATAC-seq analysis (using nfcore/atacseq pipeline implemented in Nextflow) resulted in 33,734 consensus peaks with a mean size of 2.4 kb. The overlap analyses among GWAS, eQTL, and ATAC-seq positions resulted in two, four and one putative regulatory regions that control gene expression and are associated with BFT, IMF and SF, respectively. Functional studies are still necessary to establish if identified mutations are the causal mutation. In conclusion, integration of GWAS, RNAseq, and ATAC-Seq data can result in identification of regulatory mutations associated with phenotypes of economic importance.

Keywords: ATAC-seq, GWAS, RNA-seq