



Does the Tat polymorphism originate in northern Mongolia?

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Abstract. It has been suggested that the Y-chromosomal T → C transition arose in Mongolia ~2400–4000 years ago. To test this hypothesis, we screened 2300-year-old Mongolian male specimens and ancient Yakut male specimens for this Y-chromosomal marker. Our results demonstrate that the mutation was present in Asia 2300 years ago. © 2003 Elsevier B.V. All rights reserved.

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1. Introduction

In the past few years, a large number of polymorphic markers have been identified on the Y chromosome. Among these is the T → C transition (locus RBF5) reported by Zerjal et al. [1] and later called the Tat-polymorphism. The C allele of this biallelic marker has so far been observed only in populations from Asia and northern Europe. It reaches its highest frequency in Yakuts, Buryats, northeastern Siberian populations and Finns (Table 1).

Opinions differ about whether the geographic origin of the T → C mutation lies in Asia or northern Eurasia. Zerjal et al. [1] suggested that this mutation first arose in the populations of Central Asia; they proposed Mongolia as a candidate location for the origin of the T → C polymorphism. In contrast, for Lahermo et al. [2] the wide distribution of the mutation in north Eurasian populations suggests that it arose in northern Eurasia. According to them, the estimated time of the C mutation is ~2400–4440 years ago.

In this study, we screened ancient Mongolian samples from the Egyin Gol necropolis for the Tat marker. The Egyin Gol necropolis, located in northern Mongolia, is ~2300 years old and belongs to the Xiongnu culture [3]. In addition, we genotyped

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Table 1

Frequency of the Tat C allele in some Eurasian populations including those tested in this study (references are not exhaustive)

| Language family | Population | <i>n</i> | Frequency (%) | Reference |
|-----------------|------------------|----------|---------------|------------|
| Mongolic | Buryats | 111, 81 | 57.7, 28.4 | [1,6] |
| | Mongols | 47, 148 | 2.1, 2.7 | [1,6] |
| ? | Xiongnu | 22 | 9.1 | This study |
| Chukchi | Chukchi | 24 | 58.3 | [7] |
| Eskimo-Aleut | Siberian Eskimos | 22, 33 | 50.0, 60.6 | [6,7] |
| Turkic | Yakuts | 21–35 | 85.7 | [1,8] |
| | | 5 | 100.0 | This study |
| Finno-Ugric | Finns | 41 | 61.1 | [2] |

the T → C mutation in ancient Yakut specimens excavated at two sites in the Sakha Republic (Yakutia) [4].

2. Material and methods

The ancient Xiongnu sample was composed of 22 male specimens recovered from the Egyin Gol necropolis (Lake Baïkal region) [3]. The ancient Yakut sample comprised five male specimens from two burial sites (At-Dabaan and Sytygane Syhe) located 30 km from Yakutsk, the capital of the Sakha Republic. Two of these individuals were recovered from a double grave dated from the late 18th century [4]. The three others were exhumed from three graves dated from the 15th century.

The T → C transition was amplified using the procedure reported by Zerjal et al. [1]. The loci DYS19, DYS390, and DYS39 were analysed as previously described [3]. Locus DYS388 was amplified according to Kayser et al. [5].

3. Results

All of the 5 ancient Yakut samples typed contained the C allele, whereas only 2 of the 22 ancient Egyin Gol samples tested (9.1%) showed the mutation (Table 1).

Among the seven ancient samples harbouring the T → C mutation, only four could be typed at the Y-STR loci DYS19, DYS388, DYS390 and DYS391. For all of them, the haplotype obtained was 14-12-23-11 which corresponds to the haplotype 11-11-10-11 reported by Lell et al. [7].

4. Discussion

The frequency of the Tat C allele is high in the Yakut population but the origin(s) of this allele is unclear. The Yakuts are thought to have migrated northwards from the vicinity of Lake Baïkal to the reaches of the Lena River during the 13th and 14th centuries. They were probably driven out by the Buryat and other Mongol conquerors.

All of the seven ancient Yakut individuals tested showed the C allele, confirming that the mutation occurred most probably before their migration from southern regions. Concerning the Xiongnu people, two of them harboured the mutation suggesting that the Tat polymorphism already existed in Mongolia 2300 years ago.

Amplification of four additional polymorphic markers revealed that the Xiongnu and the Yakut individuals belong to the same Tat-C haplotype (11-11-10-11 according to Ref. [7]). This haplotype is not the ancestral one but is the most frequent in native Siberian populations [7].

Since we previously obtained the mitochondrial DNA sequences of the two ancient Egyin Gol specimens bearing the C allele [3] we could also note that the HV1 sequence of one of them perfectly matched two samples from a present-day Yakut population [9].

In conclusion, our study showed that the C allele was present in parent populations to the modern inhabitants of Mongolia or Yakutia, suggesting that the mutation may have arisen in Mongolia more than 2400 years ago [1]. Moreover, our work suggests that the Xiongnu tribe under study may have been composed of some of the ancestors of the present-day Yakut population.

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