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FOR THE RECORD

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Assessment of HV1 and HV2 mtDNA Variation for Forensic Purposes in an Uruguayan Population Sample

POPULATION: 120 unrelated Caucasians living in Uruguay (Southern America)

KEYWORDS: forensic science, DNA typing, population genetics, mtDNA, Uruguay, Hv1, Hv2

In order to assess the utility of mtDNA typing for forensic purposes in Uruguay, a population sample of 120 maternally unrelated individuals were amplified and directly sequenced for the HV1 and for the HV2 segments in the control region of the human mtDNA, following previous international recommendations (1).

Blood and saliva samples were taken from 120 unrelated individuals and deposited into FTA cards (FTA card, Whatman WB 12-0205). The DNA was extracted in a 1.2 mm punch according to the manufacturer recommendations. DNA amplification and sequencing was performed according to previously published paper (2). All statistical analysis were calculated using the Arlequin Software ver. 2.000 (3).

Results are shown in Table 1. In this population sample, 107 different mtDNA sequences were observed out of 120 individuals. Of these, 97 sequences were observed once (80.8%), 8 sequences were observed twice (13.3%), one sequence was observed three times (2.5%), and another four times (3.3%). The variation of mtDNA HV1 and HV2 regions was confined to 146 positions, of which 99 was observed in the HV1 and 47 in the HV2. Sequence variation consisted of 120 transitions, 21 transversions, and 11 indels. The genetic diversity value for combined HV1 and HV2 regions was 0.998 (±0.002) in this Uruguayan population sample, and the nucleotide diversity was 0.068 (±0.035).

The complete dataset is available to any interested researcher at: http://www.lorgen.com/forense

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TABLE 1—*Mitochondrial DNA variation in a randomly selected Uruguayan population sample of 120 individuals living in various parts of Uruguay.*

ID	HAPLO-GROUPO	16039	16042	16048	16051	16059	16069	16084	16086	16092	16093	16111	16114	16124	16126	16129	16136	16140	16142	16145	16147	16148	16153	16155	16158	16163	16167	16168	16172	16173	16178	16179	16182	16183	16184	16185	16186	16187	16188	16189	16190	16192	16193	16193.1	16193.2	16209	16213	16217	16219	16220	16223	16224
CRA		G	G	G	A	A	C	G	T	T	T	C	C	T	T	G	T	T	C	G	C	C	G	A	A	A	C	C	T	C	T	C	A	A	C	C	C	C	C	T	C	C	C			T	G	T	A	A	C	T
54	A											T																																							T	
103	A											T																																							T	
66	B																																C						C				C				C			
5	B									C																						C						C								C			
39	B					C																								C		C						C								C			
52	B	A								C																						C						C								C			
58	B																																C	C				C								C			
94	B																																	C				C								C			
61	B																																	C				C								C			
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74	B																																	C				C								C			
75	B																														C			C				C								C			
93	B																																	C				C								C			
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102	B																																C	C				C											
72	B																																					C								C			
113	B																																	C				C								C			
114	B																													T			C	C				C								C			
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42	C																																																	T	
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92	C										C																																							T	
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107	C																																																	T	
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96	D																																									T								T	
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97	D																												C					C							C			C				G			
109	D																																																	T	
110	D										G																																							T	
115	D				G																																												T	
118	D																																																	T	
22	M																																																	T	
112	M										C																																							T	
55	L1a										C											T							C													T	G	C						T	
120	L3b								C							A																										T		C						T	
101	U6																									G			C																		G			
26	U6																									G			C																		G			
53	U6									C																G			C																		G			
91	U6			A																						G			C																		G			
117	U6																												C																		G			
93	H																					T																												
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81	U5				G																								C																		G			
11	K										C																																							C
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63	K																																																	C
73	K																																		T															C
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3	J						T				C				C																																		
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84	J						T								C																																		
100	J						T								C																																		
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17	T														C																																	G		
40	T														C																																	G		
47	T														C																																			
76	T														C																																			
95	T1														C											G													T			C							
16	T1														C											G													T			C			d				
18	T2														C																																			
105	T2		T												C								A																		C								
45	T2a														C																																			
71	T2a														C																																			
12	T2a														C					A																														
67	I																				G								C																			T	
108	I															A																																	T	
41	X												A			A																														A			T	
14	W																																																	T	
116	W																																																	T	
CRA		G	G	G	A	A	C	G	T	T	T	C	C	T	T	G	T	T	C	G	C	C	G	A	A	A	C	C	T	C	T	C	A	A	C	C	C	C	C	T	C	C	C			T	G	T	A	A	C	T
ID	HAPLO-GROUPO	16039	16042	16048	16051	16059	16069	16084	16086	16092	16093	16111	16114	16124	16126	16129	16136	16140	16142	16145	16147	16148	16153	16155	16158	16163	16167	16168	16172	16173	16178	16179	16182	16183	16184	16185	16186	16187	16188	16189	16190	16192	16193	16193.1	16193.2	16209	16213	16217	16219	16220	16223	16224

TABLE 1—*Continued*.

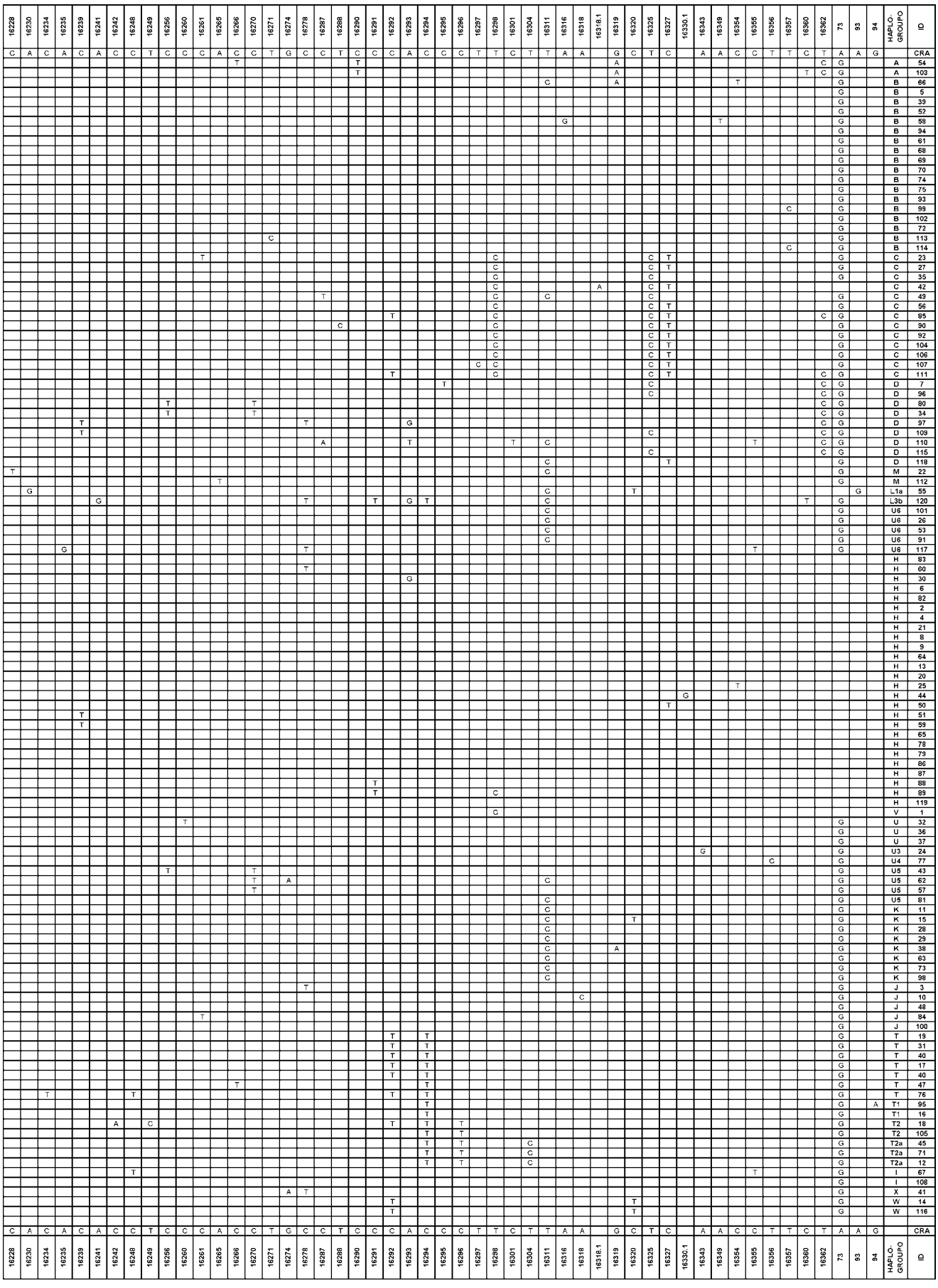

TABLE 1—*Continued*.

ID	HAPLO-GROUPO	146	150	151	152	153	182	183	185	186	188	189	194	195	198	199	200	204	207	210	215	228	235	236	243	244	247	249	250	253	263	264	279	282	290	291	295	309.1	309.2	309.3	315.1	316	320	327
CRA		T	C	C	T	A	C	A	G	T	A	A	C	T	C	T	A	T	G	A	A	G	A	T	A	A	G	A	T	C	A	C	T	T	A	A	C					G	C	C
54	A	C																					C								G							C			C			
103	A	C				G																	G								G							C			C			
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55	L1a				C							G												C			A			G										C			
120	L3b			T	C		T			A		C		C	T												A			G										C	A		
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77	U4													C							G								T		G								C		C			
43	U5																														G								C			C			
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100	J								A													A									G										C			
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31	T	C			C									C																	G		C								C			
40	T	C			C									C																	G		C								C			
17	T	C			C									C																	G		C								C			
40	T	C			C									C																	G		C								C			
47	T														T																G								C			C			
76	T																														G								C			C			
95	T1				C																										G								C			C			
16	T1																														G								C			C			
18	T2	C																													G										C			
105	T2		T																												G								C			
45	T2a																														G										C			
71	T2a																														G										C			
12	T2a													C				C													G								C			C			
67	I															C		C	A												G								C			C			
108	I				A											C		C	A										C		G										C			
41	X	C	T		C		T	G						C	T			C													G								C			C			
14	W											G		C				C	A												G								C						
116	W											G		C				C	A												G										C			
CRA		T	C	C	T	A	C	A	G	T	A	A	C	T	C	T	A	T	G	A	A	G	A	T	A	A	G	A	T	C	A	C	T	T	A	A	C					G	C	C
ID	HAPLO-GROUPO	146	150	151	152	153	182	183	185	186	188	189	194	195	198	199	200	204	207	210	215	228	235	236	243	244	247	249	250	253	263	264	279	282	290	291	295	309.1	309.2	309.3	315.1	316	320	327