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INTRODUCTION/OBJECTIVES

THE *CYP21* (STEROID 21-HYDROXYLASE GENE) IS INVOLVED IN THE SYNTHESIS OF STEROID HORMONES AND ITS MUTATIONS ARE ASSOCIATED WITH SEVERAL METABOLIC DISORDERS SUCH AS CONGENITAL ADRENAL HYPERPLASIA IN HUMANS. THE PROMOTER REGION OF THE BOVINE *CYP21* PRESENTS A SHORT INTERSPERSED NUCLEOTIDE ELEMENT (SINE) OF THE BOV-A2 WHICH OVERLAPS A PUTATIVE SP1 BINDING SITE. THE MAIN AIM OF THIS STUDY WAS TO VERIFY THE OCCURRENCE OF A *HpaII* POLYMORPHISM IN THIS SPECIFIC BOV-A2 ELEMENT IN ZEBU BREEDS, AND TO VERIFY WHETHER THE POLYMORPHIC NUCLEOTIDE IS ASSOCIATED WITH A METHYLATION POINT AT THE BOVINE *CYP21* PROMOTER.

MATERIALS AND METHODS



GIR - N = 66

GUZERÁ - N = 41

NELORE - N = 26

RESULTS

THE GENOTYPIC FREQUENCY OBSERVED IN THE DIGESTION WITH ENZYME *HpaII* OF PCR PRODUCTS OF DNA SAMPLES WERE 0.06 (CC), 0.51 (TT) AND 0.44 (CT). THE METHYLATION ANALYSIS SHOWING THAT THE DINUCLEOTIDE CPG AT THE POLYMORPHIC SITE IS METHYLATED AS IT IS PRESENT IN THE SEQUENCE.

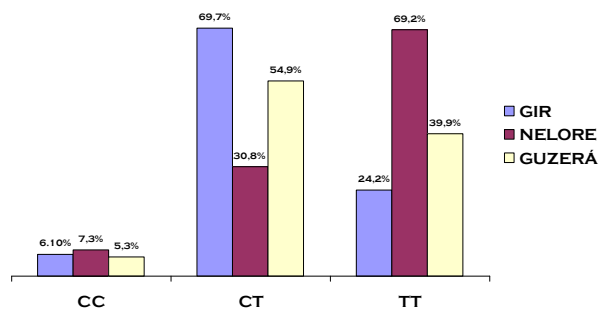


FIGURE 1. GENOTYPIC FREQUENCY PERCENTAGE (%) OF THE BOV-A2/*HpaII* POLYMORPHISM IN GIR, GUZERÁ AND NELORE BREEDS.

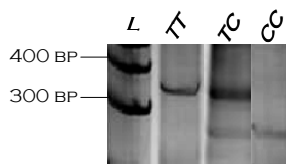


FIGURE 2. -ACRYLAMIDE GELS SHOWS POLYMORPHISM PROFILE WITH THE DIGESTION OF PCR PRODUCTS WITH ENZYME *HpaII*. L: 100BP DNA MARKER.

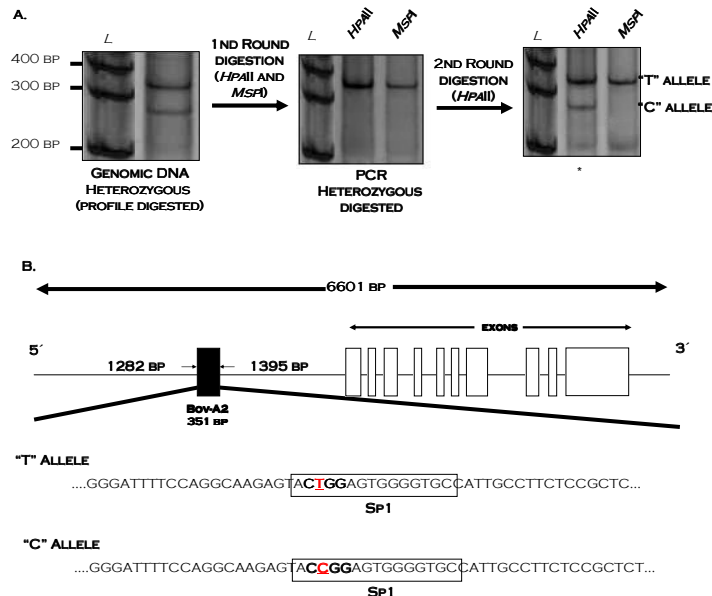


FIGURE 3 – A. ALLELE SPECIFIC METHYLATION ASSAY: PAGE RESULTS SHOWING THE METHYLATION PROFILES OF HETEROZYGOES. MAINTENANCE OF THE TWO ALLELES IN THE WELL FOR *HpaII** DIGESTION (A METHYLATION SENSITIVE ENDONUCLEASE) AND NO DETECTABLE PRESENCE OF THE "C" (METHYLATED) ALLELE AT THE *MspI* (A METHYLATION NOT-SENSITIVE ENDONUCLEASE). L: 100BP DNA MARKER. **B.** SCHEMATIC REPRESENTATION OF THE SNP (RED) OVERLAPPING A PUTATIVE SP1 TRANSCRIPTION FACTOR BINDING SITE (OPEN BOX). ([WWW.GENEREGULATION.COM/PUB/PROGRAMS/ALIBABA2](http://www.generegulation.com/pub/programs/alibaba2); [HTTP://TFBIND.JMS.U-TOKYO.AC.JP/](http://tfbind.jms.u-tokyo.ac.jp/))

CONCLUSIONS AND PERSPECTIVES

- THE FREQUENCIES (HOMOZYGOSE AND HETEROZYGOSE) OF *HpaII*/BOVA2 POLYMORPHISM IN BRAZILIAN ZEBU BREEDS IS THE FIRST ASSOCIATION OF THE GENETIC VARIABILITY OF THIS LOCUS AS A POSSIBLE SOURCE OF EPIGENETIC CHANGE AT THE SAME GENOMIC POSITION.
- THE GENETIC AND EPIGENETIC FEATURES OF THIS SNP COULD CONTRIBUTE TO A POSSIBLE FUNCTIONAL EFFECT ON THE REGULATION OF THE BOVINE *CYP21* GENE.
- FUTURE FUNCTIONAL STUDIES ON THIS REGION SHOULD CLARIFY IF THIS T>C POLYMORPHISM HAS SOME FUNCTIONAL EFFECT AND IF THERE IS SOME CORRELATION WITH THE HIGHER INCIDENCE OF THE T ALLELE.

FINANCIAL SUPPORT:

