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## SHORT COMMUNICATIONS

## Distribution of the Male Lineages of Genghis Khan's Descendants in Northern Eurasian Populations

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**Abstract**—Data on the variation of 12 microsatellite loci of Y-chromosome haplogroup C3 were used to screen lineages included in the cluster of Genghis Khan's descendants in 18 northern Eurasian populations (Altaian Kazakhs, Altaians-Kizhi, Teleuts, Khakassians, Shorians, Tyvans, Todjins, Tofalars, Sojots, Buryats, Khamnigans, Evenks, Mongols, Kalmyks, Tajiks, Kurds, Persians, and Russians; the total sample size was 1437 people). The highest frequency of haplotypes from the cluster of the Genghis Khan's descendants was found in Mongols (34.8%). In Russia, this cluster was found in Altaian Kazakhs (8.3%), Altaians (3.4%), Buryats (2.3%), Tyvans (1.9%), and Kalmyks (1.7%).

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A large-scale study of the Y-chromosome variation in 2123 subjects from 50 populations of different regions of Asia (except for its Russian part) allowed Zerjal et al. [1] to identify a group of male lineages with unique phylogenetic characteristics. These lineages formed a cluster of closely related haplotypes characterized by fanlike branching relative to the central (ancestral) haplotype and belonging to haplogroup C. The Y-chromosome lineages of this cluster had high frequencies in 16 populations living over a vast area from the Pacific to the Caspian Sea. The estimates of Ychromosome lineage diversity within this cluster showed that it emerged in Mongolia about 1000 years ago. The geographical distribution of this cluster strikingly fit the former Mongol Empire, which allowed Zerjal et al. to assume that its expansion were determined by a special form of social selection related to the preferential reproduction of Genghis Khan's dynasty in regions under Mongolian control. Thus, about 8% ( $16 \times 10^6$ ) Asian men carry the Y-chromosome lineage presumably originating from Genghis Khan. The maximum frequencies of the Y-chromosome lineages belonging to the cluster of Genghis Khan's descendants were found in Mongols and Pakistani Khazareans, which also originated from Mongols [2]. Afterwards, the Family Tree DNA (FTDNA) company studied the Y-chromosome polymorphism in 570 men from Mongolia and Kazakhstan [3] and found that 8.2% of Y-chromosome haplotypes belonged to the cluster of Genghis Khan's descendants, which confirmed the data reported by Zerjal et al. [1].

Note that the Mongolian state formed in 1206 as a result of Genghis Khan's uniting Mongolian tribes considerably expanded afterwards, as it conquered and annexed regions of China (called after that Great Khan's Ulus), Central Asia (the Chagatai Ulus), Iran (the state of Il-khan), and Russia (the Golden Horde). The power of the khans of Golden Horde (the state founded by Genghis Khan's grandson Batu in 1243 in the lower Volga region included a considerable part of the modern Russia (except for Eastern Siberian, Far Eastern, and Far Northern regions), northern and western Kazakhstan, Ukraine, part of Uzbekistan (Khorezm), and part of Turkmenistan [4]. Thus, the genetic traces of Genghis Khan's descendants may be found in ethnically differentiated populations of the modern Russia.

Here, we present data on the distribution of the Ychromosome lineages belonging to the cluster of Genghis Khan's descendants in northern Eurasian populations most of which have not been studied before.

The sample consisted of 1437 subjects from 18 ethnic groups of northern Eurasia (Altaian Kazakhs, Altaians-Kizhi, Teleuts, Khakassians, Shorians, Tyvans, Todjins, Tofalars, Sojots, Buryats, Khamnigans, Evenks, Mongols, Kalmyks, Tajiks, Kurds, Persians, and Russians). The screening of alleles *RPS4Y711-T* 

| Population      | Sample siz | Haplog | group C3 | Cluster of haplotypes<br>of Genghis Khan's descendants |       |  |
|-----------------|------------|--------|----------|--|-------|--|
|                 |            | n      | %        | N  | %     |  |
| Mongols         | 46         | 26     | 56.52    | 16   | 34.78 |  |
| Altaian Kazakhs | 36         | 18     | 50.0     | 3  | 8.33  |  |
| Altaians-Kizhi  | 89         | 14     | 15.73    | 3  | 3.37  |  |
| Buryats         | 217        | 148    | 68.2     | 5  | 2.3   |  |
| Tyvans          | 108        | 10     | 9.26     | 2  | 1.85  |  |
| Kalmyks         | 60         | 30     | 50       | 1  | 1.67  |  |
| Teleuts         | 44         | 4      | 9.09     | 0  | 0     |  |
| Khakassians     | 64         | 1      | 1.56     | 0  | 0     |  |
| Shorians        | 38         | 1      | 2.63     | 0  | 0     |  |
| Todjins         | 26         | 2      | 7.69     | 0  | 0     |  |
| Tofalars        | 30         | 0      | 0        | 0  | 0     |  |
| Evenks          | 51         | 21     | 41.18    | 0  | 0     |  |
| Sojots          | 28         | 15     | 53.57    | 0  | 0     |  |
| Khamnigans      | 51         | 28     | 54.9     | 0  | 0     |  |
| Tajiks          | 40         | 0      | 0        | 0  | 0     |  |
| Kurds           | 25         | 0      | 0        | 0  | 0     |  |
| Persians        | 78         | 0      | 0        | 0  | 0     |  |
| Russians        | 406        | 3      | 0.74     | 0  | 0     |  |
| Total           | 1437       | 321    | 22.34    | 30   | 2.09  |  |

**Table 1.** The frequencies of the Y-chromosome lineages belonging to haplogroup C3 and the cluster of Genghis Khan's descendants in the northern Eurasian populations studied

**Table 2.** The frequencies of the Y-chromosome haplotypes belonging to haplogroup C3 and the cluster of Genghis Khan's descendants in the Northern Eurasian populations studied

|           | Number of repeats in microsatellite loci |          |          |          |           |         |         |         | atell   | ite lo  | oci     |         |   |
|-----------|--|----------|----------|----------|-----------|---------|---------|---------|---------|---------|---------|---------|---|
| Haplotype | 61 SAQ                                   | DYS 385a | DYS 385b | DYS 389I | DYS 38911 | DYS 390 | DYS 391 | DYS 392 | DYS 393 | DYS 437 | DYS 438 | DYS 439 | Population  |
| 1         | 16                                       | 12       | 13       | 13       | 16        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Mongols (11), Buryats (1), Altaian Kazakhs (2), and Kalmyks (3) |
| 2         | 16                                       | 12       | 13       | 13       | 16        | 25      | 10      | 11      | 13      | 15      | 10      | 10      | Mongols (1)   |
| 3         | 16                                       | 11       | 13       | 13       | 16        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Mongols (1) and Altaian Kazakhs (1)                             |
| 4         | 15                                       | 12       | 13       | 13       | 16        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Mongols (2), Buryats (2), and Altaians (1)                      |
| 5         | 15                                       | 13       | 13       | 13       | 16        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Altaians (2)  |
| 6         | 16                                       | 12       | 13       | 13       | 17        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Tyvans (1) and Buryats (1)                                      |
| 7         | 16                                       | 12       | 13       | 14       | 17        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Tyvans (1)  |
| 8         | 16                                       | 12       | 13       | 13       | 16        | 26      | 10      | 11      | 13      | 14      | 10      | 10      | Mongols (1)   |
| 9         | 17                                       | 12       | 13       | 13       | 16        | 25      | 10      | 11      | 13      | 14      | 10      | 10      | Buryats (1)   |

Note: The number of subjects with identified haplotypes is shown in parentheses. The differences from the central haplotype 1 in the number of repeats are boldfaced. Haplotype 1 corresponds to the central haplotype of the cluster of Genghis Khan's descendants according to [1, 3].

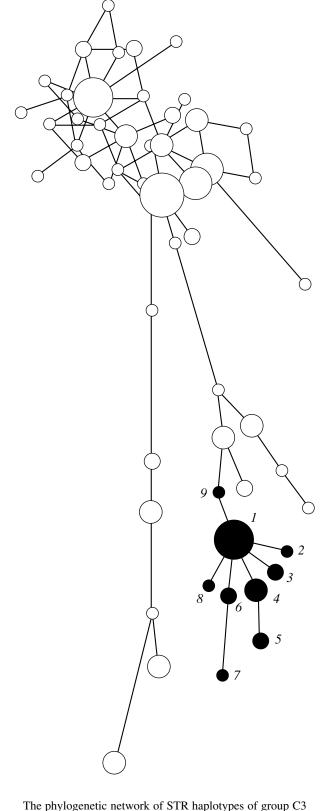
and M217-C, which define haplogroup C3, was performed as described earlier [5, 6]. Twelve STR loci of the Y chromosome (DYS19, DYS385a, DYS385b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, and DYS439) were typed by means of a PowerPlex® Y multiplex system for PCR amplification (PE Applied Biosystems) and an ABI Prism 3100 genetic analyzer (PE Applied Biosystems) with the use of allelic standards. The sizes of PCR products were determined using the GeneScan v. 3.1 and Genotyper v. 2.0 software packages (PE Applied Biosystems). The median network of the STR haplotypes of group C3 (except for subgroup C3c) was drawn using the median joining (MJ) algorithm of the Network 4.1.1.2 software [7]. To identify the STR haplotypes belonging to the cluster of Genghis Khan descendants, we used data reported in [1] and the FTDNA study [3].

The analysis showed that the Y-chromosome lineages marked with alleles RPS4Y711-T and M217-C, which define haplogroup C3, are common for most populations studied (Table 1). The C3-chromosome frequencies were high (50% of higher) in the gene pools of Mongols (56.5%), Mongolian-speaking Buryats (68.2%), Khamnigans (54.9%), Sojots (53.6%), Kalmyks (50%), and Altaian Kazakhs (50%). In the Evenk gene pool, its frequency was 41.2%. The Y-chromosome C3 lineages were found in all other Siberian populations studied except for Tofalars, their frequencies varying from 1.6% in Khakassians to 15.7% in Altaians-Kizhi. It is interesting that we also found C3 chromosomes in Russians, although with a low frequency (0.7%). Note that all Russian carriers of C3 chromosomes originated from southern Russia (the Belgorod and Saratov oblasts).

For a more detailed characterization of the C3 haplotypes of the Y chromosome found in northern Eurasian populations, we studied the variation of 12 microsatellite loci. Analysis of the phylogenetic relationships between STR haplotypes of the Y chromosome performed with the use of median networks showed the presence of a cluster of male lineages characterized by fanlike branching and a high frequency of the central haplotype, which was structurally identical to the ancestral haplotype of Genghis Khan's descendants (figure, Table 2). The highest frequencies of the haplotypes from the cluster of Genghis Khan's descendants were found in Mongols (about 35%). Among populations from Russia, the frequency of this cluster was the highest in Altaian Kazakhs (8.3%); at frequencies between 1.7 and 3.4%, this haplotype was found in Altaians, Buryats, Tyvans, and Kalmyks (Table 1). We found the central haplotype corresponding to the male lineage originating from Genghis Khan in almost every fourth Mongol (24%), 5.6% of Altaians Kazakhs, 1.7% of Kalmyks, and 0.5% of Buryats (Table 2).

Thus, our data agree with the results of earlier studies, where the highest frequencies of the cluster of male

(except for subgroup C3c). The sizes of the circles are proportional to haplotype frequencies. The lengths of the branches are proportional to the number of mutations. Haplotypes belonging to the cluster of Genghis Khan's descendants are shown in black. The numeration of haplotypes is the same as in Table 2.



lineages of Genghis Khan's descendants were found in Mongols and Kazakhs. Although southern Siberian ethnic groups have been studied in detail, the cluster of Genghis Khan's descendants was found (with low frequencies) only in populations bordering on Mongolia, from which the Mongol Empire began to expand in 1206. It is known that the Mongol Empire expanded over a considerable part of Eastern Europe by 1248 due to the khan Batu's conquests. Russian principalities were vassal states of the Mongol Empire until 1480. However, we found no genetic traces of the Mongol sovereignty over Russia (in the form of male lineages of the cluster of Genghis Khan descendants) in the Russian population. We believe that further studies on the Y-chromosome variation will substantially extend our knowledge on the evolution and history of ethnic groups of Russia, as well as the origin of individual lineages constituting them.

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