



An insertion in Caracu breed genome with a possible role in fatty acids profile

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The Caracu cattle is a Brazilian locally adapted breed known to have a good lipid composition and variable fatty acids (FA) composition in carcass. Beef is a natural source of essential FA, being the intramuscular and subcutaneous adipose tissues the most important fat deposits, and thus, associated also with meat quality traits in cattle. Among the genes already identified to be associated with FA profile, the *PTGS1* have being observed to be related with FA composition in human and cattle. We aimed to *in silico* evaluate its promoter region in Caracu cattle breed to identify possible functional variants affecting the binding of transcription factors (TF). First, whole-genome re-sequencing data from 12 animals was used. The sequencing, mapping and variant calling was performed according the 1000bulls project gold standard pipeline from which, in average, we obtained 387,156,655.1 reads with 99.65% mapped in the reference genome and a coverage of 13.04 times. Insertions and deletions (InDels) were identified based on ARS-UCD1.2 bovine genome assembly. Those variants were then classified according to their potential function (e.g. 5' UTR variant or upstream gene variance) using the Ensembl Variant Effect Predictor tool (VEP, version 84). Thus, we retrieved all InDels in the promoter sequence (3,000 bp upstream and 300 bp downstream from the transcription start site) of *PTGS1* (XM_024998322.1). FASTA files with the wild type sequence of promoter region and sequences with each identified InDel were used as input in TFM-explorer program to search for locally overrepresented TF binding sites (TFBS) using weight matrices from the JASPAR vertebrate database. A total of 4 known InDels were observed, and thus used to retrieve the FASTA sequences of the promoter regions of *PTGS1* gene containing those variants. From the promoter sequences with the identified InDels, one with an insertion of 11 base pairs between the nucleotides 93238092 and 93238093 (rs380183063) promoted the binding of *AR* transcription factor. *AR* is an androgen receptor transcription factor and studies suggest that *AR* signalling affect fatty acyl-CoAs biosynthesis in human. Besides, studies in mouse suggest a possible role in regulate fat mass. In this study, we observed an insertion in the promoter sequence of *PTGS1* gene, more precisely, in a position promoting a TFBS for *AR*. Since both *PTGS1* and *AR* have been cited to be involved in FA metabolism, we suggest that this insertion may be used for further *in vitro* and *in vivo* analyses in terms to confirm a possible role in FA profile of Caracu cattle.

Keywords: Sequencing, Indels, Genome, Transcription factors, meat quality.

Acknowledgments: We would like to thank the FAPEMIG (APQ-01834-18) and INCT-CA for the financial support.