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VARIANTS RELATED TO FEMORAL HEAD SEPARATION IDENTIFIED IN THE CHICKEN TRANSCRIPTOME

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ABSTRACT

The femoral head separation (FHS) is a condition characterized by the detachment of the articular cartilage from bone, being closely related to femoral head necrosis (FHN), one of the main locomotor problems in poultry production. This study aimed to identify variants related to FHS in chickens using RNA sequencing. To this, 8 normal and 8 FHS-affected transcriptomes from broilers femoral articular cartilage (AC) and growth plate (GP) were analyzed. Reads were submitted to quality control and mapped against the chicken genome (GRCg6a) using the two-pass mode in the STAR software. The Genome Analysis Tool kit 3.6 (GATK) was used for polymorphism identification and the Variant Effect Predictor (VEP) Ensembl tool to annotate and verify the variant effects. A total of 89,823 variants (SNPs and InDels) was identified in the analyzed samples, in which 89 differed between the normal and FHS-affected groups, located in 129 gene regions. Approximately 3% of all polymorphisms were firstly described in the current study. Most of the variants were classified as downstream, synonymous, upstream, intronic, 3' prime UTR and missense variants. Out of coding variants, 90% (24) were classified as synonymous and 10% (4) as missense. Considering the four missense variants, three were previously described in the *PRKAB1*, *SPATS2L* and T*RIM25* genes, while a novel SNP in the *SND1* was predicted as deleterious. Furthermore, SNPs were found in three genes that have already been differentially expressed in the GP between normal and FHS-broilers (*LMO2, LOXL2* and *SLC26A2).* Variants involved with FHS in chickens were described in this study and their effects should be further investigated in larger datasets to validate them as genetic markers for bone integrity in broilers.

KEYWORDS: RNA Sequencing; SNP; femoral head necrosis

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