Expression quantitative trait loci (eQTL) hotspot regions from whole genome analysis of Nellore steers

Aline S.M. Cesar[1], Luciana C.A. Regitano[2], Dante P.D. Lanna[1], Mirele D. Poleti[1], James E. Koltes[3], James M. Reecy[4], Eric Fritz-Waters[4], Luke Kramer[4], Dorian Garrick[4], Maurício A. Mudadu[2], Rymer R. Tullio[2] and Luiz L. Coutinho[1]

[1] Department of Animal Science, University of São Paulo, Piracicaba, SP, Brazil.
[2] Embrapa Southeast-Cattle Research Center, São Carlos, SP, Brazil.
[3] Department of Animal Science, University of Arkansas, Fayetteville, AR 72701, USA.
[4] Department of Animal Science, Iowa State University, Ames, IA 50011, USA.

Abstract: Detection of expression quantitative trait loci (eQTL) can help to explain how gene expression variation is associated with phenotypic variation. Such eQTL mapping studies allow for the identification of genomic regions that could be associated with gene expression regulation and might contribute to functional understanding of economically important traits in beef cattle. The aim of this study was to identify *cis* and *trans*-eQTL and discover eQTL hotspots (genetic marker directly or indirectly controlling a large amount of transcripts throughout the genome) across the bovine genome. Genotyping (BovineHD BeadChip) and RNA sequencing data from 193 samples of *Longissimus dorsi* muscle from Nellore steers were obtained using Illumina technologies. The Matrix eQTL statistical package from the R program was used to identify eQTL regions. It tests for association between genotype and gene expression levels using linear regression while controlling the false discovery rate (FDR 5%). Herein, 1,268 *cis*-eQTLs (distance ≤ 1 Mb) and 10,334 *trans*-eQTLs were identified across autosomal chromosomes. EQTL hotspots were identified by permutation test (p-value=0.05) at BTA3 (8Mb and 9Mb), BTA4 (108Mb), BTA11 (11Mb), BTA14 (73 Mb), BTA16 (59 Mb), BTA17 (55Mb) and BTA28 (20Mb and 32Mb), providing annotated genes harbored within these regions. Enrichment analysis of curated gene ontology terms was performed by DAVID v6.7 tool, which detected functional annotation clusters for transcription factor complex, transcription initiation, translational initiation, DNA binding and RNA processing. These findings provide new insight into the complex gene networks and genetic architecture that contributes to important economical traits in beef cattle.