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# The coalition between Italian goats and Italian researchers: the Italian Goat Consortium

**Paolo Ajmone Marsan and the Italian Goat Consortium**

Institute of Zootechnics

Università Cattolica del Sacro Cuore

Piacenza, Italy

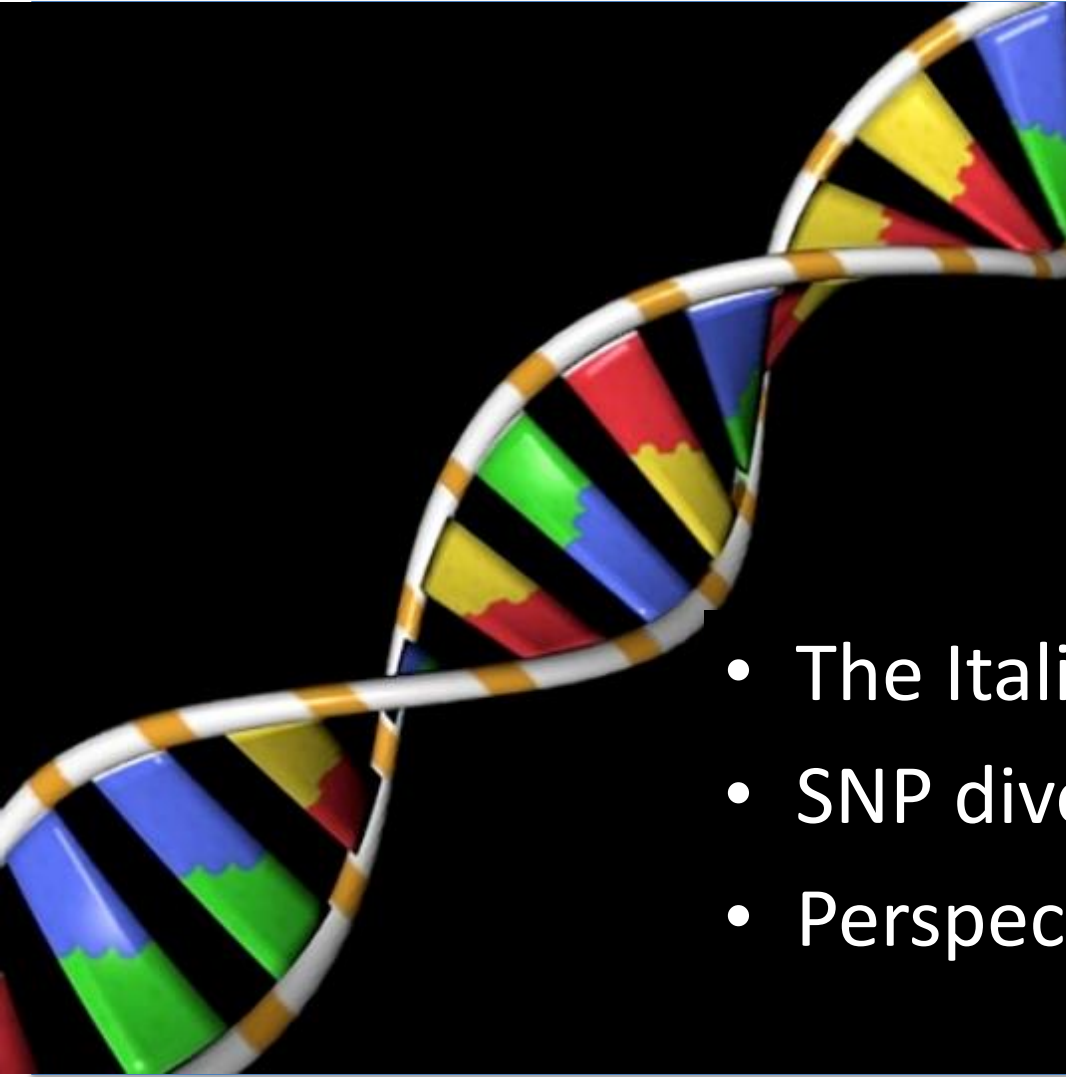
[paolo.ajmone@unicatt.it](mailto:paolo.ajmone@unicatt.it)

Cardiff, 17/06/2014



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# Outline



- The Italian Goat Consortium
- SNP diversity in Italian goats
- Perspectives



- Hard time for economy in Europe
  - Harder in Italy than in Central/Northern Europe
    - Even harder for research funding
      - Very difficult for small ruminant research
        - » No way for goat diversity!
- From crisis to opportunity
  - Modest seed funding of a project started in 2008 (Innovagen funded by the Ministry of Agriculture)
    - Coalization and Coordination.....
      - Definitely a new model for Italian scientists.....



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# Italian goat Consortium

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**Pooling local efforts and  
resources for the genomic  
characterisation of Italian  
goat breeds**

[www.italiangoaatconsortium.eu](http://www.italiangoaatconsortium.eu)



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# Sampling



Camosciata delle Alpi

Bionda dell'Adamello



Saanen

Orobica

Val Passiria



Valdostana



Teramana



Grigia Ciociara



Teramana



Sarda

Maltese

Nicastrese



Aspromontana



Sarda

Maltese

Nicastrese

Aspromontana

Girgentana



Argentata dell'Etna







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# 50K Illumina goat SNP chip

- Discovery on 6 breeds (meat, mixed and milk)
- Detection of ~12 million variations with > 10 million SNPs
- 60,000 SNPs (spaced on the genome, with >0.2 MAF, >0.8 Illumina ADT score...)
- 52,295 successful loci (tested with 288 goat DNA samples from 10 different breeds)
- Pseudochromosomes aligned on cattle
- Details on [www.goatgenome.org](http://www.goatgenome.org)
- Sequencing and novel *de novo* assembly on going at



USDA





# Dataset cleaning

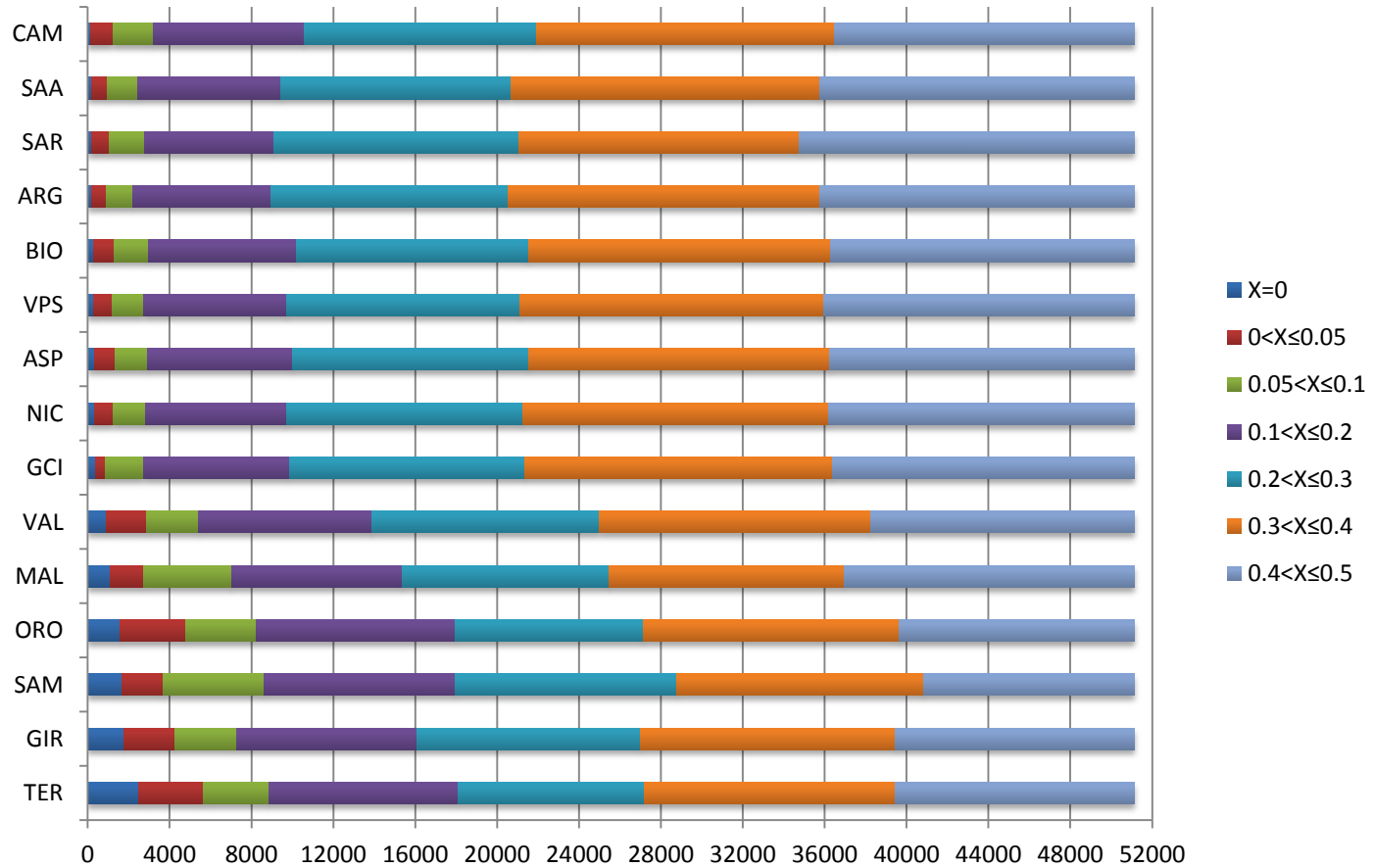
- Filtering exclusion threshold
  - $MAF \leq 1\%$
  - Missing (SNP)  $\geq 5\%$
  - Missing (animal)  $\geq 5\%$
  - HW within breed  $FDR \geq 20\%$
- Working Dataset
  - 15 breeds
  - 350 animals (15-32 per breed)
  - 51,136 SNPs

**SNPchip affected by ascertainment bias (EU Nextgen project) but highly informative for the Italian gene pool**



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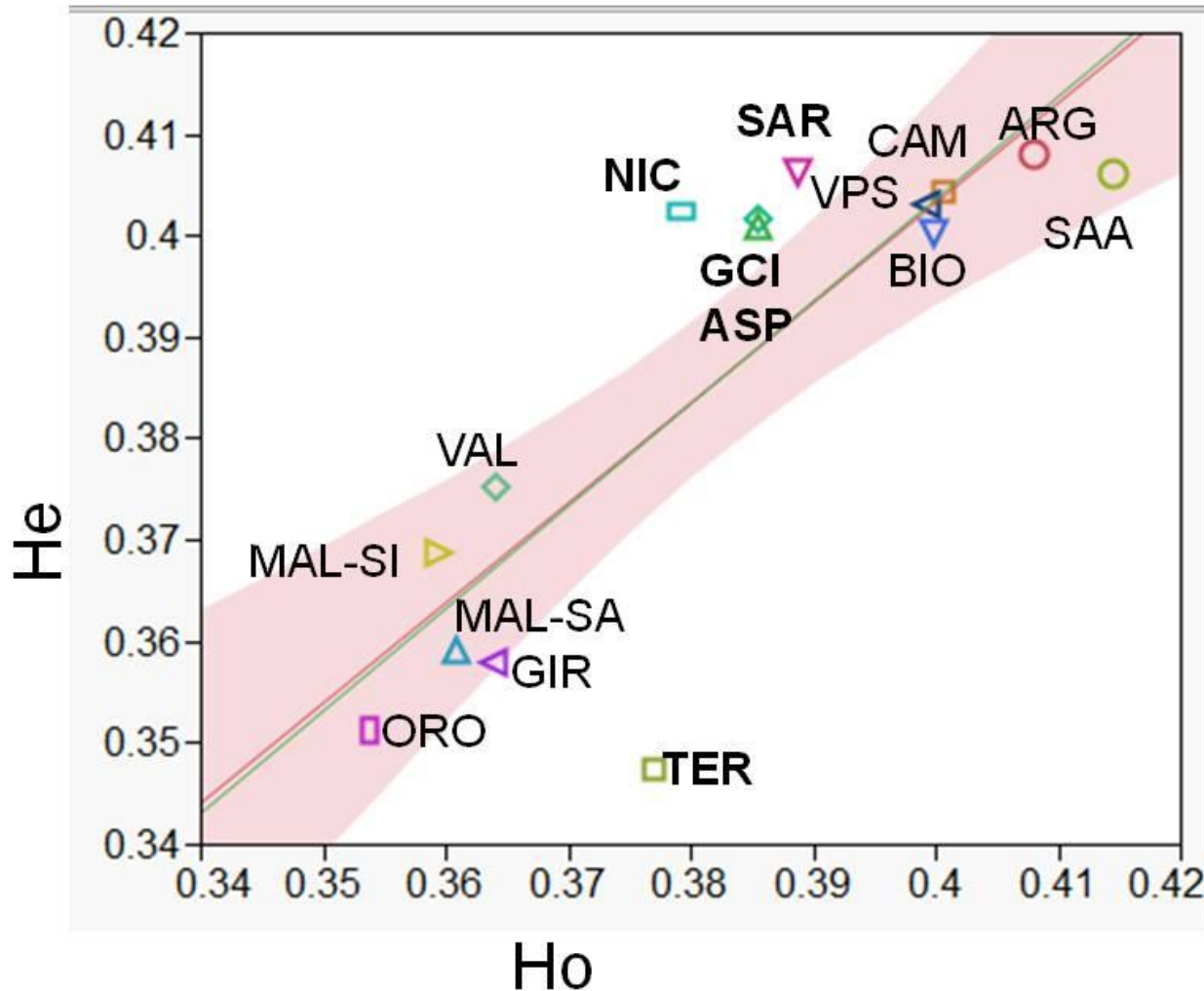
# Within breed MAF distribution





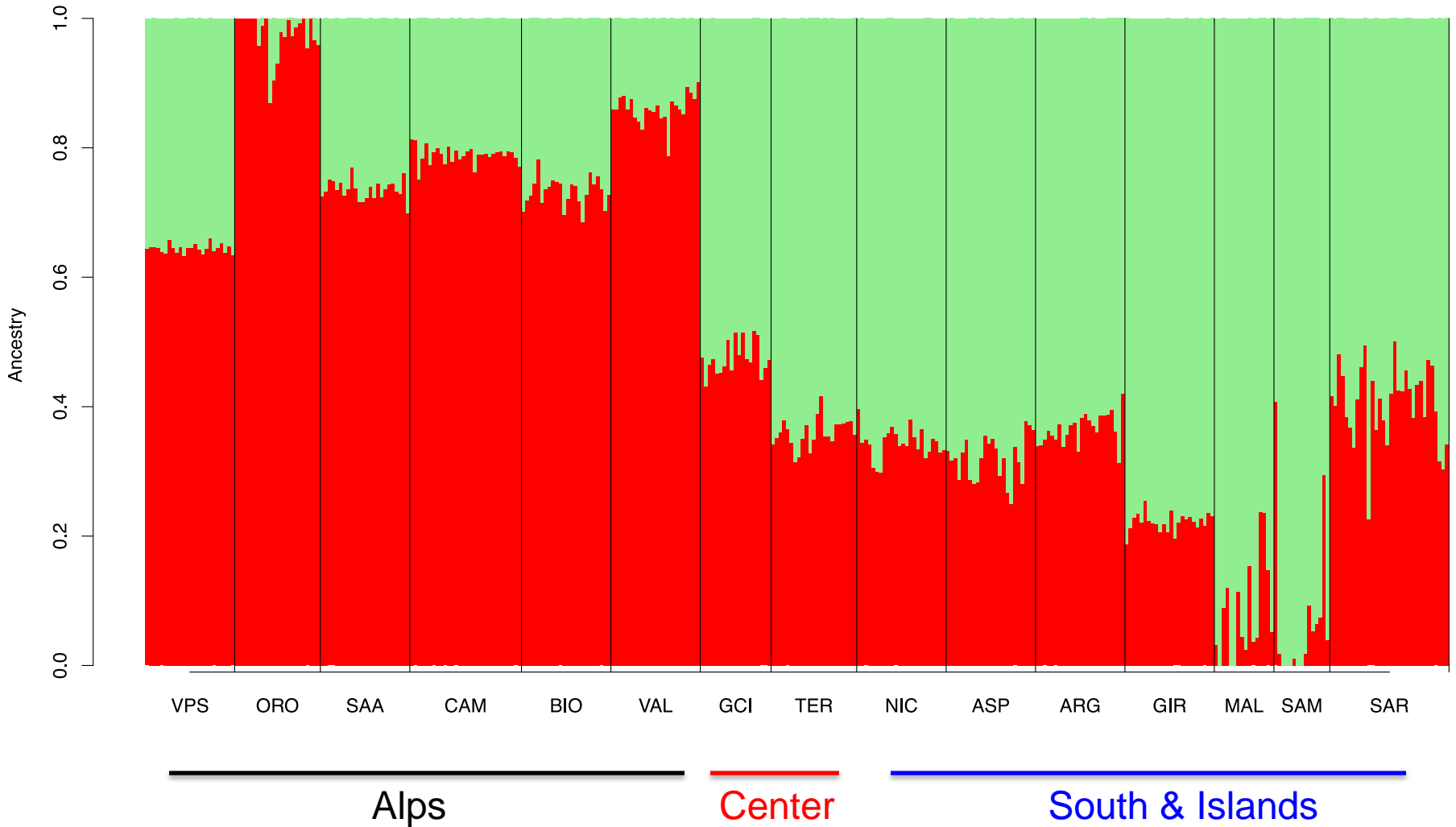


# Expected vs Observed Heterozygosity





## ADMIXTURE Goat K=2

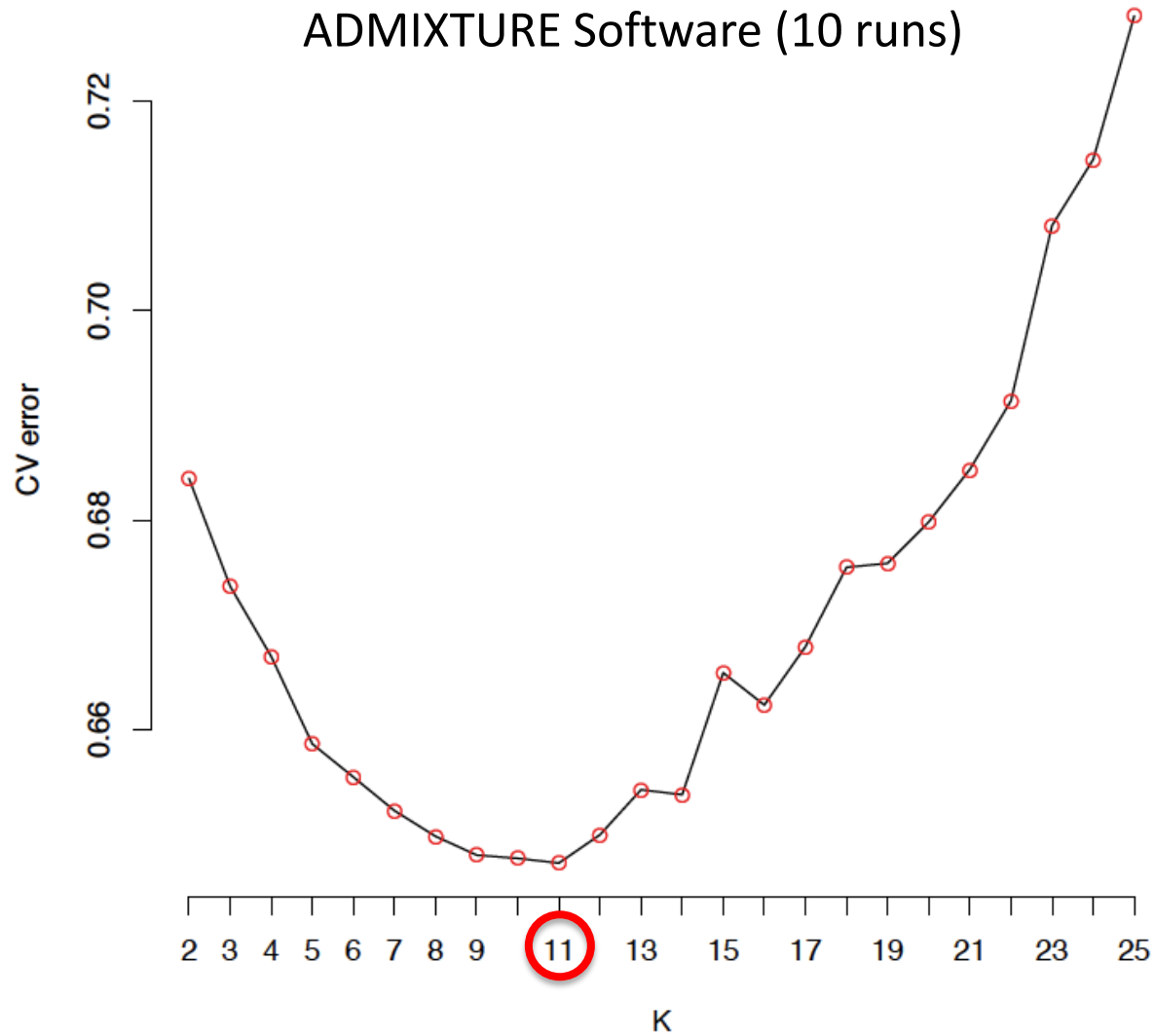




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# Cross Validation error plot

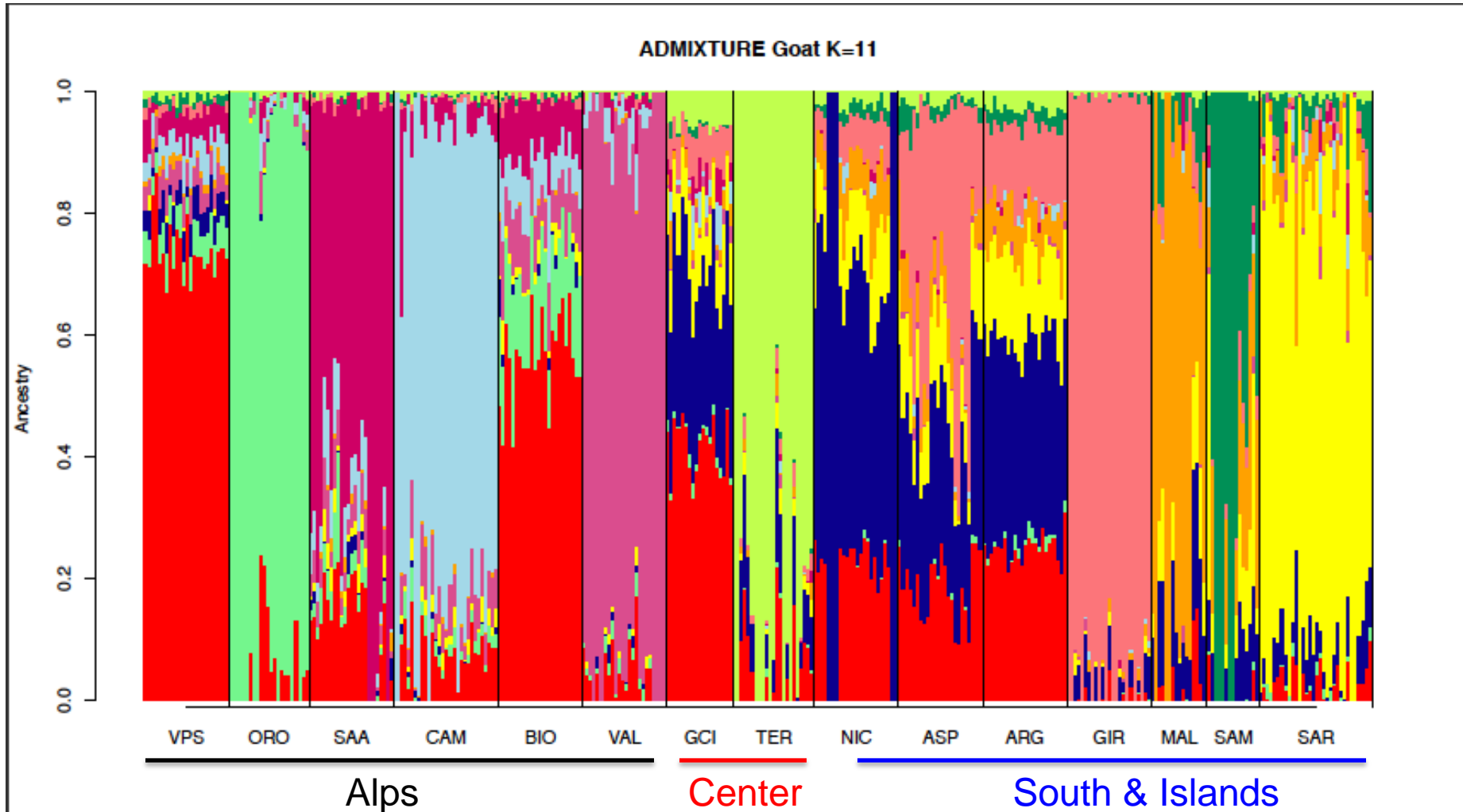
ADMIXTURE Software (10 runs)





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# The Best K (K=11)





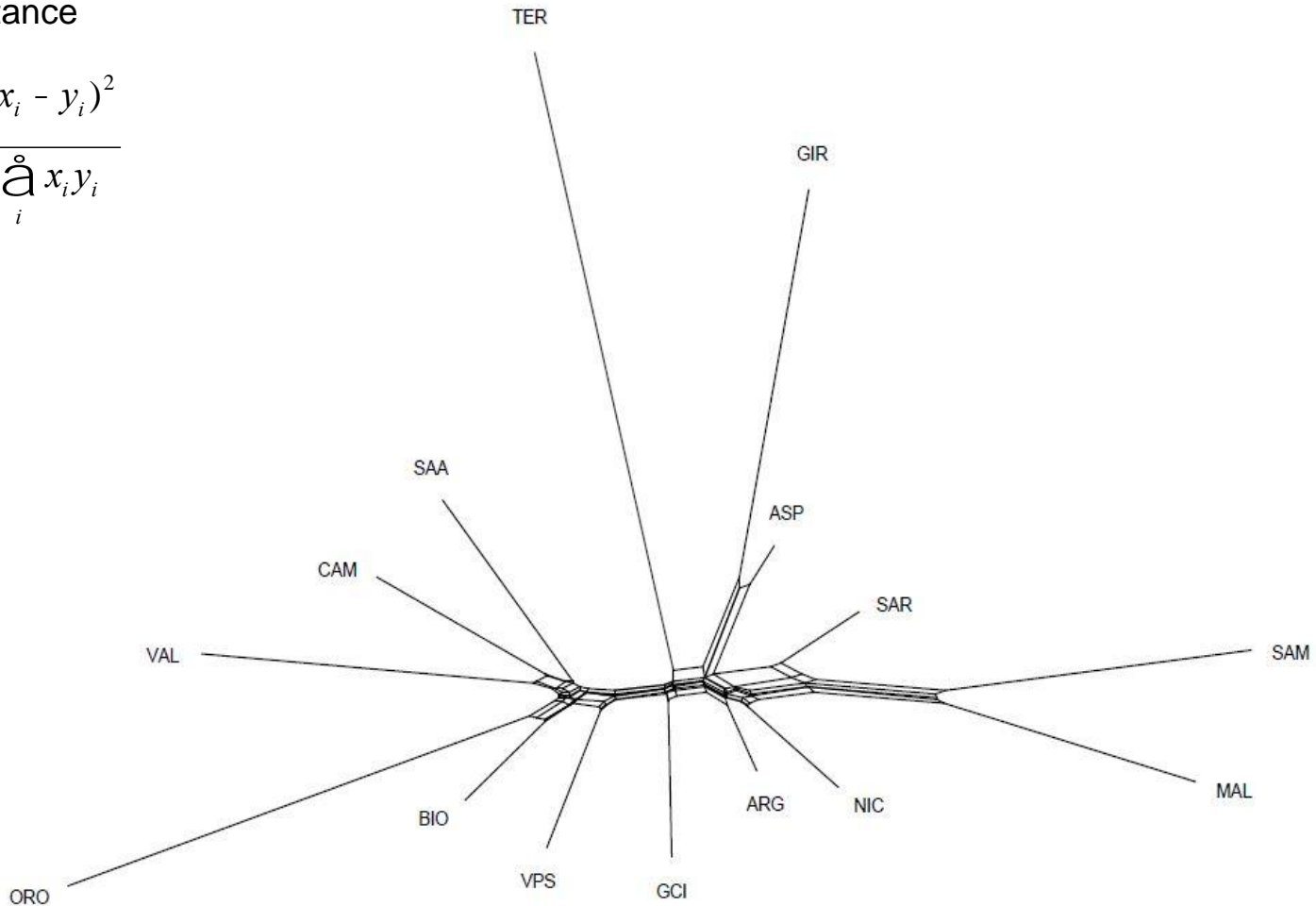


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# Neighbour Net based on Reynolds distance

Reynolds Distance

$$D_{Reynolds} = \frac{1}{2} \frac{\sum_i (x_i - y_i)^2}{1 - \sum_i x_i y_i}$$

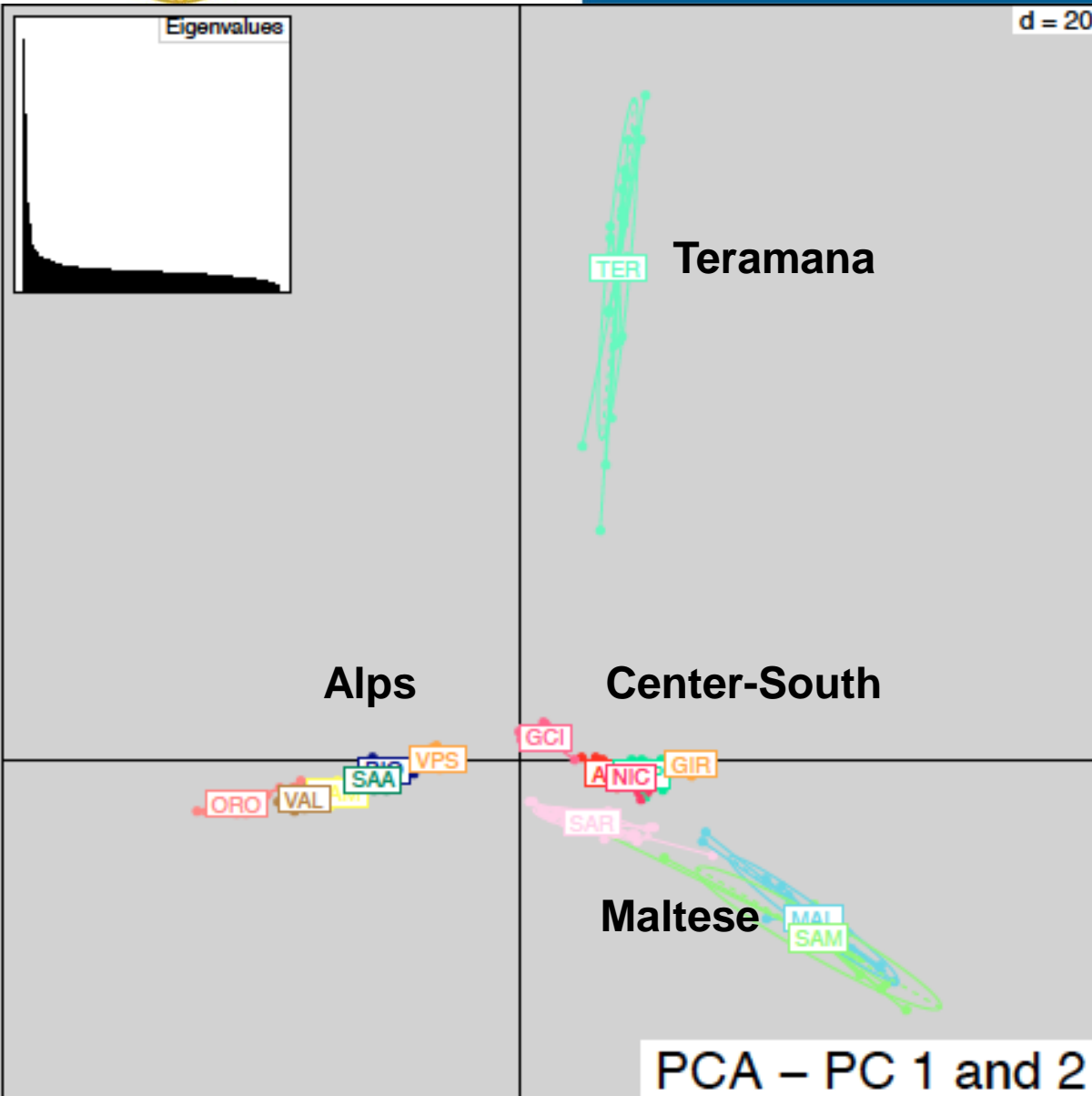






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# Principal Component Analysis

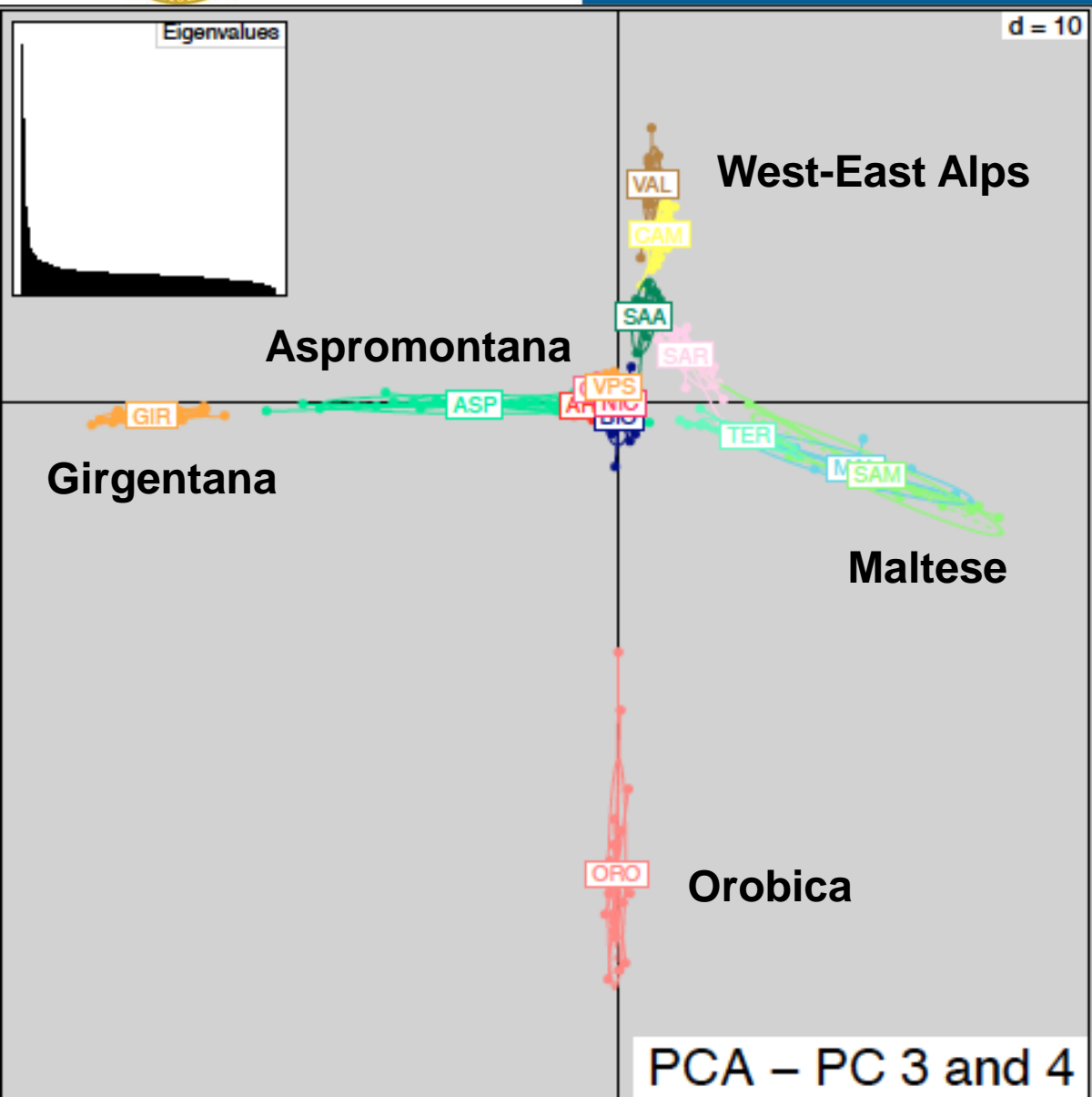


- CAM = Camosciata (Alpine)
- VAL = Valdostana
- SAA = Saanen
- BIO = Bionda
- ORO = Orobica
- VPS = Valpassiria
- GCI = Grigia Ciociara
- ARG = Argentata
- NIC = Nicastrese
- ASP = Aspromontana
- MAL = Maltese Siciliana
- GIR = Girgentana
- TER = Teramana
- SAM = Maltese Sarda
- SAR = Sarda



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# Principal Component Analysis

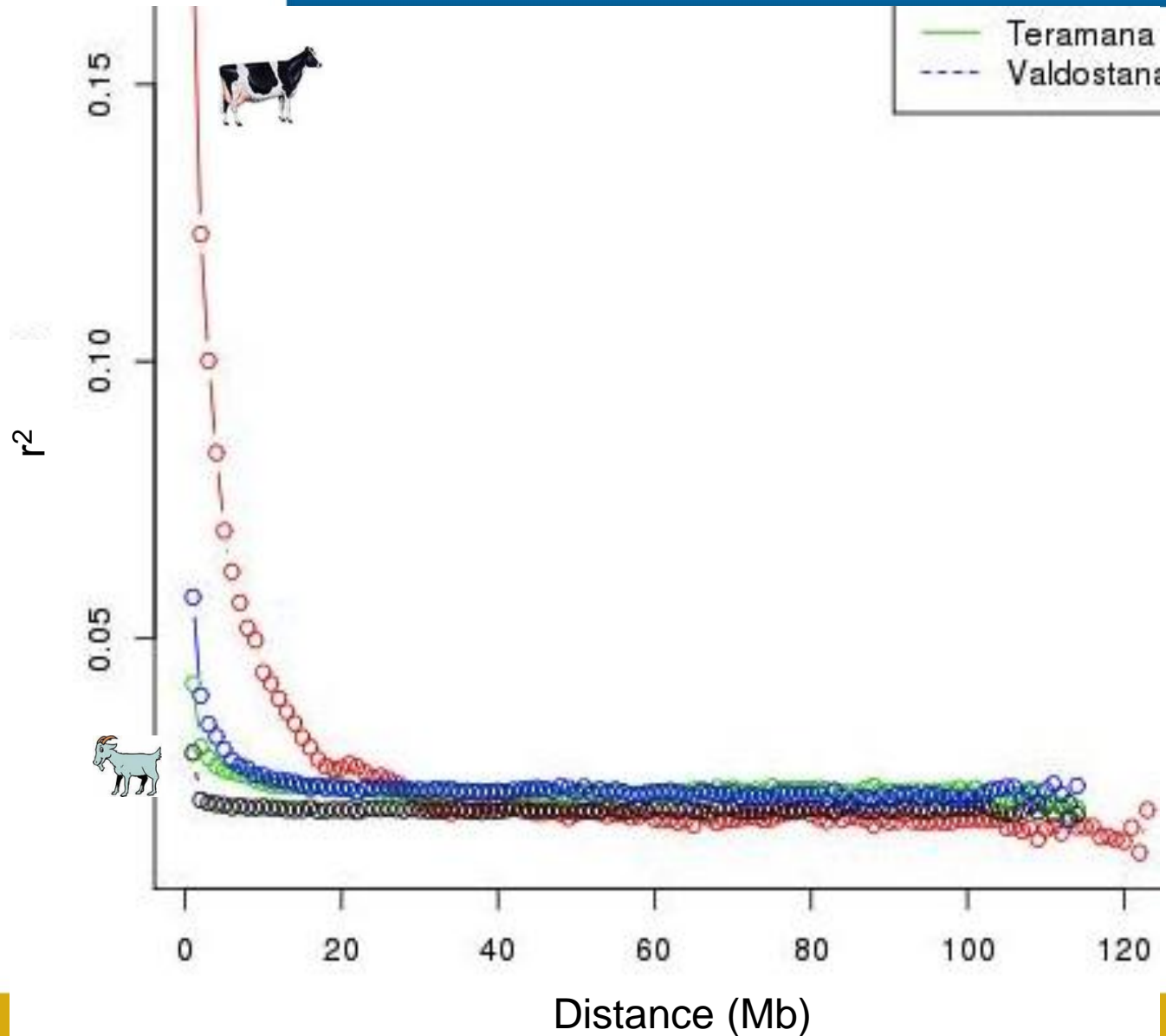


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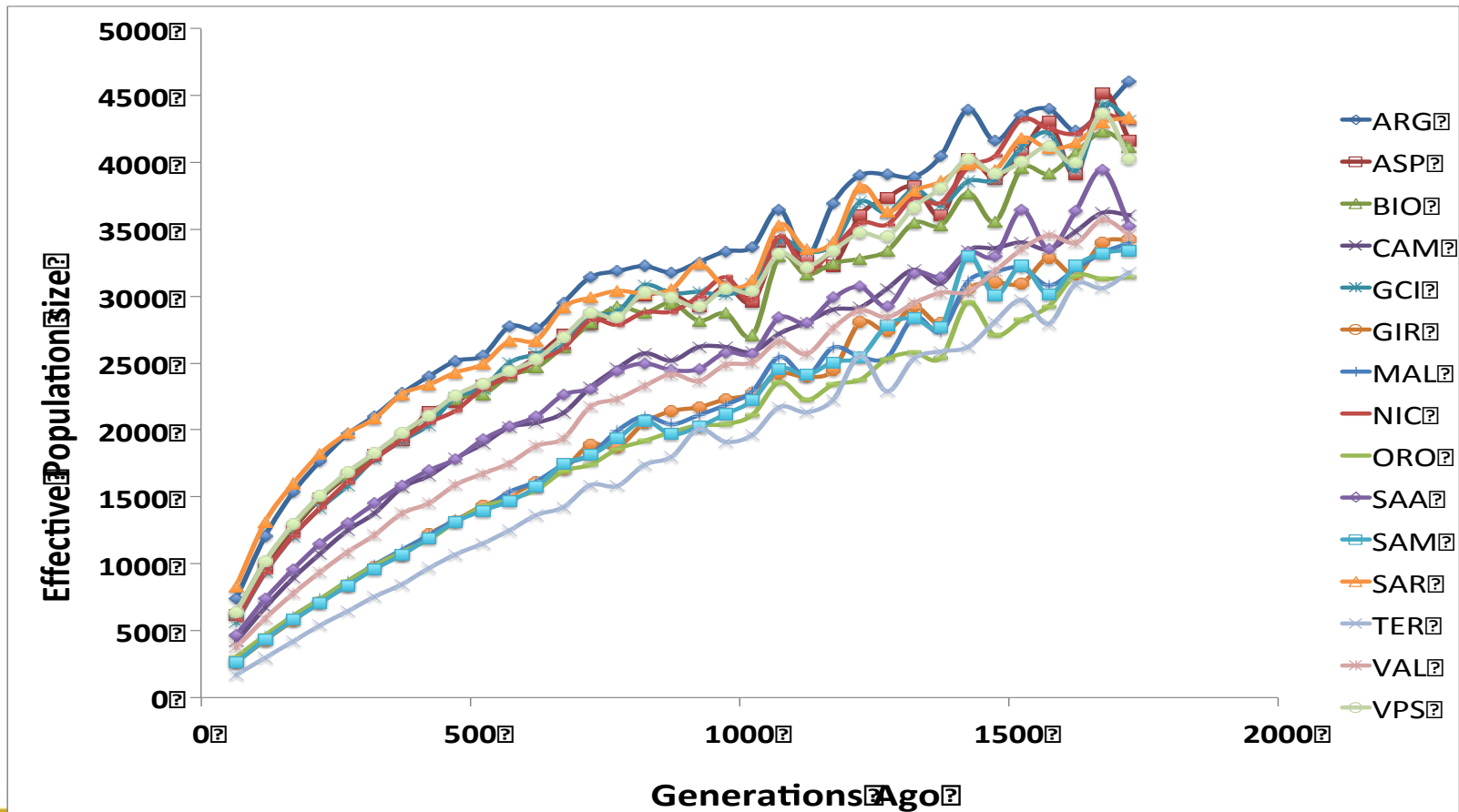
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# LD in Chromosome 6





Historical  $N_e$  of Italian goat breeds was estimated using SNeP\*. For each pair of SNPs within a chromosome the LD is calculated according to Hill & Robertson (1968) using the method of Sved (1971) and correcting for sample size and mutations (Weir & Hill 1980, Hayes 2003, Corbin et al. 2012).





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# Selection signatures

## Lositan software

Simulation of markers under the neutral model

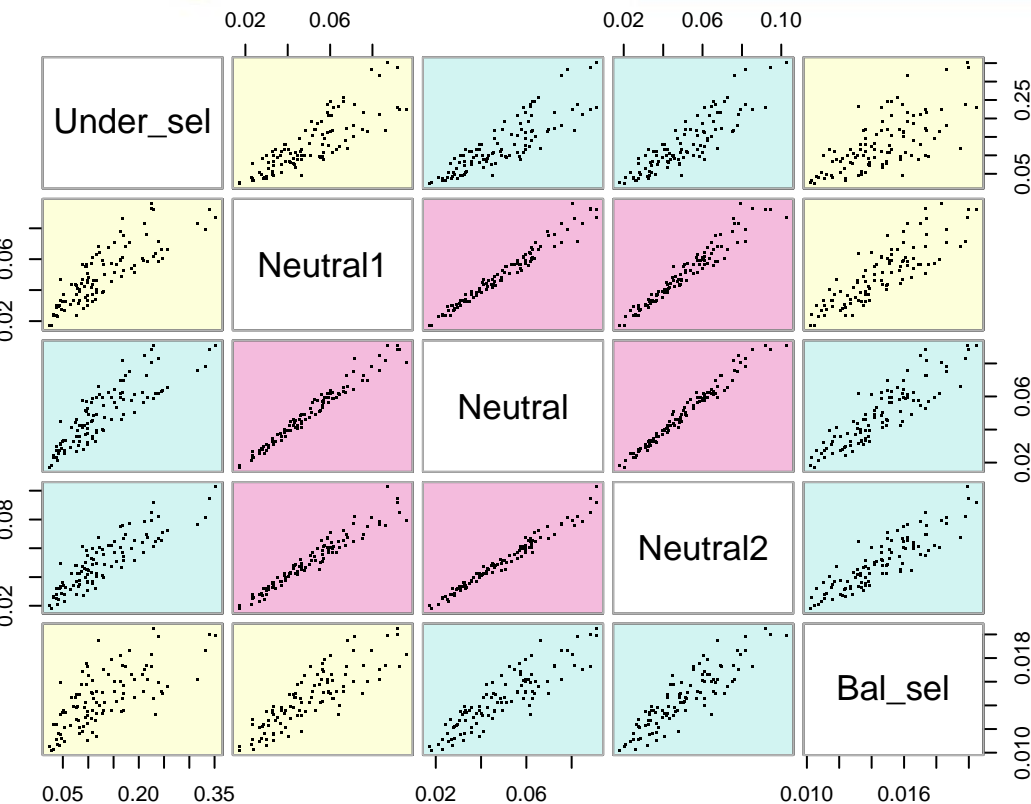
Detection of outliers having  $F_{st}$  lower than expected under a neutral model at that value of heterozygosity

- 456 markers under directional selection
- 629 under balancing selection

What about breed diversity at these loci?



# Markers under directional/balancing selection



**Pearson' correlation  
between genetic distances  
between breeds**

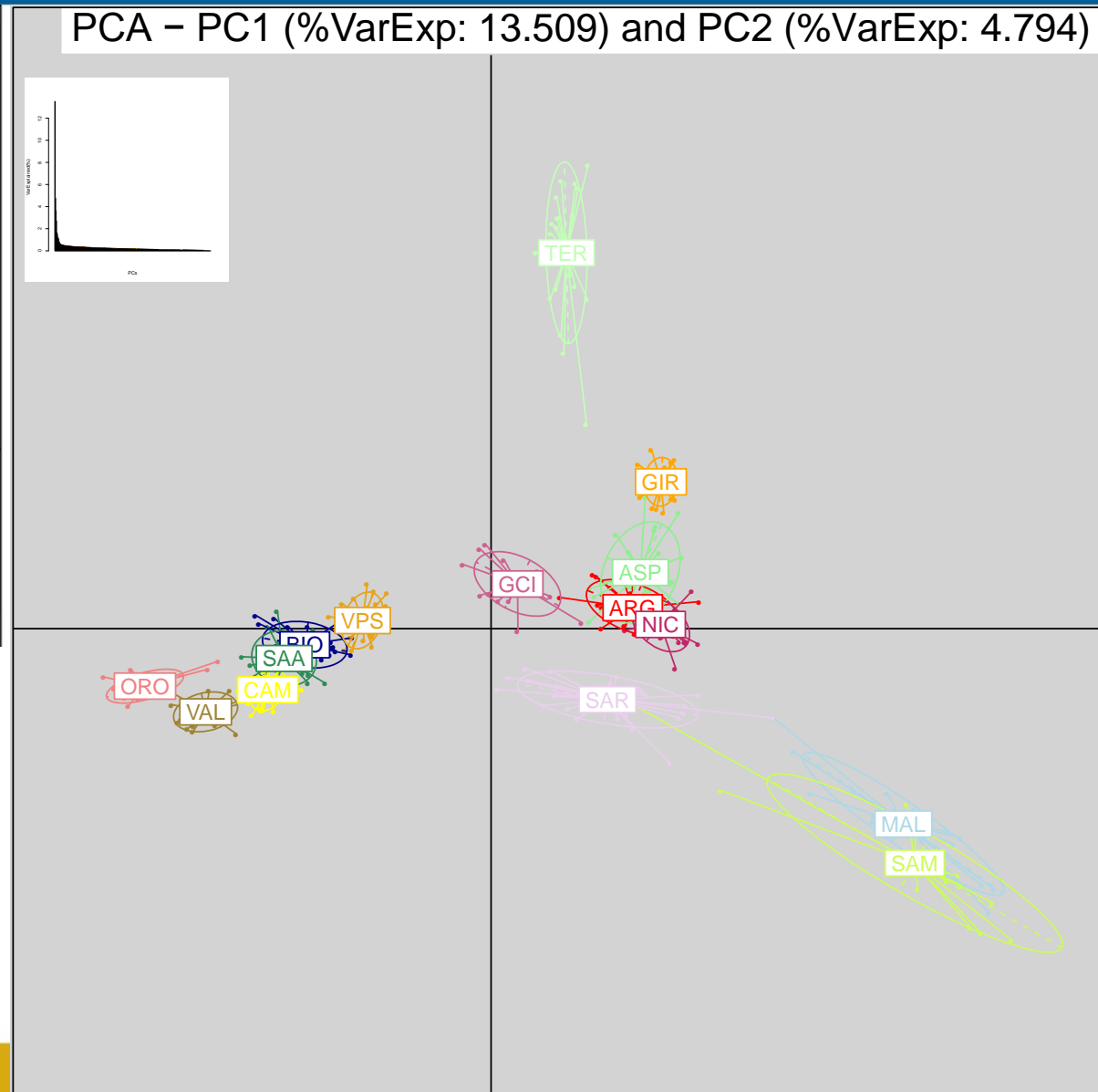
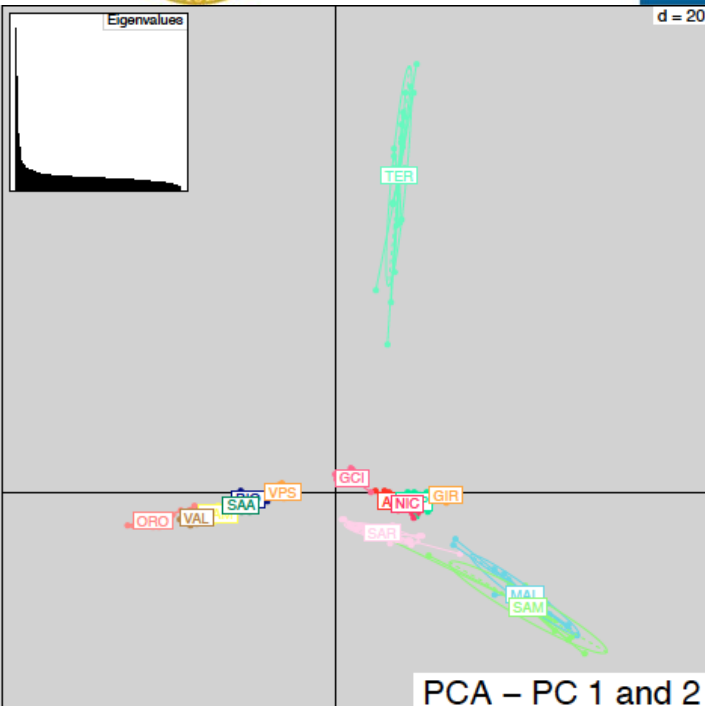
Neutral = 50051 markers  
Under\_Sel = 456 markers  
Bal\_Sel = 629 markers  
Neutral 1 = 456 markers  
Neutral 2 = 456 markers

	Ntr	Dir	Bal	Ntr1	Ntr2
Ntr	1	0.85	0.89	0.98	0.99
Dir		1	0.70	0.83	0.88
Bal			1	0.85	0.88
Ntr 1				1	0.97
Ntr 2					1





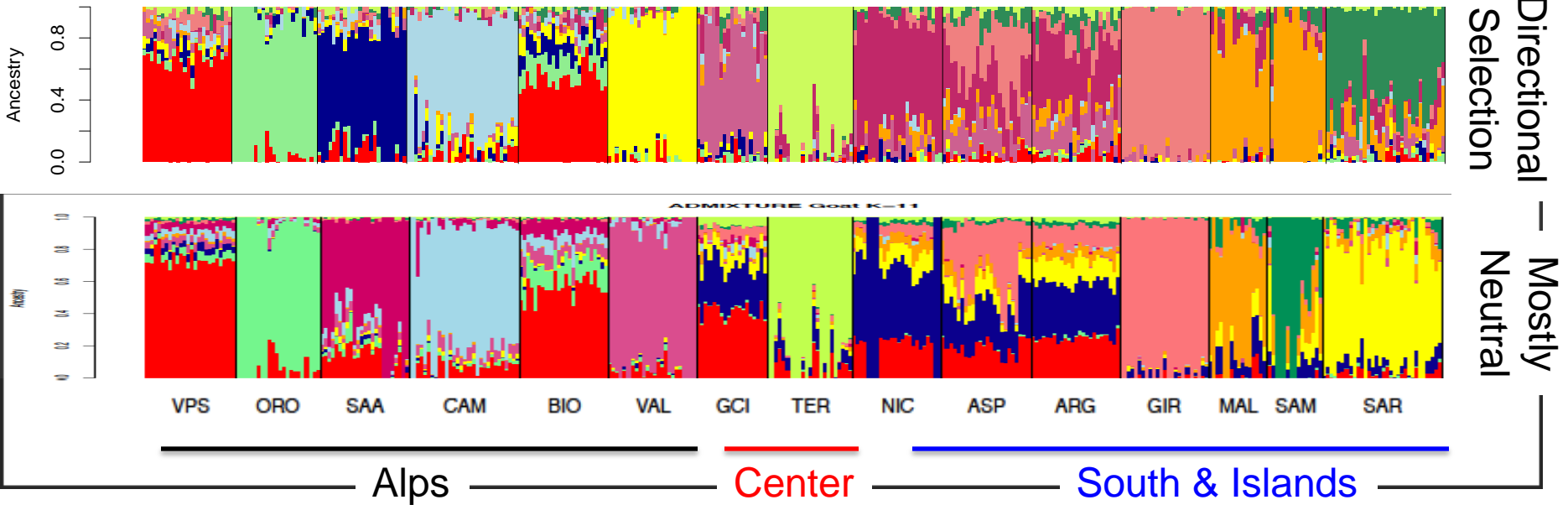
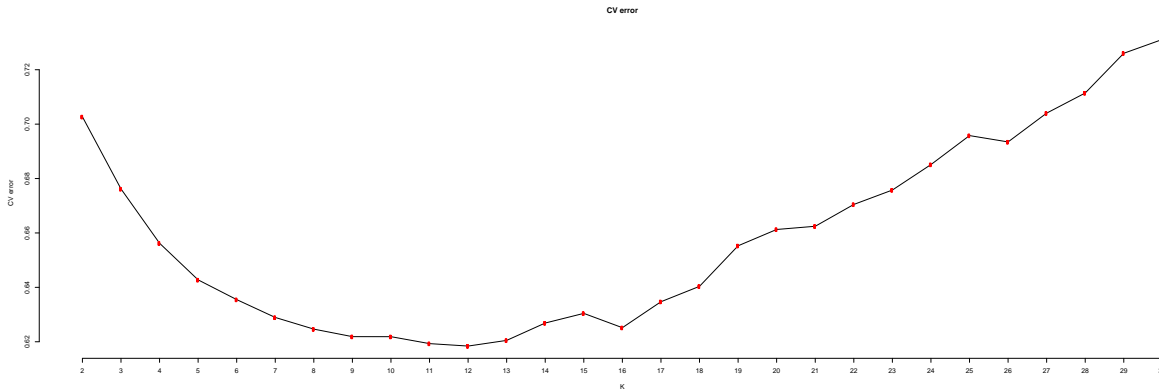
# Directional selection





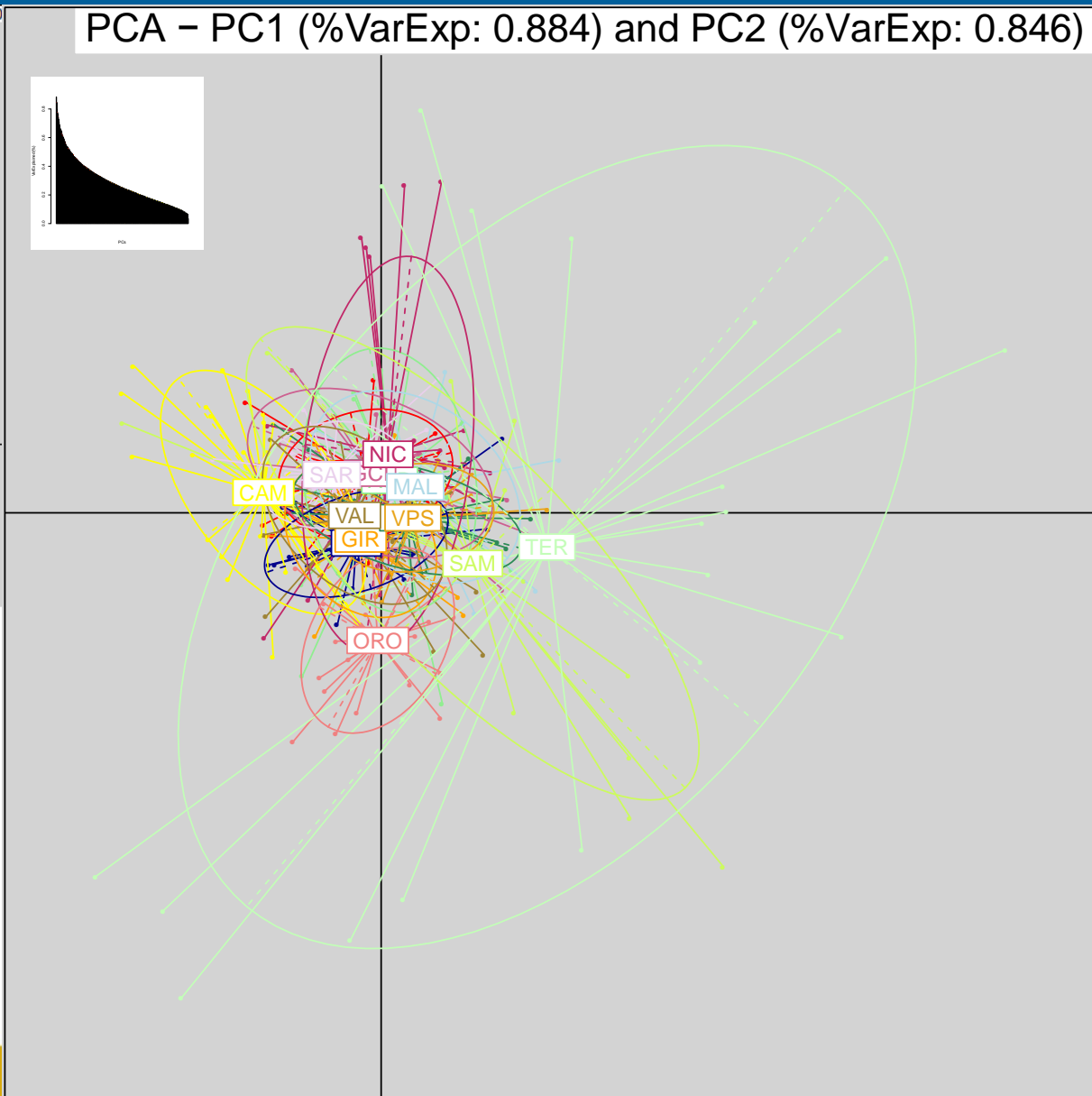
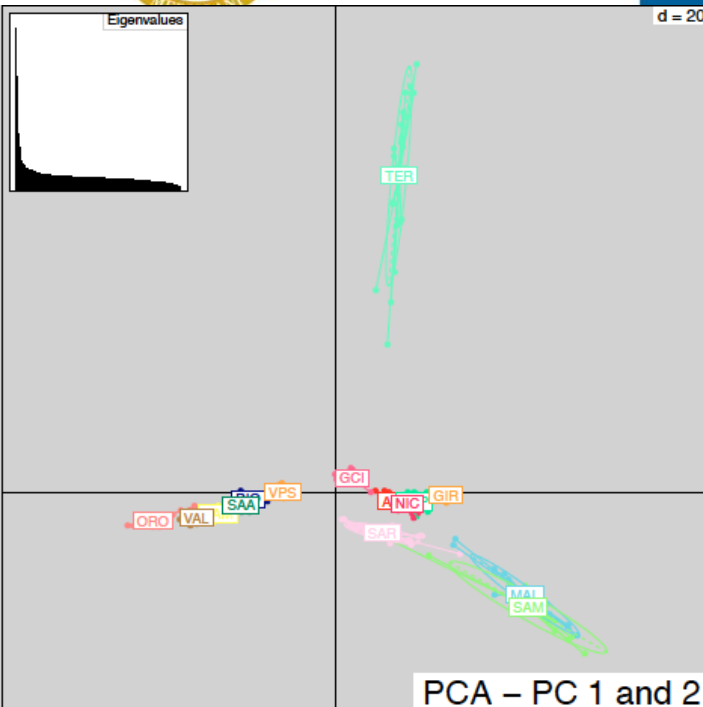
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# Directional selection





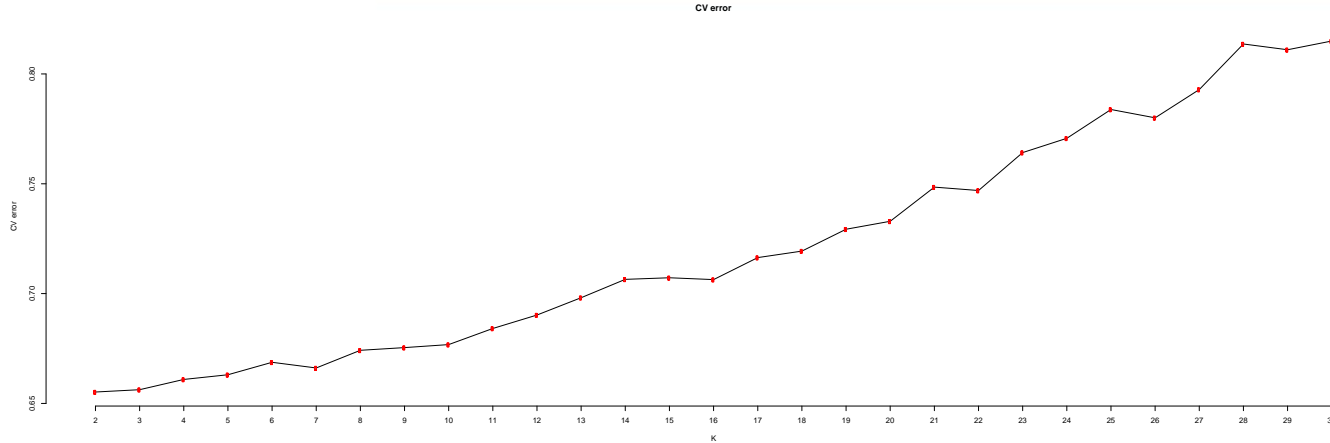
# Balancing selection



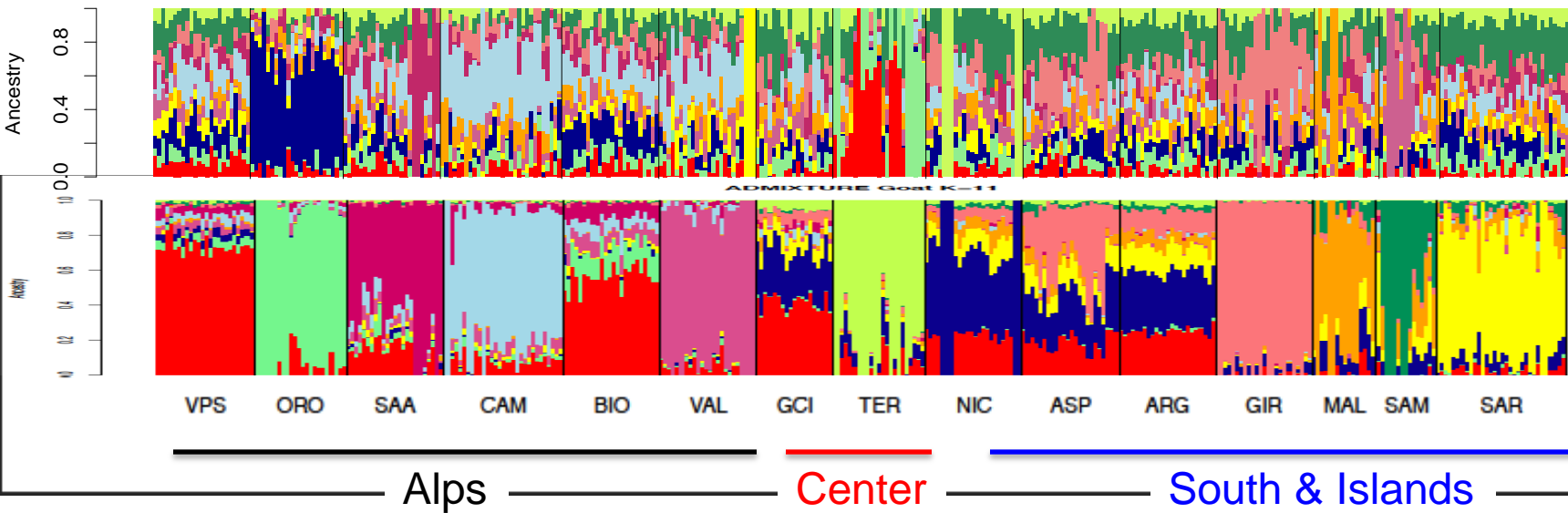


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# Balancing selection

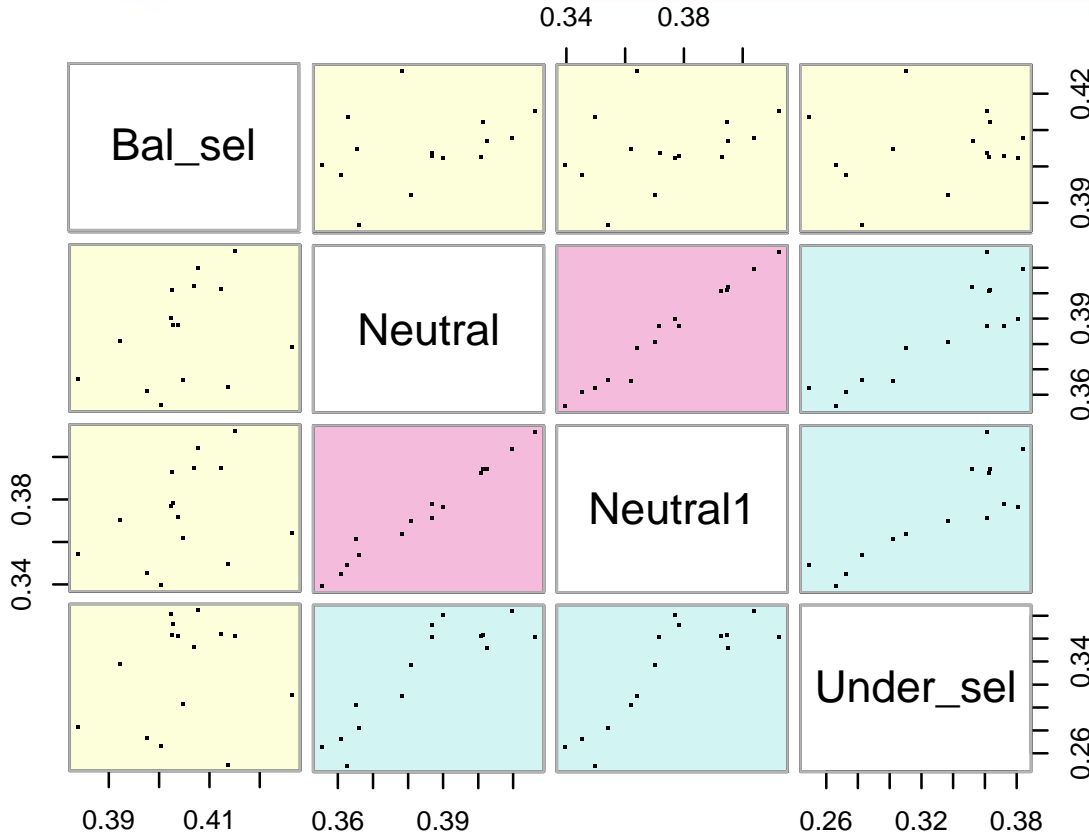


Balancing Selection  
Neutral





# Correlation between heterozygosities of breeds

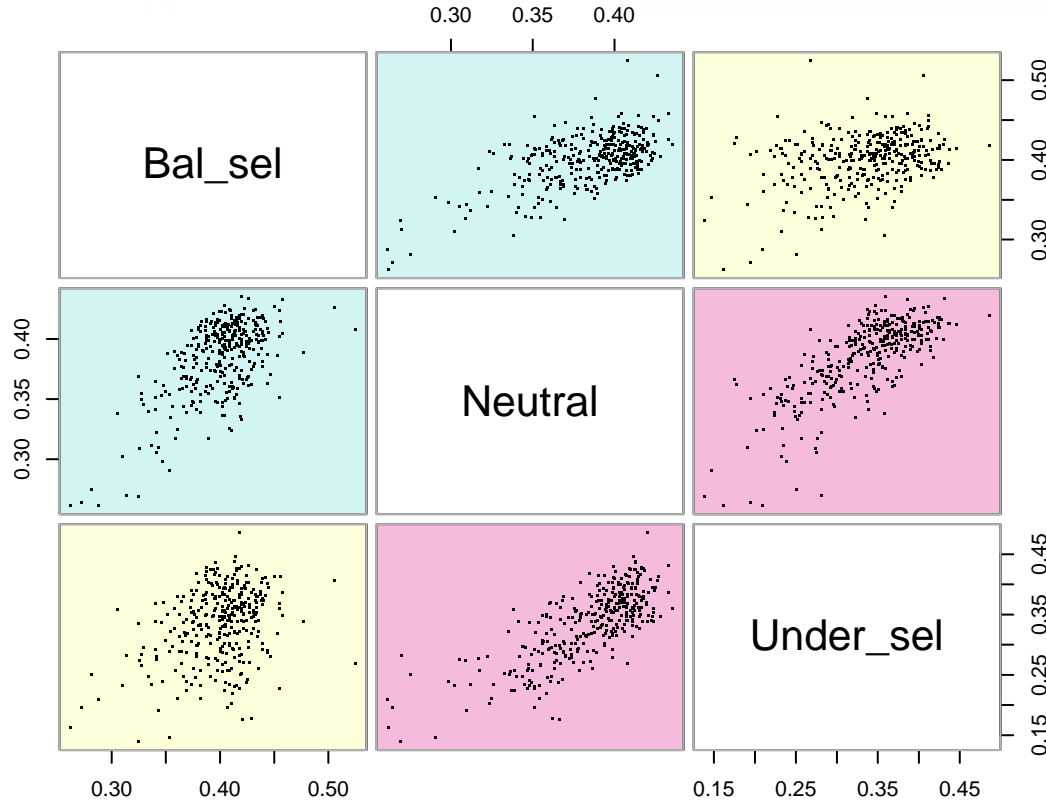


	Neutral	Directional	Balancing	Neutral 1
Neutral	1	0.87	0.33	0.99
Directional		1	0.14	0.85
Balancing			1	0.33
Neutral 1				1



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# Correlation between heterozygosities of individuals



	Neutral	Directional	Balancing
Neutral	1	0.76	0.66
Directional		1	0.33
Balancing			1





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# Conclusions



# SNPs vs others

- Much higher level of resolution (many thousand vs a few markers)
- Robust and non homoplasic
- Easier comparability across projects and data merging
- Suited to genome wide analyses (ROH, Ne, Selection signatures, GWAS, breeding applications)
- However panels should be carefully prepared and evaluated (ascertainment bias)



# Italian goats

- Little or no inbreeding.
- Variable level of admixture.
- Some distinct breeds: Girgentana, Teramana, Orobica, Maltese.
- Low Ne nowadays (bottlenecks, breeding management), higher in the past.
- Geographic partition of diversity at small geographic scale (North-South and East-West in the Alps).
- Markers under selection are valuable for conservation decisions
- Neutral marker diversity is a reasonably good proxy of diversity of markers under directional selection



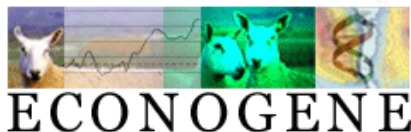
# Breeding

- Breeding will be more and more guided by molecular analyses if cost continues to decrease
- Methods customised to populations (small vs large pop. improvement, inbreeding control, maintenance of diversity)
- Knowledge of population structure is needed for any kind of application to avoid false positives



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# International Networking







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# Goat Adaptmap

➤ Traditional and novel approaches to study adaptation genomics:

➤ Selection signatures

➤ Spatial analysis

➤ Enriched SNP panel

➤ Detection of new variation

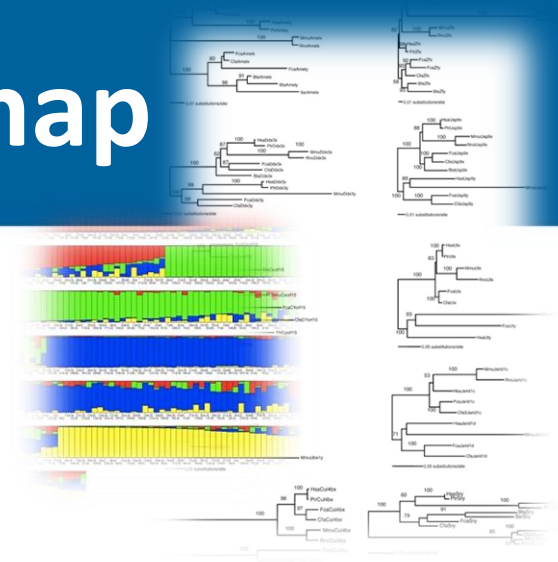
➤ To characterize the study

➤ Population studies based on Mutation Classification

Mutation effect on protein structure and function used as a tag for adaptation



Alessandra Stella



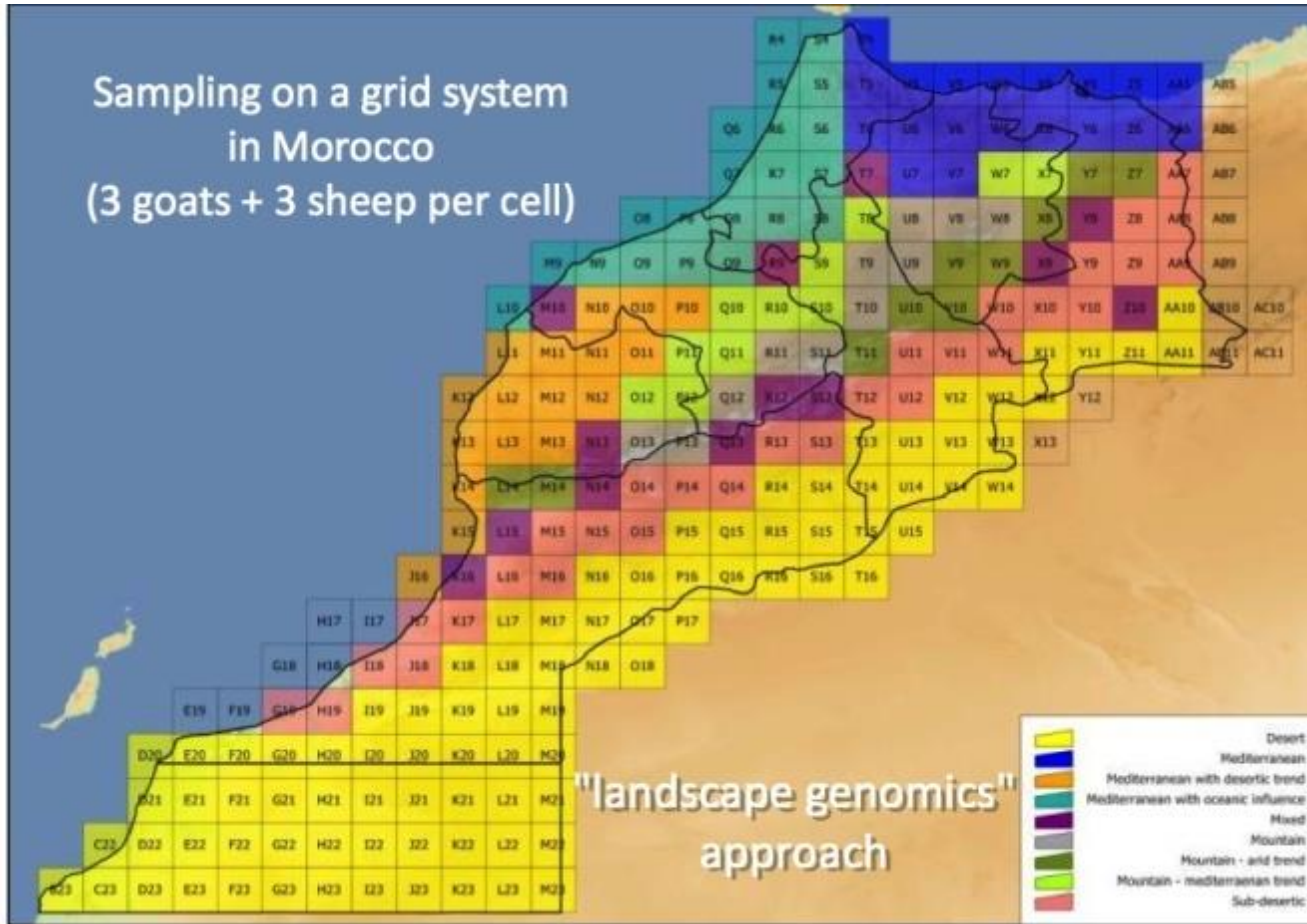
## Landscape genetics: combining landscape ecology and population genetics

Stéphanie Manel<sup>1</sup>, Michael K. Schwartz<sup>2</sup>, Gordon Luikart<sup>1</sup> and Pierre Taberlet<sup>1</sup>





# NextGen project



Pierre Taberlet



Francois Pompanon



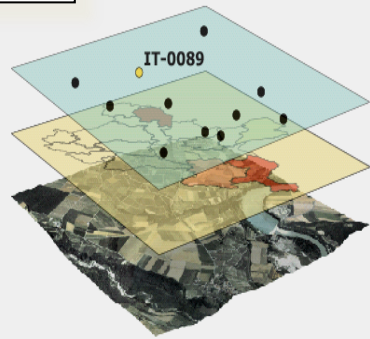
Next generation methods to preserve farm animal biodiversity by optimizing present and future breeding options.



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# Interdisciplinarity and training

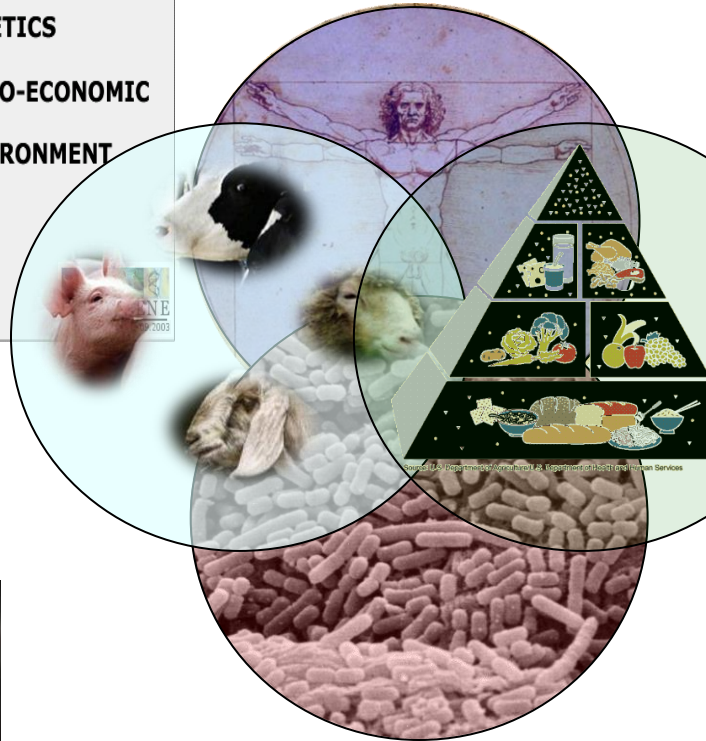
GIS



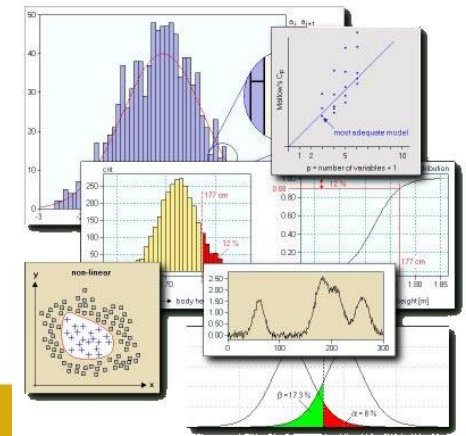
GENETICS

SOCIO-ECONOMIC

ENVIRONMENT



## Training









# Final consideration

- Very fast molecular tool development.
- Faster than our capacity to understand.

Under these circumstances any loss of diversity before characterization is a loss of unvaluable opportunity for science and agriculture



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# ACKNOWLEDGMENTS



Marco Milanese



Elia Vajana



Lorenzo Bomba



Licia Colli

## **Goat farmers**

**International Goat Consortium (SNPChip)**

**ASSONAPA (Italian small ruminant breeder association)**