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Mitochondrial DNA control region polymorphism in Han, Yao, Li, Uygure and Tibetan groups in China

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Abstract. In order to investigate the polymorphisms of mtDNA in different groups in China, we have sequenced mtDNA control region from 446 unrelated person. The polymorphic sites in Han, Li, Uygure, Tibetan and Yao groups were 269, 316, 159, 117, 141, respectively, and the pairwise difference, hyplotypes, etc. were calculated. The result showed that there were other hypervariable regions besides HVI and HVII. However, the HV III were located at different region in each group.

Keywords: mtDNA; Control region; Han group; Yao group; Li group; Uygure group; Tibetan group; China

1. Objective

The sequencing of mtDNA hypervariable region I and II has become a powerful tool to investigate biological evidence that cannot be analyzed by the use of nuclear DNA. However, the power of discrimination of mtDNA testing is limited. Studying additional diversity outside of mtDNA hypervariable regions will enhance the power and efficiency of mtDNA profiling. China is a multinational country, the living region and marriage habit of every ethnic group may influence the genetics diversity of mitochondrial DNA. In the context, the whole control region of mtDNA were studied in Han, Yao, Li, Uygure and Tibetan groups in China, in order to research the polymorphisms in different groups.

2. Materials and methods

2.1. Sample collection

100 unrelated Han samples were collected from Beijing. 105 unrelated Yao samples were collected from Du'an, Guangxi province. 100 unrelated Li samples were collected

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from Hainan province. 99 unrelated Uygure samples were collected from Kashi Xinjiang province. 42 unrelated Tibetan samples were collected from Nagqu Tibet province. The samples were collected under the rule of.

DNA was extracted by the Chelex and phenol–chloroform method. The following primers were used for amplification and sequencing: mt-1(15756-16087) H 5′-gaa tcg gag gac aac cag taa g-3′, L 5′-tag cgg ttg ttg tag ggt-3′; mt-0(16030-16481) H 5′-cat ggg gaa gca gat ttg-3′; L 5′-tta gct acc ccc aag tgt-3′; mt-2(16419-00270) H 5′-caa tat ccc gca caa gag tg-3′, L 5′-tgg aaa gtg gct gtg cag aca t-3′; mt-3(00132-00444) H 5′-ctt tga ttc ctg cct cat cc-3′, L 5′-tta gtt ggg ggg tga ctg tt-3′; mt-4(00374-00672) H 5′-cac cag cct aac cag att tc-3′, L 5′-tag aaa ggc tag gac caa acc t-3′. PCR was carried out under the following thermal cycle conditions: denature at 95 °C for 200 s; 94 °C for 30 s, 55 °C for 30 s, 72 °C for 90 s, for total 35 cycles; following 72 °C for 7 min.

PCR products were analyzed using BigDyeTerminator Cycle Sequencing Ready Reaction Kit and ABI 377.

3. Statistic analysis

The genetic diversity (GD) was calculated using the formula $GD = n(1 - \Sigma x^2)/(n - 1)$, discrimination power (Dp) was calculated according to the equation $Dp = 1 - \Sigma x^2$, where n was the number of sample, x was the haplotype frequency.

4. Result

We determined the nucleotide sequence of a 1400-bp fragment of the mt-DNA (positions 15786-16569, followed by 1–660 bp). The polymorphic sites in Han, Li, Uygure, Tibetan and Yao groups were 269, 316, 159, 117, 141, respectively. In each group, sequence

Table 1
The polymorphisms of mtDNA control region in Han, Tibetan, Li, Uygure and Yao groups in China

Region	Han ($n=100$)		Li ($n=100$)		Uygure ($n=99$)		Tibetan ($n=42$)		Yao ($n=105$)	
	N	Hyplo types	*N*	Hyplo types	*N*	Hyplo types	*N*	Hyplo types	*N*	Hyplo types
15786-16023	23	21	20	19	12	14	3	4	9	7
16024-16365	109	96	93	77	65	64	47	42	68	65
16366-00072	14	18	67	50	17	18	10	8	10	14
00073-00340	68	86	58	70	34	52	29	26	35	56
00341-00437	21	18	32	29	4	5	8	8	4	5
00438-00574	25	33	38	35	25	21	16	15	13	13
00575-00650	9	14	8	4	2	3	4	4	2	3

Region	Han ($n=100$)		Li ($n=100$)		Uygure ($n=99$)		Tibetan ($n=42$)		Yao ($n=105$)	
	Dp	GD	Dp	GD	Dp	GD	Dp	GD	Dp	GD
15786-16023	0.4628	0.4675	0.4332	0.4376	0.4122	0.4164	0.1360	0.1393	0.2901	0.2927
16024-16365	0.9892	0.9991	0.9838	0.9937	0.9723	0.9823	0.9761	0.9999	0.9738	0.9832
16366-00072	0.6794	0.6863	0.9108	0.9200	0.6472	0.6538	0.5872	0.6015	0.6362	0.6423
00073-00340	0.9850	0.9949	0.9792	0.9891	0.9497	0.9594	0.9312	0.9539	0.9669	0.9762
00341-00437	0.4726	0.4774	0.6200	0.6263	0.1345	0.1358	0.3740	0.3831	0.0923	0.0932
00438-00574	0.8838	0.8923	0.8832	0.8921	0.8218	0.8302	0.7481	0.7663	0.7510	0.7582
00575-00650	0.5174	0.5226	0.0778	0.0786	0.0398	0.0402	0.1360	0.1393	0.1092	0.1103

N: the number of nucleotide substitution.

Table 2
The variation rate (%) of mtDNA control region in Han, Tibetan, Li, Uygur and Yao groups

Position variation		Uygur ($n=99$)	Yao ($n=105$)	Han (Beijing) ($n=100$)	Li ($n=100$)	Tibetan ($n=42$)
16086	T→C	/	1.90	/	/	4.76
16093	T→C	1.01	12.38	7	2	16.67
16111	C→T	/	0.95	3	/	7.14
16129	G→A	4.04	26.67	16	14	9.52
16183	A→C	23.23	26.67	/	29	2.38
16184	C→T	1.01	5.71	5	2	2.38
16189	T→C	29.29	37.14	26	35	7.14
16223	C→T	41.41	45.71	25	43	80.95
16234	T→C	2.02	0.95	6	3	30.95
16263	C→A	1.01	2.86	/	/	/
16290	C→T	2.02	1.90	7	1	9.52
16293	A→C	/	/	2	/	7.14
16298	T→C	14.14	12.38	11	4	4.76
16304	T→C	1.05	18.09	11	28	7.14
16311	T→C	6.06	9.52	17	20	11.90
16316	A→G	1.01	/	3	/	11.90
16319	G→A	2.02	1.90	14	4	9.52
16327	C→T	16.16	6.67	5	/	4.76
16362	T→C	19.19	24.76	47	17	21.43
16390	G→A	8.08	2.86	3	8	/
16519	T→C	67.67	53.33	57	50	45.24
00073	A→G	83.83	100	100	100	97.62
00152	T→C	17.17	10.48	32	14	35.71
00190	C→T	/	/	53	/	/
00235	A→G	2.02	5.71	6	2	14.29
00249	A→d	18.18	23.81	/	12	14.29
00263	A→G	93.93	97.14	97	98	100
00309.1	C	63.63	60.95	47	54	47.62
00315.1	C	94.94	97.14	90	93	100
00489	T→C	39.39	46.66	58	43	73.81

diversity was caused mainly by nucleotide substitution. The pairwise difference, hyplotypes and the GD, Dp value for every part of control region were presented in Table 1. Table 2 shows the variation rate of some hypervariation sites in different nationals.

5. Conclusion

The result showed that there were other hypervariable regions besides HVI and HVII. However, the HV III were located at different region in each group. In Han, Uygure, Tibetan groups, the HV III were located at 00483-00574. However, in Li and Yao groups, the HV III were between HV I and HV II. Also, the variation and power of discrimination of mtDNA were enhanced by detection of the whole region than HVI or HVII. We suggested that there should be more databases established which consisted of more samples and regions of mtDNA. When mtDNA sequencing was used in forensic field, we should analyze more regions besides HV I and HV II in order to improve the discrimination power. According to the nationality of the sample, different regions should be detected.