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CONSTRUINDO SABERES, FORMANDO PESSOAS E TRANSFORMANDO A PRODUÇÃO ANIMAL

TRANSCRIPTOME ANALYSIS IDENTIFIES GENES RELATED TO UMBILICAL HERNIA IN PIGS

Mayla Regina SOUZA*[1], Adriana Mercia Guaratini IBELLI[2], Jane de Oliveira PEIXOTO[2], Marcos Antônio Zanella MORES[2], Maurício Egídio CANTAO[2], Mônica Corrêa LEDUR[1, 2]

*corresponding author: souza_mayla@hotmail.com
[1]Universidade do Estado de Santa Catarina, Chapecó, Santa Catarina, Brasil
[2] Embrapa Suínos e Aves, Concórdia, Santa Catarina, Brasil

Umbilical hernias have a great impact on animal welfare, growth and feed conversion, negatively affecting the livestock production. In the last years, studies have been developed to identify chromosomal regions related to this condition to minimize the occurrence of hernias. However, its frequency is still an issue in the herds and its etiology remains unclear. Therefore, this study aimed to identify genes related to umbilical hernia in pigs using transcriptome analysis. Thus, 10 females, 5 normal and 5 affected with umbilical hernias, were used. The normal females were chosen from families with no history of hernia. The herniary tissues were collected and the total RNA extracted and submitted to sequencing in a HiSeq2500 Illumina (2x100bp). After, the sequences quality control (QC) was performed with Seqyclean, reads were mapped against the swine genome v11.1 using STAR and features were counted using HTSeq-count. Functional analyses using DAVID and Panther databases were performed to identify the biological processes involved in the development of umbilical hernia. The differential expression analysis was performed with EdgeR. An average of 24 million reads/sample was obtained after QC, comprising 11,557 genes expressed in the herniary tissue. A total of 231 genes was differentially expressed (DE) between the control and affected groups (FDR < 0.05), being 147 downregulated and 84 upregulated in the affected compared to the control group. According to the functional analysis of the DE genes, 145 genes were recognized in biological processes on DAVID, while 166 in the Panther database. *In silico* analysis showed that a large set of genes was related to response to immune systems, cell apoptosis and extracellular matrix. In addition, 8 DE genes: *CCBE1, LGALS3, ACER2, SIT1, PTGS1, SLC2A6, KATNBL1* and *RYR3* were located in QTL regions previously associated to umbilical hernia in pigs. The expression profile obtained in this study shows possible inflammation in the herniary tissue of affected pigs, pattern that has also been described in humans, which could be a consequence of the herniary process. However, some of the DE genes, such as *CCBE1,* present functions on extracellular matrix remodeling and migration, and mutations in this gene were associated to umbilical hernia predisposition in humans. Therefore, the *CCBE1* might be a candidate gene to cause this condition in pigs. More studies are necessary to confirm the involvement of these genes in the occurrence of umbilical hernia in pigs, improving the understanding of this condition in swine and other species.

Keywords: RNA-Seq, gene expression, sequencing, swine

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