

BOOK OF ABSTRACTS

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Mutations at the caprine melanocortin 1 receptor gene are associated with coat color in Spanish goats

Zidi, A., B. Badaoui, A. Manunza, J.M. Serradilla, J. Capote, B. Urrutia, J. Carrizosa, F. Pilla, M.S. D'Andrea, J. Jordana, A. Ferrando, A. Pons, A. Sánchez, V. Landi, M. Gómez, A. Martínez, O. Vidal, M. Amills

Departament de Genètica Animal, Centre de Recerca en Agrigenòmica (CRAG), Universitat Autònoma de Barcelona, Bellaterra 08193, Spain.

Genetics of coat color in goats is not well understood. A recent survey involving the melanocortin receptor 1 (MC1R) gene revealed that black pigmentation of Murciano-Granadina goats is associated with a Trp residue at position 267, while caoba goats are homozygous for the Cys allele. Similarly, copy number variation at the agouti signaling protein (ASIP) gene has been associated with a white coat in Saanen and Girgentana breeds. Other associations have been reported, but in general they are difficult to replicate across breeds. Herewith, we have sequenced six pigmentation genes (MC1R, ASIP, TYRP1, TYRP2, KIT and TYR) and identified 21 single nucleotide polymorphisms (SNP). Genotyping of thirteen of these SNP in 590 goats from the Iberian Peninsula, Canary Islands, Italy, Morocco and Tunisia has revealed the existence of a phylogeographic pattern i.e. individuals tend to cluster according to their geographic location rather than their coat color. Principal Component Analysis of the data showed that Canarian breeds group with their North African counterparts, a feature that is consistent with historical and linguistic data. As a whole, these results suggest the existence of a marked genetic heterogeneity and low selection pressure for phenotypes related with pigmentation in goats. Besides, we have investigated the segregation of MC1R alleles at the within breed level (Murciano-Granadina, Malagueña and Canarian breeds). Photographic records were obtained to document the pigmentation status of each goat and MC1R genotypes were determined by sequencing. This approach has allowed to confirm the causality of the Cys to Trp mutation for the black/caoba coat in the Murciano-Granadina breed. We have also found two additional causal mutations that explain the inheritance of the red vs. black coat in the Canarian Palmera breed and of the red vs. blonde colour in Malagueña goats.