

R.A.

PATV



Evaluation of DUMPS allele incidence in Girolando bulls participating on Girolando Progeny Test

Pinto, ISB¹; Motta, IGB²; Pereira, GRS³, Almeida, FA³; Pereira, WP², Rettore, JVP⁴, Fonseca, I⁵; Arbex, WA⁶; Silva, MVGB⁶; Martins, MF⁶.

¹Bolsista de Apoio Técnico, FAPEMIG, ²Centro de Ensino Superior de Juiz de Fora, Juiz de Fora, MG, ³Universidade Presidente Antônio Carlos, UNIPAC, Juiz de Fora, MG, ⁴Universidade Federal de Juiz de Fora, Juiz de Fora, MG, ⁵Universidade Federal de Viçosa, UFV, Viçosa, MG, ⁶Embrapa Gado de Leite, Juiz de Fora, MG
isabellajf@gmail.com

Keywords: breeding program, hereditary diseases, PCR-RFLP, DUMPS.

Deficiency of Uridine Monophosphate Synthase (DUMPS), is one of the most studied recessive hereditary diseases affecting cattle. DUMPS is characterized by a nonsense mutation (cytosine by thymine) at codon 405 of the enzyme Uridine Monophosphate Synthase (UMPS) gene. Such enzyme is responsible for the conversion of orotic acid onto uridine monophosphate, thus being part of an essential via on pyrimidines synthesis. Once great amounts of pyrimidines are needed during embryonic development in order to synthesize nucleic acids, homozygous embryos for the studied gene usually die around the 40^o day of life. Heterozygous individuals show a normal phenotype, however showing only half of the activity of UMPS enzyme. Genotyping of cattle is needed to identify unfavorable alleles and to show the incidence of hereditary diseases in herds. On present work, 123 bulls of Girolando breed, were genotyped with the aim of assess DUMPS incidence on Brazilian herds participating on such breeding program. For that, DNA was extracted from blood and semen samples using DNeasy Blood and Tissue Kit (Qiagen, Hilden, Alemanha), quantified and valuated by spectrophotometry (Nanodrop®, Wilmington, DE, EUA). The identification of carriers was realized by PCR-RFLP technique. The amplified product was digested with *Ava* I restriction enzyme (Promega Corporation). Genotyping of the animals was performed analyzing the pattern of generated fragments, being 53 and 36 bp for normal homozygous animals (TD) and 89, 53 and 36 bp for carriers (DP). Allelic and genotypic frequencies were estimated using GENEPOP software web version 1.32 and the probability of Hardy-Weinberg Equilibrium, associated with the observed genotypic frequencies was tested by means of χ^2 test ($p < 0,05$). The frequency of TD genotype found in the studied population was 100%, thus showing that was no identification of bulls carrying DUMPS allele. These frequencies analysis showed that such population founds itself under Hardy-Weinberg Equilibrium at 5% of probability. This result may indicate that the normal allele is being fixed in this population, while DUMPS allele is being excluded. On the other hand, absence of the lethal allele may have been due to the size of analyzed sample, which was of 123 animals. By knowing the alleles that cause such disease and genotyping the animals participating on breeding programs, bulls that carries DUMPS allele have been excluded from artificial insemination programs, and that so the incidence of such disease in herds is being decreased. Financial Support: EMBRAPA/AGROFUTURO/FAPEMIG

SP 5296
P. 167