SP 6946
2015
SP-PP-SP 6946

61° CONGRESSO BRASILEIRO DE GENÉTICA
8 a 11 de setembro de 2015 Hotel Monte Real Resort, Águas de Lindóia – SP
www.sbg.org.br – ISBN 978-85-89109-06-2

GENOME-WIDE IDENTIFICATION OF COPY NUMBER VARIATION REGIONS IN GIROLANDO CATTLE

Chud, TCS[1,4];Rosa, JO[1,5]; Silva, TBR[2,4]; Oliveira, GA[3,6]; Venturini, GC[1,4]; Baldi Rey, FS[1,7]; Munari, DP[1,7]; Silva, MVGB[2,7]

1-Univ Estadual Paulista Júlio de Mesquita Filho, UNESP, Jaboticabal, São Paulo, SP.
2-Embrapa Gado de Leite, Juiz de Fora, MG.
3-Universidade de São Paulo, FZEA/USP, Pirassununga, SP.

jaqueolrosa@hotmail.com

Keywords: genomic data, single nucleotide polymorphism, synthetic breed, DGAT1, high density panel

Genomic structural variants, such as copy number variation (CNV), have been revealed to be an important source of genetic and phenotypic variation for complex traits in livestock. The aim of this study was to identify CNV regions in Girolando (Gyr X Holstein) dairy cattle. The data set contained 205 animals genotyped with the Illumina BovineHD BeadChip SNP panel (~777,962 SNPs). An algorithm based on the Hidden Markov Model was implemented using PennCNV software for CNV identification. PennCNV perl script was used in order to eliminate calls from low quality samples, based on the standard deviation of LRR (LRR > 0.30), the BAF drift (BAF drift > 0.01) and waviness factor (WF > 0.05). The final data set was composed of 197 animals. The CNV regions (CNVRs) were identified using the CNVRuler program overlapping the CNVs. Gene content of cattle CNV was assessed using Ensembl genes. We used the PANTHER classification system to test the hypothesis (P < 0.05) that GO terms of the molecular function, biological process, and pathway terms were under or overrepresented in the CNVRs. We identified 1,924 CNVR along the genome, of which 56.86% were duplications and 43.14% were deletions. A total of 2180 genes were found within these regions and they are involved in biological processes, such as development (195 genes), biogenesis (95 genes), locomotion (4 genes), immune system (165 genes), metabolic process (582 genes) and reproduction (30 genes). A deletion region on chromosome 14 harbored the gene DGAT1, which is involved with milk production and intramuscular fat deposition in cattle. This study showed that the genes found in these CNVRs might be associated with the expression of production traits in crossbred dairy cattle.

Financial Support: [4]Capes/Embrapa; [5]Capes (Programa de Pós-Graduação em Genética e Melhoramento Animal); [6]FAPESP(Processo n. 13/12097-9), [7]CNPq, [8]Embrapa, [9]FAPEMIG

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