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Analysis of the melanocortin receptor 1 (*MC1R*) gene in Sicilian goat breeds

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ABSTRACT

Mammalian coat colour is mainly determined by the distribution of two different types of melanins: pheomelanin (red/yellow pigments) and eumelanin (black pigments). Their synthesis is regulated by the melanocortin 1 receptor (*MC1R/Extension* locus) that binds the α -melanocyte-stimulating hormone (α -MSH) and the agouti signalling protein (ASIP, coded by the *Agouti* locus). In mammals, several studies have reported that loss-of-function mutations in *MC1R* lead to red/yellow pigmentation, while gain-of-function mutations lead to black/dark colours. Mutations at the *Agouti* locus exert, in general, epistatic interactions on the *Extension* locus. In goats, classical genetic studies have indicated that variations at the *Agouti* locus may be the main source of colour variability within and between breeds, while the effect of the *Extension* locus on this phenotypic trait has been only partially deduced. In order to better understand the role of the *Extension* locus on coat colour in this species, here we analysed the *MC1R* gene in three Sicilian goat breeds showing differences for this phenotype: Derivata di Siria known also as Rossa Mediterranea (solid red colour), Girgentana (cream/light grey) and Maltese (white with black spotted head). Fragments of the *MC1R* gene encompassing the 5'-untranslated region (UTR), the coding sequence and the 3'-UTR were sequenced in 10 goats for each breed. Sequence analysis showed the presence of five single nucleotide polymorphisms located in the coding region: one nonsense mutation (Q225X), three missense mutations (A81V, F250V and C267W) and one silent mutation at codon 43. The stop codon at position 225 may create a shorter *MC1R* protein whose functionality could be altered. All Girgentana goats carried this nonsense mutation that was also present in three Derivata di Siria animals. Further studies are underway to evaluate the effect of the identified mutations on coat colours and, eventually, apply them in breed traceability systems of goat products.