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Unveiling blood transcriptome regulatory variants involved in pig immunity

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Abstract

The transcriptional profile of peripheral blood in swine is known to reflect variations in immune functions. In the present work, we aimed to characterize transcriptional regulatory variants associated with innate and adaptative immune functions and health-related traits. Expression genome-wide associations studies were performed between the expression of 16,063 genes obtained from whole-blood RNA sequencing and 9,739,308 imputed single nucleotide polymorphisms in 255 healthy animals of a commercial Duroc line aged 60 ± 8 days.

A total of 12,386 expression quantitative trait loci (eQTLs) for 6,449 genes were identified. Comparison between eQTLs and genomic regions already described as associated with immunity traits revealed 358 eQTLs overlapping with 15 QTLs for 12 immunological traits. The highest number of overlapped regions were found for the relative abundance of T-helper cells and memory T-helper cells. Within these regions, immune-related genes such as members of the *CLEC*, *IGKV* and *KLR* families, *RBPJ*, and *ZAP70* were identified. Additionally, 605,069 genetic variants were associated with the expression of ten or more different genes and were considered as potential regulatory hotspots. Particularly noteworthy was the top hotspot variant rs3475331335, which regulates genes involved in immune-related biological processes such as cytokine-mediated signalling pathway (28 genes), positive regulation of response to external stimuli (25 genes) or myeloid cell differentiation (22 genes), among others.

In conclusion, this study reported regulatory variants and candidate genes associated with immune- and health-related traits in pigs, shedding new light on the regulatory mechanisms modulating pathways involved in immune functions.

Keywords

Pig, Transcriptome, Immunocompetence