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FOR THE RECORD

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Sequence Polymorphisms of the Mitochondrial DNA Control Region in 105 Chinese Han Population*

POPULATION: 105 unrelated persons, Southwest China

KEYWORDS: forensic science, DNA typing, mitchondrial DNA, hypervariable region I, hypervariable region II, population genetics, China

A total of 105 EDTA blood samples were collected from unrelated blood donors living in Chengdu, Southwest China. DNA was extracted according to the method of Miller et al. (1). The quantity of DNA was estimated by fluorometry.

The two hypervariable regions of the Mitchondrial DNA typing was carried out with 20 ng of genomic DNA in a total volume of 50 μL reaction system, containing 0.2 μM primer (2,3), 200 μm each dNTP, 1 U Taq polymerase (Eurobio, Raunheim, Germany), 1X PCR buffer, 0.5 mM $MgCl_2$. The PCR was carried out in a thermal cycler (GeneAmp PCR System 2400; Perkin Elmer Corporation, Morwalk) for 30 cycles in such conditions as denaturation at 94°C for 45 s, annealing at 55°C for 30 s, and extension at 72°C for 3 min, followed by a final extension step at 72°C for 7 min.

PCR products were purified by using the QIAquick PCR purification kit (QIAGENE, Hilden, Germany) and then sequenced by cycle sequencing using fluorescent dideoxynucleotides (ABI PRISM™ Dye Terminator Cycle Sequencing Ready Reaction Kit) followed by capillary electrophoresis in an ABI Prism 310 Genetic Analyzer (Applied Biosystems, Foster City, CA).

The genetic diversity ($h = n(1 - \Sigma x^2)/(n - 1)$ is estimated to be 0.999, and the probability of two random individuals from the studied population with the identical mtDNA haplotype ($P = \Sigma x^2$) is 1.07%

Results obtained in this study are shown in Table 1 and 2 after aligned with a reference sequence (4).

L15997	5′—CAC CAT TAG CAC CCA AAG CT—3′
And H16401	5′—TGA TTT CAC GGA GGA TGG TG—3′ for HVR-I

The HVR-I and HVR-II are amplified by two different sets of primers respectively which are shown as follows:

L00029	5′—GGT CTA TCA CCC TAT TAA CCA C—3′
And H00408	5′—CTG TTA AAA GTG CAT ACC GCC A—3′ for HVR-II

The complete dataset is available to any interested parties and researchers upon request.

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TABLE 1—*List of variable sites observed in the hypervariable region I(HVR-I) of mtDNA control region obtained from chinese donors 1~105. All nucleotides at these positions are shown together with the reference sequence.*

Haplotype	16037	16051	16054	16066	16075	16086	16092	16093	16104	16108	16126	16129	16134	16136	16140	16145	16147	16148	16157	16158	16162	16164	16172
Anderson	A	A	A	A	T	T	T	T	C	C	T	G	C	T	T	G	C	C	T	A	A	A	T
1								C															
2							C															G	C
3																							
4																							
5															C								
6														C									
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12																			C				
13								C				A											
14												A											
15																							
16																							
17																							
18																			C				
19												A											C
20												A											
21																	T						
22																							
23								C				A									G		C
24												A									G		C
25								C															
26																			C				
27																							
28																							C
29															C								
30		G																					
31				G																			
32																							
33																							
34																							
35												A											
36																							
37												A									G		C
38								C															
39							C															G	
40																							
41																							
42																							
43								C															
44							C																
45																		T					
46												A											
47																							
48														C									
49																							
50	G											A											C
51																							
52d*								C								A							
53																							
54																							

Haplotype	16175	16176	16178	16179	16181	16182	16183	16184	16185	16189	16192	16193	16194	16203	16209	16213	16217	16218	16220	16221	16223	16227	16231	16232
Anderson	C	C	T	C	A	A	A	C	C	T	C	C	A	A	T	G	T	C	A	C	C	A	T	C
1												T									T			
2						C	C			C														
3							C			C														
4														G										
5						C	C			C														
6						C	C			C														
7																					T			
8						C	C			C														
9							C			C														
10							C			C										T				
11																					T			
12																								
13																					T			
14										C											T			
15																					T			
16																					T			
17																			C			G		
18																								
19																								
20																					T			
21							C	A		C							C							
22	T										T										T			
23																								
24																								
25																					T			
26							C			C											T			
27																					T			
28																								
29							C			C														
30										C											T			
31																					T			
32																					T			
33						C	C			C							C							
34																					T			
35											T										T			
36																							C	
37																								
38				T		C	C			C														
39						C	C			C											T			
40			C				C			C			C											
41																					T			
42										C											T			
43										C											T			
44																					T			
45				T			C			C											T			
46						C	C			C							C							
47																			C			G		
48							C			C							C							
49								T													T			
50																								
51						C	C			C														
52d*						C	C			C							C							
53								T								A					T			
54						C	C			C							C							

Haplotype	16234	16235	16240	16243	16245	16249	16256	16257	16260	16261	16264	16266	16269	16270	16274	16278	16286	16287	16290	16291	16292	16294	16295	16296
Anderson	C	A	A	T	C	T	C	C	C	C	C	C	A	C	G	C	C	C	C	C	C	C	C	C
1																								
2												T												
3																								
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7																			T					
8										T														
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18																								
19																								
20																			T					
21		G																						
22																								
23																								
24																								
25																		T	T					
26																			T					
27																								
28															A									
29												A								T				
30									T						A					T				
31																								
32																								
33										T														
34																							T	
35																								
36																								
37																								
38																								
39												T												
40																								
41																			T				T	
42																								
43																								
44																								
45				C																				
46										T														
47																								
48																								
49																			T					
50																								
51																								
52d*										T														d
53																								
54										T														

Haplotype	16297	16298	16299	16301	16302	16304	16309	16311	16318	16319	16325	16327	16335	16354	16355	16356	16357	16360	16362	16365	16381	16390	16399	16400
Anderson	T	T	A	C	A	T	A	T	G	G	T	C	A	C	C	T	T	C	T	C	T	G	A	C
1								C									C							
2																			C					
3						C																		
4						C																		
5																								
6							G							T										
7								C	A															
8			G												T									
9								C											C					
10																								
11	C																							
12						C		C					G											
13												T												
14	C																							
15																								
16								C											C					
17		C																	C					
18						C																		
19						C																		
20										A														
21																								
22																								
23						C																		
24						C																		
25										A									C					
26																			C					
27	C			T																				
28																			C					
29																								
30																			C					
31											C								C					
32												T												
33						C												T						
34																								
35	C																							
36																								
37						C																		
38																								
39																			C					
40						C																		
41										A									C					
42																			C					
43											C					C			C					
44																			C					
45																			C					
46																								
47		C																	C					
48							G							T										
49																			C					
50								C																
51								C														A		
52d*																								
53								C											C					
54																								

*d: deletion.

(continues)

TABLE 1—(*Continued*)

Haplotype	16037	16051	16054	16066	16075	16086	16092	16093	16104	16108	16126	16129	16134	16136	16140	16145	16147	16148	16157	16158	16162	16164	16172	16175	16176	16178	16179	16181	16182	16183	16184	16185	16189	16192	16193	16194	16203	16209	16213	16217	16218	16220	16221	16223
Anderson	A	A	A	A	T	T	T	T	C	C	T	G	C	T	T	G	C	C	T	A	A	A	T	C	C	T	C	A	A	A	C	C	T	C	C	A	A	T	G	T	C	A	C	C
55														C															C	C			C							C	T			
56			G																																									
57							C																																					T
58																																												
59								C																																				T
60										T											G		C																					
61																																T												T
62											C																																	
63											C																																	
64					C										C															C			C											
65							C															G	C						C	C			C											T
66																																												T
67																												C	C	C			C						A	C				
68							A																										C					C						
69																														C			C											T
70																																										C		
71																												C	C	C			C						A	C				
72																														C			C							C				
73																									T																			T
74													T																		T													T
75															C															C			C											
76																																												T
77																																												
78d												A																				T	C		d									T
79																																												T
80												A																					C											T
81							C									A							C																					T
82												A			C															C			C											
83												A										G							C	C			C											T
84																																												
85								C																						C			C											
86								C																																				T
87												A									G		C																					
88																																												T
89								C																																				T
90																																												T
91						C						A																						T										T
92																																												
93																														C			C											
94								C	T																							T												T
95																																												T
96																							C										C											T
97																													C	C			C							C				
98													T																		T													T
99																																												
100																				G																								T
101																														C			C							C				
102																														C			C											
103						C																																						
104																															T													T
105						C						A																																

Haplotype	16227	16231	16232	16234	16235	16240	16243	16245	16249	16256	16257	16260	16261	16264	16266	16269	16270	16274	16278	16286	16287	16290	16291	16292	16294	16295	16296	16297	16298	16299	16301	16302	16304	16309	16311	16318	16319	16325	16327	16335	16354	16355	16356	16357	16360	16362	16365	16381	16390	16399	16400
Anderson	A	T	C	C	A	A	T	C	T	C	C	C	C	C	C	A	C	G	C	C	C	C	C	C	C	C	C	T	T	A	C	A	T	A	T	G	G	T	C	A	C	C	T	T	C	T	C	T	G	A	C
55																																														C					
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57																						T															A									C					
58																																	C																		
59	G																		T																											C					
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61												T																	C			G																		G	
62		C													T																															C					
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64							C																																			T									
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66																													C										T												
67													T											T																											
68																							T										C																		
69											A		T																						C																
70	G																												C																	C					
71													T										T								T																				
72																																			C																
73																						T															A									C					
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75							C															T																													
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80																												C																							
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85																														G			C																		
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93																																	C																		
94														T															C																						
95																																														C					
96						C																													C											C					
97													T																																						
98																													C							A															
99																							T										C																	G	
100				T												G																														C					T
101				T											T																																				
102			A						C																								C		C																
103																																	C																		
104																													C								A														
105																												C																			T				

TABLE 2—*List of variable sites observed in the hypervariable region II (HVR-II) of mtDNA control region obtained from Chinese donors 1~105.*

Haplotpye	55.1	57	59	73	94	95	103	143	146	150	151	152	153	185	189	194	195	199	200	201	202	204	205	206	207	210	214	215	217	228	235	236	237	249	257	263	272	279	281	309	309.1	309.2	310	310.1	315.1	315.2	311	317	318	320	326	338	396
Anderson	.	T	T	A	G	G	G	G	T	C	C	T	A	G	A	C	T	T	A	A	A	T	T	G	G	A	A	A	T	G	A	T	A	A	A	A	A	T	A	C	.	.	T	.	.	.	C	C	T	C	A	C	A
1				G					C																												G					C				C						
2				G						T									G																	G					C				C						
3d				G																													d		G					C				C						
4				G																															G					C				C						
5				G																						G									G					C				C						
6				G																															G					C				C						
7				G								C																			G				G					C				C				C		
8				G																					A										G					C	C			C						
9				G						T													C													G									C						G
10				G						T		C																							G					C				C						
11				G						T								C				C													G					C				C						
12d				G																											C		d		G									C						
13				G					C																								d		G	G				C				C						
14				G						T								C				C													G					C				C						
15				G				A																											G					C				C						
16	T	C	C																																																	
17d				G								C																						d		G					C	C			C						
18				G																															G									C	C						
19d				G																													d		G									C						
20				G								C							G												G				G					C				C						
21				G										A																					G					C				C						
22				G													C																		G								T				G		T			
23d				G													C																d		G					C				C						
24d				G																													d		G									C						
25				G							T	C																		G					G					C				C						
26				G			A					C							G											G					G					C				C						
27				G						T								C				C													G					C				C						
28				G												T																			G					C				C						
29				G																					G										G					C	C			C						
30				G								C																							G					C				C						
31				G						T																								G	G				C				C						
32				G								?																					d		G									C						
33				G																															G					C										
34				G															G									G							G					C				C				C		G		
35				G						T								C							A										G					C										
36				G					C																										G					C	C			C						
37d				G	A																												d		G					C				C						
38				G						T																				A				G					C	C			C						
39				G						T																									G					C	C			C						
40d				G					C																								d		G		C			C	C			C						
41				G							T	C							G												G				G					C				C						
42				G						T																								G						C	C			C						
43				G												T																			G					C	C			C						
44				G		A																													G					C				C						
45				G						T		C		A			C																			G									C						
46				G																															G	G									C						
47d				G								C																					d		G					C	C			C						
48?				G																					A										G					C				C		T	G					
49				G						T																									G					C				C						
50d				G													C																d		G					C	C			C						
51				G										A	G																				G					C				C						
52				G					C																										G					C				C						
53				G																															G					C				C					T	
54				G																															G					C				C						
55				G																															G					C	C			C						

(continues)

TABLE 2—(*Continued*).

Haplotype	55.1	57	59	73	94	95	103	143	146	150	151	152	153	185	189	194	195	199	200	201	202	204	205	206	207	210	214	215	217	228	235	236	237	249	257	263	272	279	281	309	309.1	309.2	310	310.1	315.1	315.2	311	317	318	320	326	338	396
Anderson	.	T	T	A	G	G	G	G	T	C	C	T	A	G	A	C	T	T	A	A	A	T	T	G	G	A	A	A	T	G	A	T	A	A	A	A	A	T	A	C	.	.	T	.	.	.	C	C	T	C	A	C	A
56d				G					C																									d		G					C				C									
57				G							T	C							G												G				G					C				C									
58d				G					C																									d		G			G		C	C		C									
59d				G																														d		G								C									
60				G																									G						G					C												
61d				G						T																							d		G					C												
62d				G							T	C																					d		G					C				C									
63				G					C			C													A									G					C				C									
64				G			A			T																								G					C				C									
65				G						T																								G			C		C				C									
66d				G					C																								d		G					C				C									
67				G																															G					C	C		C		T								
68d				G					C																								d		G					C	C		C									
69				G				A		T																									G					C				C									
70d				G								C																							G					C				C									
71d				G																															G				d				C									
72				G						T							C										G								G		C			C				C		T							
73				G							T	C							G												G				G		C			C				C									
74				G													C																			G					C				C									
75				G																		C													G					C	C		C									
76				G																															G					C				C									
77				G					C									C																		G					C				C									
78d				G							T	C																								G					C				C									
79				G																															G					C				C									
80				G						T								C				C		A											G					C				C									
81				G						T																									G					C	C		C									
82				G								C										C													G							C									
83				G						T								C																	G					C				C									
84d				G																													d		G					C				C									
85d				G																													d		G					C				C									
86				G													C											G							G					C				C									
87d				G																													d		G					C				C									
88				G					C			C						C																	G					C				C									
89				G				A				C																							G					C				C									
90				G				A																											G					C				C									
91				G						T								C																	G					C				C									
92d				G													C																d		G					C				C									
93d				G																													d		G					C				C									
94d				G								C					C																d		G					C				C									
95				G												T																			G					C				C									
96				G								C							G																	G					C				C									
97				G					C																										G					C	C		C									
98				G													C																		G					C				C									
99d				G																													d		G					C				C									
100				G						T		C	G																						G					C				C									
101				G																															G					C				C									
102d				G										A						G					A									d		G					C		A									
103				G																														d		G					C				C									
104				G																																G					C				C									
105				G														C			G															G					C				C									