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# Analysis of 22 mutations within milk protein genes in Italian Friesian cattle

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## ABSTRACT

The bovine milk protein caseins,  $\alpha_{s1}$ -CN,  $\beta$ -CN,  $\alpha_{s2}$ -CN, and  $\kappa$ -CN are codified by four well characterized genes, named *CSN1S1*, *CSN2*, *CSN1S2*, and *CSN3* respectively and clustered in a region of 250-kb of chromosome 6. A recent revision of milk protein nomenclature considering only protein polymorphisms indicates 8  $\alpha_{s1}$ -CN, 4  $\alpha_{s2}$ -CN, 12  $\beta$ -CN, and 11  $\kappa$ -CN variants within the genus *Bos*. Other mutations were found in the non-coding regions of the cluster, such as the promoter regions or the 3'UTR.

Many of these polymorphisms, together with others in various genes, such as the one coding for  $\beta$ -lactoglobulin (*LGB*), show important associations with different milk quality traits. Analyzing all these polymorphisms could help clarify the role of both the casein haplotype and the other polymorphisms in milk composition and cheese-making properties, and could explain which polymorphisms are really or mostly involved. The mPCR-LDR-UA approach recently developed to test simultaneously 22 SNPs in DNA regions responsible for milk protein expression was used to type 250 Italian Friesian cattle. In perfect agreement with literature, the most frequent alleles were *CSN1S1*\*B, *CSN2*\*A<sup>2</sup>, *CSN3*\*A, variant 2 of *CSN1S1* promoter, and variant A of Bov-A2 element.

A quite balanced frequency was observed for the *LGB*\*A and *LGB*\*B. No *CSN2*\*C, *CSN3*\*C, and *CSN3*\*H alleles were found. The *CSN1S1*\*C, *CSN2*\*A<sup>3</sup>, *CSN2*\*I alleles were detected only at the heterozygous condition and at a frequency lower than 2%. The method allowed also finding some unusual intragenic haplotype, such as the Bov-A2 element-*CSN3* haplotypes A-B and B-E. As to *LGB* one of the four SNPs tested was always homozygous for the same mutation, as already noticed.

This finding confirms that this synonymous SNP is probably a sequencing mistake or a rare mutation not decisive for the *LGB* typing in the Italian Friesian. Reducing cost and time for typing simultaneously many SNPs, the method will be applied to a greater number of individuals and to other breeds, aiming to find out a number of animals for each haplotype sufficient for accurate statistical analysis to give a better understanding of the significance of milk protein polymorphism.