

SNP and STR Y chromosome markers in the Canary Islands population

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Abstract. Single nucleotide polymorphism (SNP) typing was done with a PCR-based assay with co-amplification of 25 DNA fragments and detection of 35 Y chromosome SNPs by means of minisequencing with Snapshot technique. The Y SNP package can identify 34 Y chromosome haplogroups including the majority of the Y chromosome haplogroups of interest in the Canary Islands population. A total of 348 and 269 unrelated Canary Islands males were typed for 35 Y-SNPs and 8 Y-STRs, respectively. We observed 16 Y chromosome SNP haplogroups with frequencies from 0.3% to 48%. The majority of the individuals carried the Y chromosome haplogroup R1b or I of European origin while ~ 13% and ~ 7% of the Y chromosome haplogroups found indicated African and Middle Eastern heritage. Further diversity was detected when additional typing of Y chromosome STR markers was performed. © 2003 Elsevier B.V. All rights reserved.

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1. Introduction

Y chromosome-specific single nucleotide polymorphisms (SNPs) are particularly useful for identification of stable paternal lineages because of their low rate of parallel and back mutations. By testing highly polymorphic Y-STR markers in addition to the Y-SNP markers, an even more detailed DNA profile can be obtained. Here, we have typed 35 Y-SNP and 8 Y-STR markers in a population from the Canary Islands.

2. Materials and methods

DNA samples from unrelated males from Canary Islands were typed for 35 Y chromosome SNPs (348 individuals) and for 8 Y-STRs (269 individuals). SNP typing was done as previously described [1]. The minimal Y-STR haplotype was analyzed

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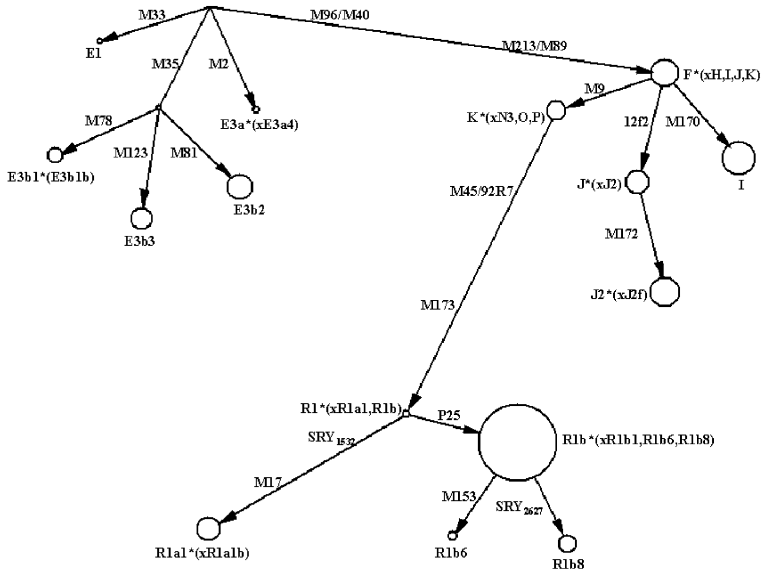


Fig. 1. Median-joining network based on 35 Y chromosome SNPs. The area of each circle is proportional to the frequency. The arrows indicate the likely direction of mutation events according to the YCC tree (2002).

following the protocol recommended by the International Forensic Y-User Group. Median-joining network was calculated using the program Network 3.111.

3. Results

We used the 35 Y-SNP package to type 348 unrelated Canary Islands males, and we observed 16 different haplogroups. Most of the haplogroups were one-step derivatives (Fig. 1). Haplogroup frequencies from 0.3% to 48% were identified. Two large haplogroups

Table 1
Observed SNP haplogroups and STR haplotypes among 269 Canary Islands people

Haplogroups	N	Frequency (%)	Number of different Y-STR haplotypes
E1	1	0.4	1
E3a*(x E3a4)	2	0.7	2
E3b*	1	0.4	1
E3b1*(E3b1b)	5	1.9	5
E3b2	19	7.1	11
E3b3	7	2.6	6
F*(xH,I,J,K)	15	5.6	12
I	30	11.2	27
J*(xJ2)	11	4.1	9
J2*(xJ2f)	15	5.6	14
K*(xN3,O,P)	10	3.7	9
R1*(xR1a1,R1b)	2	0.7	2
R1a1*(xR1a1b)	15	5.6	12
R1b*(xR1b1,R1b6,R1b8)	125	46.5	75
R1b6	3	1.1	3
R1b8	8	3.0	7

Table 2
Y-STR haplotypes that occurred three or more times in the most frequent haplogroups

Haplogroup	Allele at								Frequency
	DYS	DYS	DYS	DYS	DYS	DYS	DYS	DYS 385	
	19	389 I	389 II	390	391	392	393	I/II	
E3b2	13	14	30	24	9	11	13	13,14	5/19
E3b2	13	14	30	23	9	12	13	13,14	3/19
I	15	13	28	23	11	12	15	15,16	3/30
R1b*(xR1b1,R1b6,R1b8)	14	13	29	24	11	13	13	11,14	12/125
R1b*(xR1b1,R1b6,R1b8)	14	14	30	24	11	13	13	11,14	7/125
R1b*(xR1b1,R1b6,R1b8)	14	13	30	24	11	13	13	11,14	6/125
R1b*(xR1b1,R1b6,R1b8)	14	13	29	24	11	13	13	11,15	5/125
R1b*(xR1b1,R1b6,R1b8)	14	12	27	24	10	14	12	11,14	3/125
R1b*(xR1b1,R1b6,R1b8)	14	13	29	23	11	13	13	11,14	3/125
R1b*(xR1b1,R1b6,R1b8)	14	13	29	24	10	13	13	10,16	3/125
R1b*(xR1b1,R1b6,R1b8)	14	13	29	24	11	13	13	10,15	3/125
R1b*(xR1b1,R1b6,R1b8)	14	13	29	25	10	13	13	11,14	3/125
R1b*(xR1b1,R1b6,R1b8)	14	14	30	24	11	13	13	11,17	3/125

STR alleles are indicated by the number of repetitions.

assigned to the European lineage were found: R1b*(xR1b1,R1b6,R1b8) (47.6%) and I (9.7%). The major haplogroup E (~ 13%) and haplogroup J2*(xJ2f) (6.9%) was also detected. In addition to the SNP analysis, 269 of the samples were also typed for eight Y-STRs. A total of 190 different Y-STR haplotypes were observed; thus 79 males did not have a unique STR haplotype within this population (Tables 1 and 2). The 79 males without a unique haplotype belonged to 34 different haplotype groups. By combining SNP and STR results, the 79 males were divided into 40 haplotype groups.

4. Discussion

In the population from Canary Islands analysed here, almost half of the males belong to the haplogroup R1b and in most of the individuals where a unique haplotype could not be assigned to a sample, the individual belonged to this haplogroup. Additionally, we typed the minimal Y-STR haplotype defined in the European reference database (<http://ystr.org>). Only 70% of the 269 individuals had a unique haplotype. The combined use of Y-SNP and Y-STR markers increased the number of different haplotypes. However, it is clear that additional SNP markers are needed before the full potential of SNP markers can be exploited in forensic caseworks from Canary Islands.

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Reference

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