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ORIGINAL ARTICLE

Genetic imprint of the Mongol: signal from phylogeographic analysis of mitochondrial DNA

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Abstract Mitochondrial deoxyribonucleic acid (DNA) from 201 unrelated Mongolian individuals in the three different regions was analyzed. The Mongolians took the dominant East Asian-specific haplogroups, and some European-prevalent haplogroups were detected. The East Asians-specific haplogroups distributed from east to west in decreasing frequencies, and the European-specific haplogroups distributed conversely. These genetic data suggest that the Mongolian empire played an important role in the maternal genetic admixture across Mongolians and even Central Asian populations, whereas the Silk Road might have contributed little in the admixture between the East Asians and the Europeans.

Keywords Mitochondrial DNA · Haplotype · Mongolia

Introduction

The phylogenetic analysis of mitochondrial deoxyribonucleic acid (DNA) (mtDNA) have revealed a regional clustering of continent habitation (Herrnstadt et al. 2002) and revealed that humans had a recent African origin and migrated through the Eurasian continent (Cann et al. 1987). The mtDNA distribution of the populations who lived in the intermediate region is crucial for elucidating migrations and genetic admixture. Central Asia is located between East Asia and Europe, and there is debate regarding the origin of East Asians (Shi et al. 2005). The genetic distribution of the East Asian and European populations has specific features, such as the distribution of the mtDNA: A, B, D, G, C, F, and M7 are the main mtDNA haplogroups of East Asian populations (Ballinger et al. 1992; Yao et al. 2002), and HV, TJ, IK, U, W, and X are the European-specific haplogroups (Torroni et al. 1996). The mtDNA data from East Asians (Kivisild et al. 2002; Kong et al. 2003b) and Europeans (Ingman et al. 2000) could be a effective tool to reconstruct the origin and migration in the populations of the two continents. There was extensive genetic admixture of eastern Asians and western Eurasians in Central Asia where the Silk Road crossed between the two areas (David et al. 1998; Comas et al. 1998). Xinjiang Province in northwestern China is an important part of the Silk Road, where extensive genetic admixture was observed in local populations (Yao et al. 2004). European-specific mtDNA haplogroups were identified in all five major Xinjiang populations, and 14% of these haplogroups were found in the Xinjiang Mongolians but only 2% in the Mongolians who lived in the east part of plateau of Mongolia (Kong et al. 2003b). European-specific mtDNA haplogroups were also identified in the populations of northeast Mongolia far away the Silk Road (Kolman et al. 1996). The genetic characters of the Mongolians were important data to elucidate the origin and the late admixture history of the populations in the plateau of Mongolia.

The Mongolian empire had a great impact on the history of Europe and Central Asia and could represent an important

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role in the genetic admixture. An increasing frequency of European-specific haplogroups in the upper three regional Mongolian populations from northeastern China to Central Asia has been observed. However, no or fewer European-specific haplogroups have been found in the Han and Tibetan populations, who also live along the Silk Road (Yao et al. 2002; Qian et al. 2001). There are now nearly 8.2 million Mongolians living primarily in Mongolia, Inner Mongolia of China, and the Siberia region of Russia. Most Mongolians live outside the region of the Silk Road.

There is a Mongolian population living in Tonghai County of Yunnan Province in southwestern China. Ancestors of Tonghai Mongolians migrating to extant inhabitation at the beginning of the Mongolian empire would have carried the original genetic components of Mongolians (Du et al. 1993). These Mongolians then formed a relatively independent Mongolian group (You 1997). In this study, mtDNA haplotypes from 201 Mongolian samples were analyzed. Phylogenetic relationships among the Mongolian populations from the central plateau of Mongolia (Inner Mongolians) and Tonghai County were revealed. The genetic impact of the Mongolian empire was detected by comparing the original Mongolians with populations in the regions where the empire had explored.

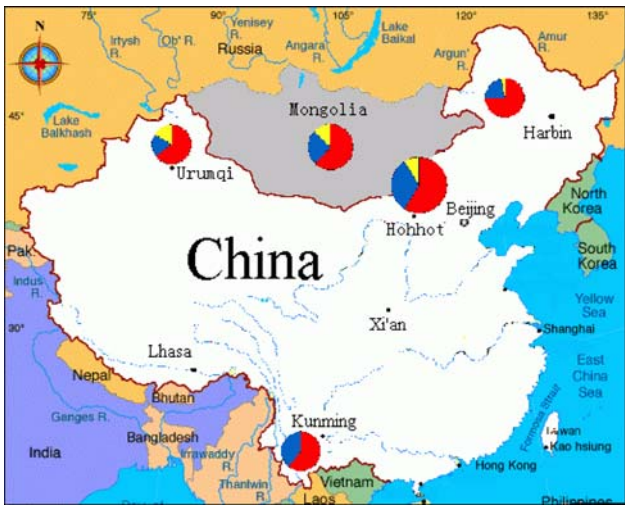

Fig. 1 Geographic location of the Mongolian samples in this study and frequency distribution of different haplogroups: *red* M haplogroups, *blue* N haplogroups, and *yellow* west-Eurasian haplogroups. Zhenglanqi is located very near Hohhot, so the two sets of data were combined

Materials and methods

Sampling

A total 201 unrelated Mongolians individuals were analyzed: 155 from central Inner Mongolia of China (107 from Hohhot City and 48 from Zhenglanqi County), and 46 from Tonghai County in Yunnan Province of China. All individuals were able to confirm their maternal origin and gave informed consent.

Previously published Mongolian mtDNA data was used for comparison. These data include 103 mtDNA from Mongolia with HVS-I and eight additional coding-region information (Kolman et al. 1996); 50 mtDNA from Xingjing Province with HVS-I and 9-bp-deletion data (Yao et al. 2004); 48 mtDNA from Xin Barage Zuoqi County in the eastern part of Inner Mongolia of China with HVS-I and HVS-II, and an additional one or two coding-regions (Kong et al. 2003b) (Fig. 1).

Genotyping of mtDNA polymorphisms

Genomic DNA was extracted by standard phenol/chloroform methods. The control region 15926-619 and four parts of the coding regions containing 2796-5441, 5126-7701, 8215-10735, and 14054-16048 were amplified using five pairs of primers, L15926/H619, L2796/H5441, L5126/H7701, L8215/H10735, and L14054/H16048, respectively. Polymerase chain reaction (PCR) was performed in a 15-μl reaction mixture that contained 20 ng DNA, reaction buffer [10 mM Tris hydrochloride (Tris-HCl), pH 8.3, 1.5 mM magnesium chloride ($MgCl_2$), 50 mM potassium chloride (KCl), 0.001% gelatin, and 0.5 μl 1 mM of each primer, 200 μM deoxynucleotide triphosphate (dNTPs), and 0.2 U *Taq* polymerase (Promega). The PCR amplification was performed in either a PE-9600 or PE-9700 (ABI Applied Biosystems, Fostor City, CA, USA) with 95°C for 2 min followed by 30 cycles of 95°C for 30 s, 58°C for 30 s, 72°C for 30 s, then 72°C 2 min. Each PCR product was diluted by adding 60 μl of water. Using upper PCR product as a template, the control region and parts of the coding regions containing 3010-3430, 4640-5204, 10170-10660, and 14478-15086 were amplified and sequenced, respectively, by six pairs of nested primers: L15996/H107, L29/H590, L3010/H3430, L4640/H5204, L10170/H10660, and L14478/H15086. Another six pairs of confirming primers, L3953/H4508, L5126/H5441, L8215/H8297, L9766/H1063, L14989/H15400, and L15391/H16048, were used when needed to unambiguously define a sample. All the primer sequences were cited from Yao et al. (2002) or designed by the primer 3 software (http://www-genome.wi.mit.edu/genome_software/other/primer3.html) (Table 1).

The nested or second PCR products were examined on a 2% agarose gel then purified with the Wizard Purification Kit (Promega, Madison, WI, USA). Each fragment was sequenced for both strands with the BigDye and Dye Terminator Cycle Sequence Kit (ABI Applied Biosystems), run on an ABI 377 DNA sequencer (ABI Applied Biosystems),

Table 1 Primers for first and nested polymerase chain reaction (PCR) and sequencing

Primer pair	Locations in CRS	Informative sites	Polymorphisms at
L15926/H619	15898-15926/639-619		
L2796/H5441	2777-2796/5463-5441		
L5126/H7701	5106-5126/7721-7701		
L8215/H10735	8196-8215/10755-10735		
L14054/H16048	14035-14054/16067-16048		
L15996/H107	15975-15996/127-107	A, B, C, Z, I, K, J, U, R9, R11	HVS I, 16519
L29/H590	8-29/619-599	M, J, M11, B4b	HVS II, 489, 499
L3010/H3430	2990-3010/3451-3430	D4, M10, D4a, M9	3010, 3172+C, 3204, 3394
4640/5204	4620-4640/5224-5204	M8, A, G, D,	4715, 4824, 4833, 4883, 5178A
10171/10659	10147-10170/10679-10660	M, F, D5, F1, F2	10400, 10310, 10397, 10609, 10535, 10586
14478/15086	14458-14478/15105-15086	M, G, D4, M10, HV	14569, 14668, 14766, 14783, 15040, 15043
L3953/H4508	3934-3953/4528-4508	R9, M9, M7bc	3970, 4071, 4491
L5126/H5441	5106-5126/5461-5441	N9	5417
L8215/H8297	8196-8215/8316-8297	B	8218-8289d
L9766/H10163	9746-9766/10181-10163	M7	9824
L14989/H15400	14969-14989/15420-15400	C4, B4b, B4d, B4c	15204, 15535, 15346,
L15391/H16048	15372-15391/16067-16048	M8, B4d	15487T, 15930

CRS Cambridge Reference Sequence

and scored using Sequence Analysis and Sequence Navigator software (Perkin–Elmer).

The sequences were analyzed by the DNA-STAR software (DNASTAR, Inc.) and compared with the Revised Cambridge Reference Sequence (rCRS) (Andrews et al. 1999). We also rechecked all polymorphisms in the Chromas 1.45 software (Griffith University, Australia). We adopted the established phylogeny of Europeans and East Asians proposed by Finnilä et al. (2001), Kivisild et al. (2002), and Kong et al. (2003a). To show genetic relationships among Mongolians in different regions, principal component analysis (PC) was conducted by haplogroup frequency using SPSS12.0 software (SPSS Inc.). Arlequin software (http://anthro.unigo.ch/arlequin) was used to calculate neutrality tests of Tonghai Mongolians (Tajima 1996) and genetic distance (F_{ST} values) of regional Mongolians and other East Asian populations (Schneider et al. 2000). Neighbor-joining (NJ) trees based on F_{ST} values were constructed in MEGA software (http://www.megasoftware.net) (Nei 1987; Kumar et al. 2001).

Result

Among all 201 northern and southern Mongolian samples, 111 samples had the 489T → C, 10400 C → T, 14783 T → C, and 15043 G → A mutations attributed to macrohaplogroup M. These samples were further subcladed to haplogroups D, G, M*, and M7–M11 according to the information in the D-loop region and encoding region. The other 90 samples did not have the above-mentioned four mutations and belonged haplogroups A, N9, F, R9b, R11, HV, K, J, or I, which belong to macrohaplogroup N.

Four parallel mutation sites were found. Deletions were detected at 16227 in four G2a haplogroup samples, 16224 in all five Z haplogroup samples, 16316 in one M9a haplogroup sample, and no 73 G → A were observed in two H haplogroup samples. Table 2 shows the distribution of haplogroup frequencies in norther Inner Mongolians and southern Tonghai Mongolians. Based on M and N macrohaplogroups, 23 haplogroups and 52 subhaplotypes were defined, including nearly all East-Asian-specific haplogroups, HV, J, K, U, and I, European-specific haplogroups (Finnilä et al. 2001), and the classification tree of the mtDNA haplogroups in Mongolians shown in Fig. 2. For Inner Mongolians, there were 39, 18, and 13 individuals belonging to haplotypes D, A, and C, respectively. The D haplotype had the highest frequency (25%), followed by A (12%) and C (9%). The combined frequency of these three haplogroups was more than 46%. F, B, and M7 haplogroups were also present, with relatively high frequencies of F (13.5%), B (7.7%), and M (76.5%), respectively. The European-specific haplogroups K, J, and I were first found in Mongolians. The combined frequencies of European-specific haplogroups K, J, I, H, and U) was 8.4%. In contrast, no European-specific haplogroup was found in Tonghai Mongolians; all 46 individuals were assigned into the East-Asian-specific haplogroups. We found 22%, 17%, and 9% of individuals belonged to haplogroups A, D, and C, respectively. The

Table 2 Estimated frequencies (%) of mitochondrial deoxyribonucleic acid (mtDNA) haplogroups in regional Mongolians

Hap	InMg (n = 155)	ThMg (n = 46)	Mg (n = 103)	XBZQMg (n = 48)	XjMg (n = 50)
M7b1	1.94	2.17	0.00	0.00	0.00
M7b	1.94	0.00	0.00	0.00	0.00
M7b2	0.65	0.00	0.00	0.00	0.00
M7c	1.94	0.00	0.00	0.00	2.00
M7	0.00	0.00	2.91	2.08	0.00
C4a	1.29	0.00	0.00	0.00	0.00
C4	4.52	2.17	0.00	0.00	0.00
C	2.58	6.52	13.59	6.25	10.00
CZ	0.65	0.00	0.00	0.00	0.00
Z	0.65	8.70	3.88	4.17	0.00
M8a	2.58	4.35	0.97	0.00	0.00
M9a	1.29	0.00	0.97	4.17	0.00
M9	0.00	0.00	0.00	0.00	2.00
M10	1.29	0.00	0.00	2.08	0.00
M11	1.94	4.35	0.97	0.00	0.00
M*	0.65	2.17	4.85	0.00	4.00
G1a	1.29	0.00	0.00	0.00	0.00
G1	0.00	2.17	0.00	0.00	0.00
G2	1.29	4.35	0.00	4.17	6.00
G2a	1.29	0.00	0.00	0.00	6.00
G3	1.29	0.00	0.00	0.00	0.00
G	0.00	2.17	5.83	0.00	0.00
D5a	3.23	2.17	0.00	2.08	0.00
D5	1.29	2.17	5.83	4.17	2.00
D4a	3.87	2.17	0.00	0.00	0.00
D4b	0.65	0.00	0.00	0.00	0.00
D4	16.13	10.87	18.45	0.00	0.00
D	0.00	0.00	0.00	33.33	22.00
A	11.61	6.52	4.85	8.33	8.00
HV	3.23	0.00	4.85	0.00	6.00
N9a	1.94	0.00	1.94	6.25	2.00
F1a	3.87	4.35	0.00	4.17	6.00
F1b	5.81	0.00	0.00	0.00	6.00
F1c	0.00	4.35	0.00	2.08	0.00
F2a	1.29	0.00	0.00	2.08	0.00
F2	1.29	0.00	0.00	0.00	0.00
F	1.29	0.00	8.74	0.00	8.00
R9a	0.00	2.17	0.00	0.00	0.00
R9b	0.65	4.35	0.00	2.08	0.00
R9	0.65	6.52	0.00	0.00	0.00
R11	1.29	0.00	3.88	0.00	0.00
B4a	0.00	0.00	0.00	2.08	0.00
B4b1	1.29	0.00	0.00	6.25	2.00
B4c	1.29	0.00	0.00	0.00	0.00
B4d	2.58	8.70	0.00	0.00	0.00
B4	0.65	4.35	0.00	0.00	0.00
B5a	0.65	0.00	0.00	2.08	0.00
B5b	1.29	0.00	0.00	0.00	0.00

Table 2 continued

Hap	InMg (n = 155)	ThMg (n = 46)	Mg (n = 103)	XBZQMg (n = 48)	XjMg (n = 50)
B	0.00	0.00	10.68	0.00	0.00
Y	0.00	0.00	0.97	0.00	0.00
K	1.94	0.00	0.97	0.00	0.00
J1b	1.29	0.00	0.00	0.00	0.00
J	0.65	0.00	0.00	0.00	0.00
I	0.65	0.00	0.00	0.00	0.00
T	0.00	0.00	1.94	2.08	0.00
U	0.65	0.00	2.91	0.00	8.00
N*	0.00	2.17	0.00	0.00	0.00
Haplotype diversity	0.9426 ± 0.0007	0.9428 ± 0.0012	0.9087 ± 0.0011	0.8580 ± 0.0057	0.9132 ± 0.0013

InMg central Inner Mongolia of China, *ThMg* Tonghai County in Yunnan Province, China. *Mg* Mongolia (Kolman et al. 1996), *XBZQMg* Xin Barage Zuoqi, eastern part of Inner Mongolia of China (Kong et al.2003b), *XjMg* Xingjiang province, western part of China (Yao et al. 2004)

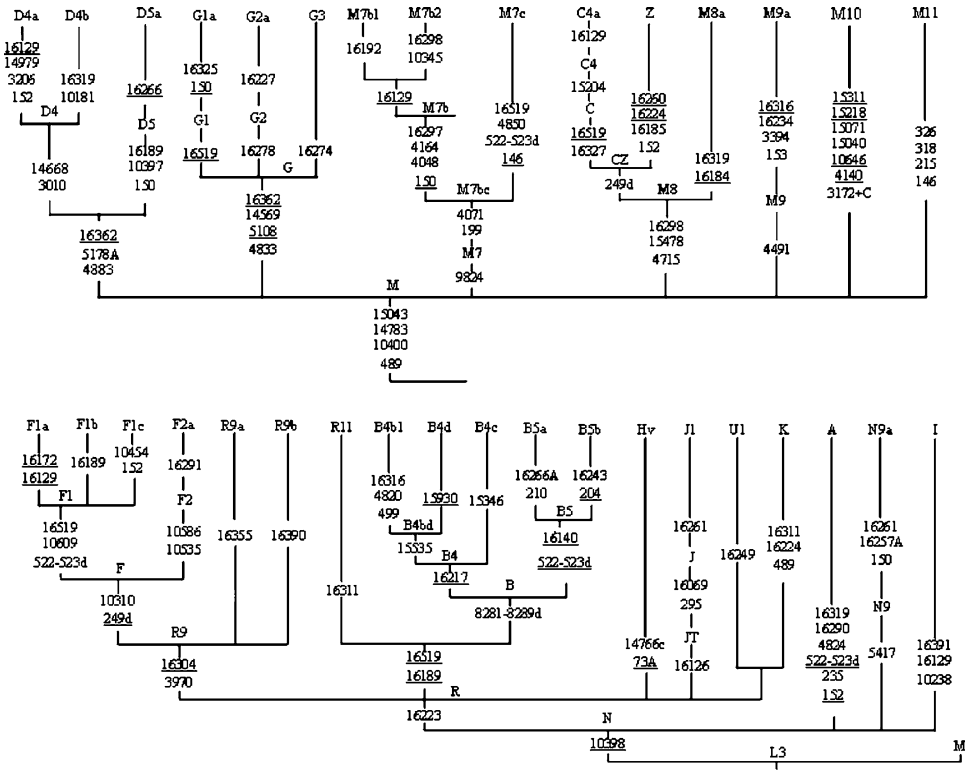

Fig. 2 Classification tree of mitochondrial deoxyribonucleic acid (mtDNA) haplogroups in Mongolians. The diagnostic mutations were compared to the Revised Cambridge Reference Sequence (CRS) and indicated on the branches: recurrent mutation is *underlined*, and *d* indicates deletion

combined frequency of these three haplogroups was nearly 48%. Haplogroup R9b had the third highest frequency (13%) in Tonghai Mongolians (Table 2). There was no significant difference in the mtDNA haplogroup frequency distribution between Mongolians from Tonghai and those from central Inner Mongolia of China. However, Fisher's exact test showed that the mtDNA haplogroup frequency distribution between Mongolians from Tonghai was significantly different from that of Mongolians in Mongolia (Kolman et al. 1996), in Xin Barage Zuoqi in the eastern part of Inner Mongolia of China (Kong et al. 2003b), and in Xingjiang Province in the western part of China (Yao et al. 2004) (Table 2).

The basal mtDNA haplogroup profiles of the five Mongolian populations were treated as input vectors for PC analyses. Figure 3a displays the PC map for the first two principal components, of which the first principal component accounts for 79% of the total variation. Southern Tonghai Mongolians and the other four northern Mongolians are separated by PC2 (accounting for 10% of the total

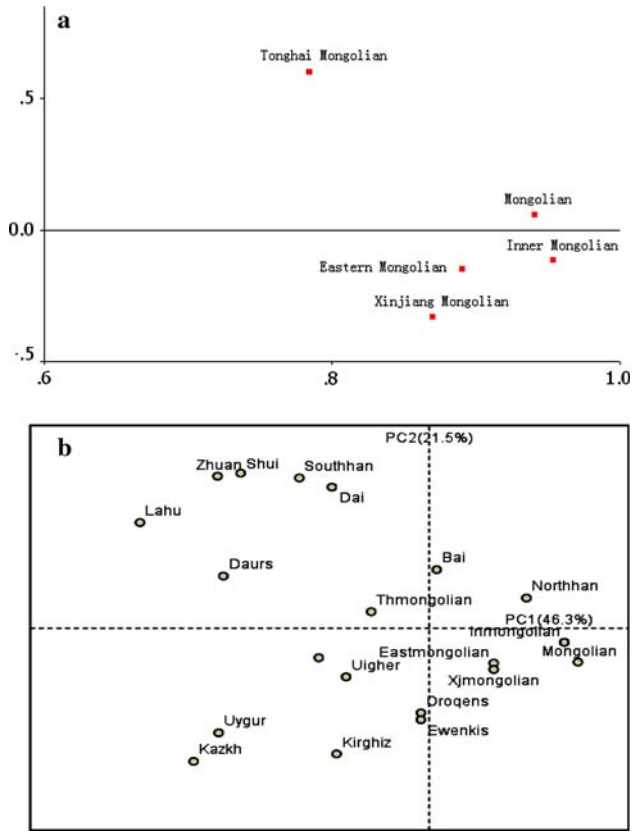

Fig. 3 **a** Principal component (PC) maps of mitochondrial deoxyribonucleic acid (mtDNA) data (with respect to the basal haplogroup profiles) of five regional Mongolians. **b** PC maps of mtDNA data of five regional Mongolians and 14 East-Asian populations. The mtDNA data of 14 different populations of eastern Asian. Kazkh, Uigher, Kirghiz, and Uygur are from the Central Asian regions (Yao et al. 2004); Daurs, Oroqens, Ewenkis, and Koreans come from northern East Asia (NEA) (Kong et al. 2003b); Dai, Shui, Zhaun, Bai, and and Lahu are from southern East Asia (SEA) (Qian et al. 2001; Yao and Zhang 2002); Northhan and Southhan divided by the Yangzi River are from China (Yao et al. 2002)

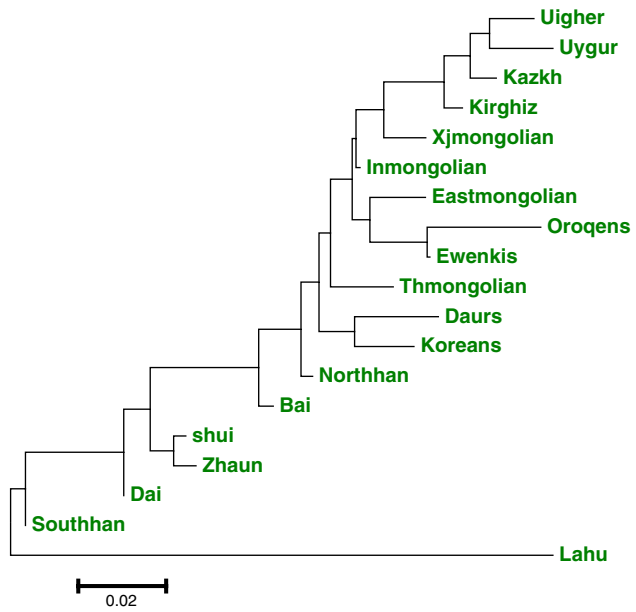

Fig. 4 Unrooted neighbor-joining (NJ) tree based on F_{st} of four regional Mongolians and 14 East Asian populations

variation). Figure 3b presents PC results conducted in regional Mongolians and other East- and Central-Asian populations. Northern East Asians, southern East Asians, and Central Asians are clearly separated by PC2 (accounting for 21.4% of the total variation). Tonghain, Inner Mongolian, and Mongolian samples fall entirely into the northern East Asian group; Xinjing and eastern Inner Mongolia samples cluster together.

The *P* value of neutrality tests in Tonghai Mongolians was 0.109. The haplotype diversity was 0.9428 in Tonghai Mongolians, 0.9426 in Mongolians from central Inner Mongolia of China, 0.9087 in Mongolians from Mongolia (Kolman et al. 1996), 0.8580 in Mongolians from Xin Barage Zuoqi, eastern part of Inner Mongolia of China (Kong et al. 2003b), and 0.9132 in Mongolians from Xingjiang Province, western part of China (Yao et al. 2004) (Table 2). An unrooted NJ tree was constructed among regional Mongolians and East Asian populations using F_{ST} distances. Since Kolman et al. (1996) only named A, B, C, and D haplogroups in Mongolian samples, we thus excluded them from the phylogenetic analysis. Fig. 4, is largely divided into two different clusters, and all four regional Mongolians grouped with the northern East Asian (NEA) cluster (Fig. 4).

Discussion

The Mongolians have four dominant haplogroups (A, D, G, and M8), including those from the regions of the Mongolia plateau or adjacent region (Yao et al. 2004), central Inner Mongolian and Monglia (Kolman et al. 1996) and eastern Xin Barage Zuoqi (Kong et al. 2003b). Those haplogroups are prevalent in the NEAs, and the prevalent southern East Asians (SEAs) haplogroups are B, M7, R9, and N9a. The European-prevalent haplogroups are HV, U, K, I, and J (Kivisild et al. 2002; Yao et al. 2002; Kong et al. 2003b). The main maternal components of these Mongolians belong to NEA-specific haplogroups A, D, G, and M8, which occupy more than 54% in eastern Xin Barage Zuoqi County samples, nearly 45% in central Inner Mongolian and Mongolian samples, and 40% in western Xingjang Mongolian samples, showing decreasing frequencies from east to west. SEA-prevalent haplogroup is nearly 28% in Inner Mongolians, 22% in eastern Xin Barage Zuoqi County, and Mongolian samples and only 16% in Xingjing Mongolians. The European-prevalent haplogroups (HV, U, K, I, J) are 14% in western Xingjang Mongolian, 10% in

Monglia, 8.4% in central Inner Mongolian samples, and only 2% in eastern Xin Barage Zuoqi County samples, showing decreasing frequencies from west to east. The NEA-specific haplogroups distributed in reverse to the European-prevalent haplogroups. The European-specific haplogroups H and U are prevalent in Mongolians, and K, J, and I haplogroups are first detected in Inner Mongolians. Mongolian genetics had a northern East-Asian orientation and admixed with the Southern East Asians. The genetic studies indicate the East Asians had single south origin, and there the early human migration through central East after modern humans came out of Africa did not occur (Shi et al. 2005). European-specific haplogroups K, J, and I indicate a late Mongolian admixture with Europeans. The Mongolian plateau lies in the heart of east Central Asia, where modern humans inhabited 20,000–25,000 years ago (Fiedel et al. 1992). Mongolians living in the Mongolia plateau were separated from Europe by Central Asia, so there must have been certain factors promoting the genetic admixture.

Mitochondrial DNA data indicated that northern East Asians were the ancestors of the Native Americans who migrated about 7,000–9,000 years ago through the Bering strait (http://www.mitomap.org/). Mitochondrial DNA analysis of the 18 Asian and 16 American groups indicate that the Mongolians were the ancestor of Native Americans, and whose mtDNA haplogroups are A, B, C, and D (Kolman et al. 1996). All studies suggested that the Mongolians did not contain the European lineage 7,000–9,000 years ago.

Mongolians migrated south to TongHai in Yunnan Province about 800 years ago (Du et al. 1993). Historically, the Mongolian army was accompanied by their wives at the beginning of the war during the Mongolian empire, and later some of them married local people (You 1997; Hu 2002). Comparison of the 14 different populations in several regions of East Asia (Yao and Zhang 2002; Yao et al. 2002, 2004; Kong et al. 2003b; Qian et al. 2001) revealed that TongHai Mongolians are close to northern Mongolians genetically and belong to the NEAs (Figs. 3b, 4). Our recent study indicated that all Mongolians clustered together in Y-STR data, including the TongHai Mongolians (our unpublished data). The extant TongHai Mongolians take the north-prevalent haplogroups (51.5%), and no European-specific haplotypes were observed. The neutrality tests showed no selective effect ($P \geq 0.05$), and there was no population expansion and bottleneck. The study also indicated that Mongolians who migrated south did not contain European-specific mtDNA haplotypes and in Native Americans—the descendants of original Mongolians—the same pattern appeared (Kolman et al. 1996).

The European haplogroups were detected in Mongolians who inhabited Central Asia, such as Xingjing Autonomous region, and central Inner Mongolian. The Silk Road (second century BC to the thirteenth century AD) would have brought the European-specific genetics into those regions. However, few European-specific haplogroups have been found in Han and Tibetan populations who lived along the Silk Road (Yao et al. 2002; Qian et al. 2001), which supports the hypothesis that the Silk Road contributed little genetic admixture between the Europeans and East Asians. In history, the Parthian empire prevented any mass migration of strange people, usually only allowing caravans of traders to pass, so population migration freely between Europe and East Asia could not occur (Dubs 1957). Thus, the genetic admixture of Europeans with northern East Asians in Central Asian can only be explained by the later conquer of the Mongolian empire (1271–1368 AD). The Mongolian populations played an important role in promoting the historical development of Eurasia. Genghis Khan (1162–1227) and his descendants waged three wars in the west and established the united multiethnic Mongolian empire crossing Eurasia, which had 3.5 million km^2 and a 100 million people. The Mongolians continued to dominate the region north of the Great Wall of China and west to the Aral Sea for two more centuries after the Mongolian empire ended in 1368. Then, some Mongolian descendants moved back to their hometowns from thousands of kilometers away. Those who migrated back carried some exotic genetic components with them. During their wars, the Mongolian conquerers incorporated some ethnic populations into their groups, and those admixtures expanded the Mongolian gene pool. The European-specific mtDNA haplogroups could be detected in different northern Mongolian populations.

Y-chromosome studies support the fact that Mongolians have kept East-Asian characteristics (Nasidze et al. 2005; Wells et al. 2001; Xue et al. 2006; Yang et al. 2005). The prevalence of the Mongolian Y chromosome in Central Asians support the inequity admixture due to Genghis Khan's military success (Zerjal et al. 2002, 2003), that is, the Mongolia conqueror contributed the dominant Y chromosome to the descendant admixture populations. This happened in the Indo-Europeans admixture in the caste establishment in India (Bamshad et al. 2001; Thanseem et al. 2006), and similar situations were found among Spanish Conquistadors and British colonialists in North America and Australia (Carvajal-Carmonn et al. 2000). The Y-chromosome data support Mongolian impact in the genetics of Central and northern East Asians, however, Mongolian impact from maternal lineage was not significant (Bamshad et al. 2001; Thanseem et al. 2006).

In summary, the dominant Mongolian mtDNA gene pool belongs to the eastern Asian lineage, which agreed with its origin. The unbalanced genetic contribution of the paternal and maternal in the admixture populations during

conquest by the Mongolian empire indicate the developmental history of Mongolian populations.

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References

Andrews RM, Kubacka I, Chinnery PF, Lightowlers RN, Turnbull DM, Howell N (1999) Reanalysis and revision of the Cambridge reference sequence for human mitochondrial DNA. Nat Genet 23:147

Ballinger SW, Theodore GS, Torroni A, Gan YY, Hodge JA, Hassan K, Chens K-H, Douglas CW (1992) Southeast Asian mitochondrial DNA analysis reveals genetic continuity of ancient Mongoloid migrations. Genetics 130:139–152

Bamshad M, Kivisild T, Watkins WS, Dixon ME, Ricker CE, Rao BB, Naidu JM, Prasad BV, Reddy PG, Rasanayagam A, Papiha SS, Villems R, Redd AJ, Hammer MF, Nguyen SV, Carroll ML, Batzer MA, Jorde LB (2001) Genetic evidence on the origins of Indian caste populations. Genome Res 11:994–1004

Cann RL, Stoneking M, Wilson AC (1987) Mitochondrial DNA and human evolution. Nature 325:31–36

Carvajal-Carmonn LG, Soto ID, Pineda N, Ortíz-Barrientos D, Duque C, Ospina-Duque J, McCarthy M, Montoya P, Alvarez VM, Bedoya G, Ruiz-Linares A (2000) Strong Amerind/white sex bias and a possible Sephardic contribution among the founders of a population in northwest Colombia. Am J Hum Genet 67:1287–1295

Comas D, Calafell F, Mateu E, Perez-Lezaun A, Bosch E, Martinez-Arias R, Clarimon J, Facchini F, Fiori G, Luiselli D (1998) Trading genes along the silk road:mtDNA sequences and the origin of Central Asian populations. Am J Hum Genet 63:1824–1838

David C, Calafell F, Mateu E, Pérez-Lezaun A, Bosch E, Martínez-Arias R, Clarimon J, Facchini F, Fiori G, Luiselli D, Pettener D, Bertranpetit J (1998) Trading genes along the Silk Road: mtDNA sequences and the origin of Central Asian Populations. Am J Hum Genet 63:1824–1838

Dubs HH (1957) A Roman city in Ancient China. Greece and Rome, 2nd Series, vol 4, pp 139–148

Du RF, Xiao CJ, Yip VF (1993) Ethnic groups in China. Science Press, Beijing

Fiedel SJ (1992) Prehistory of the Americas. Cambridge University Press, Cambridge

Finnilä S, Lehtonen MS, Majamaa K (2001) Phylogenetic network for European mtDNA. Am J Hum Genet 68:1475–1484

Fu S, Li P, Yuldasheva N, Ruzibakiev R, Xu J, Shu Q, Du R, Yang H, Hurles ME, Robinson E, Gerelsaikhan T, Dashnyam B, Mehdi SQ, Tyler-Smith C (2003) The genetic legacy of the Mongols. Am J Hum Genet 72:717–725

Herrnstadt C, Elson JL, Fahy E, Preston G, Turnbull DM, Anderson C, Ghosh SS, Olefsky JM, Beal MF, Davis RE, Howell N (2002) Reduced-median-network analysis of complete mitochondrial DNA coding-region sequences for the major African, Asian, and European haplogroups. Am J Hum Genet 70:1152–1171

Hu ZA (2002) General history of Mongolian nationality. Inner University Press, Hohhot

Ingman M, Kaessmann H, Pääbo S, Gyllensten U (2000) Mitochondrial genome variation and the origin of modern humans. Nature 408:708–713

Kivisild T, Tolk H-V, Parik J, Wang Y, Papiha SS, Bandelt H-J, Villems R (2002) The emerging limbs and twigs of the East Asian mtDNA tree. Mol Biol Evol 19:1737–1751

Kolman CJ, Sambuughin N, Bermingham E (1996) Mitochondrial DNA analysis of Mongolian populations and implications for the origin of New World founders. Genetics 142:1321–1334

Kong Q-P, Yao Y-G, Sun C, Bandelt HJ, Zhu CL, Zhang YP (2003a) Phylogeny of East Asian mitochondrial DNA lineages inferred from complete sequences. Am J Hum Genet 73:671–676

Kong Q-P, Yao Y-G, Liu M, Shen SP, Chen C, Zhu CL, Palanichamy MG, Zhang Y-P (2003b) Mitochondrial DNA sequence polymorphisms of five ethnic populations from northern China. Hum Genet 113:391–405

Kumar S, Tamura K, Jakobsen IB, Nei M (2001) MEGA2: molecular evolutionary genetics analysis software. Bioinformatics 17:1244–1245

Nasidze I, Quinque D, Dupanloup I, Cordaux R, Kokshunova L, Stoneking M (2005) Genetic evidence for the Mongolian ancestry of Kalmyks. Am J Phys Anthropol 128:846–854

Nei M (1987) Molecular evolutionary genetics. Columbia University Press, New York

Qian YP, Chu Z-T, Dai Q, Wei C-D, Chu JY, Tajima A, Horai S (2001) Mitochondrial DNA polymorphism in Yunnan nationalities in China. J Hum Genet 46:211–220

Schneider S, Kueffer J-M, Roessli D, Excoffier L (2000) Arlequin: Version 2.000 A software for population genetic analysis. Genetics and Biometry Laboratory. University of Geneva, Geneva

Shi H, Dong YL, Wen B, Xiao CJ, Underhill PA, Shen PD, Chakraborty R, Jin L, Su B (2005) Y-chromosome evidence of southern origin of the East Asian-specific haplogroup O3-M122. Am J Hum Genet 77:408–419

Tajima F (1996) The amount of DNA polymorphism maintained in a finite population when the neutral mutation rate varies among sites. Genetics 143:1457–1465

Thanseem I, Thangaraj K, Chaubey G, Singh VK, Bhaskar LV, Reddy BM, Reddy AG, Singh L (2006) Genetic affinities among the lower castes and tribal groups of India inference from Y chromosome and mitochondrial DNA. BMC Genet 7:42

Torroni A, Huoponen K, Francalacci P, Petrozzi M, Morelli L, Scozzari R, Obinu D, Savontaus ML, Wallace DC (1996) Classification of European mtDNAs from an analysis of three European populations. Genetics 144:1835–1850

Wells RS, Yuldasheva N, Ruzibakiev R, Underhill PA, Evseeva I, Blue-Smith J, Jin L, Su B, Pitchappan R, Shanmugalakshmi S, Balakrishnan K, Read M, Pearson NM, Zerjal T, Webster MT, Zholoshvili I, Jamarjashvili E, Gambarov S, Nikbin B, Dostiev A, Aknazarov O, Zalloua P, Tsoy I, Kitaev M, Mirrakhimov M, Chariev A, Bodmer WF (2001) The Eurasian heartland: a continental perspective on Y-chromosome diversity. Proc Natl Acad Sci USA 98:10244–10249

Xue Y, Zerjal T, Bao W, Zhu S, Shu Q, Xu J, Du R, Fu S, Li P, Hurles ME, Yang H, Tyler-Smith C (2006) Male demography in East Asia: a north–south contrast in human population expansion times. Genetics 172:2431–2439

Yang Z, Dong Y, Gao L, Cheng B, Yang J, Zeng W, Lu J, Su Y, Xiao C (2005) The distribution of Y chromosome haplogroups in the nationalities from Yunnan Province of China. Ann Hum Biol 32:80–87

You Z (1997) History of Yunnan Nationalities. Yunnan University Press, Kunming

Yao Y-G, Zhang Y-P (2002) Phylogeographic analysis of mtDNA variation in four ethnic populations from Yunnan Province: new data and a reappraisal. J Hum Genet 47:311–318

Yao Y-G, Kong Q-P, Bandelt H-J, Kivisild T, Zhang Y-P (2002) Phylogeographic differentiation of mitochondrial DNA in Han Chinese. Am J Hum Genet 70:635–651

Yao Y-G, Kong QP, Wang CY, Zhu CL, Zhang YP (2004) Different matrilineal contributions to genetic structure of ethnic groups in the Silk Road region in China. Mol Biol Evol 21:2265–2280

Zerjal T, Wells RS, Yuldasheva N, Ruzibakiev R, Tyler-Smith C (2002) A genetic landscape reshaped by recent events: Y-chromosomal insights into Central Asia. Am J Hum Genet 71:466–482

Zerjal T, Xue Y, Bertorelle G, Wells RS, Bao W, Zhu S, Qamar R, Ayub Q, Mohyuddin A, Fu S, Li P, Yuldasheva N, Ruzibakiev R, Xu J, Shu Q, Du R, Yang H, Hurles ME, Robinson E, Gerelsaikhan T, Dashnyam B, Mehdi SQ, Tyler-Smith C (2003) The genetic legacy of the Mongols. Am J Hum Genet 72:717–721