
HUMAN GENETICS

Restriction Polymorphism of Mitochondrial DNA in Koreans and Mongolians

M. V. Derenko¹, A. V. Lunkina¹, B. A. Malyarchuk¹, I. A. Zakharov², Ts. Tsedev³, K. S. Park⁴,
Y. M. Cho⁴, H. K. Lee⁴, and Ch. H. Chu⁵

¹ *Institute of the Biological Problems of the North, Russian Academy of Sciences, Magadan, 685000 Russia;
fax: (41322) 344-63; e-mail: mderenko@ibpn.kolyma.ru*

² *Vavilov Institute of General Genetics, Russian Academy of Sciences, Moscow, 119991 Russia*

³ *Institute of Biological Sciences, Mongolian Academy of Sciences, Ulanbatar, 510351 Mongolia*

⁴ *Seoul National University, College of Medicine, Seoul, 151-742 Korea*

⁵ *Department of History, Kangwon National University, Chunchon, 200-701 Korea*

Received February 12, 2004; in final form, July 1, 2004

Abstract—Using the data on mitochondrial DNA (mtDNA) restriction polymorphism, the gene pools of Koreans ($N = 164$) and Mongolians ($N = 48$) were characterized. It was demonstrated that the gene pools were represented by the common set of mtDNA haplogroups of East Asian origin (M*, M7, M8a, M10, C, D4, G*, G2, A, B*, B5, F1, and N*). In addition to this set, mtDNA haplogroups D5 and Y were identified in Koreans while Mongolians possessed haplogroup Z. Only in Mongolians, a European component with the frequency of 10.4% and represented by the mtDNA types belonging to haplogroups K, U4, and N1, was identified. Phylogenetic and statistical analyses of the data on mtDNA variation in the populations of South Siberia, Central, and East Asia suggested the existence of interpopulation differentiation within these regions, the main role in which was played by the geographical and linguistic factors. Analysis of the pairwise F_{ST} distances demonstrated close genetic similarity of Koreans to Northern Chinese, which in turn, were clearly different from Southern Chinese populations. Mongolians occupied an intermediate position between the ethnic groups of South Siberia and Central/East Asia.

INTRODUCTION

East Asia is one of several world regions, where the first stages of the development of anatomically modern humans took place. According to paleontological data, primary eastern race-forming area was developed on the territory of the Yellow River and the Yangtze River basins as early as during Lower Paleolithic [1]. It is suggested that later (during Upper Paleolithic) the development of Mongoloid characters, differentially expressed in the present-day populations inhabiting the vast territory of Asia began on this area. Analysis of paleontological data showed that the Central Asian race-forming area was developed later (during the Neolithic) within the territory, limited by southern steppe regions of Transbaikalia, central and eastern regions of Mongolia, and, probably, by some regions of Northern China. Beginning from the Bronze Age, Caucasoid groups started colonization of the territories of Central Asia (till Western Mongolia) and the south of Siberia. The result of this was the appearance (at the turn of the Common Era) of the South Siberian race-forming area on the territories of South Siberian steppe and the north of Central Asia. Within this area, intensive mixing between Caucasoids and Mongoloids took place. One of such local population groups has emerged in the Altai–Sayan Region [1–3].

At the same time, on the territory of China, the most important region relative to the understanding of the early stages of the Mongoloid race development, there are still no paleontological findings, covering a large time interval (from 40 to 100 thousand years ago) [4]. This is one of the reasons for such great interest to the results of molecular genetic investigations, permitting reconstruction of prehistoric (racial genetic and demographic) events based on the genetic analysis of the present-day and ancient populations. The studies of mitochondrial DNA (mtDNA) and Y-chromosome variability in East Asian populations showed that the primary colonization of this region by modern humans happened about 60 thousand years ago, during migrations of ancestral populations from Africa to the south of East Asia (in accordance with the “southern wave” model) [4, 5]. The data obtained suggested that colonization of East Asia occurred from the south to the north and that only during Upper Paleolithic these populations have reached the north of China, Mongolia, and Siberia. The existing genetic differences between southern and northern East Asian populations were probably caused by the loss of genetic diversity during the process of migration of southern populations to the north of Asia [4]. Thus, one of the main tasks of ethnic genetics is investigation of genetic differentiation of the populations of East and Central Asia with the purpose of determining the levels of the regional gene pools dif-

ferentiation. In future, this will enable localization of the areas of the formation of long-term genetic diversity, corresponding to different stages of East Asian population differentiation. Also, it will be possible to reconstruct the main stages in colonization of Central and Northern Asia.

The data on the mtDNA variation in the populations of China suggest the existence of genetic differentiation between the northern and southern Chinese populations. Southern and southeastern Chinese populations demonstrate higher diversity of the mtDNA types compared to the populations of Central, East, and Northeast China. The age of a number of mtDNA haplogroups described in the populations of China is estimated as more than 50 000 years, confirming the hypothesis on the colonization of the territory of China about 60 000 years ago [6].

At the same time, many populations of East and Central Asia, for example, Koreans and Mongolians, remain poorly studied in respect of the mtDNA markers distribution. Investigation of classical genetic markers showed that Koreans displayed maximum genetic similarity to Mongolians [7, 8]. On the contrary, the data on mtDNA variation pointed to genetic closeness of Koreans to Chinese and Japanese [9, 10]. Recent studies of the Y-chromosome markers polymorphism showed that the gene pool of Koreans contained the lineages prevalent both in Northeast and Southeast Asia. Moreover, relative to the male gene pool structure, Koreans displayed maximum genetic similarity to Manchurians, Southern Chinese from the province of Yunnan, and Vietnamese [11]. It can be thus suggested that the population of Korea was formed as a result of multiple migration events with the substantial genetic contribution from the northern Asian groups, and also from the populations inhabiting the south and north of China [11, 12]. It is also suggested that the ancestors of Koreans could have common origin with the ethnic groups of Northeast Asia, which inhabited vast territories of the Altai–Sayan and Baikal regions of Southeast Siberia [13].

Mongolians inhabit the territories proposed to represent the origin of Central Asian anthropological type, prevalent in addition to Mongols, among the majority of South and East Siberian ethnic populations [3]. Investigations of the mtDNA variation revealed higher level of the mitochondrial gene pool diversity in the populations of Mongolia compared to the populations of Siberia and Southeast Asia [14]. In the meantime, the data on biochemical markers and nuclear loci variation along with those on the polymorphism of the main non-coding region of mtDNA, point to an extremely low level of genetic differentiation characteristic of Mongolian populations [14–16]. The low level of interpopulation differentiation in Mongolians can be probably explained by specific features of their economic and cultural life style, and also by active integration and assimilation processes, which accompanied the forma-

tion of the Mongolian ethnic groups. Archaeological and anthropological data suggest that the territory of Mongolia could have been already colonized during Upper Paleolithic. These data also point to the succession of archaeological cultures observed within this region till the early Middle Ages [3, 5]. Mongolian population was formed under the influence of different groups (Hunnu, Syanbi, Jujan, Uighurs, Kidans, etc.), ethnolinguistic and anthropological affiliation of which still remains controversial. For instance, recent investigations of skeletal remains from the necropolis in the Egyin Gol Valley (Northern Mongolia) showed that the population of Xiongnu period (3rd century B.P.–2nd century A.D.) was characterized by mixed anthropological composition, since 11% of the mitochondrial lineages, identified in the study cited, belonged to western Eurasian mtDNA haplogroups [17]. Their genetic succession, however, cannot be established due to the lack of data on mtDNA polymorphism among the ancient and present-day population of Mongolia and adjacent territories.

The present study, thereby, was focused on the analysis of the structure and diversity of the mitochondrial gene pools of Mongolians and Koreans, as well as on the evaluation of the population differentiation patterns of the ethnic populations from South Siberia, Central, and East Asia.

MATERIALS AND METHODS

The sample of Mongolians (48 individuals) was formed from the inhabitants of different aimaks of Mongolia. The sample of Koreans (164 individuals) was comprised of the inhabitants of different provinces of Korea. Only individuals who were apparently unrelated at least for three generations participated in the study. Genomic DNA used as the study material was isolated from the biological tissue samples (blood and hair bulbs) using standard techniques [18].

Screening for polymorphic sites determining the main haplogroups of mtDNA types prevalent in the populations of Eurasia (Table 1) was conducted using restriction endonuclease analysis of mtDNA fragments amplified in polymerase chain reaction with the primers suggested in [19, 20]. Restriction fragments were fractionated by electrophoresis in 8% polyacrylamide gels. Gels were stained with ethidium bromide and DNA fragments were visualized in the UV light.

Polymorphism was scored by the presence (+) or absence (–) of the restriction endonuclease recognition sites. The mtDNA types were identified in accordance with the existing classification of the haplogroups of mtDNA types in the populations of Eurasia [6, 10, 21, 22]. Haplogroups and subhaplogroups of mtDNA types are defined as monophyletic clusters marked by certain polymorphism variants (Table 1). In accordance with the classification, mtDNA haplogroups are denoted by single Roman letters (excluding haplogroup HV), and