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Y-chromosome STR defined haplotypes in North Portugal

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Abstract

The Y-specific STR loci, DYS19, DYS385, DYS389I and II, DYS390, DYS391, DYS392, DYS393, DYS434, DYS437, DYS438, DYS439 and GATA A10, were studied in the Northern Portuguese population. In a sample of 212 individuals, it was possible to define 196 different haplotypes of which 182 were found only once, 12 were found in two samples and the 2 most frequent haplotypes were shared by three individuals. The observed haplotype diversity value was 0.9992. The usefulness of the inclusion of each of these new markers for forensic purposes is discussed, comparing expected and observed increase in haplotype diversity.

Keywords: North Portugal; Y-STRs; Haplotypes; Discrimination power; Haplotype diversity

1. Introduction

The Y-STR Haplotype Reference Database is the most extensive survey on European populations (available at http://ystr.charite.de). Recently, a second database was generated (Y-STR Haplotype Reference Database for US Populations; http://ystr.org/usa). These databases are very important for forensic users since it is well known that there are difficulties in the creation of large databases for the Y-linked markers [1].

The addition of new Y-STRs to the previous set will allow an increment on the haplotype diversity value and consequently a higher power of discrimination. However,

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this implies the need for re-constructing haplotypic databases [1] as well as the development of new PCR multiplex strategies. For these reasons, it is important to know quantitatively the extra information obtained when further markers are added to a previously defined set.

The aim of this work was to construct haplotypes including the nine STR loci, corresponding to the minimal haplotype of the Y-STR Haplotype Reference Databases, plus five additional markers (DYS434, DYS437, DYS438, DYS439 and GATA A10) in order to evaluate the usefulness of their inclusion in routine forensic analysis.

2. Material and methods

Samples were collected from 212 unrelated healthy blood donors from the north Portuguese population and DNA extracted as described by Valverde et al. [2].

DYS19, DYS389I and II, DYS390 and DYS393 were amplified as described by Gusmão et al. [3]. DYS391, DYS434, DYS437 and DYS439 were amplified according to Beleza et al. [4].

DYS385, DYS438 and GATA A10 were amplified in a 12.5-μl reaction volume comprising 1.5 mM $MgCl_2$, 1×buffer, 0.5U Taq Gold polymerase (PE), 200 μM dNTPs, 0.24 μM DYS385 primers [5], 0.3 μM DYS438 primers [6] and 0.12 μM GATA A10 primers [7]. After a 95 °C pre-incubation for 11 min; 10 cycles: 94 °C for 30 s, 62 °C for 30 s and 72 °C for 30 s; 20 cycles: 94 °C for 30 s, 60 °C for 30 s and 72 °C for 30 s; and 20 min final extension at 60 °C.

DYS392 was amplified in a 12.5-μl reaction volume comprising 1.5 mM $MgCl_2$, 1×buffer, 0.5U Taq DNA Polymerase recombinant (MBI Fermentas), 200 μM each dNTP and 0.2 μM of each primers (G00-456-509). Amplifications were performed in 30 cycles of 30 s at 94 °C, 30 s at 58 °C and 1 min at 72 °C.

Detection system: ABI310 and ABI377 automatic sequencers (Applied Biosystems); Genescan 2.1 Analysis software.

Allele and haplotype frequencies were estimated by gene counting. Gene and haplotype diversities were estimated according to Nei [8].

3. Results and discussion

In a sample of 212 individuals, it was possible to define 196 different haplotypes (Table 1) of which 182 were found only once, 12 were shared by two samples and the 2 most frequent haplotypes were shared by three individuals.

The observed haplotype diversity value was 0.9992. In the same sample, when combining the new markers with the classical set (DYS19, 385, 389I and II, 390, 391, 392 and 393) a 0.68% increase in haplotype diversity was obtained and the number of different haplotypes rose from 157 to 196 (Table 1).

The haplotype diversity values were calculated adding each of the five additional markers to the nine Y-STR core (Table 1). All the markers contribute to an increment on the number of different haplotypes, with DYS434 the locus that contributes the least.

Table 1
Haplotype number and diversity values in a sample of 212 individuals from a Northern Portuguese population, when combining different Y-STR sets

Haplotype composition	No. of different haplotypes	Haplotype diversity (%)
9 Y-STR set[a]	157 (157/212=74.06%)	99.25
plus DYS434	162	99.32
plus DYS437	164	99.52
plus DYS438	164	99.38
plus DYS439	175	99.72
plus GATA A10	175	99.70
14 Y-STR set[b]	196 (196/212=92.45%)	99.92
minus DYS434	196	99.92
minus DYS437	193	99.90
minus DYS438	195	99.92
minus DYS439	186	99.85
minus GATA A10	186	99.86

[a] Nine loci core set included in the Y-STR Haplotype Reference Databases.
[b] Fourteen Y-STR set studied in this work.

The informative contribution of DYS434, 437, 438, 439 and GATA A10 to an extended Y-STR database was evaluated by calculating the haplotype diversity values excluding each of the five additional markers from the whole 14 Y-STR set (Table 1). When DYS434 is not considered, the haplotype diversity is not affected. A decrease in the haplotype diversity was obtained when excluding any of the other four markers (DYS437, 438, 439 and GATA A10).

Comparing these loci, it is possible to conclude that the increase in haplotype diversity is not directly correlated to gene diversity. Indeed, when DYS438 is not considered, although it has a high gene diversity (0.6050), the haplotype diversity is the same as for the whole 14 STR core set and the number of different haplotypes is only slightly affected, with the loss of just one haplotype. With the inclusion of DYS437, the haplotype diversity is barely affected, distinguishing only three more haplotypes.

The markers that proved to be more useful for the inclusion in routine forensic analysis were DYS439 and GATA A10. Although these two markers have different gene diversity values (higher for DYS439), both contribute to the same increase in the number of different haplotypes (from 186 to 196).

In conclusion, we can state that, in the context of this population, DYS437, DYS438, DYS439 and GATA A10 contribute to a higher power of discrimination when added to the previous set of Y-STRs (those included in the Y-STR Haplotype Reference Database). The inclusion of DYS434 in routine forensic analysis will not increase the power of discrimination and, moreover, with the marginal disadvantage of an increase of the mutation rate expected for the whole haplotype [9].

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