

Prospection of polymorphisms in the MSTN gene in Bonsmara cattle

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Double muscle syndrome (DM) is observed in different cattle breeds and is characterized by muscular hypertrophy, easily recognized by the increase in muscles, especially in the hindguarter region. DM manifests itself when mutations occur in the MSTN (Myostatin) or GDF-8 (Growth Differentiation Factor - 8) gene, responsible for encoding the myostatin protein, a negative regulator of muscle development. Thus, the objective was to prospect for polymorphisms in the MSTN gene through sequencing, aiming to develop a DNA test that allows the early identification of Bonsmara animals that carry the mutated allele. For this, biological samples were collected from families in which the DM phenotype was segregating, based only on visual observations. For seguencing, eight pairs of primers were used and the electropherograms generated were edited and analyzed using the Unipro UGENE Software (version 49.1). The sequences obtained were aligned against the reference sequence of the bovine genome and the consensus sequences were compared to identify mutations. In exon 2, the Q204X mutation was found, a disruptive mutation caused by a transition from cytosine to thymine at nucleotide 610. This mutation produces a stop codon in the N-terminal latency-associated peptide at amino acid position 204, thus inactivating the myostatin protein and causing muscle hypertrophy in some breeds of cattle such as Charolais, Limousin and Blonde d'Aquitaine. To develop the DNA test four pairs of specific primers were synthesized to detect this mutation. These primers were used to genotype 818 animals using the ARMS-PCR (Amplification-refractory mutation system-Polymerase Chain Reaction) technique. The results obtained showed that in Bonsmara breed the presence of at least one Q204X mutated allele is associated with an increase in calf birth weight, muscularity and harmony score, with harmony being the main phenotypic trait used by producers to select animals. Based on this information, the process of selecting the bulls that will be the parents of the next generations will be more efficient, facilitating the task of either introducing or eliminating the mutated allele in the herds, according to the selection objectives of each producer.

Keywords: ARMS-PCR, *GDF-8* gene, muscle hypertrophy, genetic improvement, myostatin, mutation.

Acknowledgments: Cabaña Bella Alianza/PY, CNPq/PIBIC, Fundapam and UFMS.