Genome-wide SNP profiling of Greek sheep with worldwide domestic breeds revealing population and admixture patterns
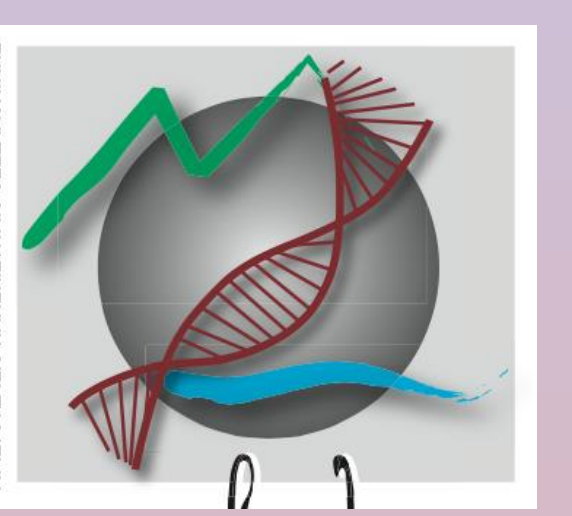
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Introduction

Greece hosts one of the largest and highly diverse pools of animal genetic resources in the Mediterranean region, where numerous sheep breeds have been regionally selected for centuries. Several factors have been involved in breed formation processes of Greek sheep such as the geographic isolation (e.g., island and mainland Greece), genetic drift, human-mediated selection, crossbreeding and transhumance. Greek sheep have been poorly studied in global scale genetic analysis. The present study focuses on the genome-wide characterization of population structure, diversity, and admixture patterns of local Greek domestic sheep collectively analyzed with 38 global breeds from the Sheep HapMap project.

Materials and Methods

◊ **Blood sampling:** 1,008 Greek domestic sheep from 5 different breeds/23 locations (Figure 1)

◊ **DNA extraction and genome-wide genotyping** using the OvineSNP50 Genotyping BeadChip v2 - **52,152 SNPs**

◊ **Joint analysis with ~2,000** published **50K sheep genotypes** of 38 breeds - **Sheep HapMap** and **iSAGE** projects

◊ **Genotype quality control & selection of representative genotypes**

◊ **Population structure:** principal component analysis (**PCA**) and model-based clustering (**Admixture**)

◊ **Genetic differentiation & diversity:** pairwise F_{ST} & heterozygosity/inbreeding levels (H_o, H_e, F_{IS}) using PLINK v1.9

Figure 1: Geographical map depicting 23 sampling locations of the five Greek sheep breeds

Results & Discussion

Global PCA

The global clustering is attributed to the geographic origin of breed formation

Greek sheep represent an intermediate genetic link between Asia and Europe, indicating cross-continent and within Europe genetic clines (Figure 2)
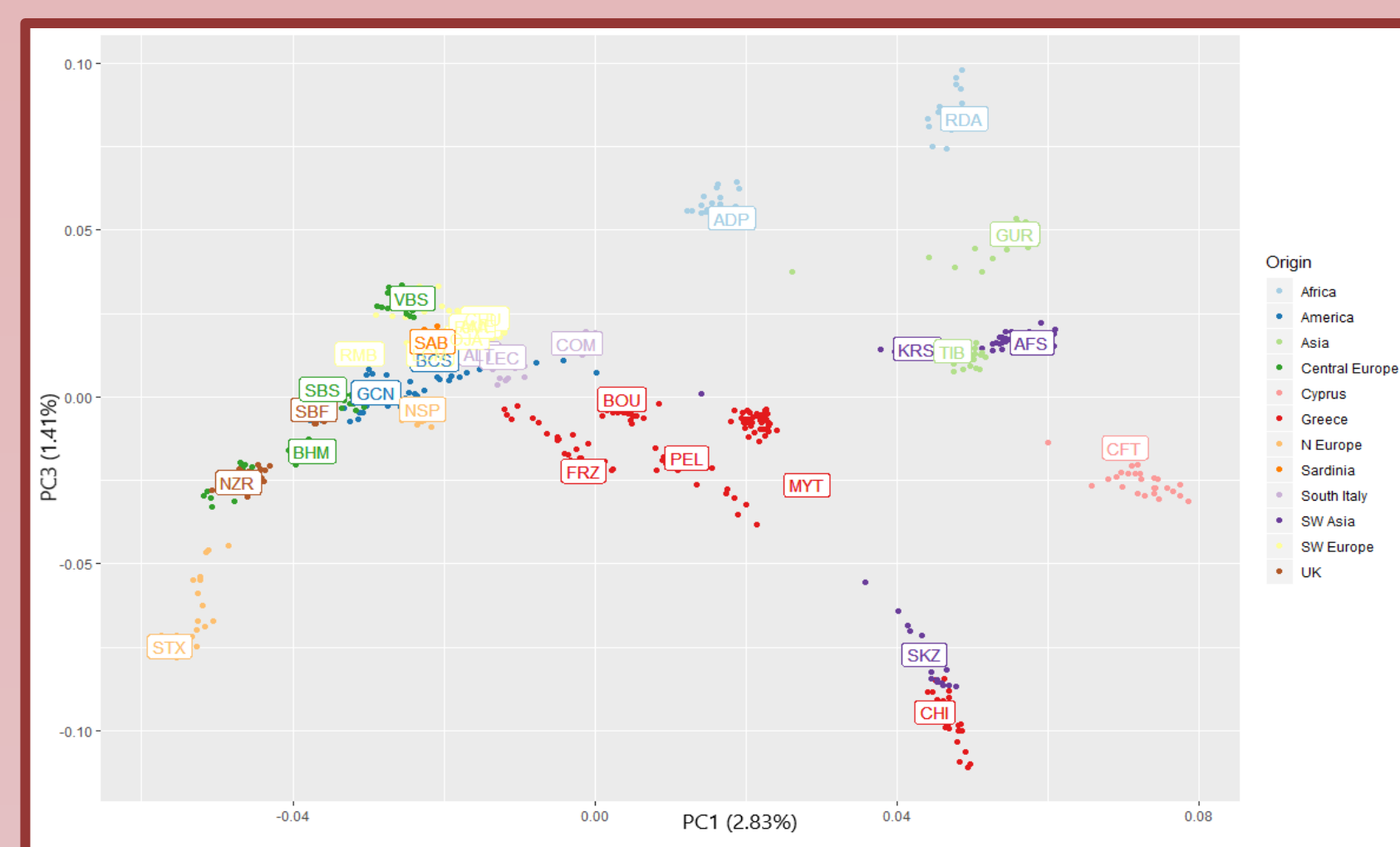

Figure 2: PCA analysis of global sheep level colored by geographic origin. First and third principal components depicted a central position of four Greek sheep breeds between Asian, African, and European sheep

Local PCA

Fat-tailed Asian origin of CHI and thin-tailed Zackel origin of BOU, PEL, MYT and FRZ may be related to the genetic differentiation between these clusters

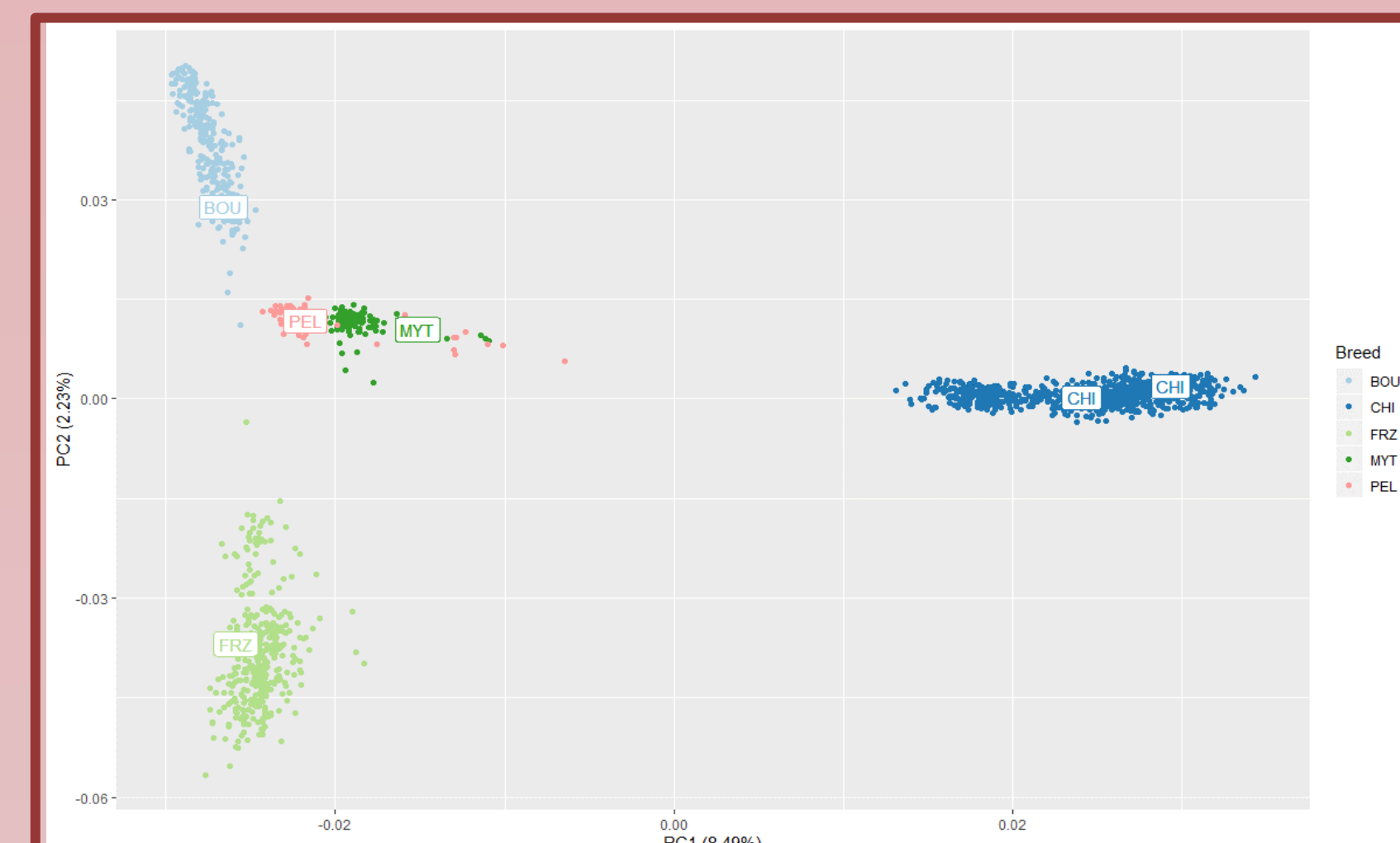MYT and PEL were not adequately differentiated due to high genetic relatedness (Figure 3)

Figure 3: PCA analysis of Greek sheep colored by breed. First and second principal components revealed the relatedness among the five Greek developed breeds.

Global Admixture

Asian and European genetic components were clearly observed (K=2) while the optimal K=35 differentiated the majority of global breeds

Mediterranean sheep were represented by a distinctive genetic cluster (K=8-14) (Figure 4)
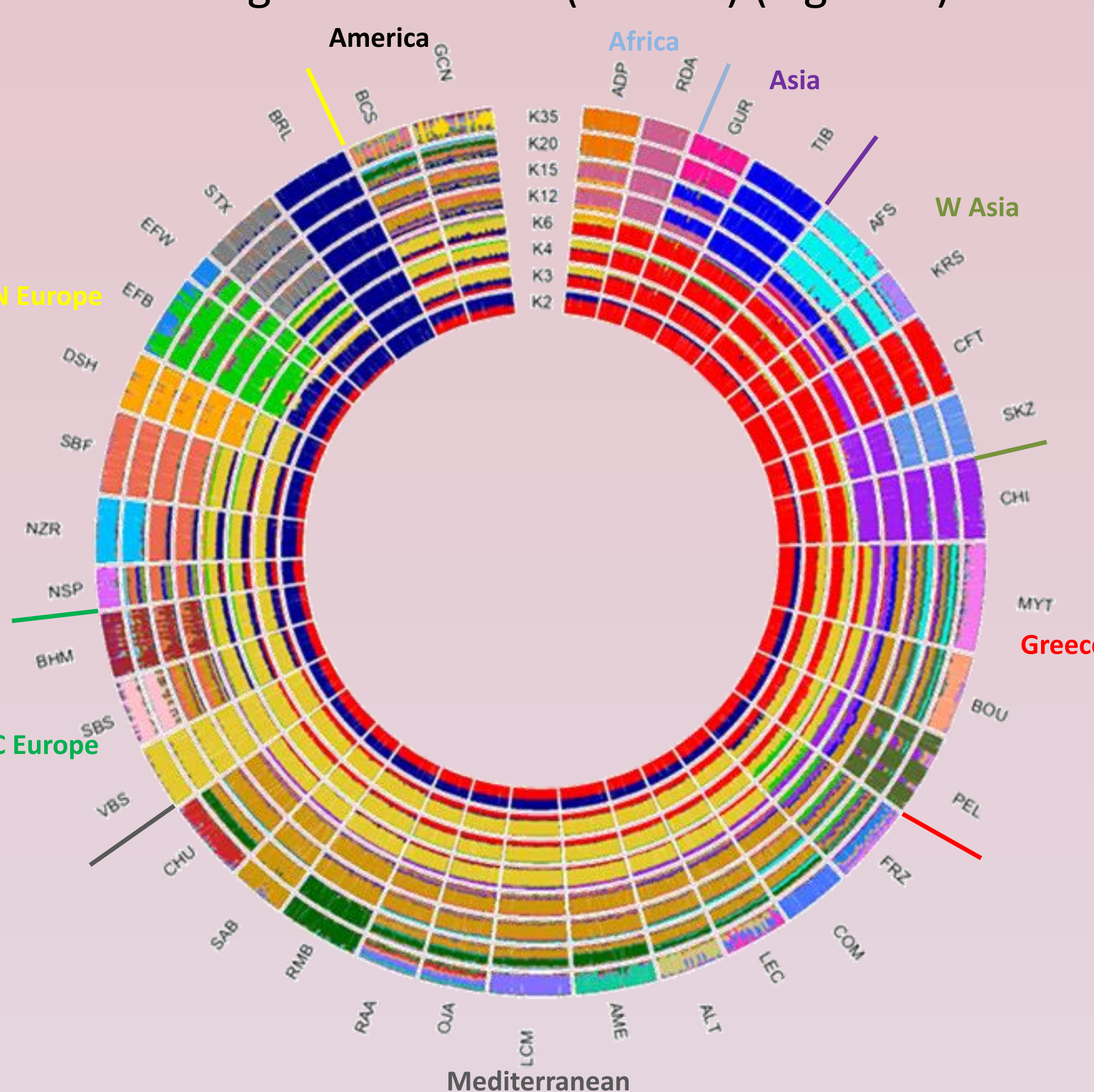

Figure 4: Circular representation of global sheep population structure results obtained with Admixture software for K = 2, 3, 4, 6, 12, 15, 20 and 35.

Local Admixture

The optimal K-value (K=5) clearly differentiated the respective five Greek breeds

Recent admixture events (K=8-13) were attributed to crossbreeding for genetic improvement purposes (Figure 5)
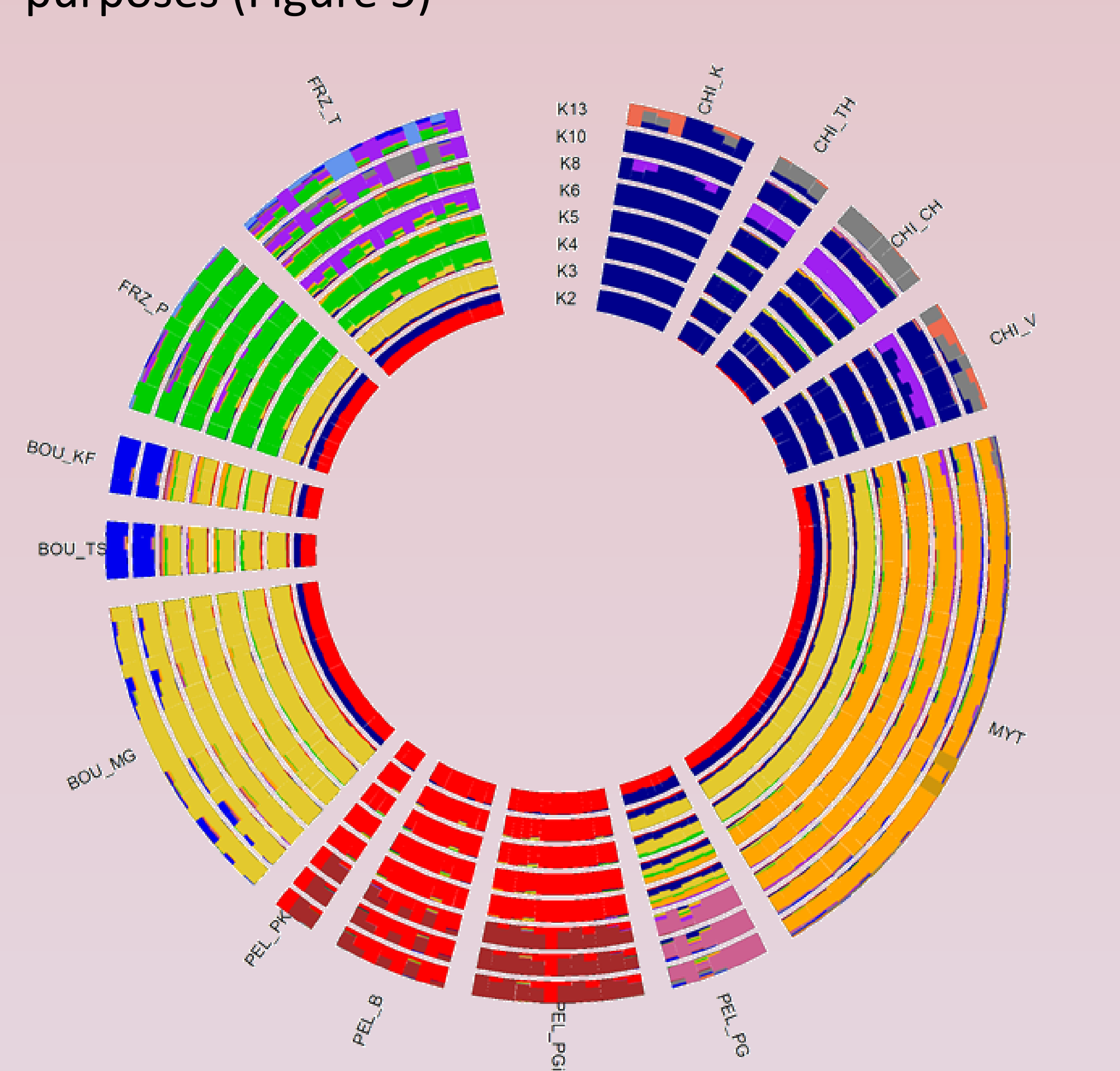

Figure 5: Circular representation of population structure results for representative local sheep obtained with Admixture software for K = 2-5, 6, 8, 10 and 13. The respective sampling sites are also depicted excluding Mytilini sheep due to highly-frequent animal exchange between farms

Global genome-wide diversity metrics

Genome-wide average diversity of Greek sheep estimated similar to previously studied Mediterranean and Asian sheep while higher from Central and North European sheep

Negative average inbreeding observed for several breeds, indicating that heterozygosity levels estimated higher than expected (Table 1)

Table 1: Average observed (Ho) and expected (He) heterozygosity and genomic inbreeding coefficient (F_{IS}) for Greek sheep breeds

Origin	Breed	No.	H_o	H_e	F_{IS}
Greece	Chios	802	0.326	0.328	0.007
	Mytilene	105	0.370	0.380	0.027
	Boutsko	291	0.366	0.374	0.021
	Pelagonia	59	0.351	0.356	0.015
	Frizarta	352	0.376	0.378	0.004
Italy	Comisana	24	0.375	0.364	-0.030
	Leccese	24	0.350	0.367	0.049
	Altamurana	24	0.363	0.365	0.006
Central Europe	Valais Blacknose Sheep	24	0.304	0.315	0.030
	Black-Headed Mutton	24	0.333	0.336	0.007
North Europe	Dorset Horn	21	0.319	0.292	-0.091
	Border Leicester	48	0.291	0.712	-0.009
	East-Friesian White	9	0.319	0.297	-0.076
	Old Norwegian Spaelsau	15	0.342	0.348	0.020
Asia, SW Asia	Tibetan	37	0.326	0.339	0.040
	Afshari	37	0.358	0.347	-0.033
	Karakas	18	0.357	0.341	-0.049
	Cyprus Fat Tail	30	0.329	0.313	-0.052

References

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