Analysis of Y-chromosomal haplotypes and haplogroups distribution in a population sample from Portugal (central area)

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Abstract. The aim of this study was to analyse the distribution of Y-chromosomal haplotypes and haplogroups found in central Portugal. In this work, we analysed 102 unrelated individuals of central Portugal. Combining the allelic state of 10 biallelic markers (YAP, SRY-8299, 92R7, 12f2, SRY-1532, SRY-2627, Tat, SY81, M9, LLY22g), we defined the haplogroup to which each sample belonged. Y-chromosomal haplotypes were defined by 16 Y-Short Tandem Repeats (STR) (DYS19, DYS385 a/b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4 and GATA H4). This population study defined 8 different haplogroups and 101 different haplotypes, where 100 haplotypes were unique and 1 was found in two apparently unrelated individuals, both belonging to the same haplogroup. © 2003 Elsevier B.V. All rights reserved.

Keywords: Y chromosome; Haplotypes; Haplogroups; STRs; Biallelic markers

1. Introduction

The comparative analysis of Y-chromosomal biallelic markers and Short Tandem Repeats (STR) in a population may be useful to verify if the distribution of diversity is well balanced [1].

For this purpose, we analysed the distribution of Y-chromosomal haplotypes defined by 16 STRs (Y DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392,

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DYS393, DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4 and GATA H4) in the eight different haplogroups defined by 10 biallelic markers (YAP, SRY-8299, 92R7, 12f2, SRY-1532, SRY-2627, Tat, SY81, M9, LLY22g) found in a population sample from Central Portugal.

In the present work, we evaluated the advantage of population analysis based in Y-chromosomal haplotypes and haplogroups (STRs and biallelic markers) in population genetics, evolutionary studies and forensic applications.

2. Materials and methods

DNA was extracted from blood stains collected from 102 unrelated Portuguese males (Central area), by Chelex 100 [3].

Ten biallelic markers (eight Single Nucleotide Polymorphisms (SNP), the Alu insertion YAP-DYS287 and the 12f2 deletion) were studied in a population from central Portugal. Biallelic markers were amplified by PCR and SNPs were also analysed by RFLP through an enzymatic digestion with the respective restriction enzyme. SRY1532, *Dra*III [4]; SRY8299, *Bsr*BI [4]; M9, *Hin*fI [5]; YAP-DYS287 [6]; 92R7, *Hin*dIII [7]; SRY2627, *Bsi*HKAI [8]; TAT, *Nla*III [9]; SY81, *Nla*III [10]; LLY22g, *Hin*dIII (Righetti and Tyler-Smith, Personal communication); 12f2 (Rosser et al., personal communication).

Fragment was detected by electrophoresis in PhastGel™ Gradient 10–15, with PhastGel™ SDS Buffer Strips (PhastSystem™-Amersham Pharmacia Biotech). Fragment length was determined through comparative analysis with 25- or 123-bp DNA ladder (Gibco®).

The Y-STRs DYS19, DYS389I, DYS389II, DYS390 and DYS393 were amplified as described by Gusmão et al. [11]. The DYS385 a/b amplification conditions complied with the methodology described by Schneider et al. [12]. Multiplex amplification of DYS391 DYS392, DYS393 was carried out according to Kloosterman et al. [13].

The Y-STR (DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4 and GATA H4) were described by Ayub et al. [14] and White et al. [15] and were amplified with two PCR tetraplex reactions (GEPY I and GEPY II), using the protocol according to Sanchez-Diz et al. [16].

PCR was performed with Applied Biosystems 9600 and 2700 thermocyclers. Electrophoresis was carried out in ABI377 automated sequencer (4% polyacrylamide denaturing sequencing gels) and in ABI310 genetic analyser. Genotype classification was done using Genescan analysis software with Local Southern Method. Samples were typed by comparison with the allelic ladders obtained using a mixture of previously amplified samples for the most common alleles.

3. Results and discussion

The frequencies of the observed SNP haplogroups (according to Jobling and Tyler-Smith [17]) and STR haplotypes are shown in Table 1.

For the Y-STRs, 101 different haplotypes were observed: 100 haplotypes were unique and 1 was found in two apparently unrelated individuals, both belonging to the same haplogroup (hg 1).

Table 1
Observed haplogroups (HG) and haplotypes among 102 Portuguese males (Central area)

HG	n	%	Different STR haplotypes
hg 1	53	51.96	52
hg 2	15	14.71	15
hg 3	1	0.98	1
hg 8	5	4.90	5
hg 9	17	16.67	17
hg 21	2	1.96	2
hg 22	1	0.98	1
hg 26	8	7.84	8

The results of this population analysis based in Y-chromosomal haplotypes and haplogroups are according to the expected. Comparisons with other Iberian samples analysed for the same markers showed a substantial similarity [18].

4. Conclusion

Population surveys based in Y-chromosomal haplotypes and haplogroups (STRs and biallelic markers loci) seem to be the best strategy for the use and validation of Y polymorphisms, for instance, in the haplotype reference databases not only for forensic applications, but also to perform comparative population genetics analysis and evolutionary studies [1,2].

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