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Reconstructing the phylogeny of African mitochondrial DNA lineages in Slavs

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To elucidate the origin of African-specific mtDNA lineages, revealed previously in Slavonic populations (at frequency of about 0.4%), we completely sequenced eight African genomes belonging to haplogroups L1b, L2a, L3b, L3d and M1 gathered from Russians, Czechs, Slovaks and Poles. Results of phylogeographic analysis suggest that at least part of the African mtDNA lineages found in Slavs (such as L1b, L3b1, L3d) appears to be of West African origin, testifying to an opportunity of their occurrence as a result of migrations to Eastern Europe through Iberia. However, a prehistoric introgression of African mtDNA lineages into Eastern Europe (approximately 10 000 years ago) seems to be probable only for European-specific subclade L2a1a, defined by coding region mutations at positions 6722 and 12903 and detected in Czechs and Slovaks. Further studies of the nature of African admixture in gene pools of Europeans require the essential enlargement of databases of African complete mitochondrial genomes.

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Introduction

It has been previously found that African mtDNA lineages represent about 1% of Eurasian mtDNAs.¹ Majority of them occurred in regions closest to North Africa – the Near East, Iberia and southern Europe.^{1–4} It has been suggested that African mtDNAs appeared in Europe mostly as a result of the Atlantic slave trade beginning in seventeenth century, with Portugal being the destination point playing predominant role in further dispersal.³ In addition, the medieval Arab/Berber conquest of Iberia might have also

contributed to this process.^{5,6} As for eastern Europe, it seems that only African children's traffic from Turkey to Russia at the end of seventeenth century might have played the leading role in African slave trade.⁷ African children were bought on the Ottoman markets by Russian traders who brought them to Russia. It is impossible to present any detailed data as no research has ever been made to determine the exact amount. However, it seems that the inflow of Africans was not significant.

Genetic studies have shown that African-specific lineages are very rare in eastern European populations. They have not been found in a large sample of indigenous eastern Europeans, including Mordva, Mari, Udmurt, Tatar, Chuvash, Komi, Bashkir, Karels, Estonians, Lithuanians and Latvians.^{8–10} Less than 0.5% of African mtDNAs belonging to different haplogroups (L1b, L3b and M1) were detected in Russians whose origin is thought to be the

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result of an admixture between the Slavs and indigenous pre-Slavonic tribes of eastern Europe that occurred in the early Middle Ages due to migrations of Slavs from their putative homeland in central Europe.^{11,12} It is noteworthy that African mtDNAs have been detected also in other Slavonic populations – in Poles, Czechs, Slovaks, Bosnians^{13–16} and Bulgarians.¹⁷ The precise ancestry of these mtDNAs is difficult to trace. Some African mtDNAs revealed in Europe are identical to those found in Africa; however some of them appear to be unique being found only in Europe.^{18,19} For this, several explanations can be proposed: the African mtDNAs detected in Europe are either present in Africa but not yet detected, or they are of African origin but have been lost among Africans, or their unique mutations have occurred exclusively in Europe. The question concerning the time of these unique mutations to originate appears to be of high importance.

In the previous studies, based on the hypervariable segment I (HVS I) variation, it has been proposed that the evolutionary age of two clusters of African mtDNAs (marked by unique mutations at 16362 within haplogroup L1b and 16294 within haplogroup L3b) do not exceed 6500 years.¹⁸ Whereas, another study has shown that unique cluster marked by the 16175 transition within haplogroup L1b has not been found in Africa but is present in Germany,¹⁷ Russia¹¹ and in medieval (twelfth–thirteenth century) population from south Iberia (Spain),¹⁹ suggesting that the ancestor of this motif arrived from Africa to Europe where the 16175 mutation occurred. The divergence time estimation for this cluster in Europe is around 20000 ± 16000 years, pointing to a prehistoric arrival of the basic African L1b-motif in Europe.¹⁹ Meanwhile, it is likely that complete mtDNA sequencing will considerably improve the phylogenetic resolution that allows us to determine the ancestry of certain European lineages belonging to the African mtDNA haplogroups with a higher accuracy. The present research was aimed at elucidating the origin of eight African-specific mtDNA haplotypes, revealed previously in different Slavonic populations: Russians, Czechs, Slovaks and Poles.

Materials and methods

In the present study, we completely sequenced eight African haplotypes of mtDNAs revealed previously in 2018 individuals from Slavonic populations of Eastern and Central Europe by means of mtDNA control region sequencing and coding region RFLP analysis (Table 1). All individuals with African variants of mtDNA identified themselves as ethnical Slavs (ie ‘indigenous’ Russians, Slovaks, Czechs or Poles) and were unaware of their African ancestry until molecular genetic studies.

Whole mitochondrial genomes were amplified and sequenced by means of the procedures described in Torroni *et al.*²¹ Sequencing reactions were run on Applied Biosystems 3130 Genetic Analyzer. Sequences were edited and aligned by SeqScape v. 2.5 software (Applied Biosystems, Foster City, CA, USA), and mutations were scored relative to the rCRS.²⁰ The eight complete mtDNA sequences have been submitted to GenBank (accession numbers EU200759–66).

For comparative purposes, a large number of published African and Eurasian mtDNA HVS I sequences and any available HVS II or RFLP data were used (see references^{1,18,19,22,23}). In addition, complete or nearly complete mtDNA sequences pooled in MitoMap mtDNA tree²⁴ were taken into analysis. The complete mtDNA tree was reconstructed manually and verified by using median-joining algorithm with NETWORK 4.1.0.9.²⁵ For phylogeny construction, the length variation in the poly-C stretches at nts 16180–16193 and 309–315 were not used. Haplogroup divergence estimates ρ and their standard errors were calculated as average number of substitutions in the mtDNA coding region from the ancestral sequence type.^{26,27} To estimate the time to the most recent common ancestor of each cluster, the evolutionary rate corresponding to 5140 years per substitution in the coding region was used.²⁸

Results and discussion

Haplogroup L1b lineage detected in Russian individual is characterized by the transition at position 16175 that is a

Table 1 mtDNA control region sequences belonging to African-specific haplogroups detected in Slavs

| Population (sample ID) | HG | HVS I sequence (–16000) | HVS II sequence | Frequency and references |
|------------------------|-----|--|------------------------------------|--------------------------|
| Russians (Tu_67) | L1b | 126 175 189 223 264 270 278 311 400 | 73 182 185GT 195 247 263 315iC 357 | 1/683 ^{11–13} |
| Slovaks (Slv_222) | L2a | 172 189 192 218 223 278 294 309 390 | 73 143 146 152 195 263 309iC 315iC | 1/207 ¹⁶ |
| Czechs (Cz_2972) | L2a | 189 218 223 247 278 294 309 390 | 73 143 152 195 263 309iC 315iC | 1/279 ¹⁵ |
| Slovaks (Slv_191) | L2a | 51 223 278 294 309 390 | 73 143 146 152 195 263 309iC 315iC | 1/207 ¹⁶ |
| Russians (Ka_69) | L3b | 124 223 278 294 362 | 59 73 152 217 263 315iC | 1/683 ^{11–13} |
| Poles (Pl_B_69) | L3d | 124 223 | 73 151 152 195 263 315iC | 1/849 ^{12,13} |
| Russians (Vo_6) | M1 | 129 182AC 183AC 189 249 311 | 73 195 263 309iC 315iC | 2/683 ^{11–13} |
| Russians (Vl_78) | M1 | 183AC 189 249 311 | 73 195 263 315iC | 1/683 ^{11–13} |

Abbreviation: HG, haplogroup; HVS, hypervariable segment.

Nucleotide positions numbers refer to transitions scored relative to the revised Cambridge reference sequence (rCRS).²⁰ Transversions are further specified, ‘i’ denotes an insertion of nucleotide. Unique mutations are shown in bold.

marker of 'European-specific' L1b-subcluster revealed also in Germany and medieval Spain (Table 1). Comparison of its complete genome sequence with the data presented in MitoMap mtDNA tree²⁴ shows that Russian sample belongs to L1b1a subcluster defined by mutations at positions 2768 and 5393 (Figure 1). This subcluster is widespread in Africa being found in 22 individuals presented in MitoMap mtDNA tree. A coalescence time estimate of subcluster L1b1a (calculated from the average sequence divergence and its standard error according to Sillard *et al*²⁷) corresponds to 8943 ± 1400 years, suggesting a relatively recent (post-Neolithic or later) arrival of the L1b1a lineage into Europe. Note that it is only the approximate lower time boundary of the actual arrival of this mtDNA lineage.

Haplogroup L2a samples were found in three individuals, two of whom are Slovaks and one comes from the Czech Republic. It is noteworthy that among them there is L2a branch defined by mutation at 16218 that is absent in published HVS I L2a data from Africa and Eurasia (Table 1). Complete genome sequencing demonstrates that these two individuals are characterized by two coding region mutations (at positions 6722 and 12903) and represent a branch that is absent among 50 L2a individuals collected together in MitoMap mtDNA tree (Figure 1). A coalescence time of this branch is estimated as 10280 ± 5140 years, though inferred only from two complete genomes. Slovak L2a1 haplotype characterized by transition at 16051 also seems to be very scarce. This control region sequence type has been recently detected in different parts of Eurasia only twice – in France³¹ and in Eastern Iran.³² Based on coding region variation data and comparison with MitoMap tree, this haplotype belongs to a specific subcluster defined by mutations at positions 3010 and 6663. Aforementioned subcluster seems to be ancient, with an estimated coalescence time of 24672 ± 3561 years for 10 mtDNA genomes analyzed.

Among Poles, the only African haplotype belonging to haplogroup L3d has been detected in population studies (Table 1). This haplotype contains a characteristic HVS I motif (16124–16223) of haplogroup L3d and complete mtDNA sequencing indicated that it belongs to the branch defined by mutation at position 6272 (Figure 1). This branch combines also two African individuals.^{30,33} The age of this subcluster is 15420 ± 5140 years.

There is also one more African mtDNA haplotype detected among Russians belonging to haplogroup L3b1. It is characterized by HVS I key mutation at position 16294 that is rare both in African and European populations being found only in one Saharawi individual from north-west Africa,³⁴ in two black individuals from Brazil²³ and in one Italian individual from Tuscany.³⁵ Complete genome sequencing demonstrates that Russian L3b sample belongs to a specific branch L3b1a defined by coding region mutations at positions 9079 and 15664 that were also

found in two African individuals.³⁶ The age of this subcluster is estimated as 20560 ± 5935 years.

It is interesting that haplogroup M1 lineages have been detected in two different Russian populations – in north-west region (two cases in Volot city) and in the Volga–Oka region (one case in Vladimir city). Haplogroup M1 is geographically restricted to East and North Africa, although it can be found at very low frequencies in populations from the Near East, the Caucasus and southern Europe.¹⁷ Meanwhile, M1 lineages revealed in Russians belong to a specific branch undergoing a back mutation at position 16223 (Table 1). Similar HVS I motifs have been detected previously in some African populations: in Chad,²² Egypt and Morocco.²⁹ Complete genome sequencing shows that all Russian M1 lineages, together with the Egyptian²⁹ and Spanish Basque³⁷ ones, appear to be a part of the subcluster M1a3b defined by coding region mutation at position 12414 (Figure 1). In addition, related M1a3a lineages have been also found in the Mediterranean area – in Italians and Berbers of Morocco.²⁹ The estimated age of subcluster M1a3b amounts 14145 ± 4262 years.

To sum up, it is striking that gene pools of different Slavonic populations include African-specific mtDNA lineages, some of which have their most related counterparts in Europe instead of Africa (such as haplotypes belonging to subclusters L1b1a and L2a1a). It is noteworthy that according to the results of phylogeographic studies, almost all African mtDNA lineages detected in the Slavs seem to be of West African origin. Only the haplogroup L2a is widespread within all regions of Africa, whereas haplogroups L1b, L3b1 and L3d are specific to West Africa.¹ In Northwest Africa, haplogroup M1 has been found at high frequencies in Algerians, and at lower frequency in Tunisians, Mozabites and Moroccan Arabs, showing a slight east–west cline.³⁴ Haplogroup M1 is rare in Iberia, but the presence of North African M1 mtDNA in the Basques remains that pre-date the Muslim invasion (eighth century) points to the prehistoric arrival of M1 lineages in Iberia.³⁸ Therefore, since most of the African lineages found in eastern European populations are present in West Africa, their migration to eastern Europe likely took them through Iberia.

In this respect, a possible explanation for presence of African mtDNA lineages in gene pools of eastern Europeans is that the Franco-Cantabrian refuge area of southwestern Europe might be the source of late glacial expansions leading to dispersal of some Northwest African mtDNAs in central and northeastern parts of Europe. It has been previously shown that ancient Iberian carriers of West Eurasian haplogroups H1, H3, V, U5b1b and U8a have participated in demographic reexpansion to repopulate Central Europe in the last interglacial periods (10000–15000 years ago).^{39–41} According to the data obtained in our study, it seems probable that Northwest Africans also contributed their mtDNA lineages to ancient Iberians, and

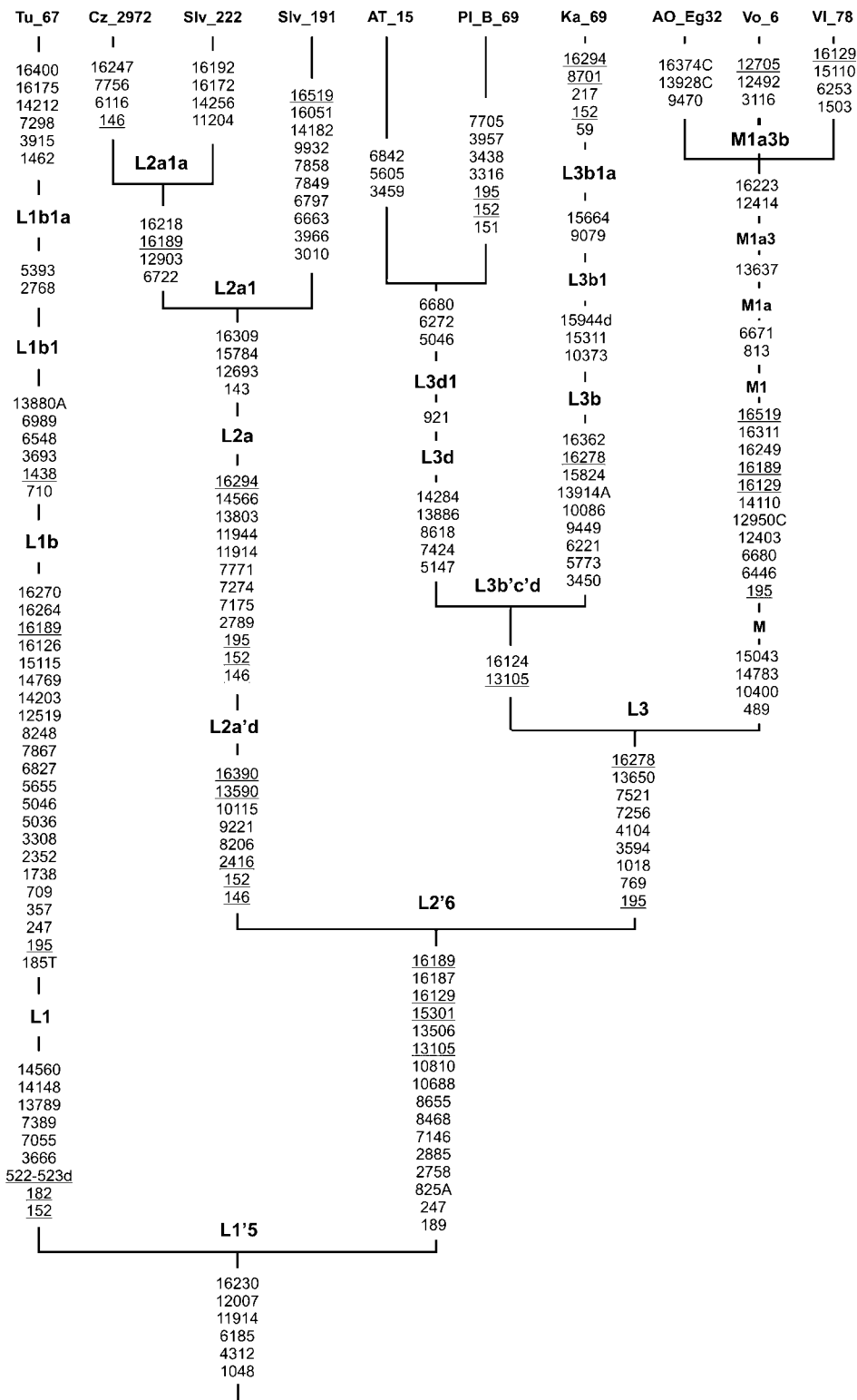


Figure 1 Complete genome-based phylogenetic tree of African-specific haplogroups found in Slavonic gene pools. The tree is rooted in macrohaplogroup L. Numbers along links refer to substitutions scored relative to the rCRS.²⁰ Transversions are further specified, recurrent mutations are underlined, 'd' denotes a deletion. Two additional complete sequences were taken from the literature^{29,30} and designated by AO and AT, respectively, followed by '_' and the original sample code.

further, via their gene pool migrations, to Europeans. Meanwhile, the strongest argument in favor of the hypothesis of the prehistoric introgression of African mtDNA lineages into the European gene pool would come from a finding of the appropriately dated European-specific subclades of the African haplogroups. However, the only two lineages that appear to represent European-specific subclades are L1b1a and L2a1a, but the lower boundary of about 9000 years for the potential introgression of L1b1a is not a strong argument for the presence of this lineage in the Franco-Cantabrian refuge area. Coalescence time estimate of $10\,280 \pm 5140$ years for European-specific subclade L2a1a is by a margin consistent with the post-Last Glacial Maximum expansion scenario. One should note, however, that absence of this subclade among Africans does not mean absence of this lineage in Africa in general due to relatively small sample size of African L2a genomes completely sequenced to date.

Therefore, the present data do not allow us to infer exactly when the African mtDNA lineages were introduced into gene pool of eastern Europeans. Both prehistoric and more recent gene flows (for instance, such as the potential Jewish L2a1 contribution⁴²) might have led to African mtDNA lineages admixture into Slavonic gene pools. More extensive studies of African populations based on complete mtDNA sequencing should provide more precise information about the genetic relationships between mitochondrial gene pools of Africans and Europeans.

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