

# Beta-lactoglobulin polymorphism in Girgentana goat breed

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## ABSTRACT

Beta-lactoglobulin ( $\beta$ -lg) is a globular protein belonging to the lipocalin family. It is the major whey protein in the milk of ruminants. It is also present in the milk of most mammals but is lacking in rodents, lagomorphs and humans. A large number of variants have been reported for cow and sheep milk. Several studies have shown association between  $\beta$ -lg variants and milk production and composition, even if the results are not always concordant. In goat, no  $\beta$ -lg variants related with amino acid change have been characterized at DNA level, but some authors described the presence of polymorphisms in the 3'UTR and in the proximal promoter region. Mutations in the promoter region could be those most likely responsible for different level of gene expression. The aim of this work was to study the genetic polymorphism at DNA level of  $\beta$ -lg gene in Girgentana goat breed. A total of 238 genomic DNA samples of Girgentana breed were genotyped. A fragment of 709 bp, including 588 bp of proximal promoter region and 121 bp of exon 1, was amplified using primers GOAPF3 and GoatE1R2. PCR-RFLP procedure was used for fast detection of two single nucleotide substitutions as described by Graziano *et al.* (2003). The base substitutions originating the polymorphic sites consist of: 1. a transition T:C at position -341 and 2. a transition C:T at position -60. A FspBI PCR-RFLP protocol was used to detect the mutation -341 (T/C) and a SmaI PCR-RFLP protocol for the mutation -60 (C/T) of the proximal promoter region. The allelic frequencies and the Hardy-Weinberg equilibrium were estimated using the GENEPOP software. Girgentana goat breed shows no significant deviation from Hardy-Weinberg equilibrium for the allele frequencies found in both polymorphic sites considered. The genotypic frequencies for both mutations resulted in 0.65 (T/T), 0.33 (T/C) and 0.02 (C/C) for the position -341, and 0.82 (C/C), 0.17 (C/T) and 0.01 (T/T) for the position -60. These results are in agreement with the previous obtained by Graziano *et al.* (2003) in the same breed. Further analysis are in progress to investigate the possible effect of these variants on the expression of  $\beta$ -lg gene, on the milk protein composition and on milk production traits.

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