

Between Europe and the Mediterranean: High Resolution Y-Chromosome Structure of Italy - Preliminary Results

S. Sarno¹, A. Boattini¹, D. Yang Yao¹, S. De Fanti¹, M. Sazzini¹, G. Ciani¹, A. Useli²,
B. Martinez-Cruz³, J. Bertranpetit³, D. Comas³, D. Luiselli¹, D. Pettener¹

¹ Area di Antropologia, Dip. di Biologia ES, Univ. di Bologna. E-mail: alessio.boattini2@unibo.it

² Dip. di Zoologia e genetica Evoluzionistica, Univ. di Sassari

³ Institute of Evolutionary Biology, UPF, Barcelona

KEY WORDS: Y chromosome, Italy, genetic variation.

Introduction

The Italian Peninsula, due to its pivotal geographical position, located in the heart of Mediterranean Sea, but, at the same time, tightly connected with Europe, was involved in all the principal colonization phases of the continent, thus offering a unique opportunity to test hypotheses on the peopling of the northern Mediterranean coast and southern Europe. Nevertheless, despite its geographical and historical importance, only few studies (Di Giacomo *et al.*, 2003, Capelli *et al.*, 2007) have been specifically focused on the genetic history of Italian population, since Cavalli-Sforza pioneering study (1994) about the “geography and history of human genes”. Within the worldwide “Genographic Project” workframe, this research aims to increase and update our knowledge about Italian population genetic history, by increasing the specificity of sampling strategy and the resolution power of Y-chromosome uniparental molecular markers analyzed. Here we are going to present the preliminary results.

Materials and Methods

A total of 1016 samples from 8 sampling areas and 25 populations were collected. The sampling strategy was built on a preliminary surname-based analysis (Boattini *et al.*, in press). Groups of homogeneous provinces were aggregated following the surname-based clusters, and, within them, a specific set of provinces was selected taking into account Italian geographic and historical background. For each province, samples were selected based on the standard grandparents criterion, taking also into account the presence of founder surnames. DNA samples were extracted from fresh blood by a Salting Out modified protocol. Samples were genotyped with a set of 90 Y-SNP, using TaqMan Assays (Applied Biosystems). All samples were additionally amplified for 19 Y-STR, 17 typed with the standard Y-filer Kit (Applied Biosystems), and the remaining two loci (DYS388, DYS426) typed in a separate multiplex (Multiplex 2), along with 6 out of 90 SNP

(M91, M139, M60, M175, M186, M17). Both the Y-SNPs and Y-STRs genotyping were performed in collaboration with the Human Genome Diversity Group at the Institut de Biologia Evolutiva, PRBB-UPF (Barcelona, Spain).

Haplogroup frequencies were estimated by chromosome counting. Standard measures of genetic diversity and pairwise F_{st} genetic distances were carried out with Arlequin v3.5.

The genetic structure of Italian samples were investigated by Principal Component Analysis, performed on HGs frequencies using R software. Synthetic maps were obtained superimposing the first three principal component contour maps (PC1 red, PC2 green, PC3 blue), created with the Surfer Golden software following the Kriging interpolation procedure.

Results and Discussion

A total of 989 Y-chromosomes were successfully genotyped for all markers, and clustered in 50 different haplogroups following the Y chromosome phylogeny (Karafet *et al.*, 2008).

The gene diversity (h), calculated using HGs frequencies, highlights the genetic peculiarity of the Sardinian samples which reach the lowest values, with a south-north decreasing pattern (from 0.8752 of Cagliari-Carbonia-Iglesias to 0.8110 of Olbia-Tempio). The same trend is also observed for peninsular samples – excluding Sardinia – with a linear decrease for h values with increasing latitudes ($R^2=0.2691$, P -value=0.01598) (data not shown).

The outgroup position of Sardinia is also remarked by the PCA analysis (Fig.1). This pattern is probably due to the “autochthonous” haplogroup I2a1-M26, which is rare or absent outside of the island. Removing the Sardinian samples the genetic landscape within the Italian Peninsula becomes more complex (Fig. 2). The Italian samples are distributed along the PC1 axis following a North-South direction, but highlighting a different behavior between a more homogeneous cluster of south-central populations, and a heterogeneous group of northern ones. Our results – besides confirming the presence of general clines across the Italian Peninsula along the NW-SE direction – highlight a more complex genetic framework resulting from the

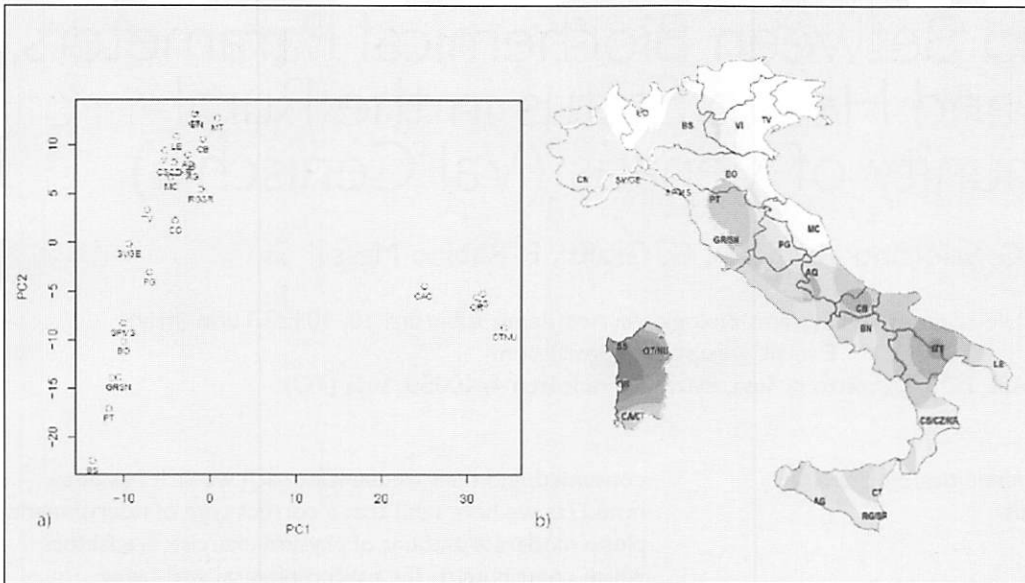


Fig. 1. a) Plot of the principal component analysis conducted on the Hg frequencies of the Italian Samples (PC1 - 36.2% vs PC2 - 19.5%). b) Synthetic map the first three PCs.

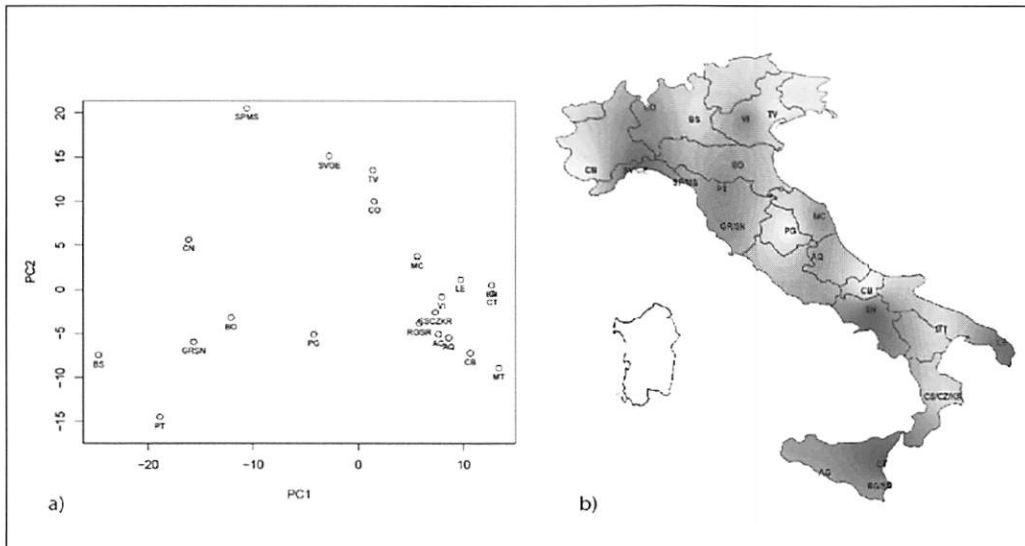


Fig. 2. a) Plot of the principal component analysis conducted on the Hg frequencies of Italy without Sardinia (PC1 - 33.3% vs PC2 - 18.3%). b) Synthetic map the first three PCs.

several historically stratified evolutionary events that, during the history, have shaped the genetic population differentiation. A higher inter-population variability is observed in the northern populations, while a greater intra-population diversity emerges in the southern ones. In addition, some populations have shown local specific pattern of genetic variation.

Thanks to the availability of such a detailed and complete molecular dataset, the completion of Y-Chromosome analysis, so as the typing of mtDNA, will consent to investigate and clarified the complex history of Italy, as well as to shed light on the central role of Italian Peninsula within the European and Mediterranean genetic context.

References

Boattini A., Lisa A., Fiorani O., Zei G., Pettener D., Manni F. General method to unravel ancient population structures through surnames. Final validation on Italian data. *Hum. Biol.*, in press.

- Capelli C., Brisighelli F., Scarnicci F., Arredi B., Caglià A., Vetrugno G., Tofanelli S., Onofri V., Tagliabracci A., Paoli G., Pascali V.L., 2007. Y chromosome genetic variation in the Italian peninsula is clinal and supports an admixture model for the Mesolithic-Neolithic encounter. *Mol. Phylogenet. Evol.*, 44(1): 228-239.
- Cavalli-Sforza L.L., Menozzi P., Piazza A. 1994. *The history and geography of human genes*. Princeton University Press. Princeton (New Jersey).
- Di Giacomo F, Luca F, Anagnou N, Ciavarella G, Corbo RM, Cresta M, Cucci F, Di Stasi L, Agostiano V, Giparaki M, Loutradis A, Mammi' C, Michalodimitrakis EN, Papola F, Pedicini G, Plata E, Terrenato L, Tofanelli S, Malaspina P, Novelletto A., 2003. Clinal patterns of human Y chromosomal diversity in continental Italy and Greece are dominated by drift and founder effects. *Mol. Phylogenet. Evol.*, 28(3): 387- 395.
- Martínez-Cruz B., Ziegler J., Sanz P., Sotelo G., Anglada R., Plaza S., Comas D., The Genographic Consortium, 2011. Multiplex single-nucleotide polymorphism typing of the human Y chromosome using TaqMan probes. *Investig. Genet.*, 2(1): 13.