

Complex interactions of the Eastern and Western Slavic populations with other European groups as revealed by mitochondrial DNA analysis

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Abstract

Mitochondrial DNA sequence variation was examined by the control region sequencing (HVS I and HVS II) and RFLP analysis of haplogroup-diagnostic coding region sites in 570 individuals from four regional populations of Poles and two Russian groups from northwestern part of the country. Additionally, sequences of complete mitochondrial genomes representing K1a1b1a subclade in Polish and Polish Roma populations have been determined. Haplogroup frequency patterns revealed in Poles and Russians are similar to those characteristic of other Europeans. However, there are several features of Slavic mtDNA pools seen on the level of regional populations which are helpful in the understanding of complex interactions of the Eastern and Western Slavic populations with other European groups. One of the most important is the presence of subhaplogroups U5b1b1, D5, Z1 and U8a with simultaneous scarcity of haplogroup K in populations of northwestern Russia suggesting the participation of Finno-Ugrian tribes in the formation of mtDNA pools of Russians from this region. The results of genetic structure analyses suggest that Russians from Velikii Novgorod area (northwestern Russia) and Poles from Suwalszczyzna (northeastern Poland) differ from all remaining Polish and Russian samples. Simultaneously, northwestern Russians and northeastern Poles bear some similarities to Baltic (Latvians) and Finno-Ugrian groups (Estonians) of northeastern Europe, especially on the level of U5 haplogroup frequencies. The occurrence of K1a1b1a subcluster in Poles and Polish Roma is one of the first direct proofs of the presence of Ashkenazi-specific mtDNA lineages in non-Jewish European populations.

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1. Introduction

The Slavs are the most numerous and one of the most rapidly expanding groups of Indo-European-speaking people of central and eastern Europe [1]. Despite their demography and territorial extent, the origin and early development of Slavs are still poorly comprehended and thus extensively discussed by linguists, archeologists, anthropologists and historians. Among the Indo-European languages the Slavic group is most closely related to the Baltic languages. This has led some linguists to suggest the former existence of a common Proto-Balto-Slavic language, from which the Slavic and Baltic groups

later differentiated [2]. In the period A.D. 400–900, Proto-Slavic collapsed and differentiated into modern Slavic languages. Subsequent expansions to the east and northeast from A.D. 500 to 1000 carried Slavic speakers into territories previously occupied by Balts and Finno-Ugrians [3]. From A.D. 500 Slavic tribes also pushed south and west into the Byzantine world to settle in the Balkans and central Europe while other tribes moved down the Dniepr river or pressed east towards the Urals and beyond [2].

Genetic studies on Western (Poles, Czechs and Slovaks) and Eastern Slavic populations (Russians, Byelorussians, Ukrainians) still have not left a phase of data collection. Although there are several examples of research on Polish populations using mtDNA variability, no detailed mtDNA studies have been performed to analyze the differentiation of regional populations of Poles. MtDNA data from Russian populations appears to be

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more numerous, although scarce results have been obtained for the regional populations living in the northwestern part of European Russia. For better understanding of the complex interactions between the Slavs and non-Slavic populations of Europe, we present here mtDNA data from four regional populations of Poles and two Russian groups from northwestern part of the country.

2. Materials and methods

2.1. Population samples

A total of 570 blood samples from maternally unrelated individuals were collected from the following six populations: Pomerania-Gdansk ($n = 166$) and Kaszuby ($n = 87$) from northern Poland; Suwalszczyzna ($n = 73$) from northeast Poland; Upper Silesia ($n = 87$) from southern Poland, Velikii Novgorod ($n = 79$) and Volot ($n = 78$) from northwestern part of European Russia (Novgorod oblast). Appropriate informed consent was obtained from all participants in this study.

2.2. Sequencing and RFLP analysis of mtDNA

Hypervariable segments I and II (HVS I and II) of the mtDNA noncoding control region were amplified and sequenced as described elsewhere [4]. The nucleotide sequences of HVS I from position 15999 to 16400 and HVS II from position 30 to 407 were determined and compared with the Cambridge reference sequence [5,6]. Nucleotide positions showing point indels and transversions located between positions 16180–16193 and 303–315 were excluded from further analysis.

All Polish samples were subjected to RFLP analysis of haplogroup-specific coding region sites (Table S1) as described elsewhere [7,8]. Russian samples had been screened previously for haplogroup-diagnostic RFLP markers by Lunikina et al. [9]. Sample classification into mtDNA clusters and subclusters follows [10–18] and the authors' unpublished data on complete mitochondrial genome sequences. Complete sequencing of four mitochondrial genomes representing K1a1b1a subhaplogroup in Polish (individuals Gd168 and Kat118) and Polish Roma populations (individuals Gy17 and Gy22) [19] was performed using the methodology described by Torroni et al. [20].

2.3. Phylogenetic and statistical analysis

Median joining (MJ) networks were created with the computer program Network 4.1.1.1 (A. Röhl; © Fluxus Technology; www.fluxus-engineering.com). Most parsimonious tree of the complete haplogroup K1a1b1a mtDNA sequences was reconstructed manually. The statistical significance of population differences with respect to the frequencies of mtDNA haplogroups was evaluated using the chi-square test. The population genetic structure was analyzed using methods implemented in the Arlequin v. 3.0 software [21]. The statistical significance of F_{ST} values was estimated by permutation analysis using 10,000 permutations. Multidimen-

sional scaling (MDS) analysis of pairwise interpopulation F_{ST} values was performed with the use of the software package STATISTICA v. 7.1 (StatSoft, Inc., Tulsa, USA). Spatial analysis of molecular variance (SAMOVA) [22] was performed using geographical coordinates characterizing the studied populations and the computer program SAMOVA v. 1.0 (web.unife.it/progetti/genetica/Isabelle/samova.html). Variations between positions 16001 and 16400 of HVS I region were considered in the genetic structure analyses. mtDNA HVS I data from the following northeast European populations were used for comparisons (sample sizes are given in parentheses): Russians from the Stavropol (62), Orel (76), Saratov (63), Tula (73), Kaluga (71), Vladimir (72), Yaroslavl (41) and Pskov (68) regions [4,23]; Russians from the Kursk (34), Kostroma (55) [24] and Rostov (25) regions [11]; Poles from the Pomerania Kujawy (436) region [4]; Byelorussians (92) [25], Latvians (299) [26], Lithuanians (180) [27,28], Estonians (47), Karelians (83) and Finns (403) [29,30].

3. Results and discussion

Four hundred and thirteen different haplotypes were identified in a total sample of 570 Polish and Russian individuals (Table S2). Haplogroup frequency patterns revealed in Poles and Russians (Table 1) are similar to those characteristic of other Europeans. Below we discuss these characteristics of Polish and Russian mtDNA pools which have probably arisen relatively recently and thus could be helpful in the reconstruction of some details of putative “Slavic identity”.

3.1. Phylogenetic analysis of haplogroup U in populations of northeastern Poland and northwestern Russia

Analysis of haplogroup (hg) frequency patterns (Table 1) reveals the presence of significant differences between frequencies of hg U in some regional groups of Poles and Russians. The highest frequencies of U were observed in Suwalszczyzna and Upper Silesia. The latter exhibits the presence of seven U subhaplogroups (U1–U5, U8 and K) which accounts for the greatest diversity within U out of all Polish and Russian populations studied. In the population of Suwalszczyzna sub-hgs U1, U2 and U8 have not been observed, U3 and U4 occurred with low frequencies and the vast majority of the hg U pool was distributed into U5 (20.5%) and K (9.6%). Significant differences of U5 frequencies were observed between Suwalszczyzna and Kaszuby ($P < 0.05$) as well as between Suwalszczyzna and Pomerania-Kujawy region ($P < 0.01$) [4]. In European Russia, high frequencies of U5 were observed in northwestern populations of Velikii Novgorod (19%, Table 1) and Pskov (20.6%) [23]. Frequency of hg U5 was significantly higher in Pskov than in the Pomerania-Kujawy ($P < 0.01$) [4,23]; comparably, significant differences ($P < 0.01$) were observed between frequencies of this hg in Pomerania-Kujawy and Velikii Novgorod. U5, the most ancient and abundant subclade of hg U in Europe [7,11,31] occurs with the highest frequencies in some Finno-Ugrian populations of northern Europe, such as Finns and Estonians [26] or Saami

Table 1
mtDNA haplogroup distributions (percentage, with number of individuals in parentheses) in regional populations of Poland and Russia

Haplogroup	Pomerania-Gdańsk (n = 166)	Upper Silesia (n = 87)	Kaszuby (n = 87)	Suwalszczyzna (n = 73)	Velikii Novgorod (n = 79)	Volot (n = 78)
H	43.4 (72)	43.7 (38)	42.5 (37)	37.0 (27)	41.8 (33)	41.0 (32)
HV	1.2 (2)	0	2.3 (2)	0	1.3 (1)	6.4 (5)
HV*	0.6 (1)	0	0	0	1.3 (1)	1.3 (1)
HV3	0.6 (1)	0	2.3 (2)	0	0	5.1 (4)
R0	0.6 (1)	0	0	0	0	0
HV0	0	0	0	1.4 (1)	0	2.6 (2)
V	3.6 (6)	3.4 (3)	9.2 (8)	4.1 (3)	6.3 (5)	3.8 (3)
J	6.0 (10)	6.9 (6)	11.5 (10)	8.2 (6)	5.1 (4)	9.0 (7)
T*	8.4 (14)	1.1 (1)	5.7 (5)	2.7 (2)	6.3 (5)	9.0 (7)
T1	3.0 (5)	2.3 (2)	1.1 (1)	1.4 (1)	0	0
U	25.3 (42)	32.2 (28)	16.1 (14)	32.9 (24)	26.6 (21)	12.8 (10)
K	2.4 (4)	3.4 (3)	4.6 (4)	9.6 (7)	0	3.8 (3)
U1	0	2.3 (2)	0	0	0	0
U2	0.6 (1)	4.6 (4)	2.3 (2)	0	2.5 (2)	0
U3	0.6 (1)	2.3 (2)	0	1.4 (1)	1.3 (1)	0
U4	8.4 (14)	5.7 (5)	1.1 (1)	1.4 (1)	0	1.3 (1)
U5	11.4 (19)	11.5 (10)	8.0 (7)	20.5 (15)	19.0 (15)	5.1 (4)
U8	1.8 (3)	2.3 (2)	0	0	3.8 (3)	2.6 (2)
R	0	0	0	2.7 (2)	1.3 (1)	3.8 (3)
I	1.2 (2)	0	4.6 (4)	4.1 (3)	3.8 (3)	0
W	2.4 (4)	5.7 (5)	4.6 (4)	2.7 (2)	1.3 (1)	2.6 (2)
X	1.8 (3)	2.3 (2)	1.1 (1)	2.7 (2)	1.3 (1)	0
N1a	0.6 (1)	0	1.1 (1)	0	0	1.3 (1)
N1b	1.2 (2)	1.1 (1)	0	0	0	1.3 (1)
N1c	0.6 (1)	0	0	0	0	0
N*	0	0	0	0	1.3 (1)	0
A	0	0	0	0	1.3 (1)	0
C	0	1.1 (1)	0	0	0	0
Z	0	0	0	0	1.3 (1)	1.3 (1)
D	0	0	0	0	0	1.3 (1)
D5	0	0	0	0	1.3 (1)	0
G2a	0.6 (1)	0	0	0	0	0
M1	0	0	0	0	0	2.6 (2)
M10	0	0	0	0	0	1.3 (1)

(27–58%); in the latter, however, U5 is represented almost exclusively by the haplotypes with the “Saami motif” 16144–16189–16270 (sub-hg U5b1b1) [32]. Phylogenetic reconstruction of sub-hg U5b in Polish and Russian populations (Figure S1) shows that haplotypes with “Saami motif” occur mainly in Poles from northern and northeastern parts of the country but also in northwestern part of European Russia. In particular, it is worth noting that frequencies of U5b1b1 haplotypes in populations of Suwalszczyzna and Velikii Novgorod were almost identical (2.7 and 2.6%, respectively) and comparable to those observed in Lithuanians (2.2%) [32]. U5b1b1 was observed previously also in northern Russians from the Archangielsk region (6.6%), Karelians (6%), Finns (2%) and in a sample of 134 individuals from unspecified northern Russian populations (3%) [25,30,32]. It is worth noting that Russians from northwestern Pskov also present a high frequency (35%) of Y-chromosomal haplogroup N3, which is distinctive of all Uralic- and Baltic-speaking populations north and east of the Baltic Sea [23]. Interestingly, elevated frequencies of N3 (11%) were also observed in northeastern Polish population from Suwalszczyzna [33]. There were also differences in hg K frequency distribution in some regional populations of Poles and Russians. In northwestern Russian

groups from Velikii Novgorod (Table 1) and Pskov [23], hg K has not been observed at all, whereas in northern Polish (Pomeranian) populations it was found with relatively low frequencies (2.4–3.4%). It is noteworthy that hg K is relatively scarce in both Baltic- and Volga Finno-Ugrian people [26,34]. In this respect, northwestern Russians are again situated closer to their Finno-Ugrian neighbors.

Another mtDNA component which appears to be characteristic mainly of the populations of northern Poland and northwestern Russia is sub-hg U8a. It was observed in northwestern Russians from Velikii Novgorod and Volot (Figure S2), with frequencies of 3.8 and 2.6%, respectively. Previously, it was also identified in Pskov (1.5%), another population from northwestern Russia [23]. In Poland, U8a haplotypes were observed only in Pomerania-Gdansk (1.8%) and Upper Silesia (2.3%). Analysis of U8a distribution shows that frequencies of this sub-hg in the Russian populations of Novgorod oblast and in Poles from Upper Silesia are the highest among the observed so far in Europe. U8a is a component characteristic probably of mtDNA pools of northern and eastern European populations, since it was earlier detected (with frequencies <2%) in Lithuanians, Finns and Estonians.

3.2. Selected East Eurasian components in mtDNA pools of northwestern Russian populations

Northwestern Russian populations from Velikii Novgorod and Volot are distinctive of a relatively high proportion of East Eurasian hgs—3.9% (A, Z, D5) and 6.5% (Z, D, M1, M10), respectively. It is worth noting that both Z and D5 are characterized by a similar phylogeographic pattern in north-eastern European populations. Z-haplotypes observed in the populations of Novgorod oblast belong to subclade Z1. It was noted with relatively high frequencies in Saami (1.0–7.2%) and Finns (1.5%) as well as in some Finno-Ugrian and Turkic-speaking groups of the Volga Basin, with frequencies 1.8–2.7% [29,32,34]. D5, represented in Russians from Velikii Novgorod by the haplotypes with the HVS I motif 16126–16136–16189–16223–16360–16362 (Table S2) occurs in Europe predominantly in northeastern Finno-Ugrian populations—Saami, Finns, Karelians and Estonians. It was also observed with appreciable frequency (5.2%) in other northwest populations from European Russia [32].

3.3. mtDNA picture of northeastern Poles and northwestern Russians revealed by genetic structure analysis

Previous analyses of mtDNA differentiation in Russians exhibit the presence of a considerable heterogeneity of Russian populations [23]. In this study, we aim at verifying possible genetic structure which would be visible on the level of regional populations of Poles and Russians. Statistically significant differences ($P < 0.05$) were observed between classical (based on hg frequencies) F_{ST} distances computed for Pomerania-Kujawy and Suwalszczyzna. MDS analysis performed on the basis of classical pairwise F_{ST} values revealed that northwestern Russian populations from Velikii Novgorod and Pskov were grouped together with northeastern Polish Suwalszczyzna in the first dimension of MDS and separated from remaining Polish and Russian populations (Fig. 1). The separation of northeastern Poles and northwestern Russians from other regional Polish and Russian populations is even better illustrated in SAMOVA analysis, performed with the use of geographical coordinates and mtDNA HVS I sequences obtained for the studied populations. The results of SAMOVA performed for Polish and Russian populations (Table S3) show that populations of Suwalszczyzna and Pskov are always grouped together when 2, 3 and 4 groups are *a priori* defined. Moreover, with *a priori* definition of 3 and 4 groups, Suwalszczyzna is grouped together with northwestern Russians from Pskov and Velikii Novgorod (Table S3).

Since phylogenetic analysis suggests that northwestern Russians and northeastern Poles bear some similarities to Baltic and Finno-Ugrian groups of eastern Europe, especially on the level of hg U5 frequencies, we have performed MDS analysis of pairwise molecular F_{ST} distances based on HVS I haplotypes belonging to U5 haplogroup. The results of this analysis (Figure S3) show that the first dimension of MDS sharply separates Karelians, Finns and Byelorussians from the remaining

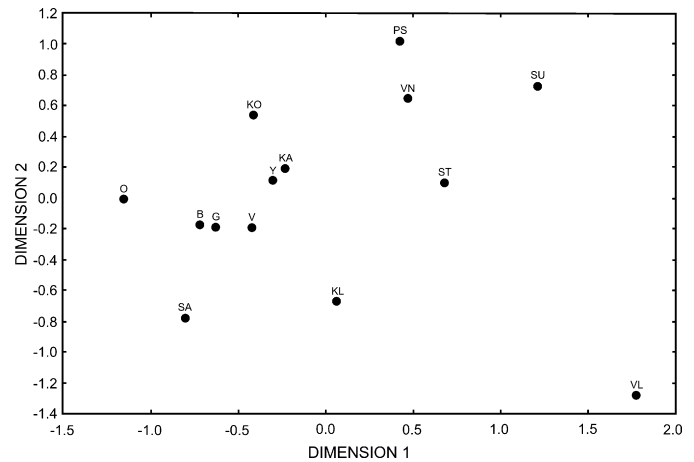


Fig. 1. Two-dimensional MDS scaling plot of Polish and Russian regional populations based on pairwise F_{ST} values derived from frequencies of mtDNA haplogroups. Populations are designated as: G—Pomerania-Gdańsk; KA—Upper Silesia; KO—Kaszuby; SU—Suwalszczyzna; VN—Velikii Novgorod; V—Volot [this study]; B—Pomerania-Kujawy; ST—Stavropol; O—Orel; SA—Saratov [4]. KL—Kaluga; PS—Pskov; VL—Vladimir; Y—Yaroslavl [23]. For better resolution, the population of Tula (T) characterized by extremely high frequency of haplogroup H [23] was excluded from analysis. Stress value = 0.0806.

populations. Northwestern Russians (Velikii Novgorod, Pskov) and northeastern Poles (Suwalszczyzna) are again grouped together and located in proximity to Latvians and Estonians in the second dimension of MDS (Figure S3).

3.4. Ashkenazi-specific mtDNA lineages in Polish populations

Recent study by Behar et al. [35] based on the analysis of complete mtDNA genomes showed that the most dominant Ashkenazi-specific mtDNA lineage was K1a1b1a. From a global set of 13,359 samples analyzed, the only non-Jewish K1a1b1a mtDNA lineage was found in a subject of unknown ethnicity from Western Ukraine. Interestingly, we have found K1a1b1a mtDNA lineages in both Poles and Polish Roma, previously studied by Malyarchuk et al. [19]. HVS I haplotype 16223–16224–16234–16311 (identical to that found in Western Ukrainian individual) has been revealed in two individuals from Pomerania-Gdansk and Upper Silesia populations (at frequencies of 0.6% and 1.1%, respectively, Table S2). Haplotype 16224–16234–16311 was found in 3 out of 69 Roma individuals studied (4.3%). Complete sequencing of K1a1b1a mtDNAs found in Polish populations (Fig. 2) shows that these genomes completely match with previously reported K1a1b1a mtDNAs found in Ashkenazi Jews [35]. In addition, these lineages were previously revealed in samples collected in the United States, in subjects of unknown ethnic or religious affiliations [36]. There are some phylogeographic peculiarities of the K1a1b1a mtDNA differentiation in Polish populations. Haplotype 16223–16224–16234–16311 is present in populations of southeast Poland—besides of Pomerania-Gdansk and Upper Silesia, it was also revealed in Lublin population (1%) [37]. Moreover, this haplotype appears to be the most frequent

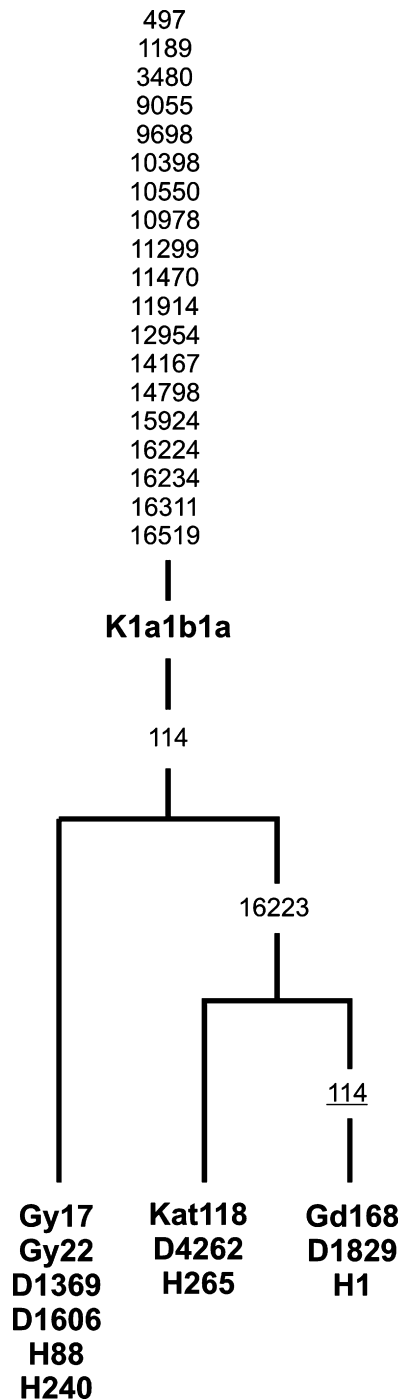


Fig. 2. Most parsimonious tree of the complete haplogroup K1a1b1a mtDNA sequences. The tree is rooted in hg U* and includes 12 mtDNAs, out of which four are novel and eight (designated as D and H) were reported in Fig. 1 of Behar et al. paper [35]. Mutations are shown on the branches and are transitions. Underlined nucleotide position indicates a back mutation. The hypervariable nucleotides (the indels) at nucleotides 309 and 315 in HVS II were excluded. The haplogroup K1a1b1a root sequence differs from the revised mtDNA reference Cambridge sequence [6] at sites: 73, 114, 263, 497, 750, 1189, 1438, 1811, 2706, 3480, 4769, 7028, 8860, 9055, 9698, 10398, 10550, **10978**, 11299, 11467, 11470, 11719, 11914, 12308, 12372, **12954**, 14167, 14766, 14798, 15326, 15924, 16224, 16234, 16311, and 16519 (K1a1b1a-specific mutations are shown in bold).

in Polish as well as German Jews (at frequencies of 16.4% and 18.8%, respectively) [38]. Meanwhile, haplotype 16224–16234–16311 found in the Polish Roma from western part of Poland was revealed at higher frequency in German and French Jews (18.8% and 17.4%, respectively) than in Polish Jews (7.3%) [38]. Therefore, this haplotype might have been introduced into Polish population during the major Roma immigration from Germany to Poland in the 16th century [39]. It is worth noting that in the middle of 17th century, Poland became the largest world center of Ashkenazi Jewry—half a million Jews living there constituted roughly five per cent of the total population of Poland and the Grand Duchy of Lithuania [40].

4. Conclusion

The results of this study show that there are several attributes of Slavic mtDNA pools seen on the level of some regional populations which help to understand complex interactions of the Eastern and Western Slavic populations with other European groups. The most important include the presence of sub-hgs U5b1b1, D5, Z1 and U8a with simultaneous scarcity of hg K in populations of northwestern Russia. The results of analyses of pairwise distances and SAMOVA suggest that Russians from Velikii Novgorod and Pskov areas and Poles from Suwalszczyzna differ from all remaining Polish and Russian samples. Accordingly, northwestern Russians and northeastern Poles bear some similarities to Baltic (Latvians) and Finno-Ugrian groups (Estonians) of northeastern Europe, especially on the level of U5 hg frequencies. Such picture can be explained by relatively recent (early medieval) contacts of Slavic tribes with their Finno-Ugrian and Baltic neighbors. Indeed, according to archeological and historical data, by the middle of 9th century Slavs took over the zone of the former Long Barrows Culture lying in areas of Finno-Ugrian hydronymy and this became territory of a tribe later known as the Pskov Krivichi, centered on the stronghold of Pskov [1]. The northernmost Slavic neighbors of Krivichi called themselves Slovienie. They found themselves in the areas of Finno-Ugrian and Baltic peoples (in the area of the Sopki mounds) and established stronghold Novgorod on the shores of Lake Ilmen [41]. Suwalszczyzna in turn witnessed a very long history of Baltic settlement (beginning from Jatviagian invasions in 5th and 6th centuries) [41] which can account for some genetic similarities of the present-day Polish populations from this area to Baltic-speaking groups.

The occurrence of K1a1b1a subcluster in Poles and Polish Roma seems to be one of the first direct proofs of the presence of Ashkenazi-specific mtDNA lineages in non-Jewish European populations. One may suggest that Ashkenazi maternal lineages were introduced at different times into gene pools of the Polish Roma and Poles.

One may also envisage that in the near future complete mitochondrial genome analyses will contribute more to our understanding of complex interactions between Slavic, Baltic and Finno-Ugrian groups of northeastern Europe.

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Appendix A. Supplementary data

Supplementary data associated with this article can be found, in the online version, at doi:10.1016/j.fsigen.2007.01.010.

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