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Y-chromosome STR haplotypes in two population samples: Azores Islands and Central Portugal

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Abstract

The Y-chromosome haplotypes defined by nine STRs (DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393) were studied in 207 unrelated individuals from Central Portugal and 63 from Azores Islands. The most common haplotype in Central Portugal was shared by 3.4% of the males, while 160 haplotypes were unique. In Azores Islands the most common haplotype was shared by 6.4% of the males, while 40 haplotypes were unique. The values of haplotype diversity were 0.993 for Central Portugal and 0.976 for Azores Islands.

Keywords: Y-chromosome STRs; PCR multiplex; Forensic genetics; Population genetics

Population: Blood samples were obtained from 207 healthy unrelated males from Central Portugal and 63 from Azores Islands.

Extraction: DNA was extracted using Chelex 100 protocol described by Walsh et al. [1]. The quantity of recovered DNA was determined using QuantiBlot® Human DNA Quantitation Kit (Applied Biosystems).

PCR: DYS19, DYS389I, DYS389II, DYS390 and DYS393 were amplified as described by Gusmão et al. [2]. The DYS385 amplification conditions complied with the methodology described by Schneider et al. [3] and multiplex amplification of DYS391, DYS392, DYS393 was carried out according to Kloosterman et al. [4]. PCR was performed with an Applied Biosystems 9600 thermocycler.

The allele nomenclature system was proposed by Kayser et al. [5] and de Knijff et al. [6] with the exception of the DYS389 locus. The nomenclature of this locus was defined in accordance with Roewer et al. [7].

Typing: Electrophoresis was carried out on 4% polyacrylamide denaturing sequencing gels in a 377 automated sequencer (Applied Biosystems). Genotype classification was done using Genescan analysis software with local southern method and by side-to-side comparison with allelic ladders. These ladders were kindly provided by Dr. Carracedo (Institute of Legal Medicine of Santiago de Compostela), Dr. P.M. Schneider (Institut für Rechtsmedizin, Mainz, Germany) and Dr. Kloosterman (The Forensic Science Laboratory, Rijswijk, The Netherlands).

Results: The results are shown in Tables 1 and 2.

Quality control: Proficiency testing of GEP-ISFG WG (http://www.usc.es/gep-isfg).

Data analysis: The haplotype diversity was calculated according to Nei [8]. Analysis of molecular variance (AMOVA) was performed with the Markov test using the Arlequin software 1.1 [9]. The genetic distance matrix between populations was obtained by using the pairwise difference genetic distance.

Access to data: geneforense@dcinml.mj.pt.

Other remarks: In Central Portugal a total of 178 different haplotypes were observed (Table 1), 160 of them

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Table 1
Y STR haplotypes in Central Portugal population

Haplotype	n[a]	F[b]	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
h1	1	0.0048	13	11.14	10	26	24	10	14	13
h2	1	0.0048	13	11.14	10	26	24	11	12	13
h3	1	0.0048	13	11.14	10	26	24	11	13	13
h4	1	0.0048	13	11.14	10	26	25	11	13	13
h5	1	0.0048	13	11.15	10	26	24	11	13	13
h6	1	0.0048	13	13.14	10	26	24	9	11	13
h7	1	0.0048	13	13.14	11	27	24	9	10	13
h8	2	0.0097	13	13.14	11	27	24	9	11	13
h9	1	0.0048	13	13.14	11	27	24	9	11	14
h10	1	0.0048	13	13.15	11	27	22	10	13	13
h11	1	0.0048	13	14.14	10	26	24	9	10	13
h12	1	0.0048	13	14.14	10	26	25	9	13	13
h13	1	0.0048	13	14.15	11	26	24	9	11	13
h14	1	0.0048	13	14.16	11	27	23	10	13	13
h15	1	0.0048	13	14.17	10	29	25	10	11	13
h16	1	0.0048	13	15.17	10	27	24	10	11	13
h17	1	0.0048	13	16.16	10	29	24	10	11	13
h18	1	0.0048	13	16.17	11	27	24	9	11	14
h19	1	0.0048	13	16.18	10	27	24	10	11	12
h20	1	0.0048	13	16.18	10	27	24	10	11	14
h21	1	0.0048	13	16.18	10	27	24	10	12	13
h22	2	0.0097	13	16.18	10	27	25	10	11	13
h23	1	0.0048	13	17.18	10	27	24	11	11	13
h24	1	0.0048	14	10.13	10	23	25	11	12	13
h25	1	0.0048	14	10.14	10	26	23	10	13	13
h26	1	0.0048	14	10.14	10	26	24	11	13	13
h27	2	0.0097	14	10.14	10	27	24	10	13	14
h28	1	0.0048	14	10.15	10	26	24	10	14	13
h29	1	0.0048	14	11.11	10	26	24	11	12	13
h30	1	0.0048	14	11.11	10	26	24	11	15	13
h31	1	0.0048	14	11.11	11	27	24	11	13	13
h32	1	0.0048	14	11.13	9	25	24	10	13	14
h33	1	0.0048	14	11.13	9	25	24	11	14	13
h34	1	0.0048	14	11.13	10	23	25	11	12	13
h35	1	0.0048	14	11.13	10	26	24	10	13	13
h36	1	0.0048	14	11.13	10	26	24	11	13	12
h37	1	0.0048	14	11.13	10	26	24	11	13	13
h38	1	0.0048	14	11.13	10	26	25	11	11	13
h39	1	0.0048	14	11.13	10	26	25	11	12	13
h40	1	0.0048	14	11.13	10	26	25	11	14	13
h41	2	0.0097	14	11.13	10	27	24	11	13	13
h42	1	0.0048	14	11.14	9	24	24	11	13	13
h43	1	0.0048	14	11.14	9	25	24	11	13	13
h44	1	0.0048	14	11.14	10	24	23	11	13	13
h45	1	0.0048	14	11.14	10	25	24	10	12	13
h46	1	0.0048	14	11.14	10	25	24	10	13	13
h47	1	0.0048	14	11.14	10	25	24	11	13	13
h48	2	0.0097	14	11.14	10	25	24	11	14	13
h49	1	0.0048	14	11.14	10	26	23	10	13	13
h50	1	0.0048	14	11.14	10	26	23	11	13	13
h51	1	0.0048	14	11.14	10	26	24	9	13	13
h52	1	0.0048	14	11.14	10	26	24	10	13	12
h53	6	0.0290	14	11.14	10	26	24	10	13	13
h54	7	0.0338	14	11.14	10	26	24	11	13	13
h55	1	0.0048	14	11.14	10	26	24	11	13	14
h56	1	0.0048	14	11.14	10	26	25	10	13	13
h57	1	0.0048	14	11.14	10	26	25	11	11	13

Table 1 (*Continued*)

Haplotype	n[a]	F[b]	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
h58	2	0.0097	14	11.14	10	26	25	11	13	13
h59	1	0.0048	14	11.14	10	26	27	11	13	12
h60	1	0.0048	14	11.14	10	27	22	10	13	12
h61	1	0.0048	14	11.14	10	27	24	11	13	13
h62	1	0.0048	14	11.14	10	27	25	11	13	13
h63	1	0.0048	14	11.14	11	27	23	11	13	13
h64	1	0.0048	14	11.14	11	27	24	10	13	13
h65	2	0.0097	14	11.14	11	27	24	10	14	13
h66	1	0.0048	14	11.14	11	27	24	11	12	13
h67	4	0.0193	14	11.14	11	27	24	11	13	13
h68	1	0.0048	14	11.14	11	27	24	11	13	14
h69	1	0.0048	14	11.14	11	28	23	11	13	14
h70	1	0.0048	14	11.14	11	28	24	11	12	13
h71	2	0.0097	14	11.14	11	28	24	11	13	13
h72	1	0.0048	14	11.15	10	25	24	11	10	13
h73	1	0.0048	14	11.15	10	26	24	10	12	13
h74	2	0.0097	14	11.15	10	26	24	10	13	13
h75	1	0.0048	14	11.15	10	26	24	11	12	13
h76	1	0.0048	14	11.15	10	26	24	11	13	12
h77	1	0.0048	14	11.15	10	26	24	11	13	13
h78	1	0.0048	14	11.15	11	27	24	11	13	13
h79	1	0.0048	14	11.16	10	26	24	11	13	13
h80	1	0.0048	14	11.16	11	27	23	12	13	13
h81	1	0.0048	14	12.12	10	26	24	10	13	13
h82	2	0.0097	14	12.14	10	26	24	10	13	13
h83	1	0.0048	14	12.14	10	26	24	10	13	14
h84	1	0.0048	14	12.14	10	26	24	11	13	13
h85	2	0.0097	14	12.14	10	27	24	10	13	13
h86	1	0.0048	14	12.14	10	28	22	10	13	13
h87	1	0.0048	14	12.14	11	27	24	12	12	13
h88	1	0.0048	14	12.14	11	28	25	10	13	13
h89	1	0.0048	14	12.15	10	26	25	11	14	13
h90	1	0.0048	14	12.16	10	27	24	11	13	14
h91	1	0.0048	14	12.16	11	27	25	10	13	13
h92	1	0.0048	14	13.13	9	25	22	11	13	13
h93	2	0.0097	14	13.14	9	25	22	10	11	13
h94	1	0.0048	14	13.14	9	25	22	10	12	13
h95	1	0.0048	14	13.14	10	26	22	10	11	13
h96	1	0.0048	14	13.14	10	26	25	10	11	12
h97	1	0.0048	14	13.14	11	27	24	9	11	13
h98	1	0.0048	14	13.15	10	27	22	10	11	12
h99	1	0.0048	14	13.15	10	28	24	10	11	12
h100	1	0.0048	14	13.15	12	29	23	10	11	12
h101	1	0.0048	14	13.16	10	26	24	11	13	13
h102	1	0.0048	14	13.16	10	27	22	10	11	12
h103	1	0.0048	14	13.17	10	26	25	11	12	14
h104	2	0.0097	14	14.14	9	25	22	10	11	13
h105	1	0.0048	14	14.15	9	25	22	10	11	13
h106	1	0.0048	14	14.17	10	27	24	11	13	13
h107	1	0.0048	14	14.18	9	28	22	10	11	12
h108	1	0.0048	14	14.18	10	26	23	10	14	12
h109	1	0.0048	14	14.18	10	26	25	10	11	12
h110	1	0.0048	14	14.18	10	27	22	10	11	12
h111	1	0.0048	14	14.18	10	28	22	10	11	12
h112	1	0.0048	14	14.18	11	27	24	12	11	12
h113	1	0.0048	14	14.19	10	28	22	10	11	12
h114	1	0.0048	14	15.17	11	27	23	10	14	12
h115	1	0.0048	14	15.19	10	28	22	10	12	12

Table 1 (*Continued*)

Haplotype	n[a]	F[b]	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
h116	1	0.0048	14	16.18	9	25	24	10	13	12
h117	1	0.0048	14	16.18	10	28	23	10	11	13
h118	1	0.0048	14	17.18	11	27	23	10	13	12
h119	1	0.0048	15	10.16	11	26	24	10	11	13
h120	1	0.0048	15	11.12	11	27	23	11	13	14
h121	1	0.0048	15	11.13	10	26	25	11	13	13
h122	1	0.0048	15	11.13	11	27	24	10	13	13
h123	1	0.0048	15	11.14	9	26	24	10	13	14
h124	1	0.0048	15	11.14	10	24	23	11	12	13
h125	2	0.0097	15	11.14	10	26	23	11	13	13
h126	2	0.0097	15	11.14	10	26	24	10	13	13
h127	1	0.0048	15	11.14	10	26	24	11	13	13
h128	1	0.0048	15	11.14	11	26	23	11	12	13
h129	1	0.0048	15	11.14	11	27	23	9	12	13
h130	1	0.0048	15	11.14	11	29	24	11	11	13
h131	1	0.0048	15	11.15	10	26	24	10	13	13
h132	1	0.0048	15	11.15	10	26	24	11	13	13
h133	1	0.0048	15	11.15	10	27	24	11	13	13
h134	1	0.0048	15	12.13	10	26	24	11	12	13
h135	1	0.0048	15	12.14	11	27	23	11	13	13
h136	1	0.0048	15	12.15	11	27	24	10	14	13
h137	1	0.0048	15	12.16	9	25	22	10	11	14
h138	1	0.0048	15	13.14	10	26	24	11	13	12
h139	1	0.0048	15	13.15	9	24	22	10	11	13
h140	1	0.0048	15	13.15	10	27	23	10	14	12
h141	1	0.0048	15	13.15	10	27	23	11	11	12
h142	1	0.0048	15	13.16	10	26	24	9	11	12
h143	1	0.0048	15	13.17	9	26	21	10	11	15
h144	1	0.0048	15	13.17	10	26	23	9	11	12
h145	1	0.0048	15	13.18	9	26	25	10	11	12
h146	1	0.0048	15	13.18	10	26	23	9	11	12
h147	1	0.0048	15	13.18	10	26	24	10	14	12
h148	1	0.0048	15	14.14	11	28	23	10	13	12
h149	1	0.0048	15	14.15	9	25	22	10	11	14
h150	1	0.0048	15	14.15	9	26	23	10	11	14
h151	1	0.0048	15	14.15	9	26	23	10	12	13
h152	1	0.0048	15	14.15	9	26	23	10	13	14
h153	1	0.0048	15	14.15	9	27	22	10	11	14
h154	1	0.0048	15	14.16	11	29	23	10	12	13
h155	1	0.0048	15	14.17	11	28	23	10	11	14
h156	1	0.0048	15	15.15	9	26	22	10	10	14
h157	1	0.0048	15	15.15	9	26	22	10	11	14
h158	1	0.0048	15	15.16	9	26	22	10	11	14
h159	1	0.0048	15	15.17	10	26	24	10	11	12
h160	1	0.0048	15	15.17	10	28	24	10	11	13
h161	1	0.0048	15	16.16	11	28	23	10	12	14
h162	1	0.0048	15	16.17	10	28	21	10	14	13
h163	1	0.0048	15	9.16	10	26	22	10	14	12
h164	1	0.0048	16	11.14	10	27	25	11	11	13
h165	1	0.0048	16	11.15	10	26	24	11	13	13
h166	1	0.0048	16	12.12	10	25	23	11	11	13
h167	1	0.0048	16	12.15	10	26	21	9	11	13
h168	1	0.0048	16	13.13	10	26	23	9	11	12
h169	1	0.0048	16	13.15	10	26	25	10	14	14
h170	1	0.0048	16	13.15	10	28	21	10	14	14
h171	1	0.0048	16	13.16	10	26	23	9	11	12
h172	1	0.0048	16	14.14	10	28	21	10	11	14
h173	1	0.0048	16	15.15	10	26	23	11	11	14

Table 1 (*Continued*)

Haplotype	n[a]	F[b]	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
h174	1	0.0048	16	15.16	9	27	22	10	11	13
h175	1	0.0048	17	11.13	10	29	24	11	13	13
h176	1	0.0048	17	12.12	10	25	23	10	11	13
h178	1	0.0048	17	9.16	11	27	23	10	16	12
GD[c]/HD[d]			0.595	0.883[d]	0.522	0.687	0.646	0.583	0.664	0.486

[a] The number of haplotypes.
[b] Frequency of each haplotype in 207 individuals.
[c] Gene diversity.
[d] Haplotype diversity.

Table 2
Y STR haplotypes in Azores Islands population

Haplotype	n[a]	F[b]	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
h1	1	0.0159	12	11.13	11	27	23	11	14	13
h2	2	0.0317	13	13.14	11	27	24	9	11	13
h3	1	0.0159	13	17.17	11	28	24	9	11	13
h4	1	0.0159	14	11.12	10	25	23	11	13	13
h5	1	0.0159	14	11.13	10	26	23	10	13	13
h6	1	0.0159	14	11.13	10	26	24	11	13	12
h7	1	0.0159	14	11.13	10	26	24	11	13	14
h8	1	0.0159	14	11.14	9	25	24	10	13	13
h9	1	0.0159	14	11.14	10	25	24	11	13	13
h10	2	0.0317	14	11.14	10	26	23	10	13	13
h11	1	0.0159	14	11.14	10	26	23	12	13	13
h12	1	0.0159	14	11.14	10	26	24	10	11	13
h13	1	0.0159	14	11.14	10	26	24	10	13	13
h14	1	0.0159	14	11.14	10	26	24	11	13	14
h15	1	0.0159	14	11.14	10	26	24	12	13	13
h16	1	0.0159	14	11.14	10	27	23	10	13	13
h17	1	0.0159	14	11.14	11	27	25	11	13	13
h18	1	0.0159	14	11.14	11	28	24	11	13	13
h19	1	0.0159	14	11.15	10	26	24	10	13	13
h20	1	0.0159	14	11.15	10	26	24	11	13	13
h21	1	0.0159	14	11.15	10	27	23	11	13	13
h22	1	0.0159	14	12.12	10	26	25	10	11	12
h23	1	0.0159	14	12.13	11	27	24	10	14	13
h24	2	0.0317	14	12.14	10	26	24	11	13	13
h25	1	0.0159	14	12.14	10	27	25	11	13	13
h26	1	0.0159	14	12.15	9	25	22	10	11	13
h27	2	0.0317	14	13.14	9	24	22	10	11	13
h28	1	0.0159	14	13.14	9	25	22	10	11	13
h29	1	0.0159	14	13.15	10	27	23	9	11	12
h30	2	0.0317	14	13.16	10	28	23	11	11	12
h31	1	0.0159	14	13.17	11	29	23	10	11	12
h32	1	0.0159	14	14.15	11	27	23	10	11	13
h33	1	0.0159	14	18.18	11	28	25	9	12	14
h34	1	0.0159	15	11.12	10	26	24	11	13	12
h35	1	0.0159	15	11.13	11	27	24	11	13	13
h36	2	0.0317	15	11.14	9	25	24	11	13	13
h37	1	0.0159	15	11.14	10	26	24	10	13	13
h38	4	0.0635	15	11.14	10	26	24	11	13	13
h39	1	0.0159	15	12.16	11	27	25	10	11	13
h40	2	0.0317	15	12.19	10	26	22	10	11	13
h41	2	0.0317	15	13.14	9	25	22	10	11	13

Table 2 (*Continued*)

Haplotype	n[a]	F[b]	DYS19	DYS385	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
h42	1	0.0159	15	13.16	10	27	24	10	11	12
h43	1	0.0159	15	14.14	9	26	22	10	13	13
h44	3	0.0476	15	14.14	11	27	23	10	11	12
h45	1	0.0159	15	14.15	10	27	24	10	12	15
h46	1	0.0159	15	14.15	10	27	25	9	11	13
h47	1	0.0159	15	14.18	9	25	24	10	13	12
h48	1	0.0159	15	9.14	10	26	24	11	13	13
h49	1	0.0159	16	11.14	11	25	24	11	13	13
h50	1	0.0159	17	16.16	11	28	21	11	12	13
GD[c]/HD[d]			0.572	0.862[d]	0.587	0.720	0.664	0.608	0.560	0.404

[a] The number of haplotypes.
[b] Frequency of each haplotype in 63 individuals.
[c] Gene diversity.
[d] Haplotype diversity.

being unique. The most common haplotype (DYS19:14, DYS385:11/14, DYS389I:10, DYS389II:26, DYS390:24, DYS391:11, DYS392:13, DYS393:13) had a frequency of 3.4% (h54). The frequency of each haplotype is shown in Table 1. The haplotype diversity for all nine Y-specific STR loci was calculated to be 0.993.

In Azores Islands a total of 50 different haplotypes were observed (Table 2), 40 of them being unique. The most common haplotype (DYS19:15, DYS385:11/14, DYS389-I:10, DYS389II:26, DYS390:24, DYS391:11, DYS392:13, DYS393:13) had a frequency of 6.4% (h38). The frequency of each haplotype is shown in Table 2. The haplotype diversity for all nine Y-specific STR loci was calculated to be 0.976.

AMOVA analysis, in all the analyzed systems except DYS385, using the two Portuguese populations studied and other European and African populations [10–14] shows that Central Portugal and Azores Islands populations are distant from African populations ($P < 0.05$). Another significant difference was founded between Central/Southern Italy and Central Portugal ($P = 0.000 \pm 0.000$) and Azores Islands ($P = 0.004 \pm 0.010$) populations.

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