

PHYLOGENETIC ANALYSIS OF PUTATIVE BALKAN SPECIFIC MTDNA LINEAGES



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Introduction: Available studies on human populations in Europe demonstrate that increased levels of genetic diversity are present in populations from the Balkan Peninsula. In this region, frequent admixture and differential introgression of various gene pools were rather common during the entire history of humans. Furthermore, it has been shown recently that complex demographic history of human populations from the Balkans was characterized also by microdifferentiation of particular mitochondrial DNA (mtDNA) lineages that are potentially specific for this region.

Methodology: In order to provide new insights into the presence and abundance of putative Balkan's mtDNA lineages we completely sequenced 164 mitogenomes from the Serbian population which occupies the central part of the Balkan Peninsula. We analyzed them phylogenetically against available complete mtDNAs of modern and ancient Western Eurasians. Coalescence ages of subclades were estimated using the mutation rate for the entire mtDNA molecule.

Results: Detailed analysis of complete mtDNAs found in Serbian population allowed us to describe several new mtDNA lineages and to delineate those that are potentially specific for the Balkan Peninsula. Along with K1a13a1 and X2q1, which have been identified previously as putative Balkan's lineages, we detected additional lineages potentially specific to this region - H6a2b, H7j, K1a4l, T1a1l, U1a1c2, U4c1b1, U5b3j and W3b4. The sample sizes of H6a2b, U1a1c2, U4c1b1, U5b3j and K1a4l, however, were too small, and thus, further analyses, with increased sample sizes, are required to support their Balkan's origin. Most the detected mtDNA lineages occur exclusively in south-Slavic populations of the Balkan Peninsula with their coalescence age estimates ranging from 0.9 to 3.9 kya. This suggests that some of these lineages most likely emerged after the settlement of Slavs in the Balkan Peninsula while the older ones probably evolved in indigenous populations which were present in this region prior to the Slavic settlement.

Conclusion: The obtained results further support the view that microdifferentiation of mtDNA lineages was present in the Balkans over time, and that further studies, with increased sample of complete mitogenomes from this region, are required to shed more light on evolutionary processes that shaped contemporary genetic landscape of Balkan's populations.

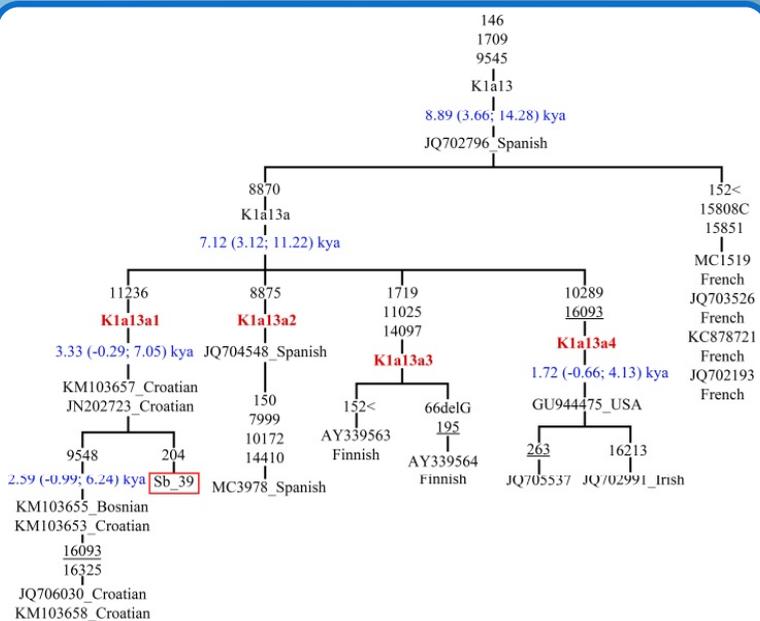


FIGURE 1. The phylogenetic tree of sub-haplogroup K1a13 with Serbian haplotype based on complete mitochondrial genome sequences. Several new subclades belonging to haplogroup K1a13 (K1a13a1, K1a13a2, K1a13a3 and K1a13a4) were delineated. Lineage K1a13a1, dated to 2.6–3.3 kya, is exclusively found in South-Slavic-speaking populations from the Central Balkans. Thus, it may have originated within the Balkan Peninsula and may represent a genetic fingerprint of Iron Age Balkan populations.

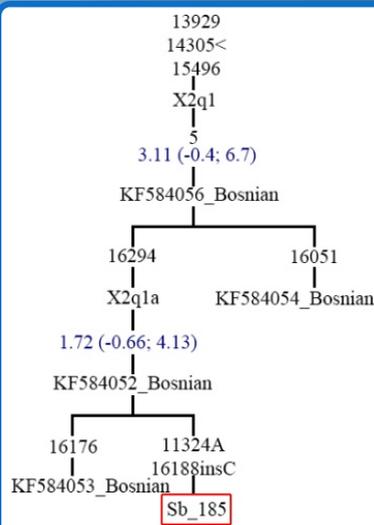


FIGURE 2. The phylogenetic tree of sub-haplogroup X2q1 with Serbian haplotype based on complete mitochondrial genome sequences. It is proposed that X2q1 lineages are southeast European variants of the X2 clade and that they are result of one or more local microdifferentiation processes that have occurred in the Balkan Peninsula.

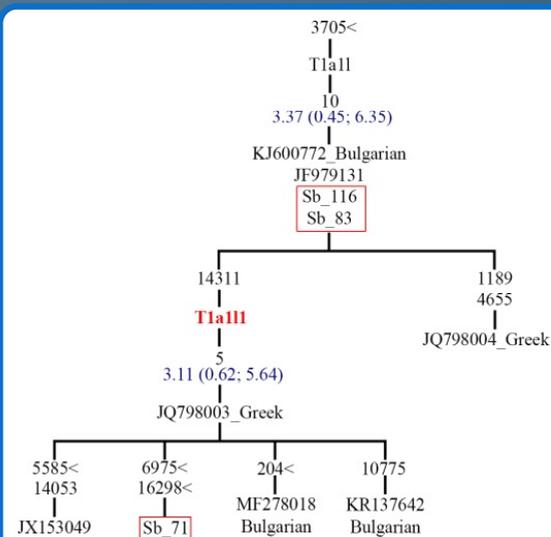


FIGURE 3. The phylogenetic tree of sub-haplogroup T1a1l with Serbian haplotypes based on complete mitochondrial genome sequences. New sub-clade T1a1lI belonging to sub-haplogroup T1a1l was defined. Sub-haplogroup T1a1lI is exclusively found in the populations of Balkan Peninsula (Serbians, Greeks and Bulgarians). Thus it is possible that this sub-clade is Balkan specific which is in accordance with previous observations that have identified southeast Europe as a place from which some of the T sub-clades have expanded to central Europe and Mediterranean during Neolithic.

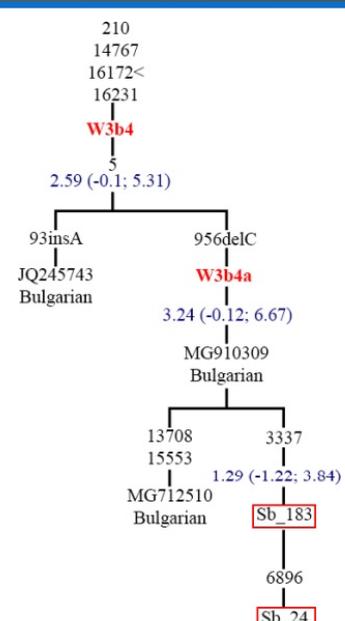


FIGURE 4. The phylogenetic tree of sub-haplogroup W3b4 with Serbian haplotypes based on complete mitochondrial genome sequences. We have defined new sub-haplogroup W3b4 and its sub-branch W3b4a. This sub-haplogroup encompasses haplotypes found in Serbian and Bulgarian populations.