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Bloody RNA! Identification of blood transcriptional regulatory elements in the pig genome

Jové-Juncà Teodor[1], Crespo-Piazuelo Daniel[1], González-Rodríguez Olga[1], Hernández-Banqué Carles[1], Pascual Mariam[1], Ballester Maria[1], Quintanilla Raquel[1]

[1]Institut de Recerca i Tecnologia Agroàlimentaries, Torre Marimon, 08140, Caldes de Montbui, Spain
teodor.jove@irta.cat

The present work is part of the METAPIGEN project, whose objective is to characterize the genetic determinants of porcine immunometabolism and identify selection markers to improve immunocompetence in pigs. In the present study we aimed to identify regulatory elements in blood transcriptome by expression genome-wide association studies (eGWAS). Thus, blood samples of 255 animals from a commercial Duroc population were collected in Tempus™ blood RNA tubes at 60±8 days of age. Total RNA was sequenced on the Illumina NovaSeq6000 platform. Counts were calculated using RSEM/1.3.0 and normalized by trimmed mean M-values. Transcripts that did not show expression in at least 5% of the animals were removed. After filtering, 16,063 genes remained for further analysis. Genotyping of these animals was carried out with the GGPSNP70 commercial array and then imputed at whole genome level with Beagle using a multiple-breed reference population of 1,602 WGS data. A total of 9,739,308 polymorphisms remained, after filtering out those with minor allele frequency below 5% and more than 10% missing genotype data. eGWAS were conducted using GCTA on the normalized data and filtered imputed genotypes. After Bonferroni correction, 29,107,899 associations were found between 6,569,199 variants and the expression of 7,223 transcripts, resulting in 12,386 expression quantitative trait loci. A total of 4,469,388 (68.03%) variants were found in proximity (at 1Mb or less) to their associated transcript and were annotated as *cis*-regulatory elements. Furthermore, 605,069 variants were associated to 10 or more transcripts and were considered hotspot regulatory elements. The most significant association was found for *ACBD5* (p-value: 2.02472e-74). This gene encodes an acyl-CoA binding protein which is related to platelet formation and megakaryocyte differentiation. Our results identified key regulatory elements associated with gene expression in blood. Further analyses are being performed to study the impact of these regulatory elements in the variation of immunity traits.

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