

# Y-SNPs genetic variation in the Caucasian population living in southern Portugal

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## Intoduction

- The Y chromosome has unique characteristics, namely the genetic non- recombination and the direct father/ son transmission [1].
- Binary polymorphisms located on the non-recombining portion of the human Y chromosome (Y-SNPs) have been widely used for characterizing and classifying paternal lineages in various fields such as genealogy, anthropology and forensics [2]. In anthropological context, Y-SNPs have importance for ancestry analysis, allowing the study of migration phenomena and also human evolution history reconstruction [3].
- The application of these markers in biogeographical studies implies the characterization of haplogroups with different markers, according to geographic region under study. In this research was selected a set of 9 Y-SNPs (92R7, M70, M22, Tat, P25, SRY1532, M173, M213, M9), that determine the most frequent haplogroups in European populations. [4].

## Material and Methods

- A sample of 103 unrelated Caucasian individuals, living in southern Portugal, was selected for this study. Bloodstains were extracted by Chelex® method [5].
- The Y -SNPs multiplex PCR reaction was performed with QIAGEN® Multiplex PCR kit using an Veriti® 96-Well Thermal Cycler amplification system (Applied Biosystems). The amplified products were purified with Exo-SAP-it® (USB®), followed by minisequencing reaction performed with the SNaPshot Multiplex kit™ Kit (Lifetech) according to manufacturers protocol. Sequenced products were purified with SAP (USB®), analyzed in an automatic sequencer ABIPrism®3130 Genetic Analyzer and analyzed with GeneMapper 3.2.1 software. (Applied Biosystems) [4].
- The nomenclature was established according to the YCC [6], Jobling and Tyler-Smith [7] and Karafet [8]. Haplogroup frequencies were calculated with SPSS IBM v 21.0.

## Results

In this population sample 3 haplogrupos were detected: R1\* which represented 67% of the sample (Fig.1), followed by haplogroup F (xK) represented by 22% (Fig.2) and the haplogroups BCD or E with 11% representation (Fig. 3).

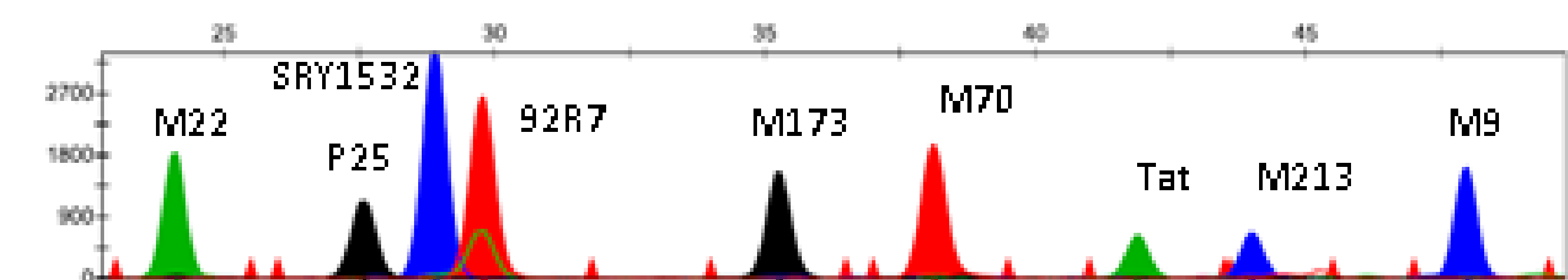


Fig. 1. Haplogroup R1\*.

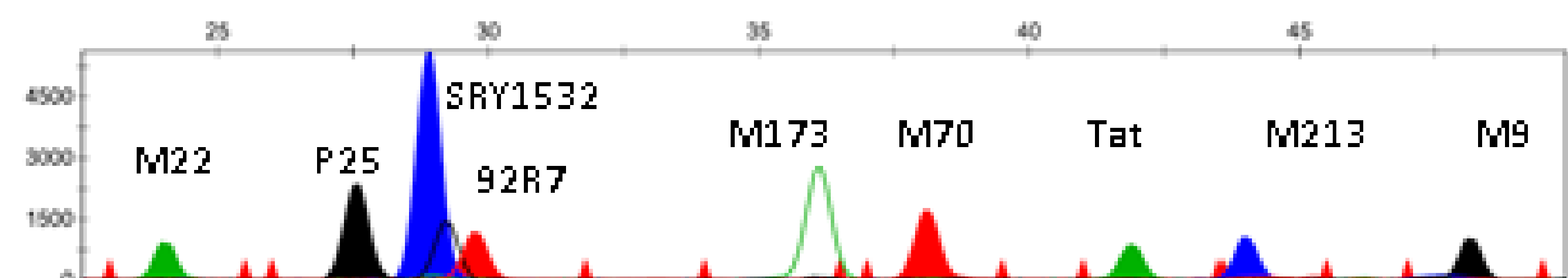


Fig. 2. Haplogroup F.

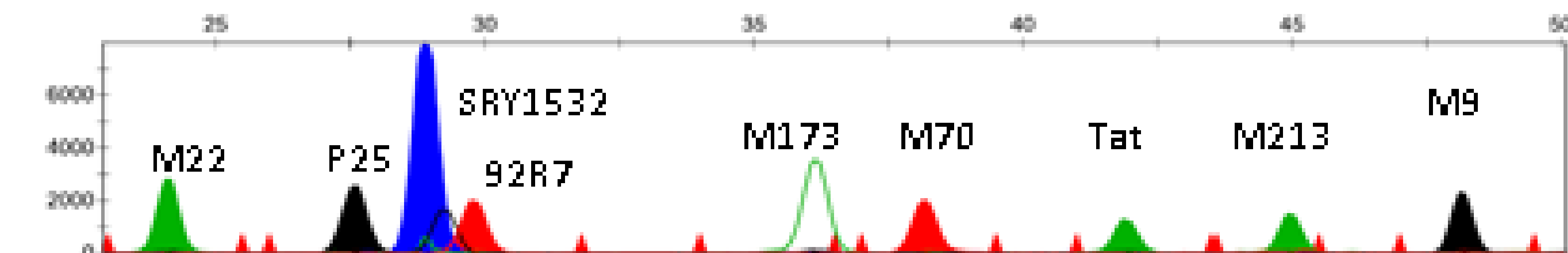


Fig. 3. Haplogroup BCD ou E.

Table 1: Frequencies of haplogroups present in the Caucasian population.

| Haplogroup | Frequencies | Percentage (%) |
|------------|-------------|----------------|
| R1*        | 69          | 67%            |
| F (xK)     | 23          | 22%            |
| BCD ou E   | 11          | 11%            |

HAPLOGROUPS OF CAUCASIAN POPULATION

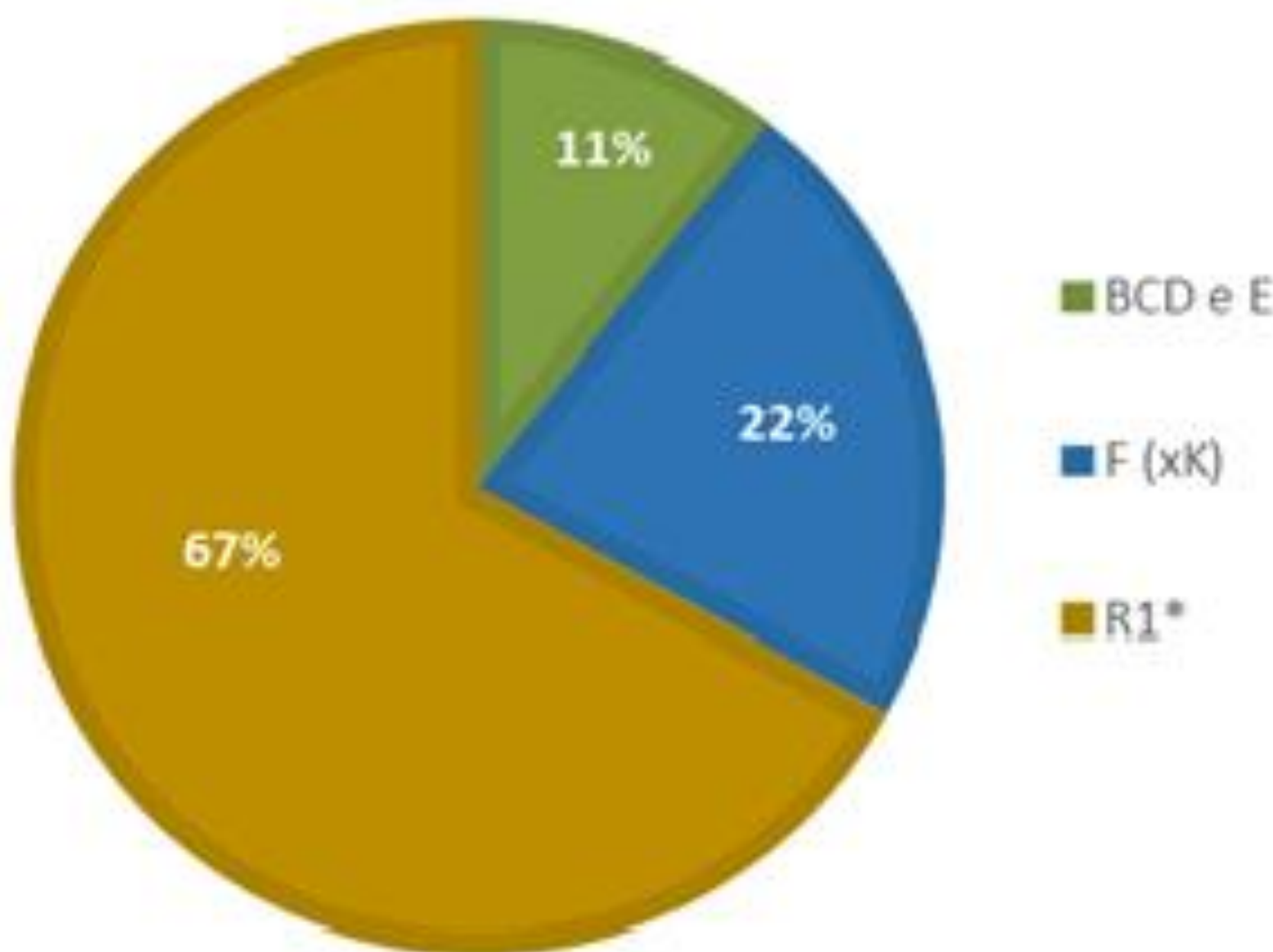


Fig. 4. Haplogroup frequencies in a sample from Causasian population (Portugal).

## Discussion

- The Y chromosome lineage most frequent in Portugal is the represented by haplogroup R1\*. This haplogroup is the most frequent among European populations, being present all over the world [8].
- The presence of haplogroup F could be a reflection of human evolution, effect of the Paleolithic gene pool, resulted from Human expansion that followed the exit of the African continent in direction to Asia and the rest of the world.
- The detection of haplogroups BCD or E are a consequence of the colonization of African continent (haplogroup B and E are typically African), which contributed to the presence of a large number of African immigrants in Portugal. Consequently, Portuguese Caucasians have genetic information about African ancestral Y chromosome lineages.

## Conclusion

- The application of Y- SNPs allowed the characterization of the Caucasian population residing in southern Portugal. These Y-SNPs revealed importance in the ancestry analysis as well as the study of human migration phenomena.
- According to published studies [9], as expected, the results show up in line, revealed that most of the male population from south Portugal belong to haplogroup R1\*.
- For more conclusive results, further studies must be performed in this population to different multiplexes so that these new results can then be used by forensic laboratories.

## Referências

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