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The origin of Mosuo people as revealed by mtDNA and Y chromosome variation

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Abstract The Mosuo, living in the Lugu Lake area in northwest Yunnan Province, China, is the only matriarchal population in China. The Mosuo was officially identified as Naxi nationality although its relationship with Naxi remains controversial. We studied the genetic relationship between the Mosuo and five other ethnic groups currently residing in northwest Yunnan, i.e. Naxi, Tibetan, Bai, Yi and Pumi, by typing the genetic variations in mtDNA HVS1 and 21 Y chromosome markers (13 SNPs & 8 STR markers). We showed that the maternal lineages of the Mosuo bear the strongest resemblance with those found in Naxi while its paternal lineages are more similar to those that are prevalent in Yunnan Tibetan. The marked difference between paternal and maternal lineages may be attributable to the genetic history, matriarchal structure, and visiting marriage.

Keywords: Mosuo, mtDNA, Y chromosome, polymorphism.

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The Mosuo is the only living matriarchal population in China, and is one of the most important model communities for anthropological studies of matriarchal society[1]. The national censor conducted in 1990 indicates that there are about 15000 Mosuo people who are mainly distributed in the Lugu Lake area at Ninglang County in the northwest Yunnan. Historical studies showed that the Mosuo was derived from an ancient tribe Maoniu Qiang, which is a branch of Di-Qiang group. The Di-Qiang group, the ancestor of all Tibetan-Burman language subfamily, originally lived in northwest China and migrated to southwest China about 2700 years ago[2,3]. The Mosuo was identified as a subgroup of the Naxi in the 1950s. However, the Mosuo believe that they are a unique ethnic group or should be a branch of Tibetan instead of the Naxi since they bear stronger cultural similarity with the former than the latter[4]. A study of genetic relationship of the Mosuo, Tibetan, and Naxi may shed light on the understanding of the origin of the Mosuo.

In the past two decades, genetic markers have been widely used to infer the origin, migration and admixture of human populations[5—8]. Among them, mtDNA and non-recombining portion of Y chromosome were shown to be more informative in tracing human evolutionary history since they only transmit through maternal and paternal lineages, respectively[5,9]. For mtDNA markers, the HVS1 segment in the D-loop region has higher mutation rate than the rest of mtDNA, therefore becomes the most studied marker in inferring genetic relationships among different populations with plethora of data from worldwide populations for comparison. Recently, the utility of Y chromosome markers for resolving genetic history of human populations has been recognized[9—11]. Haplotypes derived from single nucleotide polymorphisms (SNPs) in the non-recombining portion of the Y chromosome, which occurred in an ordered time series, have been widely used for inferring the history of the given populations. Of special relevance to this study, Y SNPs have been used effectively in revealing the origin and prehistoric migrations in East Asia and Pacific[12—19]. Short tandem repeats (STRs) are another type of genetic markers in Y chromosome and they have been extensively used to reconstruct phylogenic relationship among closely related populations and estimate the age of evolutionary events, due to their higher mutation rates[20,21].

The Mosuo has rich culture and unique custom, and the cultural influence from the Tibetan, Naxi, Pumi and Yi is evident[4]. In this study, we analyzed the polymorphisms in the mtDNA HVS1 region, 13 Y-SNPs and 8 Y-STRs in the Mosuo and five other populations currently residing in the northwest Yunnan (Naxi, Tibetan, Yi, Bai, and Pumi). Principle component (PC) analysis and phylogenic analysis were employed to reveal their genetic relationship of both paternal and maternal lineages.

1 Materials and methods

1.1 Samples

Samples of all six populations in this study were collected with proper informed consent. They were sampled from the populations in the following locations: Ninglang County (Mosuo and Pumi), Dali (Bai), Lijiang (Naxi), Shuangbai (Yi) and Zhongdian County (Tibetan). Efforts were made to ensure that the subjects in this study are not related and all of the four grandparents of each subject are from the same ethnic group as the subject himself. 5 mL blood was extracted for each individual, and anticoagulated by ACD. Genomic DNA were extracted by standard phenol-chloroform method and stored at –20℃ after extraction. Only male subjects were analyzed for Y chromosomes and most of the samples that have been analyzed for mtDNA are males, except for 5 unrelated female individuals in the Naxi.

1.2 DNA analysis

The HVS1 region of mtDNA was amplified by the primers L15996 and H16401[22]. PCR products were purified by resin. The purified PCR fragments were subsequently sequenced using ABI Big-Dye sequencing Kit and ABI PRISM 377 DNA sequencer (Applied Biosystems, Forster City, CA, USA). Sequence Analysis 3.3 software (Applied Biosystems) was used to extract sequences. All genotyping results were confirmed with the sequences from both directions. HVS1 sequences of the Mosuo have been submitted to GenBank (Accession number: to be added).

We selected 13 Y-SNPs (YAP, M7, M9, M15, M45, M88, M89, M95, M110, M119, M122, M130 and M134) that are most informative for East Asian populations, following Su et al.[14]. The length variation of YAP and M15 was directly observed on 3%—4% NuSieve agarose (FMC, Bioproducts, Rockland, ME, USA). The other 11 loci were genotyped by PCR-RFLP assay. Restrict enzyme sites were engineered into one primer for all the loci, except for M130, which carried one natural restriction site around the SNP. Primers, amplification conditions and restriction enzymes for the 13 sites are shown in table 1.

We genotyped 8 STRs in Y chromosome (DYS19, DYS388, DYS389-1, DYS389-2, DYS390, DYS391, DYS392 and DYS393). Primers and size information

Table 1 Primers, PCR condition, restriction enzymes and allele calling for Y-SNPs

Locus	Annealing temperature/℃	Primer sequence (5′—3′)	Restriction enzymes	Allele calling (Size of fragment bp)	
YAP	53	CAGGGGAAGATAAAGAAATA ACTGCTAAAAGGGGATGGAT	/	non-Alu (150)	Alu insertion (400)
M15	56	ACA AATCCTGAACAATCGC GTCTGGGAAGAGTAGAGA AAAG	/	9 bp insertion (151)	none insertion (142)
M89	52	GAAAGTGGGGCCCACAGAAGGA GCAAATCAGGCAAAGTGAGACAT	*Nla* III	C (79+21)	T (100)
M9	55	GAAACGGCCTAAGATGGTTGGAT AAACTGAATCTTTTTTCCTCATTTTTG	*Bam* HI	C (190+20)	G (210)
M119	55	AGGTAAATGACTCACCCTAAGGAAG GGGTTATTCCAATTCAGCATACACGC	*Bst*u I	A (161)	C (135+26)
M95	55	ATAAGGAAAGACTACCATATTAGCG TTTGAAGGCCCCAGTTGTGAG	*Hha* I	C (178+24)	T (202)
M122	55	TAGAAAAGCAATTGAGATACTAATTCA GCGATGCTGATATGCTAGTTCAG	*Nla* III	C (100+22)	T (122)
M134	55	AAGGACCAGGAAAGTATGATCG TTTGATGATTCTTCTTTGGGCTTC	*Nla* III	none deletion (100+22)	1 bp deletion (122)
M7	56	TGTACCCTTGACCAATGCCTT TTGTAGTTGAGTTACTGTTCTTCTA	*Bfa* I	C (103+23)	G (126)
M130	56	TATCTCCTCTTCTATTGCAG CCACAAGGGGGAAAAAACAC	*Bsl* I	T (205)	C (162+43)
M110	55	AACATTCTCTGTAGACTCACTGG ATTTAGCACTTCTTTTCCCC	*Nla* III	T (200)	C (88+122)
M88	55	TCTTATTCCTGCTTCTTCCGC CATGTGATGGTTCAGTAGGTGTGA	*Bst*u I	A (146)	G (125+21)
M45	55	ATTGGCAGTGAAAAATTATAGCTA TGCCTTTGCTACAACTCTCCTA	*Bfa* I	G (140+22)	A (162)

for those Y-STRs can be found in the GDB database (http://www.gdb.org). Forward primers were labeled with 6-FAM (DYS388, DYS389), NED (DYS390, DYS392, DYS393) and HEX (DYS19, DYS392), respectively. Y-STRs were amplified by multiplexing and electrophoresized at ABI PRISM 377 sequencer as described in the previous report[21]. Genescan™2.0 and Genotype™1.1 software (Applied Biosystems) were used for sizing and allele calling. Two standard samples, whose fragment sizes were confirmed by directly sequencing, were added to each electrophoresis to calibrate the allele calls from different gels.

1.3 Data analysis

The HVS1 sequences of 360 bp (16024—16083) were aligned by ClustalW software, in reference to the CRS[23]. Fst and Nei's net genetic distance d_A[24] were calculated by ARLEQUIN[25]. The statistical significance of Fst was examined based on 3000 permutations, under the null hypothesis assuming no difference between the compared populations. Haplotype and haplogroup assignment for each sample were inferred following Yao et al. based on diagnostic sites in HVS1 region[26]. Inference of Y-SNP haplotypes was achieved following Su et al. and the nomenclature therein[14]. For Y-STRs, Rst[27] between populations was estimated using ARLEQUIN, its statistical significance was examined based on 3000 permutations assuming no difference between the populations. Multidimensional scanning (MDS) of distance matrix, principal component (PC) analysis and correlation analysis were conducted using SPSS. Neighbor joining (NJ) phylogenic tree was constructed using MEGA[28].

In the PC analysis of YSNP haplotypes, we included the data of Tibetan[15], Yunnan Han[18], Sichuan Han[18], Zhuang[14], Mongolian[14], Buyi[19], and Uyghur (unpublished). And mtDNA data of Yunnan Han[26], Guangdong Han[26], Qingdao Han[26], Xinjiang

Table 2 Y-SNP haplotype frequency

Population	Size	Diagnostic mutation and haplotype												
		M130	YAP	M15	M89	M9	M122	M7	M134	M119	M110	M95	M88	M45
		H1	H2	H3	H4	H5	H6	H7	H8	H9	H10	H11	H12	H13
Bai	50	6.0	2.0	4.0		18.0	8.0	4.0	34.0	6.0		12.0	2.0	4.0
Yi	50	8.0	2.0	10.0	38.0	16.0	10.0		10.0	2.0		4.0		
Mosuo	47	6.4	23.4	4.3	2.1	31.9	4.3	6.4	12.8	8.5				
Naxi	40	2.5	37.5			10.0			2.5			47.5		
Pumi	47	2.1	70.2	2.1	4.3	6.4	2.1		6.4	4.3				2.1
Tibetan-YN	50	4.0	28.0	8.0		12.0	6.0	4.0	34.0			4.0		

Han[26], Uyghur[29], Kazak[29], Tu[30], Mongolian[30], Dai[30], Zhuang[30] and Wa[30] were used in this study.

2 Results

2.1 Frequency of Y-SNP haplotype and mtDNA haplogroup

Table 2 presents Y-SNP haplotype frequencies in the six populations. Overall 10 haplotypes were observed in this study with 9 haplotypes in the Mosuo, Bai and Yi, 8 in Tibetan and Pumi, and only 5 in the Naxi. H5, H2 and H8 are the major haplotypes observed in the Mosuo and Tibetan, which account for 68% and 74% in the two populations, respectively. H2 and H11 are the most prevalent haplotypes in Pumi (70.2%) and Naxi (47.5%). Haplotype frequencies in Bai and Yi are relatively dispersed, indicating higher diversity in these two populations. Of special interest is that H11, the most frequent haplotype in the Naxi (47.5%) was not observed in the Mosuo.

Among 293 individuals analyzed, 177 HVS1 mtDNA haplotypes were observed, with 39 haplotypes in the 47 Mosuos. Among the 39 haplotypes found in the Mosuo, 29 types are unique, 5 with the Naxi; 3 with the Pumi, and 2 with the Bai, Tibetan and Yi, respectively. The haplotypes were classified into haplogroups based on HVS1 sequences following Yao et al.[26]. Majority of the haplotypes (87%) can be assigned to one of the haplogroups, whose frequencies are listed in table 3. B, D and F are the most frequent haplogroups in the Mosuo (73.8%), and the frequencies of Haplotype B, F in the Naxi are very close to that of the Mosuo. F and G are the main haplogroups in the Bai, and Haplotype A, D are most frequent in the Tibetan, Pumi and Yi (frequency of haplogroup C is very high in Pumi).

2.2 Genetic distance

We estimated Rst distance using Y-STR data (table 4). As shown in the matrix of pairwise Rst based on 8 Y-STRs (table 4), the distance between the Mosuo and Tibetan is the smallest among all pairwise comparisons, and the hypothesis of no difference cannot be rejected. The Rst value between the Mosuo and Naxi is as six-fold as that between the Mosuo and Tibetan. For mtDNA HVS1 data, two genetic distance estimators, Fst and Nei's net genetic distance (d_A) were used and listed in table 5. The two estimators for mtDNA data showed a perfect correlation ($r = 0.974$, $p < 0.01$). Interestingly, the distance between the Mosuo and Naxi are much smaller than those between the Mosuo and others for both distance measures. The distances estimated using mtDNA data showed a different pattern from those estimated using Y-STR data ($r = 0.298$, $p = 0.28$), indicating a marked differential genetic history of maternal and paternal lineages of the Mosuo.

2.3 Principal component (PC) analysis

The PC plot based Y-SNP haplotype frequency is presented in fig. 1. The first three PCs account for 70.3% of the total variance. Four clusters are evident: A (Yunnan Tibetan, Tibetan, Mosuo, Pumi), B (Bai, Dai, Sichuan Han and Yunnan Han), C (Mongolian, Uyghur and Yi), D (Zhuang, Buyi and Naxi). The second principle component (PC2) separates the 5 populations from northwest Yunnan (populations in Cluster A and Naxi) from the rest. This PC is signi-

Table 3 Haplogroup frequency estimated by HVS1 variation motif

mtDNA haplogroup	YN Han[a)]	QD Han[a)]	XJ Han[a)]	GD Han[a)]	Bai	Mosuo	Naxi	Pumi	YN Tibetan	Yi
	(43)	(50)	(47)	(69)	(37)	(46)	(45)	(35)	(36)	(40)
A	4.7	4.0	10.6		5.4	4.3	8.9	14.3	13.9	19.5
B*	2.3	2.0								
B4	11.7	10.0	2.1	29.0		13.0	17.8	2.9	5.6	12.2
B5*					5.4					
B5a	4.7		4.3			17.4	6.6		11.1	
B5b	2.3		2.1	1.4						
B total	21.0	12.0	8.5	30.4	5.4	30.4	24.4	2.9	16.7	12.2
C	4.7		6.4			13.0	8.9	22.9	8.3	2.4
D4k	9.3	26.0	19.1	10.1	10.8	13.0	6.7	17.1	19.4	14.6
D5*	2.3	3.9	2.1	5.8						
D5a	2.3	6.0	4.3			8.7		5.7	5.6	2.4
D total	13.9	35.9	25.5	15.9	10.8	21.7	6.7	22.8	25.0	17.0
F*	2.3		2.1	1.4						
F1a	11.6	4.0	4.3	17.4	16.2		17.8	2.9		4.9
F1b	4.7	4.0	2.1	1.4	5.4	17.4	4.4	2.9		4.9
F1c		2.0	2.1	1.4	2.7					
F2*				2.9						
F2a	2.3	2.0	4.3	1.4	8.1	4.3	2.2	5.7	2.8	2.4
F total	20.9	12.0	14.9	25.9	32.4	21.7	24.4	11.5	2.8	12.2
G2		6.0	2.1	1.4	10.8		2.2			2.4
M10	2.3	2.0		2.9	8.1		8.9	2.9	2.8	9.8
M7*	2.3		2.1	1.4						
M7B	16.3	4.0	6.4	8.7	5.4		2.2			12.2
M7c			2.1	1.4						
M7 total	18.6	4.0	10.6	11.5	5.4		2.2			12.2
M8a		8.0	4.3	2.9						
M9		4.0	4.3	0.0	5.4			5.7	5.6	7.3
N9a	7.0	6.0		1.4						
R9a	2.3		4.3	1.4			2.2			
Y		2.0	2.1							
Z			2.1						2.8	
Others[b)]	4.6	4.0	4.2	5.7	16.2	8.6	11.1	17.1	25	4.9

a) Data from ref. [26]; b) containing undefined M^*, N^* and R^* lineages.

ficantly correlated with the frequency of H2(r=0.844, p<0.01), indicating high frequency of H2 is the primary characteristics of the northwest Yunnan populations. The first principle component (PC1) and the third principle component (PC3) separate Cluster A and D, and they have significant correlation with the frequency of H8 (r = 0.810, p < 0.01) and H11 (r = 0.898, p < 0.01), demonstrating that differential in H8 and H11 frequencies marks the genetic difference between the Mosuo and Naxi.

Table 4 Y-STR Rst distance (below diagonal) and *P* value (above diagonal)

	Bai	Yi	Mosuo	Naxi	Pumi	Tibetan-YN
Bai		0.003	0.012	0.000	0.000	0.005
Yi	0.048		0.001	0.000	0.000	0.009
Mosuo	0.043	0.076		0.000	0.000	0.101
Naxi	0.260	0.191	0.138		0.003	0.006
Pumi	0.410	0.334	0.238	0.111		0.000
Tibetan-YN	0.065	0.057	0.021	0.084	0.193	

In the PC analysis of mtDNA haplogroups, sub-groups under B, D, F and M7 were combined respectively (fig. 2). The first two PCs explained 76.5%

Table 5 mtDNA HVS1 sequence Fst (below diagonal) and net genetic distance d_A (above diagonal)

		1	2	3	4	5	6	7	8	9	10	11	12	13	14
1	Bai		0.845	0.313	0.451	0.499	0.196	0.264	0.199	0.269	0.349	0.497	0.508	0.188	0.529
2	Mosuo	0.079		0.211	0.779	0.515	0.492	0.441	1.329	0.517	0.504	1.045	0.312	0.687	1.131
3	Naxi	0.031	0.019		0.569	0.353	0.203	0.108	0.630	0.201	0.214	0.572	0.137	0.263	0.572
4	Pumi	0.052	0.071	0.054		0.438	0.305	0.661	0.939	0.680	0.445	0.660	0.927	0.376	0.504
5	Tibetan-YN	0.057	0.048	0.034	0.048		0.273	0.514	1.162	0.481	0.271	0.642	0.540	0.394	0.624
6	Yi	0.022	0.046	0.020	0.033	0.030		0.215	0.702	0.282	0.049	0.250	0.400	0.140	0.579
7	Han-YN	0.032	0.043	0.011	0.073	0.058	0.024		0.486	–0.019	0.145	0.545	–0.012	0.245	0.685
8	Wa	0.028	0.130	0.068	0.117	0.141	0.086	0.064		0.541	0.722	0.976	0.783	0.584	0.965
9	Zhuang	0.035	0.058	0.025	0.082	0.060	0.036	–0.002	0.072		0.217	0.573	0.019	0.229	0.580
10	Tu	0.045	0.050	0.022	0.055	0.034	0.006	0.018	0.105	0.029		0.275	0.242	0.098	0.426
11	Mongolian	0.059	0.085	0.046	0.072	0.070	0.022	0.061	0.147	0.070	0.039		0.766	0.211	0.605
12	Dai	0.062	0.031	0.014	0.104	0.063	0.046	–0.002	0.106	0.003	0.033	0.093		0.380	0.874
13	Uyghur	0.026	0.071	0.030	0.049	0.050	0.018	0.032	0.086	0.031	0.014	0.029	0.051		0.195
14	Kazak	0.069	0.108	0.059	0.063	0.076	0.068	0.084	0.141	0.075	0.062	0.089	0.111	0.029	

Note: Underlined Fst $P > 0.05$

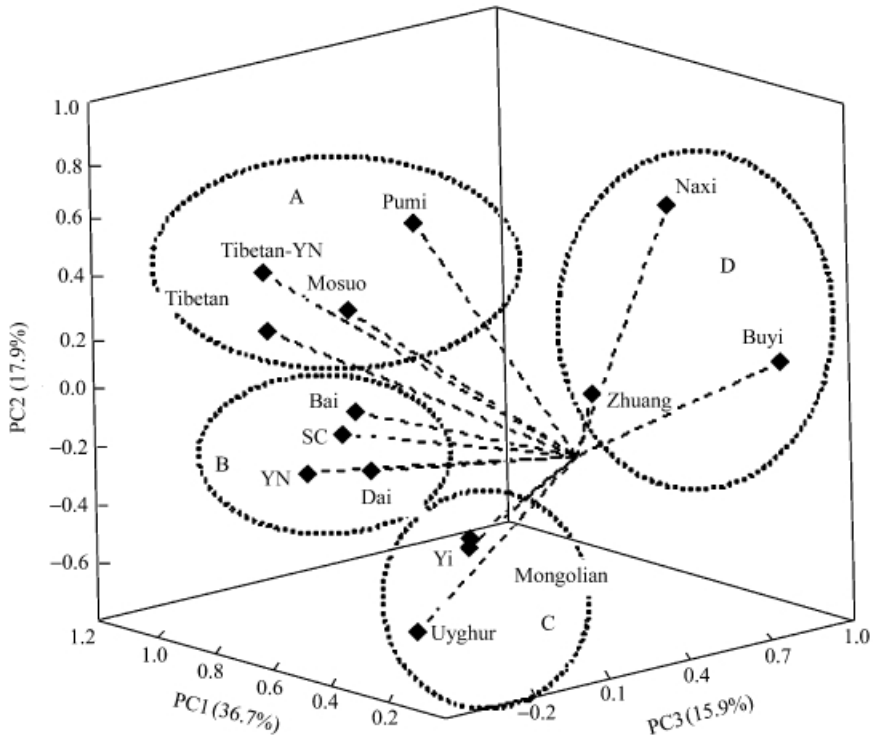

Fig. 1. YSNP haplotype PC plot. YN= Han Yunnan; SC= Han Sichuan.

of the total variation. Two clusters were observed: the northern cluster (N) which encompasses the Yunnan Tibetan, Pumi, Yi and two northern Han populations (Qingdao, Xinjiang), whereas Naxi, Bai and two southern Han populations (Yunnan and Guangdong) were located in the southern cluster (S). The PC1 is significantly correlated with the frequency of Haplogroup F ($r = 0.799$, $p < 0.01$) and B ($r = 0.654$, $p < 0.05$), respectively, and the PC2 shows significant correlation with Haplogroup D ($r = 0.721$, $p < 0.05$) strong

negative correlation with Haplogroup F ($r = -0.876$, $p < 0.01$). This indicates that Haplogroup B, D and F attribute to the difference between the N and S clusters. Interestingly, the Mosuo lies between theN and S clusters, and its PC1 and PC2 are closer to the S and N cluster, respectively. Furthermore, we conducted PC analysis using the Fst of mtDNA (fig. 3) and the result is very similar to that observed in PC analysis using haplogroup frequencies. The S and N clusters are separated by PC2 with the Mosuo lying between the two.

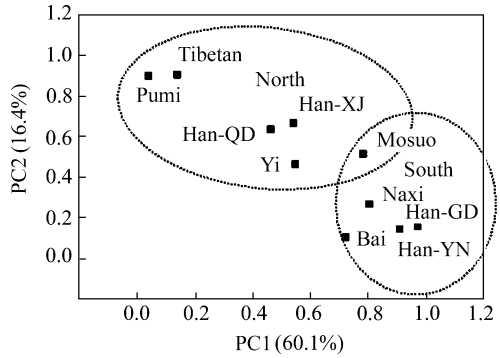

Fig. 2. PC plot conducted by mtDNA haplotype frequency.

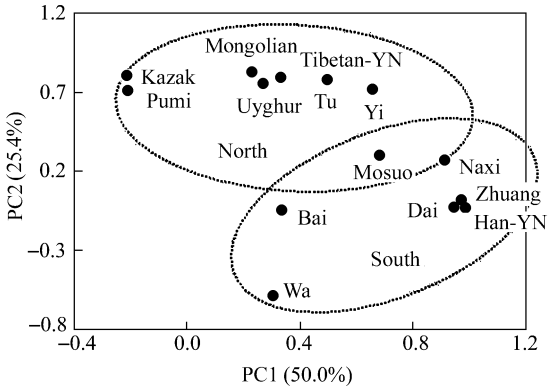

Fig. 3. PC plot based on mtDNA Fst distance matrix.

2.4 Phylogenetic analysis

The phylogenetic tree (NJ tree) and MDS plot constructed by Y-STR Rst matrix are presented in fig. 4, both reveal a similar structure with the Naxi and Pumi forming one cluster, and the Mosuo, Yunnan Tibetan, Yi and Bai forming another. The Mosuo showed a closer distance with the Yunnan Tibetan supporting the observation based on genetic distance.

Similarly, the phylogeny based on mtDNA Fst matrix is consistent with the result based on PC analysis (fig. 5). Populations can be grouped into two clusters. The northern cluster contains the Tu, Uyghur, Kazak, Yi, Pumi and Yunnan Tibetan, whereas the others are in the southern cluster. The Mosuo showed the closest relationship with the Naxi in the southern cluster.

3 Discussion

Based on Y SNPs, the major Y chromosome haplotypes in the Mosuo are H2, H5 and H8. The Alu insertion, i.e. YAP (H2 and H3), was believed to be derived from Central Asia while H8 is a typical East Asian haplotype[15]. The significant correlation between the Y-SNP haplotype frequency distribution of the Mosuo and that of the Yunnan Tibetan ($r = 0.642$, $p<0.05$) indicates a shared genetic history of their paternal lineages. H11 and H9 are widely distributed in southern East Asia populations, but very rare in the north, therefore, their presence marks the characteristics of southern populations. Only 8.5% of H9 was observed in the Mosuo, and H11 is completely absent, indicating a lack of contribution of southern Y chromosome lineages in the Mosuo. Contrarily, the Naxi carry high frequency of H11 (47.5%) which attributes primarily to the different paternal genetic structures between the Mosuo and Naxi, as supported by both PC and phylogenetic analyses.

For mtDNA representing the maternal lineages, Haplogroup B (30.4%), F (21.7%) and D (21.7%) are the most frequent in the Mosuo. The frequency distribution of Haplogroup B and F showed a south to north decline in East Asia, implying that high frequencies of B and F are the characteristics of southern populations[26,31]. The frequencies of these two haplogroups in the Mosuo and Naxi are very similar, and the distributions of all haplogroups are significantly correlated in these two populations ($r = 0.827$, $p< 0.01$), implying significant similarity and therefore shared genetic history of their maternal lineages. The frequency distribution of Haplogroup D among popu-

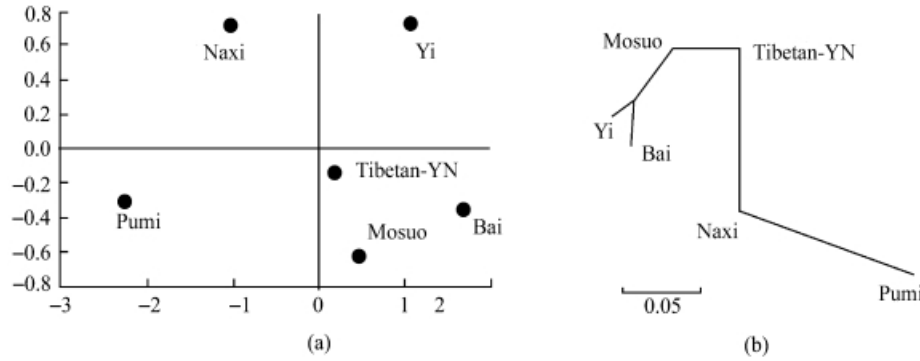

Fig. 4. (a) MDS plot; (b) NJ tree based on YSTR R_{ST} distance.

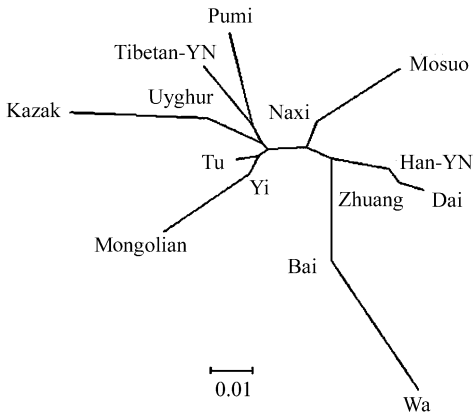

Fig. 5. NJ tree based on mtDNA Fst distance matrix.

lations is significantly negatively correlated with that of Haplogroup F ($r = –0.785$, $p < 0.05$), and a decrease of frequency from northern to southern Han was observed (see table 4 in ref. [26]). This observation suggests that high frequency of haplogroup D might be one of the characteristics of the northern populations. The frequency distribution pattern of mtDNA in the Mosuo indicated a dual influence of southern and northern lineages in the mtDNA gene pool of the Mosuo, which was also shown by both PC analysis and phylogenetic tree.

To synthesize the observation made on both paternal and maternal lineages, we conclude that the paternal lineage of the Mosuo shares a stronger genetic affinity with the Yunnan Tibetan with limited presence of the southern lineages, whereas the maternal lineages of the population share a stronger genetic affinity with Naxi with primarily southern lineages and appreciable amount of northern lineages. There is distinct difference in the genetic characteristics, therefore, genetic history of the paternal and maternal lineages of the Mosuo. Neither the Naxi nor the Yunnan Tibetan shows sufficient genetic similarity with the complete spectrum of the genetic characteristics of the Mosuo.

Haplogroup (haplotype) assignment is essentially a way to classify mtDNA or Y chromosome types based on their respective genetic affinities, and its reliability depends on the consistency between the criteria and true phylogenetic relationship of the haplotypes. To further confirm the results revealed by haplogroup (haplotype) analysis, we conducted PC and phylogenetic analyses directly using the haplotypes of Y-STR and HVS1. And the results are perfectly consistent with those obtained by haplogroup analysis. With the confirmatory results based on different types of genetic markers and analysis methods, the reliability of our conclusion is therefore strongly enhanced.

The observation of the difference between maternal and paternal genetic history in human populations were made earlier[32,33], but the significant differential found in the Mosuo is remarkable. Sex-specific migration pattern is the most likely reason for this. According to historic records, the Tibetan-Burman languages speaking populations in Yunnan Province, including the Mosuo, were derived from the ancient Di-Qiang tribal group originally lived in northwestern China, and redistributed after the long

journey into Yunnan began some 2700 years ago[34]. Different waves and groups of the migrations and gene flow between the populations and between the migrants and early settlers likely attributed to the complex pattern of the genetic diversity in the present-day Tibetan-Burman languages speaking populations. The Mosuo and Naxi shared the same clan name in history records (Mosha), and both were derived from the ancient Maoniu Qiang. Their geographic regions were by large overlapped during most periods of their settlements[35]. Furthermore, they all speak Naxi though in different dialects. The similarity of their maternal lineages is consistent with the shared language and history. And the southern mtDNA lineages in their gene pools might have been introduced during the earlier settlement of Mosha when interaction with original settlers in the area. The similarity of paternal genetic structure between the Mosuo and Yunnan Tibetan was probably due to the interplay of two factors: shared genetic history and gene flow. The Mosuo and Tibetan were originated from different branches of ancient Di-Qiang, the latter was descendent of Fa-Qiang[34]. And their languages belong to Lolo-Burmese and Himalayish sub-families, respectively. Even though the Mosuo and Tibetan showed close genetic relationship with their paternal lineages, historic and linguistic evidence does not support the notion that the age of common ancestor of the Mosuo and Tibetan was younger than that of the Mosuo and Naxi.

The Mosuo is the only matriarchal population in China, and still reserves the unique visiting-marriage system, in which a man goes to his lover's house in the evening, and returns to his mother's home at dawn. According to a survey in 1998, about 75% of Mosuos still practiced this marriage pattern[4]. This social structure led to an increased contribution of the males compared with that observed in other matriarchal and matrilocal communities, and thus increased the complexity of paternal genetic structure. Little has been known about the exact history of the visiting marriage system, but some thought that it was a tradition left from ancient Di-Qiang tribes[3]. Thus it is likely that this system has been practiced for a long time. The culture of Tibetan has made substantial impact on the Mosuo culture, and the Lamaism, which was introduced from the Tibetan, has been the dominant religion in the Mosuo society[36]. And visiting marriages between Lamas with Mosuo women have been practiced as a religious ceremony[4]. The paternal genetic structure might become closer to that of Tibetan with the influx of male lineages, though limited from Tibetan. In summary, gene flow from the other ethnic groups might be the leading cause that enhanced the complexity of the genetic structure in the Mosuo, along with the southern mtDNA lineages derived from the early admixtures, and more recent influx of the paternal lineages.

Studies of human populations should be based on multidisciplinary approaches, and genetic studies only provide one of many aspects therefore bear limited importance in elucidating the ethnicity of the population. A complete picture of the history of a population can only be obtained with the synthesis of the knowledge learnt from history, linguistics, ethnology, anthropology, archaeology and genetics. The aim of this study is to provide genetic evidence on the genetic history of the Mosuo, nothing more nothing less.

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