



## Portuguese population study with 16 Y-STR loci

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**Abstract.** The aim of this study was to present a Portuguese Population Database of 214 Southern Portugal samples studied with 16 Y-STR loci including the eight minimal Y-STR haplotype—DYS19, DYS390, DYS391, DYS392, DYS393, DYS389I/II, DYS385 and other Y-STR loci—GATA A7.1 (DYS460), GATA C4, GATA A7.2 (DYS461), GATA A10, GATA H4, DYS437, DYS438 and DYS439. In this study, 209 different haplotypes could be defined and 205 haplotypes were found just once. This database has a discrimination power of 97.7%, a haplotype diversity of 99.6% and 2.3% of shared haplotypes. Some samples do not match any sample in the Y-STR Haplotype Reference Database which assigned specific region characteristics to these population samples. Forty-three alleged father–child non-exclusion cases were also studied and two possible mutations have been detected in DYS439 and DYS393. Complex paternity investigation cases without alleged father have also been studied. In an autosomal STR exclusion case, the same 16 Y-STR haplotype was obtained between the excluded alleged father and the child. © 2003 Elsevier B.V. All rights reserved.

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### 1. Introduction

Although autosomal STRs are commonly used for forensic investigations, Y-chromosome STRs provide very useful information in paternity investigation cases without alleged father and in other investigation cases where paternal lineage identification can be assessed.

In Forensic Genetics, it is necessary to have population databases concerning the main populations which the Forensic Laboratory used to work with. The aim of this study is to present a Portuguese Population Database mainly from Southern Portugal studied with 16 Y-STR loci including the eight minimal Y-STR haplotype—DYS19, DYS390, DYS391, DYS392, DYS393, DYS389I/II, DYS385 and other Y-STR loci—GATA A 7.1 (DYS460), GATA C4, GATA A 7.2 (DYS461), GATA A10, GATA H4, DYS437, DYS438 and

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DYS439, included in the Y-Chromosome Quality Control Group of the Spanish and Portuguese Group (GEPY) of the ISFG.

## 2. Materials and methods

After DNA extraction with the Chelex method, Y-STR amplifications of 214 samples from Southern Portugal population were carried out in a 9600, 9700 or 2400 Perkin-Elmer thermocycler in five multiplex reactions: Pentaplex—DYS393, DYS390, DYS19, DYS389I/II [1]; Triplex I—DYS391, DYS437, DYS439 and Triplex II—DYS392, DYS385, DYS438 [2]; GEPY I—GATA A7.2 (DYS461), GATA C4, DYS437, DYS438 and GEPY II—GATA A7.1 (DYS460), GATA A10, DYS439, GATA H4 [3,4]. Samples were analysed in an ABI Prism 3100 DNA Sequencer with Genescan (v. 3.7).

## 3. Results and discussion

Table 1 shows some results of the 16 Y-STR Haplotype Portuguese Population Database, mainly haplotypes shared by several individuals. A total of 214 males were analysed for all 16 Y-STR loci—209 different haplotypes could be defined and 205 haplotypes were found just once. This database has a discrimination power of 97.7%, a haplotype diversity of 99.6% and 2.3% of shared haplotypes.

When considering the eight minimal Y-STR haplotype, we can detect haplotypes shared by several individuals making haplotype clusters up to seven non-related individuals. With 16 Y-STR loci, the discrimination power was successfully increased, with haplotype clusters involving only two or three non-related individuals as h15, h82, h169 and h173 in Table 1.

Minimal haplotypes of all samples have been compared with the Y-STR Haplotype Reference Database. When considering the eight minimal Y-STR haplotype, seven individuals shared the most frequent haplotype also detected in different populations in

Table 1  
Some 16Y-STR haplotypes detected in a Southern Portuguese population

Haplotype	N	DYS 19	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 385	DYS 437	DYS 438	DYS 439	DYS 460	DYS 461	A10	GATA C4	GATA H4
h1	1	12	13	30	24	10	13	13	11/15	15	12	13	11	12	14	23	27
h2	1	13	12	28	24	10	11	13	17/17	14	10	11	10	13	14	21	27
h15	3	13	14	30	24	9	11	13	13/14	14	10	10	11	13	14	21	28
h16	1	13	14	30	24	9	11	13	13/15	14	10	10	11	13	14	21	28
h82	2	14	13	29	24	11	13	13	11/14	15	12	13	11	12	15	23	27
h83	1	14	13	29	24	11	13	13	11/14	15	12	13	11	12	15	24	27
h99	1	14	13	30	22	9	11	12	13/18	15	9	12	11	13	15	24	27
h133	1	14	14	30	24	11	13	13	11/15	15	12	12	11	13	14	23	28
h134	1	14	14	30	24	11	13	13	11/15	15	12	11	11	12	14	23	28
h169	2	15	13	29	24	11	13	13	11/14	15	12	12	11	12	15	23	28
h173	2	15	13	30	22	11	11	13	14/14	16	10	12	10	12	14	21	27
h201	1	16	15	32	22	10	12	15	16/17	15	10	12	11	12	15	21	27
h208	1	17	13	28	23	10	11	13	12/12	15	10	11	10	11	16	22	28
h209	1	17	13	29	24	11	11	12	14/18	15	9	13	10	13	14	21	27

the Y-STR Haplotype Reference Database (351 matches)—h78 to h83. Only two non-related individuals shared the corresponding extended 16 Y-STR haplotype—h82. Three non-related individuals shared the most frequent haplotype—h15 in the Southern Portuguese population database.

Haplotypes differing by just one or two repeat units such as h15–h16, h82–h83 or h133–h134 can be encountered in this database. Some minimal haplotypes have no matches in the Y-STR Haplotype Reference Database as, for example, h99 and h201.

### 3.1. Forensic cases

Studying 43 alleged father–child non-exclusion cases with 17 autosomal STRs performed with SGMPlus and Powerplex16, two Y-STR genetic incompatibilities have been detected in DYS439 (12 → 13) and in DYS393 (14 → 13), in cases with  $W = 99.9999992\%$  and  $W = 99.999999998\%$ , respectively.

Otherwise, in 14 alleged father–child exclusion cases, 5 to 14 genetic incompatibilities were detected in each case, being DYS385, DYS389II, DYS439 and DYS460 the loci with more exclusions.

The 16-YSTR loci were also used to investigate complex forensic cases without alleged father as in an exclusion paternity case with two alleged half-brothers and a child detecting two different paternal lineage and in an non-exclusion paternity case including an alleged half-brother and a child sharing the same 16 Y-STR haplotype. In an autosomal STR exclusion case with a  $W = 99.995\%$  (without considering the exclusion systems), the same Y-STR haplotype was obtained between the excluded alleged father and the child, suggesting the same paternal lineage.

The 16 Y-chromosome STR haplotype seems to be extremely helpful in population studies to determine specific region characteristics and in forensic cases to obtain unique paternal lineage profiles.

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