

Chromosome Y haplotypes database in a Venezuelan population

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Abstract. The non-recombining portion of human Y chromosome is a source of polymorphic regions for the analysis of male DNA. The PowerPlex® Y System consisting of 12 Y-STR markers (DYS19, DYS385a/b, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS438, DYS439 and DYS437) is a useful tool for database creation. In turn these databases allow us to determine and correlate different haplotype distributions, and the reconstruction of phylogenetic relationships among human populations. In this work, a chromosome Y database corresponding to 62 individuals living in Caracas, Venezuela is presented. This is an important step leading to the implementation of this important forensic tool in our country. As in other studies, our results indicate that the 12 Y-STRs makers here studied are useful markers for forensic and paternity testing. © 2005 Published by Elsevier B.V.

Keywords: Y-STR; Venezuelan database; PowerPlex® Y System

1. Introduction

The core of a forensic genetic laboratory is the use of autosomal markers analysis; however, mtDNA and Y-STRs haplotypes represent exceptional tools that provide information from a non-recombining lineage [1]. Since a large portion of crimes is committed by men, Y-STRs haplotypes are essential in the identification mixed samples such as in the case of rapes. Numerous populations have already been investigated; however, there is scarce information about Y-chromosomal haplotypes in a Venezuelan population. The use of PowerPlex® Y System permitted us to construct a database for using in routine practice that includes the minimal and extended recommend Y-STRs haplotypes [1].

2. Materials and methods

Population: The population sample consisted of 62 unrelated individuals from Caracas City, all of them with four grandparents born in Venezuela. Following recommendations of the bioethics committee from Venezuelan Government agency (FONACIT), an informed consent form was signed by all participants.

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Table 1
Frequencies for 12 Y-STR loci of unrelated males from Caracas (N=62)

Allele	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439	DYS385
9					0.0968				0.1290		10–14 0.0161
10					0.4677	0.0323			0.2258	0.1129	11–13 0.0161
11					0.4193	0.4032			0.2097	0.2903	11–14 0.2742
12	0.0161	0.1935			0.0161	0.0161	0.1452		0.4355	0.4194	11–15 0.1129
13	0.1129	0.5645				0.4677	0.7258	0.0161		0.1452	12–12 0.0161
14	0.4677	0.2258				0.0806	0.0645	0.5000		0.0323	12–13 0.0323
15	0.2581	0.0161					0.0645	0.4194			12–14 0.0161
16	0.0645							0.0645			12–15 0.0161
17	0.0806										13–13 0.0484
18											13–14 0.0806
19											13–15 0.0323
20											14–16 0.0161
21				0.1452							14–17 0.0161
22				0.0484							14–18 0.0484
23				0.3065							14–19 0.0161
24				0.4194							15–16 0.0323
25				0.0806							15–17 0.0323
26											16–17 0.0484
27											16–18 0.0161
28			0.1129								16–19 0.0323
29			0.3548								17–17 0.0323
30			0.2871								17–18 0.0161
31			0.1290								17–20 0.0161
32			0.0161								18–18 0.0161
GD	0.6909	0.5926	0.6946	0.7003	0.5958	0.6108	0.4438	0.5697	0.6988	0.7050	0.8892

GD= gene diversity.

DNA extraction: A blood drop was collected on FTA® cards (GIBCO-BRL) by pricking subjects' fingers. Samples were processed with the FTA™ Purification Reagent from GIBCO-BRL following the manufacturer's recommendations.

DNA amplification: STR amplification was carried out according to the PowerPlex® Y System kit (Promega) protocol using a PTC-100 thermocycler device (MJ Research®).

Typing: STR analysis was performed on an ABI Prism™ 310 Genetic Analyzer, and automated allele sizing was performed using Genotyper® Software v. 3.7 containing PowerTyper Y Macro [2].

Quality control: Proficiency testing of the GEP-ISFG WG [3].

Data analysis: Statistical evaluations were performed with Arlequin version 2000 software [4]. Gene diversity value was calculated according to Nei [5].

3. Results and discussion

The allele frequencies and D of the 12 Y-STRs in the Caracas, Venezuela population are shown in Table 1. Table 2 shows the haplotypes found for the 12 Y-STRs loci. The haplotype diversity was 0.9979 (S.D.=0.0034) and haplotype frequencies were 0.0161 and 0.0322 for those found once or twice, respectively.

Acknowledgements

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Table 2
Y-STR haplotypes

H	N	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439
C1	1	12	16–18	13	30	24	10	11	13	14	10	11
C2	1	13	14–19	12	30	23	10	14	12	15	11	12
C3	1	13	13–14	13	30	24	9	11	13	14	10	10
C4	1	13	15–17	13	30	24	10	14	13	15	11	13
C5	1	13	15–16	14	30	22	10	14	13	14	12	13
C6	1	13	13–14	14	30	24	9	11	13	14	10	10
C7	1	13	13–14	14	29	25	9	11	13	14	10	10
C8	1	13	14–18	14	31	23	11	11	13	14	10	12
C9	1	14	13–14	12	28	23	10	11	13	16	10	11
C10	1	14	11–15	12	29	24	11	13	13	15	12	12
C11	1	14	11–15	13	28	24	10	13	13	15	12	12
C12	1	14	11–14	13	29	23	10	12	13	14	12	12
C13	1	14	11–14	13	29	23	10	13	13	15	12	11
C14	1	14	12–14	13	29	23	11	13	13	15	12	12
C15	1	14	12–15	13	29	23	11	13	14	14	12	12
C16	1	14	10–15	13	29	23	12	13	13	15	12	12
C17	1	14	10–14	13	29	24	11	13	13	14	12	12
C18	1	14	11–14	13	29	24	11	13	13	14	12	13
C19	1	14	11–14	13	29	24	11	13	13	15	12	12
C20	1	14	11–14	13	29	24	11	13	13	15	12	13
C21	2	14	11–15	13	29	24	11	13	13	15	12	13
C22	2	14	11–15	13	29	24	11	13	12	15	12	12
C23	1	14	11–14	13	29	25	10	13	13	15	10	13
C24	1	14	11–14	13	30	23	11	13	13	15	12	14
C25	1	14	16–17	13	30	24	10	11	13	14	11	12
C26	1	14	14–18	13	32	23	10	11	13	14	10	12
C27	1	14	13–13	14	29	22	10	13	14	15	9	11
C28	1	14	11–14	14	30	23	11	13	13	14	12	12
C29	1	14	11–14	14	30	24	11	13	13	14	12	11
C30	1	14	13–13	14	30	23	10	11	12	15	9	11
C31	1	14	11–14	14	30	23	11	13	13	15	12	12
C32	1	14	13–14	14	30	24	9	11	13	14	10	10
C33	1	14	11–13	14	31	24	10	13	13	15	12	13
C34	1	14	13–13	14	31	24	10	13	13	15	12	11
C35	1	14	11–14	15	31	24	11	13	13	14	12	11
C36	1	15	13–15	12	28	23	10	10	12	15	9	11
C37	2	15	16–19	12	28	24	10	11	12	14	9	12
C38	1	15	14–17	12	28	24	11	11	12	16	9	12
C39	1	15	13–15	12	29	23	10	11	14	15	10	12
C40	1	15	11–14	12	29	23	11	13	13	15	12	13
C41	1	15	14–16	12	29	24	10	14	13	14	9	11
C42	1	15	15–16	12	30	22	10	10	14	16	10	12
C43	1	15	11–14	13	29	23	10	13	13	15	12	11
C44	1	15	12–13	13	29	24	11	13	13	14	12	12
C45	1	15	11–14	13	29	25	10	13	13	14	11	11
C46	2	15	16–17	13	30	21	11	11	13	14	11	10
C47	1	15	11–14	13	30	24	11	13	13	14	10	11
C48	1	15	15–17	13	31	21	11	11	13	14	11	11
C49	1	15	14–18	13	31	23	10	14	13	14	9	11
C50	1	16	12–13	12	28	21	9	11	12	16	10	11
C51	1	16	11–14	13	29	23	11	13	13	15	12	12
C52	1	16	17–20	13	30	21	11	11	15	14	11	11
C53	1	16	17–17	14	31	21	10	11	13	14	11	14
C54	1	17	12–12	14	29	25	9	11	13	15	10	12
C55	1	17	17–18	13	30	21	10	11	15	13	11	12
C56	1	17	18–18	13	30	21	10	11	15	14	11	12
C57	1	17	11–14	13	30	25	10	11	13	14	11	10
C58	1	17	17–17	13	31	21	10	11	15	14	11	11

H=number of haplotypes, N=individuals per haplotype.