



Portuguese Population Study with 16 Y-STR Loci



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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 investigation cases where paternal lineage identification can be assessed.

The aim of this study is to present an **extended 16 Y-STR Portuguese Population Database of 214 Southern Portugal individual samples** for forensic purpose, including the 8 Y-STR minimal haplotype – **DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385** (Gusmão et al, 2001, D Syndercomb-Court, personal communication) and Y-STR loci included in the Y-Chromosome GEP-ISFG collaborative exercise – **DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4** (Paula Sánchez-Diz et al, Results of the GEP-ISFG collaborative study on two Y-STR tetraplexes – GEPYI and GEPY II, 2002).

Results and Discussion

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

Haplotype	N	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS385	DYS437	DYS438	DYS439	DYS460	DYS461	GATA 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
h3	1	13	13	29	24	9	12	13	16/18	14	10	11	11	12	14	24	27
h4	1	13	13	29	24	10	11	13	18/18	14	10	12	9	12	14	21	28
...
h12	1	13	13	30	25	10	11	13	16/18	14	10	12	9	12	15	22	28
h13	1	13	13	31	23	10	11	13	17/17	14	10	12	10	11	15	20	26
h14	1	13	13	31	24	10	11	13	17/17	14	10	12	10	12	14	22	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
h17	1	13	14	30	24	10	11	13	13/14	14	10	9	12	13	15	22	28
h18	1	13	14	30	25	9	11	13	13/14	14	10	10	11	13	14	21	29
...
h79	1	14	13	29	24	11	13	13	11/14	15	12	11	10	12	15	23	28
h80	1	14	13	29	24	11	13	13	11/14	15	12	12	10	13	15	23	28
h81	1	14	13	29	24	11	13	13	11/14	15	12	12	12	15	23	29	
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
h84	1	14	13	29	24	11	13	13	11/15	15	12	12	11	12	14	24	28
h85	1	14	13	29	24	11	13	13	11/15	15	12	13	11	12	14	23	28
...
h130	1	14	14	30	24	11	13	13	09/14	15	12	11	11	12	15	25	28
h131	1	14	14	30	24	11	13	13	11/14	14	12	12	10	12	15	23	28
h132	1	14	14	30	24	11	13	13	11/14	15	12	12	11	12	15	23	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
h135	1	14	14	30	24	11	13	13	12/14	15	12	12	10	12	15	23	28
h136	1	14	14	30	24	11	13	13	12/14	15	12	12	11	13	15	23	29
h137	1	14	14	30	24	12	13	13	11/16	16	12	11	11	12	15	21	28
...
h166	1	15	13	29	23	11	12	15	16/16	14	10	12	12	12	15	21	27
h167	1	15	13	29	24	10	13	13	11/15	15	12	12	11	12	15	23	28
h168	1	15	13	29	24	10	13	14	12/16	14	9	11	11	12	14	23	26
h169	2	15	13	29	24	11	13	13	11/14	15	12	12	11	12	15	23	28
h170	1	15	13	29	24	11	13	13	11/14	15	12	13	11	12	14	24	28
h171	1	15	13	29	24	11	13	13	12/14	15	12	12	11	11	15	23	28
h172	1	15	13	29	24	11	13	14	11/14	15	12	12	11	12	14	24	28
h173	2	15	13	30	22	11	11	13	14/14	16	10	12	10	12	14	21	27
h174	1	15	13	30	24	10	11	13	19/19	14	10	11	11	12	14	20	28
h175	1	15	13	30	24	11	13	13	11/14	15	12	11	11	12	14	23	29
h176	1	15	13	30	24	11	13	13	11/14	15	12	12	11	12	15	23	28
h177	1	15	13	30	24	11	13	13	11/15	15	12	11	11	12	15	23	27
h178	1	15	13	31	21	10	11	14	15/18	14	11	13	10	13	15	21	27
...
h212	1	16	14	32	22	10	11	12	13/15	14	9	11	10	13	15	21	28
h213	1	16	15	32	22	10	12	15	16/17	15	10	12	11	12	15	21	27
h214	1	17	13	28	23	10	11	13	12/12	15	10	11	10	11	16	22	28
h215	1	17	13	29	24	11	11	12	14/18	15	9	13	10	13	14	21	27

HAPLOTYPE POPULATION DATABASE

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**
- A total of **209 different haplotypes** could be defined and **205 haplotypes were found just once** – **Discrimination - 97,7%** **Shared Haplotypes – 2,3%** **Haplotype Diversity – 99,6%**

Considering the 8 minimal Y-STR haplotype, the most frequent haplotype shared by 7 individuals was:

N **DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385**
7 **14 13 29 24 11 13 13 11-14 15 12 13 11 12 15 23 27** **h78 to h83**

and can also be detected in different populations in the Y STR Haplotype Reference Database with **351 matches**



Considering these 7 samples, one **16 Y-STR haplotype** was shared by **2 non-related individuals** – **h82**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
14 13 29 24 11 13 13 11-14 15 12 13 11 12 15 23 27

Two other different haplotypes were shared by **2 non-related individuals** – **h169 and h173**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
15 13 29 24 11 13 13 11-14 15 12 12 11 12 15 23 28
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
15 13 30 22 11 11 13 14-14 16 10 12 10 12 14 21 27

The **most frequent haplotype h15** was shared by **3 non-related individuals**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
13 14 30 24 9 11 13 13-14 14 10 10 11 13 14 21 28

Some haplotypes differ by just one, two or three repeat units – **h15-h16, h82-h83, h133-h134**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
13 14 30 24 9 11 13 13-14 14 10 10 11 13 14 21 28
13 14 30 24 9 11 13 13-15 14 10 10 11 13 14 21 28

Some minimal haplotypes have no matches in the Y STR Haplotype Reference Database – **h99, h207**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
14 13 30 22 9 11 12 13-18 15 9 12 11 13 15 24 27
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
16 15 32 22 10 12 15 16-17 15 10 12 11 12 15 21 27

FORENSIC CASES

In **43 non-exclusion alleged father-child cases** - studied with 17 autosomal STRs SGM Plus and Powerplex16 with paternity probability (W) between 99,9999% and 99,99999999%, **two genetic incompatibilities were detected:**

One genetic incompatibility in **DYS439** in a paternity case with a **W = 99,9999992%**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
13 13 31 24 10 11 13 17-17 14 10 12 10 12 14 22 27
13 13 31 24 10 11 13 17-17 14 10 13 13 10 12 14 22 27

One genetic incompatibility in **DYS393** in a paternity case with a **W = 99,99999998%**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
14 13 29 23 11 13 14 11-14 14 10 12 10 12 14 22 27
14 13 29 23 11 13 13 11-14 14 10 13 10 12 14 22 27

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

In an **exclusion case** with **W value - 99,995%** without considering the exclusion systems, the **same Y-STR haplotype** was obtained **between the alleged father and the child**
DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
14 13 29 25 11 13 13 11-13 15 11 13 10 12 16 23 28
14 13 29 25 11 13 13 11-13 15 11 13 10 12 16 23 28

COMPLEX FORENSIC CASES

- Exclusion paternity case

Two alleged half-brothers and a child

DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
14 13 29 24 10 13 14 11-15 14 12 12 11 12 16 23 27
14 13 29 24 10 13 14 11-15 14 12 12 11 12 16 23 27
15 12 29 22 10 11 12 12-14 15 10 12 10 12 16 25 27

- Non-exclusion paternity case

An alleged half-brother and a child

DYS19, DYS389 I/II, DYS390, DYS391, DYS392, DYS393, DYS385, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4
14 14 30 25 10 14 13 11-14 15 12 13 10 12 14 23 28
14 14 30 25 10 14 13 11-14 15 12 13 10 12 14 23 28

Conclusions

- When performing a 16 Y-STR study the **genetic diversity was successfully increased**. With the 8 Y-STR minimal haplotype, haplotype clusters can be detected involving several non-related individuals, whereas with a 16 Y-STR haplotype, **haplotype clusters involve only 2 or 3 non-related individuals**.
- The **16 Y-chromosome STR haplotypes** seems to be **extremely important to solve complex forensic cases** and in **population studies** to obtain **unique paternal profiles** and to determine **specific population characteristics**.