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## Genetic analysis of six pigmentation genes in european and north african goat populations

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We have characterized the genetic variability of the caprine melanocortin 1 receptor (MC1R), agouti signaling protein (ASIP), tyrosinase (TYR), v-kit Hardy-Zuckerman 4 feline sarcoma viral oncogene homolog (KIT) and tyrosinase-related proteins 1 and 2 (TYRP1 and TYRP2) genes. A total of 21 single nucleotide polymorphisms (SNP) were found, being distributed as follows: MC1R (7 SNP, 5 missense and 1 non-sense), ASIP (4 SNP, 2 missense), TYR (2 SNP, 1 missense), KIT (3 SNP, 1 missense), TYRP1 (2 SNP) and TYRP2 (3 SNP, 1 missense). Thirteen of these SNP have been selected to perform the genotyping of 590 goats from the Iberian Peninsula (Murciano-Granadina, Malagueña, Rasquera and Azpi-Gorri), Balearic Islands (Ibizan), Canary Islands (Majorera, Tinerfeña and Palmera) Italy (Cilentana, Garganica, Derivata di Siria, Girgentana, Jonica, Maltese), Morocco and Tunisia (local breeds) as well as two unrelated breeds (Cashmere and Saanen). Filtering of non-neutral SNP left 10 valid markers that were employed to perform population studies. Principal component analysis based on Reynolds distances as well as STRUCTURE and factorial correspondence analyses consistently showed that Canarian and North African goats are closely related. This agrees very well with linguistic and genetic evidences demonstrating that the pre-Hispanic settlers of the Canary Islands had a Berber ancestry. In contrast, the high  $F_{ST}$  value ( $F_{ST} = 0.27$ ) observed between North African and Iberian goat populations might suggest the absence of a significant gene flow between these two geographical locations, although additional analyses need to be performed to confirm this hypothesis.

# GENETIC ANALYSIS OF SIX PIGMENTATION GENES IN EUROPEAN AND NORTH AFRICAN GOAT POPULATIONS

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## Introduction

- Demographic history of livestock populations can be inferred from the analysis of genetic data. In goats, studies based on autosomal genetic markers have been able to detect population structure with more efficiency than those based on mitochondrial DNA.
- Herewith, we aimed to investigate the genetic relationships between goat populations from the Iberian Peninsula and Balearic Islands, Canary Islands, Italy and North Africa by using an array of single nucleotide polymorphisms (SNP) located at six pigmentation genes. Two additional breeds (Cashmere and Saanen) were used as “outgroups” in population analyses.

## Experimental design

### • Animal material:

590 goats from 18 breeds



### • Polymorphism identification and typing:

- Six pigmentation genes (see **Table 1**) were partially sequenced and polymorphic sites were identified.
- A Sequenom Mass ARRAY iPLEX device was used to type 13 SNP.

### • Population analyses:

- Reynolds genetic distances were calculated with Arlequin 3.5. GenAIX 6.3 provided a graphical summary, as a PCA plot, of the resulting distance matrix.
- Population structure was analysed with Structure v2.1.

## Results and Discussion

### • Variability of six goat pigmentation genes:

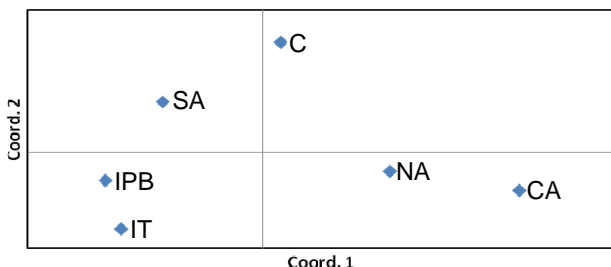
A total of 21 SNP were found in the six analysed genes (**Table 1**). The melanocortin receptor 1 gene was the one that displayed higher levels of genetic variability. Most of *MC1R* polymorphisms were non-synonymous (**Nsyn**) and *in silico* tools predicted that four of them might be functionally relevant.

### • Genetic relationships amongst populations:

- Multilocus genetic analyses consistently showed a **close relationship between Canarian and North African goats (Figures 1 and 2)**. These results agree well with genetic, archeological, anthropological and linguistic studies demonstrating that the Canary Islands were settled in the first millennium BC by human and livestock populations coming from North West Africa.
- Iberian and Balearic goats did not seem to have a North African origin, being more related with their Italian counterparts. Currently, there is a considerable debate about the occurrence of bidirectional human and livestock migrations between North Africa and Southern Iberia in ancient times. Our data, rather than supporting this scenario, suggest that **Iberian goats have a European genetic background.**

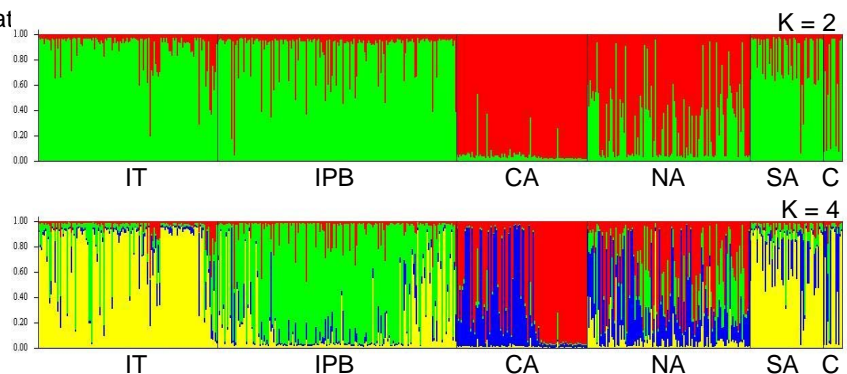
**TABLE1.** Polymorphism of 6 goat pigmentation genes.

Gene	SNP	Nsyn SNP
<i>MC1R</i>	7	5
<i>ASIP</i>	4	2
<i>TYRP1</i>	2	0
<i>TYRP2</i>	3	1
<i>TYR</i>	2	1
<i>KIT</i>	3	1



**FIGURE 1.** PCA analysis based on Reynolds genetic distances amongst goat populations.

Italy (**IT**: Cilentana, Garganica, Derivata di Siria, Girgentana, Jonica, Maltese), Iberian Peninsula and Balearic Islands (**IPB**: Murciano-Granadina, Malagueña, Rasquera, Azpi-Gorri, Ibizan), Canary Islands (**CA**: Majorera, Tinerfeña and Palmera), North Africa (**NA**: Moroccan and Tunisian local breeds), Cashmere (**C**) and Saanen (**SA**).



**FIGURE 2.** Structure-based clusterization analysis.

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