

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 fractioned 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 accordance with the Cambridge Reference Sequence of mtDNA [20]. Restriction site gain or loss is defined as + or –, respectively.
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</i> / <i>HincII</i>
L1/2	+3592 <i>HpaI</i>

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 *Tag* polymorphism in the 5416–5419 fragment. To identify the samples within haplogroups L1 and L2, a scheme of restriction analysis described in [25] was used. 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 differentiation [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

Table 2. Frequency distribution patterns of mtDNA in Czechs in comparison with Slavic populations (the sample sizes are in brackets)

MtDNA haplogroup	Czechs (279)	Poles (436)	Slovenians (104)	Bosnians (144)	Russians (201)
H	46.2 (129)	45.2 (197)	47.1 (49)	47.9 (69)	42.3 (85)
HV*	1.4 (4)	0.9 (4)	0	0.7 (1)	2.0 (4)
HV0	2.9 (8)	4.8 (21)	6.7 (7)	6.3 (9)	5.5 (11)
J	12.2 (34)	7.8 (34)	9.6 (10)	6.9 (10)	8.0 (16)
T*	9.7 (27)	9.4 (41)	4.8 (5)	3.5 (5)	9.0 (18)
T1	2.9 (8)	2.1 (9)	1.0 (1)	1.4 (2)	2.0 (4)
K	3.6 (10)	3.4 (15)	3.9 (4)	4.2 (6)	3.0 (6)
U*	14.3 (40)	16.1 (70)	19.2 (20)	19.4 (28)	17.9 (36)
R0a	0	0	0	1.4 (2)	0.5 (1)
R*	0	0.5 (2)	0	0	0.5 (1)
W	0.4 (1)	3.7 (16)	4.8 (5)	1.4 (2)	2.0 (4)
X	1.1 (3)	1.8 (8)	1.0 (1)	1.4 (2)	3.5 (7)
N1*	1.1 (3)	0.2 (1)	0	0.7 (1)	0
I	2.2 (6)	1.8 (8)	1.9 (2)	2.8 (4)	2.5 (5)
N9a	0.4 (1)	0	0	0	0
A	0.4 (1)	0	0	0	0
M	1.1 (3)	1.8 (8)	0	1.4 (2)	1.5 (3)
L	0.4 (1)	0.2 (1)	0	0.7 (1)	0
<i>h</i>	0.74 ± 0.02	0.75 ± 0.02	0.73 ± 0.04	0.72 ± 0.03	0.77 ± 0.02

Note: The data for Russians and Poles are elicited from [12], and for Bosnians and Slovenians, from [13]. *h*, mitochondrial gene pool diversity.

differences in all pairs of comparison by means of *F* statistics ($F_{st} = 0$; $P = 0.68$), as well as by means of exact test ($P = 0.3$ (0.08)).

Analysis of mtDNA variation in European populations showed that the gene pools of different Slavic populations almost always contained DNA lineages of eastern-Eurasian origin (Table 3). The causes of this are unclear. It can be hypothesized that rather diverse Mongoloid component was the part of pre-Slavic gene pool. Alternatively, the appearance of this component in the gene pools of different Slavic groups has a “cumulative” character, i.e., it is associated with gradual assimilation of Mongoloid DNA lineages as a result of the interaction between the Eastern European and Asian populations in different historical periods.

According to linguistic, archaeological, and paleogeographic data, the decay of Balto-Slavic linguistic community happened 2000–3000 years BP, which resulted in the appearance of proto-Balts and proto-Slavs [33]. Analysis of mtDNA variation in Slavs and Balts showed that their gene pool were different with respect to the frequency of eastern-Eurasian component (Table 3). Low frequency of Mongoloid mtDNA variant in Letts and Lithuanians suggests that Mongoloid component was probably not typical of Balto-Slavic protogene pool. Thus, it seems reasonable that accumu-

lation of Mongoloid mtDNA lineages in Slavs and their ancestors was intensified only in the last 4000 years.

Investigations of mtDNA variation in Russian population of Eastern Europe showed that regional groups of Russian populations were different in terms of frequencies and composition of mtDNA haplogroups having the eastern-Eurasian origin. The data in Table 3 demonstrate that the frequency of Mongoloid component is increased northwards. The highest frequencies of the Mongoloid component are typical of the Russian populations from Russian Pomor’e and Northwestern region. These populations, however, differ in the mtDNA haplogroup composition. It was established that assimilation of the indigenous pre-Slavic population of Eastern Europe by true Slavs was of great importance to the process of the development of Russian population. Therefore, interregional differences between the groups of Russians seem likely to be associated with specific genetic features of assimilated populations. It was demonstrated that northern-European Finno—Ugric populations (Saami, Finns, and Karelians) were basically characterized by the presence of two mtDNA haplogroups, Z and D5 [29, 31]. Based on the data of phylogeographic analysis, it is suggested that these haplogroups could be introduced to the north of Europe from Asia as early as during the early Neolithic [31]. Therefore, the high frequency of haplo-

Table 3. Frequency of eastern-Eurasian mtDNA lineages in the gene pools of different ethnolinguistic groups of Europe

Population	Frequency, %	MtDNA haplogroup composition
German-speaking populations		
Germans (333) ¹	0.6	D
Norwegians (397) ¹	0.5	Z
Finno-Ugric populations		
Estonians (409) ¹	0.2	D
Karelians (83) ²	4.8	D
Finns (580) ¹	1.8	Z, D
Hungarians (98) ³	1.7	B, M
Maris (136) ⁴	7.4	A, M*, C, Z, D
Mordovians (102) ⁴	2.9	C, D
Udmurts (101) ⁴	20.8	A, C, Z, D
Komi (136) ⁴	11.8	A, C, Z, D, G
Baltic-speaking populations		
Letts (299) ¹	0.3	G
Lithuanians (180) ¹	0.6	A
Slavic-speaking populations		
Eastern Slavs (Russians from southern and central regions) (458) ^{5, 6}	1.3	M*, C, D, G
Eastern Slavs (Russians from northwestern region) (228) ^{6, 7}	4.0	A, Z, D, M*, G
Eastern slavs (Russian Pomors) (134) ⁸	5.2	D5, Z
Western Slavs (Poles and Czechs) (808) ^{5, 9, 10}	1.6	A, N9a, M*, C, D, G
Southern Slavs (Bosnians, Croatians, Herzegovinians, Serbs, Macedonians, and Sloven) (1705) ^{11, 12}	1.2	A, F, D, M*

Note: In brackets are the sample sizes. Frequencies of mtDNA haplogroups are presented according to: ¹, [28]; ², [29]; ³, [30]; ⁴, [27]; ⁵, [12]; ⁶, [16]; ⁷, [32]; ⁸, [31]; ⁹, present study; ¹⁰, [17]; ¹¹, [15]; ¹², [13].

groups Z and D5 in Russian Pomors, and to a lesser extend, in Russians from the northeastern part of Russia, can be explained by assimilation of mainly northern-European Finno-Ugric populations. At the same time, the repertoire of eastern-Eurasian haplogroups in the populations from most of the European regions of Russia, including its northwestern part, is much wider, than in the population of Russian Pomor'e [32]. These findings indicate that other populations, genetically closer to the modern East European populations, like Mordovians, Maris, Udmurts, and Komi were subjected to assimilation. Complication of the Mongoloid component structure in the gene pools of these populations is believed to be associated with the long-lasting interactions with the populations of Siberia and Central Asia. One of the main historical periods of intensification of this interaction was the early Middle Ages, when the waves of migrations of a number of steppe populations (Huns, Avars, Bulgars, and Mongols) passed through Eastern Europe. For instance, in the 4th century AD, Avars even the reached the territories of Central Europe (Wallachia, Pannonia, Transylvania, and Bohe-

mia), where they formed the Avar Khaganate, which existed until the 19th century AD.

It is suggested that after the decay of Avar Khaganate the populations included into it were assimilated by Slavic tribes [34]. It is thereby suggested that rather high frequencies of eastern-Eurasian mtDNA lineages observed in the gene pools of some populations of Western and Southern Slavs (especially on the territory of former Avar Khaganate) can be considered as a consequence of the process described.

Concerning the population of Eastern Europe, it should be noted that the forest zone of Eastern Europe was the area of intense population admixture [35]. It seems likely, that formation of the complex of Mongoloid traits happened not later than in Upper Paleolithic. For this reason, it is suggested that East Siberian populations could have much time for migration to Eastern Europe [35]. The number of such migrations still remains unclear, since in the northwest of Eastern Europe Mongoloid component is detected 10000–8000 years ago; in Dnepr–Donetsk tribes, 7000–6000 years ago, and on the territory of Ivanovo oblast (Sakhtysh), 6000–5000 years ago [35, 36]. The data on mtDNA variation in Russian populations are

consistent with anthropological data, since they point to the substantial differences in the frequencies of Mongoloid mtDNA lineages between the Russian populations of the Russian North, Northwest, and the central/southern regions of the European part of Russia (Table 3). We hope that future investigations would provide the possibility of analyzing the chronology of the development of the Mongoloid component diversity in the gene pools of Russians and other Slavic populations.

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