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Mutations in the bovine prolactin receptor (PRLR) gene: allele and haplotype frequencies in the Reggiana cattle breed

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ABSTRACT

Prolactin receptor (PRLR) is a member of the cytokine receptor superfamily. PRLR exerts its functions binding three types of ligands (prolactin, placental lactogen and growth hormone) involved in a large number of biological processes, like development, fetal growth, pregnancy, lactation and response to stressors. In mice and pigs, mutations in the *PRLR* gene have been indicated to affect mammary gland development or were associated with litter size and reproduction traits. The bovine *PRLR* gene has been assigned to chromosome 20, a few cM from the growth hormone receptor (*GHR*) gene, where several QTL for milk production traits have been mapped in a few dairy breeds. Recently, a missense mutation identified in exon 3 of this gene (*S18N*, named according to the amino acid exchange) has been associated to milk, fat and protein yield in Finnish Ayrshire. Another missense mutation (*L186P*) in exon 7 of this locus has been identified by us and others. Here we analysed these two polymorphisms in the Reggiana cattle breed as a first step of association studies between the *PRLR* locus and production traits in this breed. DNA was extracted from semen that was possible to collect from 124 Reggiana sires. Two PCR-RFLP protocols were set up to analyse the mutations mentioned above. Sequencing of amplified fragments was carried out to confirm the results of the PCR-RFLP genotyping methods. At the *S18N* missense mutation, allele *N*, that was suggested to positively affect milk, protein and fat yield, was the less frequent (0.39). At the *L186P* mutation, allele *L* was observed with the higher frequency (0.75). For 102 animals it was possible to infer the haplotype structure using only the genotype information. All four possible haplotypes were deduced. The most and less frequent ones were *S18-L186* (0.44) and *N18-P186* (0.06), respectively. The other two haplotypes (*N18-L186* and *S18-P186*) showed a frequency of 0.32 and 0.18, respectively.