



# Y-chromosome lineage in five regional Mongolian populations



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## ARTICLE INFO

### Article history:

Received 24 September 2013

Accepted 2 October 2013

### Keywords:

Y-STR  
Y haplogroup  
Mongolian  
Region  
Frequency  
Network

## ABSTRACT

We analyzed 17 Y-STRs and Y haplogroups (Y-hgs) for about 493 Mongolian male samples from 5 regions in Mongolia, and compared with the results from Japanese male samples. Only 358 Y-STR haplotypes (Y-HTs) were observed in Mongolian males, and the haplotype diversity and discrimination capacity were calculated as 0.9934 and 0.7099 whose values were very low by comparing with Japanese males (0.9989 and 0.9565, respectively). About 63% of Mongolian males had C3 groups ranging from 50% in Ulaanbaatar to 69% in Ulaangom. The 96% of C3 was C3\*, which is C3 except C3a to C3f, in Dalandzadgad, alternatively the 78% of C3 was C3c in Ulaangom. The network analysis with regional information and Y-hgs revealed that at least 4 major “star” or “star”-like cluster exist although the degrees of influence were variable. These clusters suggested that at least 4 major male ancestors with Y-HG-C3 have affected the gene pool of Mongolian males at the different periods.

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## 1. Introduction

It is said the Y-chromosomal diversity is lower around Mongolia, from northern east to central Asia than the other areas of the world due to the observation of “star” cluster chromosome, likely to be originated from the male-line descendants such as Genghis Khan [1–3]. In the present study, Y-STRs and Y-haplogroups (Y-hgs) were analyzed to investigate the genetic diversity in Mongolian male lineage and the paternal genetic relationship between Mongolian and Japanese by constructing a Network with regional information and the results of Y-hgs.

## 2. Materials and methods

### 2.1. DNA samples

Buccal swab samples were collected from unrelated healthy males among 5 regional Mongolian populations (Ulaangom ( $n = 95$ ), Dalandzadgad ( $n = 100$ ), Ulaanbaatar ( $n = 97$ ), Undurkhaan ( $n = 84$ ), and Choibalsan ( $n = 117$ ); total  $n = 493$ ) with written informed consents. DNA samples were extracted using QIAamp DNA Mini kit (QIAGEN) according to the protocol.

### 2.2. Y-STRs haplotyping

17 Y-STRs were haplotyping using a AmpFISTR Yfiler kit according to the user's manual.

### 2.3. Y haplogrouping

Y-hgs were determined mainly by a method using a SNaPshot multiplex kit [4], and additionally an allele specific PCR method by ourselves was performed for Y-hg-C1, -J, -O, -N and R.

### 2.4. Network analysis

We constructed a median-joining network based on the 14 Y-STR haplotypes except DYS389II and DYS385 loci with the Y-hgs data and region information using the program Network 4.6.1.1 by adding with 138 Japanese male data.

## 3. Results and discussion

We observed 358 and 132 haplotypes in Mongolian and Japanese males, respectively. The frequencies for Y-hgs in each regional Mongolian and Japanese male population were calculated as shown in Table 1. The haplotype diversity and discrimination capacity were 0.9934 and 0.7099 in Mongolian, and 0.9989 and 0.9565 in Japanese, respectively. These values show low diversity in Mongolian males. About 63% of Mongolian males had C3 groups from 50% in Ulaanbaatar to 69% in Ulaangom. The 96% of C3 was C3\*, which is C3 except C3a to C3f, in Dalandzadgad, meanwhile

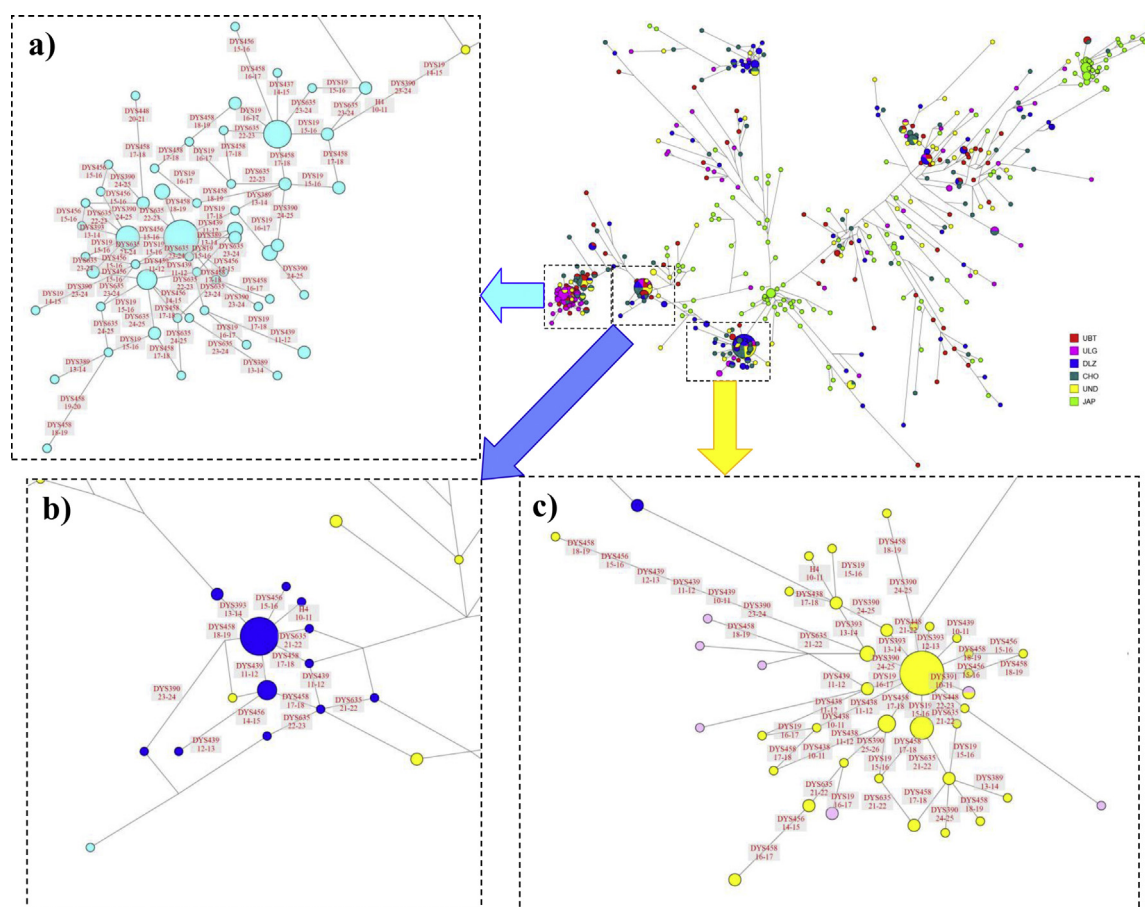
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**Table 1**

Y-hg frequencies (%) in each 5 regional Mongolian and a Japanese male population.

Y-hg	Mongolian						Japanese
	Ulaangom (n = 95)	Dalandzadgad (n = 100)	Ulaanbaatar (n = 97)	Undurkhaan (n = 84)	Choibalsan (n = 117)	Total (n = 493)	
C1	0.0	0.0	0.0	0.0	0.0	0.0	1.4
C3	68.4	61.0	49.5	57.1	59.0	59.0	10.1
C3c	53.7	2.0	23.7	16.7	17.1	22.6	0.0
C3d	3.2	8.0	11.3	10.7	8.5	8.4	0.0
C3e	0.0	3.0	0.0	1.2	3.4	1.5	0.0
C3*	11.6	48.0	14.4	28.6	29.9	26.5	10.1
D	1.1	4.0	4.1	4.8	3.4	3.5	34.8
O	4.2	11.0	25.8	13.1	15.4	13.9	47.8
N	8.4	14.0	8.2	15.5	10.3	11.3	5.1
R	13.7	7.0	3.1	3.6	3.4	6.2	0.0
Q1b	3.2	2.0	3.1	2.4	3.4	2.8	0.7
E	0.0	0.0	0.0	1.2	0.0	0.2	0.0
J	0.0	1.0	3.1	0.0	2.6	1.3	0.0
F-J	1.1	0.0	1.0	1.2	0.9	0.8	0.0
K-T	0.0	0.0	2.1	1.2	1.7	1.0	0.0

**Fig. 1.** A median-joining network for 14 Y-STRs constructed by the data from 493 Mongolian and 138 Japanese males with regional information. Panels (a)–(c) were “star (-like) clusters” expanded from each part in the network with Y-hgs data (C3c in light blue, C3d in blue, C3\* in yellow).

the 78% of C3 was C3c in Ulaangom. The network analysis (Fig. 1) revealed that at least 4 major “star” or “star”-like cluster exist although the degrees of influence were variable. These clusters suggested that at least 4 major male ancestors with Y-hg-C3 have affected the gene pool of Mongolian males at the different periods.

#### Conflict of interest

None.

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