

Analysis of the Y Chromosome in The Population of a Mountain Village, Postua

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Introduction

Postua is a small village (around 500 inhabitants) in the Sessera Valley, province of Vercelli, on the slopes of the Biellese pre-Alps. The name Postua derives from the Latin *post*, in the sense of the last settlement of the foothills. Its geographical position, far from the main byways of the area, has led to a certain isolation of the community.

Postua was settled in ancient times, presumably in the Celtic era, since the zone contained iron deposits. The first written documentation of the village's existence dates to the Middle Ages: a document from 1279 refers to Postua and its forges.

Materials and methods

The sample consisted of 102 individuals native to Postua for at least 3 generations.

We analysed 23 biallelic polymorphisms (SNPs) of the Y chromosome, the most diffuse in Europe. Six of them were discovered by Underhill in March 2002.

To complete the picture provided by the SNPs, we also analysed 4 Y chromosome microsatellites.

Results

There are only 6 haplotypes in Postua, 2 of which are fairly diffuse in the population.

The Postua data were then compared with those of other Italian and European populations (Fig. 1).

We then calculated the genetic diversity to clarify the position of Postua with respect to other populations from northern and central Italy (Trino Vercellese, Asiago and Murlo in Tuscany). The values are high for Asiago and Murlo (from 0,73 to 0,79), but lower for Postua (0,53) and Trino (0,55), which appear to be more closed populations with lower genetic diversity.

The genetic distances were used for Delaunay triangulation, and first- and second-order genetic barriers were identified between the populations (Fig. 2). A first-order barrier separates Asiago and Murlo from the Piedmont populations, while a second-order barrier isolates Postua from the other Piedmont populations. The identified barriers and the isolated position of Postua are supported by the results of the AMOVA test: the variance

among groups is 6,73%, with $P=0,05$, which confirms the validity of the clusters.

For the Piedmont populations, the study of SNPs was combined with an analysis of several microsatellites: the trinucleotide DYS 392 and the tetranucleotides DYS 19, DYS 391, DYS 393. The distribution of the allele frequencies is very similar in the three populations.

The results of the microsatellite analysis were combined with those of the SNPs to form 23 haplogroups (marked by 23 biallelic polymorphisms), including 74 haplotypes. The AMOVA revealed a significant difference among the haplogroups: the variance is rather high (25,79%) with a very low P value (0,0001). There are several population differences: Trino is well represented in the M269 haplogroup, with many haplotypes only present in this population, while it is almost absent in M173. In contrast, Postua is almost absent in M269 but well represented in M173.

The AMOVA did not show a very great difference among the three populations when all 23 haplogroups were considered, but the variance among populations increased to 11,13% when only the M269 and M173 haplogroups were considered. This indicates that the greatest differences among the populations are between these 2 haplogroups.

Conclusions

In this study, the microsatellites allowed us to achieve a more thorough and accurate analysis than with the SNPs alone.

The results for Postua seem to support the hypothesis of genetic isolation, probably due to a restricted number of founder haplotypes and limited gene flow.

References

- [1] Bosch E. et al. 2001. High-resolution analysis of human Y-chromosome variation shows a sharp discontinuity and limited gene flow between northwestern Africa and the Iberian Peninsula. *Am. J. Hum. Genet.*, 68: 1019-1029.
- [2] Lell J.T. et al., 2002. The dual origin and Siberian affinities of Native American Y chromosomes. *Am. J. Hum. Genet.*, 70: 192-206.
- [3] Malaspina P. et al. 2001. A multistep process for the dispersal of a Y chromosomal lineage in the Mediterranean area. *Ann. Hum. Genet.*, 65: 339-349.

[4] Nebel A. et al., 2001. Haplogroup-specific deviation from the stepwise mutation model at the microsatellite loci DYS388 and DYS392. Eur. J. Hum. Genet., 9: 22-26.

[5] Semino O. et al., 2000 The genetic legacy of Paleolithic Homo sapiens in extant Europeans: a Y chromosome perspective. Science: 290: 1155-1159.

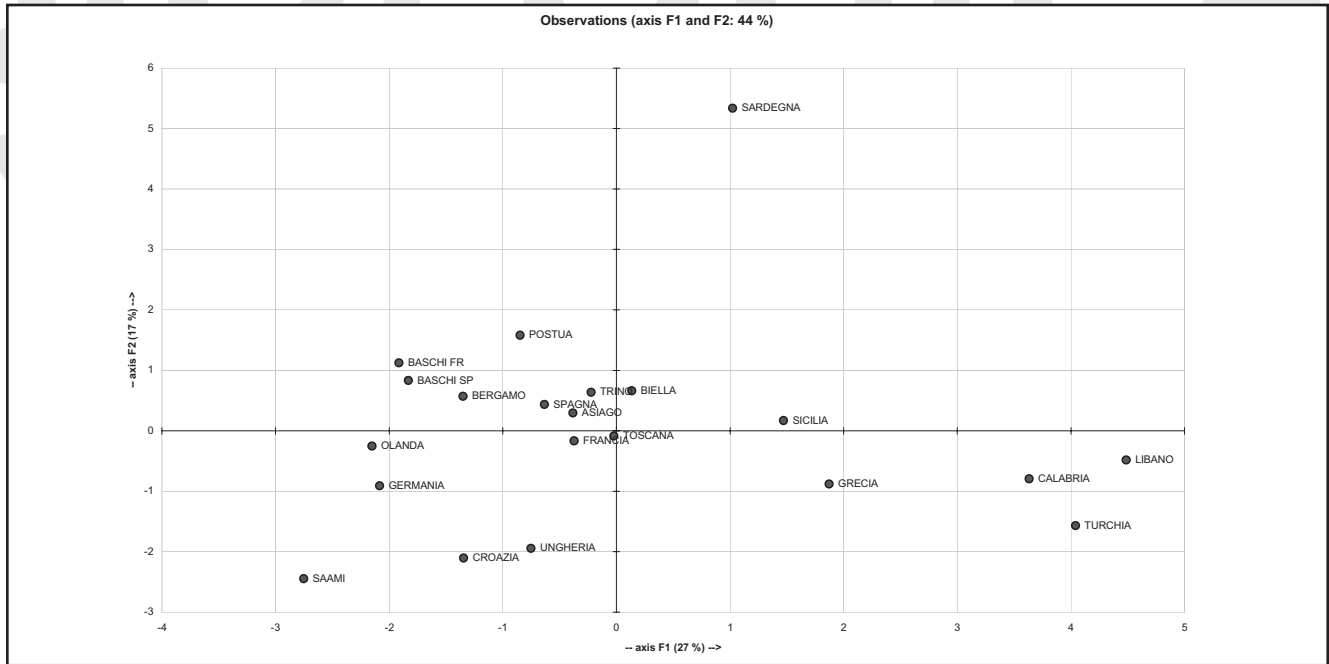


Fig. 1. Principal Components Analysis: Postua is close to the other northern Italian populations and to the Basque and Spanish populations

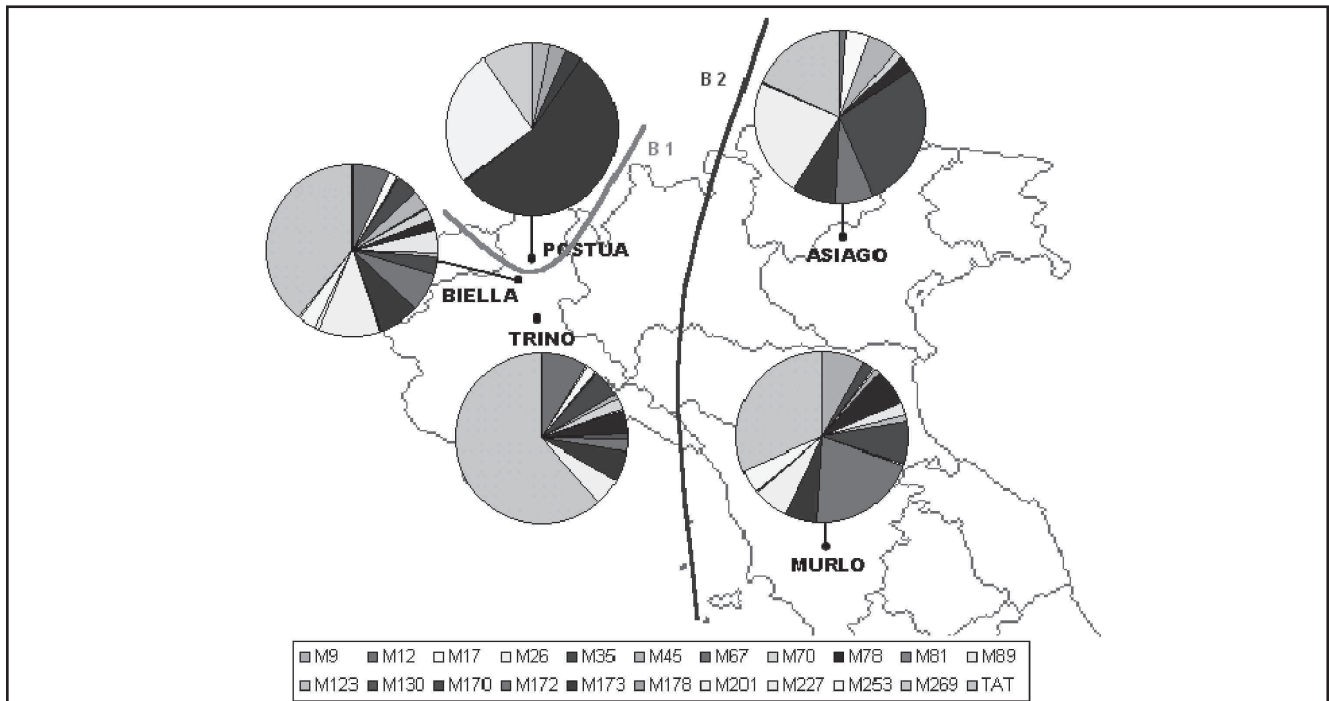


Fig. 2. Haplotype frequencies and Delaunay triangulation: the most diffuse haplotype at Biella, Trino Vercellese and Murlo is the M269 mutation (according to Underhill's nomenclature), while at Asiago it is M170 and at Postua M173. There are only 6 haplotypes at Postua, a lower number than in the other populations.