
HUMAN GENETICS

On the Origin of Mongoloid Component in the Mitochondrial Gene Pool of Slavs

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Abstract—The data on mitochondrial DNA (mtDNA) restriction polymorphism in Czech population ($n = 279$) are presented. It was demonstrated that in terms of their structure, mitochondrial gene pools of Czechs and other Slavic populations (Russians, Poles, Slovenians, and Bosnians) were practically indistinguishable. In Czechs, the frequency of eastern-Eurasian (Mongoloid) mtDNA lineages constituted 1.8%. The spread of eastern-Eurasian mtDNA lineages belonging to different ethnolinguistic groups in the populations of Europe was examined. Frequency variations of these DNA lineages in different Slavic groups was observed, with the range from 1.2 and 1.6% in Southern and Western Slavs, respectively, to 1.3 to 5.2% in Eastern Slavs, the Russian population of Eastern Europe. The highest frequency of Mongoloid component was detected in the mitochondrial gene pools of Russian populations from the Russian North and the Northwestern region of Russia. This finding can be explained in terms of assimilation of northern-European Finno-Ugric populations during the formation of the Russian population of these regions. The origin of Mongoloid component in the gene pools of different groups of Slavs is discussed.

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INTRODUCTION

Genetic aspects of the European population development are still poorly understood, although different issues in this area have been already investigated [1–7]. These studies focused on analysis of highly polymorphic uniparentally inherited genetic systems, mitochondrial DNA (mtDNA) and Y chromosome, are among the most intensely developed. Using mtDNA polymorphism analysis the investigation of the populations of Eastern, Western, and Southern Slavs have been examined [8–18]. Analysis of mtDNA variation in European populations, including Slavic populations, showed that Slavs have a common origin, the central position among these populations is occupied by Western Slavs, and genetic differences between the groups of Slavs are mostly determined by the degree of admixture with pre-Slavic population inhabiting the contemporary ethnic area of Slavs, as well as by the intensity of their interactions with the neighboring populations [19]. The latter conclusion follows from the fact that the neighbors of Russians are western-Finnish populations, characterized by rather high genetic similarity to Russians, Germans, which are close to western Slavs, and Balkan populations, which are close to Southern Slavs [19].

It should be noted, however, that not all Slavic groups are equally characterized. Mitochondrial gene pools diversity has been thoroughly investigated in Southern and Eastern Slavs, while Western Slavs in these investigations were represented by virtually one ethnic group, Poles [5, 11–16]. Czechs and Slovaks, also belonging to the group of western-Slavic popula-

tions remain poorly investigated [17]. The present study was focused on investigating mtDNA diversity in Czech population, as well as on the comparative analysis of mtDNA distribution patterns in Slavs and neighboring European populations.

MATERIALS AND METHODS

Biological material (whole blood samples) was collected in the Departments of Internal Medicine of the hospitals from individuals with no hereditary pathologies. The sample tested ($n = 279$) was represented by Czech individuals born in different regions of Czech Republic.

Genomic DNA for the analyses was isolated from the blood cells with the help of standard methods, including cell lysis with proteinase K (Sigma, United States) in the presence of 1% sodium dodecyl sulfate, DNA purification by means of phenol/chloroform extraction, and DNA precipitation with ethanol.

Screening of polymorphic sites, determining the main groups of mtDNA haplotypes, spread in the populations of Eurasia (Table 1), was performed by means of the analysis of the mtDNA fragments amplified in polymerase chain reaction with the primers described in [21, 22]. Restriction fragments were fractionated by use of electrophoresis in 8% polyacrylamide gels. For DNA detection, gels were stained with ethidium bromide, and DNA was visualized in UV light. Polymorphism was scored as the restriction site gain (+) and loss (–).

Table 1. Scheme of identification of the main mtDNA haplogroups using restriction analysis

mtDNA haplogroup	Restriction polymorphism variants
HV	–14766 <i>MseI</i>
H	–14766 <i>MseI</i> , –7025 <i>AluI</i>
HV0a	–14766 <i>MseI</i> , +15904 <i>MseI</i> , +4577 <i>NlaIII</i>
HV0b	–14766 <i>MseI</i> , –15904 <i>MseI</i> , +4577 <i>NlaIII</i>
V	–14766 <i>MseI</i> , +15904 <i>MseI</i> , –4577 <i>NlaIII</i>
R	+12704 <i>MboII</i>
U	+12308 <i>HinfI</i>
K	+10394 <i>DdeI</i> , +12308 <i>HinfI</i> , –9052 <i>HaeII</i>
J	+10394 <i>DdeI</i> , –13704 <i>BstNI</i>
T	+13366 <i>BamHI</i> , +15606 <i>AluI</i>
T1	+13366 <i>BamHI</i> , +15606 <i>AluI</i> , –12629 <i>AvaII</i>
N1	–12498 <i>NlaIII</i>
I	–4529 <i>HaeII</i> , +8249 <i>AvaII</i> , +10032 <i>AluI</i> , +10394 <i>DdeI</i>
W	+8249 <i>AvaII</i> , –8994 <i>HaeIII</i>
X	–1715 <i>DdeI</i> , +14465 <i>AccI</i>
M	+10394 <i>DdeI</i> , +10397 <i>AluI</i>
C	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , –13259 <i>HincII</i> /+13262 <i>AluI</i>
D	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , –5176 <i>AluI</i>
G	+10394 <i>DdeI</i> , +10397 <i>AluI</i> , +4830 <i>HaeII</i> /+4831 <i>HhaI</i>
A	+663 <i>HaeIII</i>
F1	–12406 <i>HpaI/HincII</i>
L1/2	+3592 <i>HpaI</i>

Note: Restriction polymorphism variants are given according to the Cambridge Reference Sequence of mtDNA [20]. Restriction site gain or loss is defined as + or –, respectively.

The mtDNA haplotypes were typed based on the existing classification of mtDNA in human populations [7, 23]. In accordance to this classification, mtDNA haplogroups, except for haplogroup HV, were designated using Latin single-letter code. Taking into consideration the recommendations given in recent study [24], cluster pre-HV was designated as R0; (pre-HV)1, as R0a; pre-V, as HV0; pre-VI, as HV0b; and pre-V2, as HV0a.

To identify DNA samples, which were impossible to classify using the scheme of the analysis presented in Table 1, screening of additional markers, determining haplogroups L1 and L2 (within cluster L), and N9a (within cluster N, with exception of R) was performed. Identification of the samples within haplogroups L1 and L2 was done using the scheme of restriction analysis, described in [25]. Haplogroup N9a was identified with the help of the analysis of *TasI* polymorphism in the 5416–5419 fragment. mtDNA haplotypes characterized by the presence of the +5416*TasI* variant were defined as N9a [23].

Statistical significance of the among-population differences in terms of mtDNA haplogroup frequencies was evaluated using the exact test for population differ-

entiation [26]. Indices of mtDNA diversity in the populations, as well as F statistics values were computed with the help of the ARLEQUIN 2.0 software package [26], designed for the analysis of molecular variation and population genetic structure.

RESULTS AND DISCUSSION

Analysis of mtDNA variation in Czechs revealed that their gene pool was characterized by typical European composition of mtDNA haplogroups and subhaplogroups. Similarly to other Slavic populations, the dominant clusters in Czechs were H, U, T, and J (Table 2). The overwhelming majority of mtDNA clusters, identified in Czechs, were of the western-Eurasian origin. The frequency of eastern-Eurasian (Mongoloid) mtDNA lineages in this population constituted 1.8% (haplogroups A, N9a, and M). African lineage (with the frequency of 0.4%) belonging to haplogroup L2a and marked by the +13803*HaeIII* variant was also detected. In terms of their structure, mitochondrial gene pools of the Slavic population groups examined were very similar the composition of mtDNA haplogroups and subhaplogroups (Table 2). Analysis of population genetic differentiation showed the absence of the population