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ANALYSIS OF ALLELE FREQUENCIES OF THE GROWTH HORMONE RECEPTOR (*GHR*) F279Y MUTATION IN SEVERAL CATTLE BREEDS

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Several studies have indicated that bovine chromosome 20 (BTA20) harbors quantitative trait loci (QTL) for milk yield and composition. A combined linkage and linkage disequilibrium analysis positioned a major QTL in the middle of BTA20 and, for its map position, the growth hormone receptor (*GHR*) gene was considered a strong positional and functional candidate gene, knowing the major role played by the growth hormone axis in the initiation and maintenance of lactation. Analysis of polymorphisms in this gene identified a mutation in exon VIII (a T>A transversion, that causes the replacement of an amino acid, a phenylalanine to tyrosine substitution, in a highly conserved transmembrane domain of the GHR protein at position 279), indicated as F279Y, with highly significant effects mainly on milk yield, protein percentage and fat percentage (Blott *et al.*, Genetics 163:253-266; 2003). Allele F increases the percentage of protein and fat in the milk while has a negative effect on milk yield. On the other hand, allele Y has a positive effect on milk yield and a negative effect on protein and fat percentage. Moreover, these effects seem consistent across populations and breeds.

As a first step in order to further evaluate the effects of the F279Y polymorphism and to investigate its possible use in marker assisted selection (MAS) plans in dairy and dual purpose cattle breeds reared in Italy we studied the distribution of these two alleles at the *GHR* locus in a total of 679 animals (Italian Holstein-Friesian, 108; Italian Brown, 104; Italian Simmental, 104; Jersey, 104; Reggiana, 108; Modenese, 66; Rendena, 85). DNA was extracted from semen, blood, milk or hair bulbs. A new PCR-RFLP method was set up to analyse the T>A point mutation of exon VIII of the *GHR* gene inserting an artificial restriction site by means of a mismatched forward primer. The genotypes obtained from a few animals were confirmed by sequencing of the region containing the polymorphism.

In all breeds investigated allele F was the most frequent and ranged from ~0.95 (Italian Brown and Jersey) to ~0.73 (Italian Holstein-Friesian). In Reggiana and Modenese it was ~0.92 while in Rendena was ~0.82. Frequency of the FF genotype varied between breeds ranging from ~0.89 (Italian Brown and Jersey) to ~0.51 (Italian Holstein-Friesian). Genotype FY was observed with the highest frequency in Italian Holstein-Friesian (~0.43) and the lowest in Reggiana (~0.08). A few animals with genotype YY were identified in Italian Holstein (~0.06), Reggiana and Rendena (~0.04) and Italian Simmental (~0.01). For all breeds no significant deviation from the Hardy-Weinberg equilibrium was observed for this polymorphism.

From these data, it is interesting to note the highest presence of allele Y in the Italian Holstein-Friesian breed compared to the other breeds. This may be due to the fact that this allele, that is suggested to have a positive effect on milk yield, could have been indirectly selected because the selection strategies in this breed have been mainly towards an increase in the level of milk production for many years.

Further studies are needed to confirm the effects of this polymorphism in the investigated breeds and to evaluate its application in MAS programs.