

## SHORT COMMUNICATION

Min Yu · Yongli Zhang · Yali Xue · Feng Chen · Qi Wang  
 Xiaoyi Huang · Baiqiu Wang · Yang Yu · An Liu  
 Linlin Ma · Rongqian Shi · Fuqu Lu · Zhongcheng Shi  
 Yu Zhang · Wenhong Cheng · Qionghua Ai · Fang Xu  
 Chengbin Huang · Baibin Chen · Huanjie Yang  
 Xianghua Kang · Yanyang Sun · Guiyin Zhang · Pu Li  
 Songbin Fu

## A new haplogroup pattern displayed in Fujian Han in China

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**Abstract** Human Y-chromosomal binary polymorphisms have been considered to preserve the paternal genetic legacy and provide evidence on human evolution and the genetic relationships among and demographic history of different populations. To reveal the genetic origin and immigration of the Fujian Han, 13 binary markers on the Y chromosome were used to screen Fujian Han by allele-specific polymerase chain reaction. The results indicated that the M<sub>9</sub>G marker was highly prevalent (96.20%), suggesting a significant genetic drift. In addition, M<sub>122</sub>C frequency was only 22.78%, and M<sub>45</sub>A and M<sub>103</sub>T were default. The distinctive haplogroup frequencies (H<sub>1</sub>, H<sub>5</sub>, and H<sub>6/7/8</sub>) imply that the haplogroup pattern is a relatively ancestral and interim type.

**Key words** Binary polymorphism · Y chromosome · Haplogroup · Fujian Han · Population

### Introduction

An increasing number of Y-chromosomal binary markers have been widely accepted as a powerful tool for unraveling human evolution (Hammer and Zegura 1996;

Hammer et al. 1997; Underhill et al. 1996, 1997; Lell et al. 1997; Bianchi et al. 1998; Karafet et al. 1999). Y-chromosomal binary markers are single base-pair substitutions or small sequence insertion/deletion mutations with a low mutation rate (Underhill et al. 1997). The large numbers available and the low mutation rate of binary markers have given them an important role in studies of human origin and relationships among different populations (Wang et al. 1998). Nineteen markers polymorphic in the individuals of eastern Asian origin were selected by Underhill et al. (2000) from 166 biallelic Y-chromosome markers. Ten out of the 19 Y-chromosomal binary markers are East Asian specific and generally absent from other world populations; therefore, those markers are expected to be highly informative for studying the migrations of East Asian populations (Su et al. 1999, 2000). In the present paper, 13 of the 19 binary markers were genotyped in the Fujian Han population of China to reveal the characteristic distribution of the loci and the relatively ancient haplogroup pattern.

### Subjects and methods

#### DNA samples

Eighty healthy unrelated male individuals were randomly selected from the Han population of Fujian Province in southeastern China. All persons gave informed consent prior to their inclusion in the study. DNA samples were prepared from peripheral blood samples by using the standard phenol–chloroform extraction method.

#### Genotyping of Y-chromosome binary markers

A total of 13 Y-chromosome biallelic loci were screened: M<sub>1</sub> (Alu insertion, also called YAP), M<sub>15</sub> (9-bp insertion), M<sub>89</sub> (C → T), M<sub>9</sub> (C → G), M<sub>119</sub> (A → C), M<sub>50</sub> (T → C), M<sub>110</sub> (T → C), M<sub>103</sub> (C → T), M<sub>95</sub> (C → T), M<sub>88</sub> (A → G), M<sub>111</sub> (2-bp deletion), M<sub>45</sub> (G → A), and M<sub>122</sub> (T → C). For each base

M. Yu · Y. Zhang · Y. Xue · F. Chen · Q. Wang · X. Huang ·  
 B. Wang · Y. Yu · A. Liu · L. Ma · R. Shi · F. Lu · Z. Shi · Y. Zhang ·  
 C. Huang · B. Chen · H. Yang · X. Kang · Y. Sun · G. Zhang · P. Li ·  
 S.B. Fu (✉)

Laboratory of Medical Genetics, Department of Biology, Harbin  
 Medical University, Harbin 150086, China  
 Tel./Fax +86-0451-6677243  
 e-mail: fusb@ems.hrbmu.edu.cn

W. Cheng  
 Hailaer Sanitation and Epidemic Prevention Institute, Hailaer, China

Q. Ai  
 Yining Health School of Xinjiang Uighur Ethnic Municipality,  
 Xinjiang Yining, China

F. Xu  
 Ningxia Hui Ethnic Municipality Medical University, Yinchuan,  
 China

**Table 1.** Variation frequencies of 13 binary markers in five Chinese groups (%)

Groups (size)	M <sub>1</sub> <sup>+</sup>	M <sub>15</sub> <sup>+</sup>	M <sub>89</sub> T	M <sub>9</sub> G	M <sub>119</sub> C	M <sub>50</sub> C	M <sub>110</sub> C	M <sub>103</sub> T	M <sub>95</sub> T	M <sub>88</sub> G	M <sub>111</sub> <sup>+</sup>	M <sub>45</sub> A	M <sub>122</sub> C
Fujian Han (80)	1.25	2.53	76.25	96.20	13.92	8.75	11.39	0	11.25	1.25	11.39	0	22.78
Mongolia (45)	—	0	48.89	44.44	0	0	0	0	0	0	0	0	—
Daur (36)	—	0	72.22	64.52	5.56	0	0	0	0	0	0	0	—
Xibe (42)	—	2.38	70.73	61.9	7.14	0	0	0	0	0	0	0	—
Hezhe (45)	—	0	66.67	73.33	0	0	0	0	0	0	0	0	—

For M<sub>1</sub><sup>+</sup>, M<sub>88</sub>T, M<sub>50</sub>C, M<sub>95</sub>T and M<sub>88</sub>G in the Fujian Han, *N* = 80; For M<sub>9</sub>G, *N* = 78; and for all others, *N* = 79  
M<sub>1</sub><sup>+</sup>, YAP<sup>+</sup>; M<sub>15</sub><sup>+</sup>, 9-bp insertion; M<sub>111</sub><sup>+</sup>, 2-bp deletion

**Table 2.** Distribution of M<sub>89</sub>T and M<sub>9</sub>G in different populations of China

Population (size)	M <sub>89</sub> T			M <sub>9</sub> G		
	Frequency (%)	$\chi^2$	<i>P</i>	Frequency (%)	$\chi^2$	<i>P</i>
Fujian Han (80)	76.25			96.20		
Mongolian (45)	48.89	10.39	<0.05	44.44	46.77	<0.05
Daur (36)	72.22	0.36	>0.05	64.52	22.89	<0.05
Xibe (42)	70.73	0.63	>0.05	61.90	27.03	<0.05
Hezhe (45)	66.67	1.63	>0.05	73.33	16.44	<0.05

$\chi^2$  value is incomparison with the Fujian Han

substitution locus, two allele-specific primers were synthesized to recognize two different alleles. The primer sequences were kindly provided by Li Jin, University of Texas (the primer sequences are available on request). An allele-specific genotyping assay was used to type the 13 Y-chromosome binary markers as described in previous reports (Su et al. 1999; Ke et al. 2000).

## Results and discussion

The variation frequencies of the 13 Y-chromosome binary markers for the 80 samples from the Fujian Han are shown in Table 1. For the purpose of comparison, their frequencies in four northern Chinese groups (Mongolian, Daur, Xibe, and Hezhe) are included (Feng Chen, Harbin Medical University of China, unpublished data). The Fujian Han display a much greater diversity of loci than do the other four ethnic groups.

Among the loci, M<sub>9</sub>G had extremely high frequency, 96.20% on average (91.98%–100%). M<sub>89</sub>T frequency was also high: 76.25% (66.92%–85.58%). It has been reported that M<sub>9</sub>G and M<sub>89</sub>T are generally absent in Africans, but very common in non-African populations (Su et al. 1999; Underhill et al. 2000). Our data show that M<sub>9</sub>G occurred with much greater frequency in the Fujian Han than in the other four groups (*P* < 0.05), but that M<sub>89</sub>T occurred with a relatively average frequency compared with the Daur, Xibe, and Hezhe, *P* > 0.05; compared with the Mongolian, *P* < 0.05) (Table 2). This high prevalence of M<sub>9</sub>G might suggest a significant genetic drift during the evolution of the Fujian Han.

The data in Table 1 indicate that the C allele in M<sub>122</sub> is common in Fujian Han, having a medium frequency (22.78%). It is well known that the M<sub>122</sub>C allele is East

Asian specific and absent in Africans, Europeans, and Oceanians (Su et al. 1999). Moreover, recent studies of extant Siberian and Central Asian populations reveal only sporadic occurrences of M<sub>122</sub>C (Su et al. 2000). The average frequency of M<sub>122</sub>C in Sino-Tibetan populations is 52.2% (28.6%–100%) (Su et al. 2000). The distinctive frequency at this locus in the Fujian Han probably reflects their demographic history.

M<sub>1</sub>(YAP) is an ancient polymorphism occurring both in Africans and Asians, but generally absent in other populations (Hammer et al. 1997). The highest frequency of YAP<sup>+</sup> has been revealed among the black populations from Africa (79%), followed by Europeans, Oceanians, and Asians (Spurdle et al. 1994). In Japan, the YAP<sup>+</sup> frequency was found to be 42% (Spurdle et al. 1994). However, ten Indian ethnic groups were monomorphic for the deletion allele at the YAP locus (Bhattacharyya et al. 1999). Similarly, Wang et al. (1999) reported that four Northeastern ethnic groups in China were all YAP-negative. Xu et al. (1998) got the same result from the Guangdong Han (south China) and Shanxi Han (north China). In the current study, only one YAP<sup>+</sup> among 80 Fujian Han individuals was found. Taken together with results of some previous investigations that showed low frequencies of YAP<sup>+</sup> in Jiangsu Han (1.8%), Shanghai Han (3.3%), Sichuan Han (7.1%), and Jiangxi Han (4.8%) (Su et al. 2000), it is reasonable to suggest that YAP<sup>+</sup> is present in Chinese Han populations with low frequency.

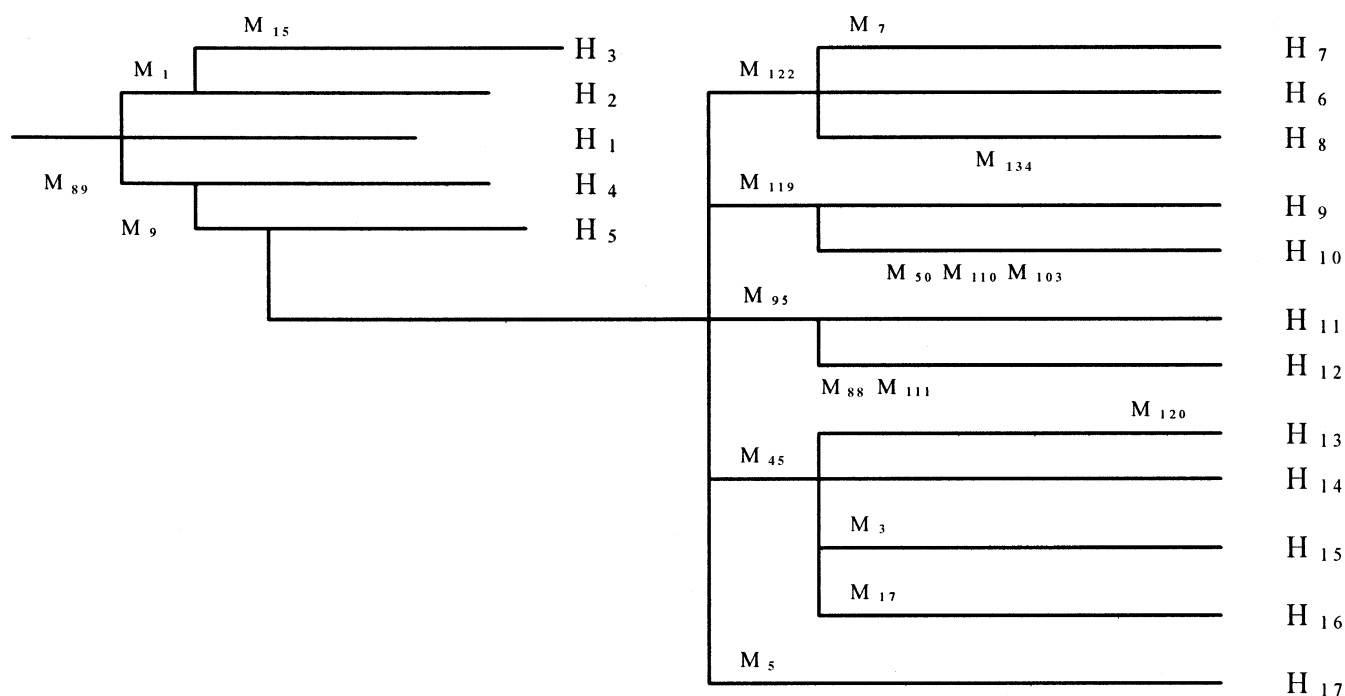
As shown in Table 1, the Fujian Han are monomorphic for M<sub>103</sub>C and M<sub>45</sub>G. These results are the same as for the four northern groups (Mongolia, Daur, Xibe, and Hezhe). This is consistent with the findings of Su et al. (1999) that M<sub>45</sub>A is relatively rare in East Asian populations.

According to the phylogenetic tree constructed by Su et al. (1999) (See Fig. 1), the haplogroup pattern of the Fujian Han population is as shown in Table 3. Seven of the 17

**Table 3.** Y-chromosome haplogroup distribution in Fujian Han and world populations

Population (size)	Frequency (%)														
	H <sub>1</sub>	H <sub>2</sub>	H <sub>3</sub>	H <sub>4</sub>	H <sub>5</sub>	H <sub>6/7/8</sub>	H <sub>9</sub>	H <sub>10</sub>	H <sub>11</sub>	H <sub>12</sub>	H <sub>13</sub>	H <sub>14</sub>	H <sub>15</sub>	H <sub>16</sub>	H <sub>17</sub>
East Asia:															
Northern Han (82) <sup>a</sup>	8.5			2.4	22.0	52.5	9.8				4.9				
Southern Han (280) <sup>a</sup>	7.9	0.4		1.4	12.9	55.1	16.8		3.6	0.7	1.4				
Fujian Han (80)	22.1		1.3		48.1	9.1	11.7		7.8						–
Africa:															
African (24) <sup>a</sup>	20.8	79.2													
Europe:															
European (39) <sup>a</sup>	10.3			12.8	25.6							51.3			
Oceania:															
Oceanian (100) <sup>a</sup>	16			2	40							4			38

<sup>a</sup>From Su et al. (1999)



**Fig. 1.** Phylogenetic tree of 17 Y chromosome haplogroups defined by 19 biallelic markers. This is a revised version of that presented by Bing Su et al. (1999). H<sub>1</sub> was considered to be the ancestral haplogroup

because of its appearance in chimpanzees. The others were established according to the derived type of the corresponding markers

haplogroups were present in the Fujian Han population: H<sub>1</sub>, H<sub>3</sub>, H<sub>4</sub>, H<sub>5</sub>, H<sub>6/7/8</sub>, H<sub>9</sub>, and H<sub>11</sub>. They all showed the characteristic East Asian signature (Su et al. 1999). H<sub>1</sub>, the ancestral haplogroup, which is the root of the phylogenetic tree, has a frequency of 22.1% in the Fujian Han, corresponding to that in the African population (20.8%). H<sub>5</sub>, the predecessor of H<sub>6</sub>–H<sub>17</sub> (derived from H<sub>5</sub>, with additional mutations), characterizes 48.1% of the Fujian Han Y chromosomes, which is close to the frequency Oceanians (40%). Haplogroup H<sub>6/7/8</sub>, which shares a T to C substitution at locus M<sub>122</sub>, has been found to be predominant in East Asians and absent in Africans, Europeans, and Oceanians (Su et al. 1999). Surprisingly, this finding is not consistent with the Fujian Han results (in whom H<sub>6/7/8</sub> accounts for only 9.1%). In comparison with the southern Han and the north-

ern Han, the Fujian Han displayed a relatively lower frequency of H<sub>6/7/8</sub>, but much higher frequencies of H<sub>5</sub> and H<sub>11</sub>. The characteristics of the Y-chromosome loci are not compatible with those of either the southern Han or the northern Han, although Fujian Province is located in southeast China. In conclusion, the genetic evidence of Y-chromosome haplogroup distributions present an interesting picture regarding the origin of the Fujian Han. The distinctive frequencies of haplogroups H<sub>6/7/8</sub>, H<sub>1</sub>, and H<sub>5</sub> in the Fujian Han imply that the haplogroup pattern is a relatively ancestral and interim type. If the ancestors of the Sino-Tibetan population in China were originally from southern East Asia (Su et al. 1999), they might have reached the Fujian region first, with a part of them continuing to reside there, and the others making their way northward.

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