

For the Record

Sequence Polymorphisms of the Mitochondrial DNA Control Region in 100 German Caucasians

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Population: 100 unrelated persons, western Germany

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DNA from 100 unrelated individuals was prepared as described previously (1). The amount of DNA was quantitated photometrically.

A total of 10 ng of genomic DNA was used as template for the PCR amplification reactions. The two hypervariable segments (HV I and HV II) within the control region were amplified as listed in Table 1. PCR was performed in a total volume of 25 μ L consisting of 0.5 μ M primer, 200 μ M each dNTP, 2.5 μ L 10 \times PCR buffer, 0.5 mM MgCl₂, 1 U Taq polymerase (ganRES[®]PLUS, Serac, Germany). Amplification was carried out on a Biometra, Triothermoblock at 94°C for 1 min, 50°C for 1 min and 72°C for 3 min for 25 cycles.

The solid phase sequencing procedure on an A.L.F. express (Amersham Pharmacia Biotech) and sequence analysis was performed as described previously (1). Genetic diversity was calculated according to Tajima (2).

Results obtained from the 100 unrelated individuals (Rhine area) are shown in Table 2 after alignment with a reference sequence (3). From this population data, we estimate the genetic diversity ($h = n(1 - \sum \chi^2)/n - 1$) to be 0.99. The probability of two randomly selected individuals from a population having identical mtDNA types ($P = \sum \chi^2$) is 0.6%.

The complete data set is available to any interested researcher upon request.

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TABLE 1—Oligonucleotides used for PCR amplification and sequencing of mitochondrial DNA. 'L' and 'H' stand for the light and heavy strand of the mtDNA, respectively. For L-strand synthesis of HV I biotinylated L15997 with M13-H16401, for H-strand synthesis M13-L15997 with biotinylated H16401 was used. For L-strand synthesis of HV II biotinylated L00029 with M13-H00408, for H-strand synthesis M13-L00029 with biotinylated H00408 was used.

Amplification Primer	Nucleotide Sequence
Universal M13	5'-CGACGTTGTAAAACGACGGCCAGT-3'
L15997	5'-M13/Bio-CACCATTAGCACCCAAAGCT-3'
H16401	5'-M13/Bio-TGATTTACGGAGGATGGTG-3'
L00029	5'-M13/Bio-GGTCTATCACCTATTAACCAC-3'
H00408	5'-M13/Bio-CTGTAAAAGTGCATACCGCCA-3'
Sequencing Primer	Nucleotide Sequence
Universal M13	5'-CY5-CGACGTTGTAAAACGACGGCCAGT-3'

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References

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TABLE 2—Continued.

Haplotype	72	73	93	119	146	150	152	153	161	*174.1	185	188	189	195	199	200	204	207	217	225	227	228	239	242	250	263	282	293	295	*309.1	*309.2	310	315	*315.1	*315.2	316	317	325	340					
Reference	T	A	A	T	T	C	T	A	T	*	G	A	A	T	T	A	T	G	T	G	A	G	T	C	T	A	T	T	C	*	*	T	C	*	*	C	C	C	C					
mtDNA Nr.																																												
51	G	G	.	.	.	C	C	.	.	C			
52	.	.	.	C	G	C		
53	.	G	.	.	T	G	.	.	.	C	C		
54	.	.	G	C	C	.	G	C		
55	.	G	.	.	T	G	C		
56	G	C		
57	.	G	C	G	C	.	.	C		
58	.	G	C	G	C	.	.	C		
59	.	G	G	C	.	.	C	
60	.	G	.	.	T	C	.	.	C	G	C	.	.	C	
61	.	G	G	C	.	.	C	
62	C	G	C	.	.	C		
63	C	G	C	.	.	C		
64	C	G	C	.	.	C	
65	.	G	A	A	.	G	.	.	T	C	.	.	C	
66	C	C	G	C	C	.	.	C	.	.	.	
67	C	G	C	.	.	C	
68	G	C	.	.	C	
69	G	C	.	.	C	
70	G	C	C	C	.	.	.	C	.	.	C		
71	.	G	G	C	.	.	C	
72	.	G	G	C	.	.	C	
73	.	G	G	C	.	.	C	
74	G	C	.	.	C	
75	.	G	C	G	C	.	.	C	
76	.	G	.	.	T	G	C	C	.	.	C	
77	.	G	G	C	.	.	C	
78	.	.	G	G	C	.	.	C	
79	G	C	.	.	C	
80	A	G	C	C	.	.	C	
81	.	G	.	.	.	C	C	G	C	.	.	C	
82	.	G	A	A	.	G	.	.	T	C	.	.	C	
83	.	G	.	.	.	C	C	.	.	.	G	C	.	.	C	.	.	.	T	
84	G	C	.	.	C	
85	G	C	.	.	C	
86	G	C	.	.	C	
87	.	G	G	C	.	.	C	
88	G	C	C	C	.	.	C	.	.	.	
89	.	G	.	.	T	G	C	C	.	.	C	.	.	.	
90	.	G	.	.	.	C	C	G	C	.	.	C	
91	G	C	.	.	C
92	.	G	.	C	G	C	.	.	C	A	G	C	.	.	C	
93	.	G	G	C	.	.	C
94	.	G	.	.	C	G	C	.	.	C
95	G	C	.	.	C
96	G	C	C	.	.	C	.	.	.
97	C	G	C	.	.	C
98	G	C	.	.	C
99	.	G	G	G	C	.	.	C
100	.	.	.	C	C	G	C	.	.	C