

## Polymorphic distribution of Y-chromosome haplotype and mitochondrial DNA in the Bouyei people in China

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In the evolution of humans, many kinds of mutations in the human genome have been accumulated, providing credible genetic evidence for the study of human origins and migrations. The "out-of-Africa" hypothesis of modern human evolution and the genetic origin of the Japanese has come about by studying mitochondrial DNA.<sup>1,2</sup> Recently, researchers have recognized the power of Y-chromosome markers in resolving migratory patterns of modern humans as more and more Y-chromosome single nucleotide polymorphism markers have been found. The markers on the nonrecombinant part of the Y-chromosome allows for the reconstruction of intact haplotypes which are probably the best genetic tools to study human migrations. We can analyze the paternal history of some people in different areas by Y-chromosome haplotypes.<sup>3,4</sup>

The Bouyei people is a minority of southern China. Most of them live in Guizhou Province. Their language is similar to the one spoken by the Zhuang, Li, Dong and Yao peoples. In studying the origin and migration of the Bouyei people, the Y-chromosome haplotypes and polymorphism of mitochondrial DNA were tested.

### METHODS

#### DNA samples

The samples were taken from the purebred Bouyei people who lived at Zhengfeng, an autonomous county of Bouyei and Miao people in Guizhou Province of China. Blood relationships did not exist amongst the samples. The peripheral blood samples were collected, and the genomic DNA was abstracted from leucocytes by phenol-chloroform. Forty-five male and 35 female samples were obtained in this study.

#### Genotyping of Y-chromosome haplotype

A total of 13 Y-chromosome single nucleotide polymor-

phisms were screened, including M7 (C-G mutation), M9 (C-G mutation), M15 (9-bp insertion), M45 (A-G mutation), M88 (G-A mutation), M89 (T-C mutation), M95 (C-T mutation), M110 (C-T mutation), M119 (A-C mutation), M122 (T-C mutation), M130 (T-C mutation), M134 (1-bp deletion), and YAP (DYS287). For genotyping, the polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP) assay was used for testing the Y-chromosome biallelic markers. According to Su and Ke et al,<sup>5,6</sup> the sequences of the primers and their PCR conditions and restriction enzymes were used. The Y-chromosome haplotype was judged by the phylogenetic tree of the Y-chromosome haplotype.

#### Detection of mtDNA polymorphism

The polymorphism of mtDNA region V in the samples were detected by DNA sequencing after PCR. The sequences of the primers and their PCR conditions were designed according to the previous results.<sup>7,8</sup>

### RESULTS

#### Frequency distribution of Y-chromosome haplotypes

Nine Y-chromosome haplotypes (H1, H4, H5, H6, H7, H8, H9, H11, and H12) were found in 45 male Bouyei

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**Table.** Y-chromosome haplotype frequency distribution in China and world populations

Region	Population	n	Frequency of haplotype																
			H1	H2	H3	H4	H5	H6	H7	H8	H9	H10	H11	H12	H13	H14	H15	H16	H17
China																			
Northern Han																			
	Liaoning	6	16.7					50.0					16.7			16.7			
	Hebei	6						16.7	50.0			33.3							
	Shandong	32	9.4			3.1		18.8	28.1			28.1	9.4			3.1			
	Henan	28	7.1		3.6	25.0		32.1				14.3	10.7			7.1			
Southern Han																			
	Zhejiang	50	12.0			6.0		24.0				26.0	26.0		6.0				
	Jiangsu	55	7.3	1.8		1.8		18.2	23.6	3.6		21.8	16.4		3.6			1.8	
	Hubei	18	5.6					11.1	27.8	5.6		33.3	16.7						
	Hunan	15						13.3	33.3			26.7	13.3		13.3				
	Guangdong	15						40.0				26.7	20.0			6.7	6.7		
	Shichuan	14	7.1					28.6	7.1	7.1		35.7	7.1		7.1				
	Yunnan	27	11.1			3.7		3.7	18.5			55.6	3.7		3.7				
	Yao	10	20.0		30.0				10.0						40.0				
	Dong	10	20.0						10.0			20.0	20.0	10.0	20.0				
	Li	11										9.1	27.3		54.6	9.1			
	Zhuang	28	3.6		3.6	7.1		3.6	3.6			25.0	17.9		25.0	10.7			
	Tujia	10	10.0					20.0	30.0	10.0			20.0			10.0			
	Yi	14			14.3			42.9	21.4			7.1			14.3				
	Bouyei	45	7.0			4.4		17.7	4.4	2.2		2.2	4.4		46.7	11.1			
Asia																			
	Cambodian	26	3.8		3.8	11.5		11.5	3.8			15.4	3.8	3.8	32.1	11.5		3.8	3.8
	Thai	20				5.0		5.0	5.0	5.0			5.0	5.0	45.0	20.0		5.0	
	Malaysia	13				7.7		7.7	30.8			15.4	7.7	23.1	7.7				
Others																			
	African	24	20.8	79.2															
	American	26															3.8	96.2	
	European	39	10.3			12.8		25.6									51.3		

The Y-chromosome haplotype frequency distribution in Bouyei people are calculated in this study. The other data are quoted from previous records.<sup>5-7</sup>

samples. The frequency distribution of these haplotypes in Bouyei people and other populations are listed in Table.

**mtDNA polymorphism in Bouyei people**

Only two mtDNA polymorphisms ( standard and short type ) were observed in all samples. The frequency distribution of short type ( 9-bp deletion ) in the Bouyei people was 33.3%.

**DISCUSSION**

Nine Y-chromosome haplotypes ( H1 , H4 , H5 , H6 , H7 , H8 , H9 , H11 , and H12 ) found in the Bouyei people were compared with those in worldwide populations. The two ancient haplotypes , H1 and H2 , were found in all worldwide populations. This may indicate that the mutations at these loci had occurred before humans moved out from Africa. It is confirmed that haplotype H5 is the predominant haplotype of other geographical haplotypes ( H6 – H17 ). Furthermore , haplotypes H6 – H12 are

essentially eastern-Asian specific.<sup>5-7</sup> The frequency distribution of these haplotypes in the southern Chinese ( including the Han people and other minorities ) is similar to that in southeastern-Asian populations. In addition , H7 , H11 , and H12 haplotypes only exist in southern populations in China , but not in northern populations. We conclude that the ancestors of the Chinese arrived in southern areas of China at first , then moved northward.

In this study , the southern-Chinese specific haplotypes ( H7 , H11 , and H12 ) were found in the Bouyei people. The frequency of haplotypes H11 and H12 ( mutation at M95 locus ) in the Bouyei people was the highest. The total frequency distribution of 9 haplotypes in the Bouyei people were very similar to that in Daic speaking populations , including the Zhuang , Li , Dong and Yao people from Jinxiu. Therefore , it is quite clear that the Bouyei people belong to southern-Chinese populations. This coincides with the Bouyei people 's geographical character. Furthermore , the same conclusion was reached

by comparing the polymorphism of the mtDNA region V between the Bouyei people and other Chinese populations.<sup>8,9</sup> These results indicate a closer genetic relationship amongst the Bouyei people and Daic speaking populations mentioned above.

According to historical exploration, the Bouyei people were descended from the ancient Pu and Baiyue peoples. They had to move to the Guizhou and Yunnan areas because they were defeated by the ancient Chu Han people. In the evolution of the Bouyei people, the Han Chinese continuously amalgamated into their population.<sup>10</sup>

In conclusion, the results presented in this report suggest that the Baiyue people make up the main population in the paternal history of the Bouyei people. In addition, the Pu and Han Chinese people amalgamated into the Bouyei population. Gene intercourse between the Bouyei people and other populations create the genetic characteristics of today's Bouyei people. Furthermore, the study of single nucleotide polymorphisms (SNPs) is a useful method, not only in the evolution of humans, but also in medical research. It has been confirmed that mtDNA is related to some diseases, such as senility. Also, the mutation in some chromosomes plays an important role in the occurrence of some diseases. Therefore, our findings may explain particular diseases in the Bouyei people.

#### REFERENCES

1. Vigilant L, Stoneking M, Harpending H, et al. African

- populations and the evolution of human mitochondrial DNA. *Science* 1997 ;253 :1503-1507.
2. Horai S, Murayama K, Hayasaka K, et al. mtDNA polymorphism in east Asian populations, with special referable to the peopling of Japan. *Am J Hum Genet* 1996 ; 59 :579-590.
3. Qian Y, Qian B, Su B, et al. Multiple origins of Tibetan Y chromosomes. *Hum Genet* 2000 ;106 :453-454.
4. Kim W, Shin DJ, Harihara S, et al. Y chromosome DNA variation in east Asian populations and its potential for inferring the peopling of Korea. *J Hum Genet* 2000 ;45 :76-83.
5. Su B, Xiao JH, Underhill R, et al. Y-chromosome evidence for a northward migration of modern humans into eastern Asia during the last ice age. *Am J Hum Genet* 1999 ;65 :1718-1724.
6. Ke YH, Su B, Xiao JH, et al. The frequency distribution of Y-chromosome haplotypes in Han Chinese and the origin and migration of Chinese population. *Sci Chin ( Life Sci )* 2000 ; 30 :614-620.
7. Anderson S, Bankier AT, Barrell BG, et al. Sequence and organization of the human mitochondrial genome. *Nature* 1981 ;290 :457-467.
8. Harihara S, Hirai M, Suutou Y, et al. Frequency of a 9-bp deletion in the mitochondrial DNA among Asian population. *Hum Biol* 1992 ;64 :161-166.
9. Mellon T, Peterson R, Rudd AJ, et al. Polynesian genetic affinities with southeast Asian populations as identified by mtDNA analysis. *Am J Hum Genet* 1995 ;57 :403-414.
10. Wang XF, Wang RS, Wei GY. In: Wang XF. History of nationality: Bouyei people lived by North-Pan River. Guiyang: Guizhou Publishing House ;1985 :4-12.

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