Y-Chromosome variation and inter-haplotype mutational distances in 111 unrelated individuals from Tuscany, Italy

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Abstract

The phylogenetic median-joining network algorithm applied to 111 Y-chromosomes from Tuscany typed at 10 STR loci showed the existence of at least two well-separated clusters of haplotypes. We suggest that they correspond to different haplogroups.
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1. Introduction

Blood samples from 111 unrelated individuals were typed for the following Y-chromosome STR loci DYS19, DYS389-I, DYS389-II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS439, DYS438, DYS385 (a and b).

2. Materials and methods

The amplification protocol involved three multiplex PCR reactions [1], the amplified fragments being separated by an ABI Prism 310 (PE Applied Biosystems).

Mutational distance was calculated for each pair of haplotypes as the absolute difference between the number of repeats observed at each locus, summed across loci; each haplotype was paired all others (6105 pairs).

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Median-joining network analysis was computed by the program NETWORK 3.0 [2] (http:// www.fluxus-engineering.com/sharenet.htm), using the phylogenetic median-joining network algorithm.

3. Results

A total of 106 haplotypes were unique, 1 haplotype was observed twice and another was observed three times (for a total of 108 different haplotypes in 111 chromosomes).

For calculating the inter-haplotype mutational distances we excluded locus DYS385, since its dual product cannot be resolved into alleles of different loci. The number of unique haplotype dropped to 90 (81%).

The distribution of the number of mutational steps was included between 0 (identical haplotypes) and 19. The distribution was bimodal, with a first peak at $m = 5$ mutational
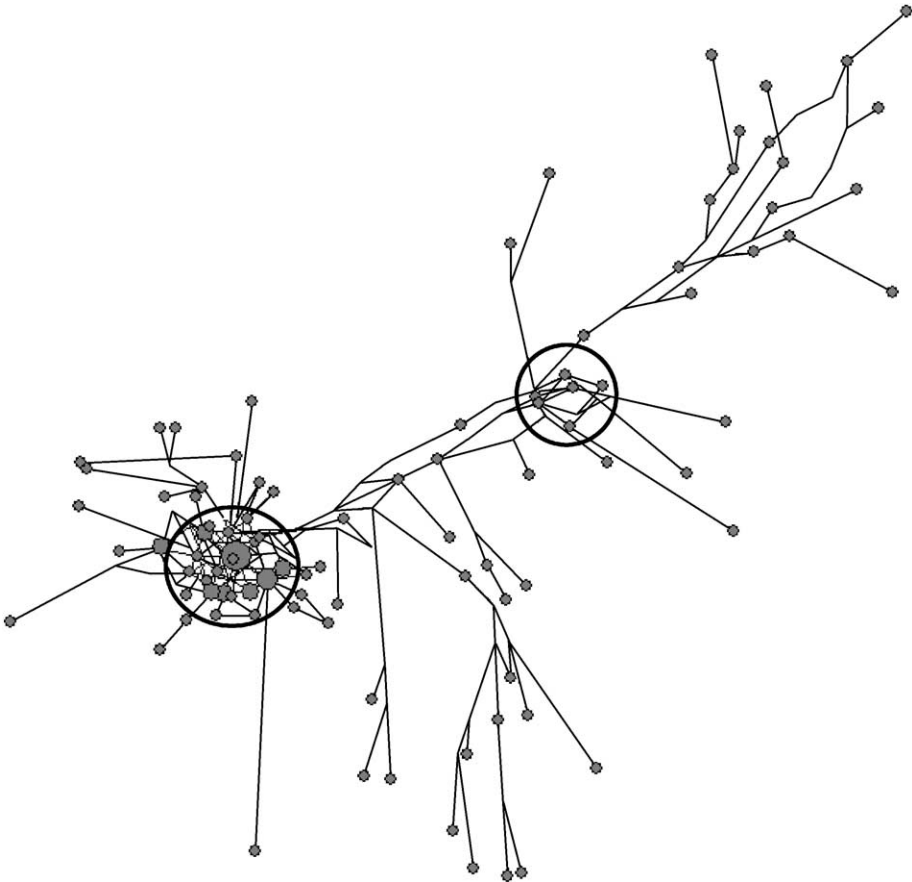

Fig. 1.

steps and a second peak at $m = 12$. This suggested that the sample was composed of at least two clusters of haplotypes, both connected to a number of related haplotypes (small m), but separated one from the other by a large value of m. Interestingly, all haplotypes represented by more than one chromosome were separated by a small number of steps, indicating their probable derivation from the same ancestor haplotype.

To verify the hypothesis of the existence of multiple clusters of haplotypes, each separated by a small number of mutational steps, we applied to our data the phylogenetic median-joining network algorithm (see Fig. 1). It may be seen that a large number of haplotypes, including those represented by more than one individual are separated by a small number of mutational steps, and that a second cluster of haplotypes is topologically well separated from the first.

4. Discussion

Among 111 unrelated subjects from Tuscany typed at 11 STRs, we found all Y-chromosome haplotypes different from each other, with the exception of three individuals sharing the same haplotype. A comparison with another population sample from Northern Italy [1], typed at the same loci, showed a similar pattern of variation. The more common haplotype in this series (found in 4 out of 122 individuals) was also the same haplotype shared by three Tuscan subjects.

The DYS385 locus was responsible of a significant part of the discriminating power, but we did not consider it in the analysis of mutational distance.

We analyzed the differences between haplotypes in term of total number of mutational steps, and found that a large number of haplotypes were separated from the commonest haplotype by a single step. In addition, we noticed a second cluster of haplotypes, all of them being unique, whose members were separated one from the other by a single step. The median-joining network analysis confirmed this hypothesis, showing that a smaller cluster of haplotypes was clearly identifiable. We are evaluating the hypothesis that these two cluster of haplotypes correspond to different haplogroups, as defined by SNPs.

References

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