Mitochondrial Genome Diversity in the Tubalar, Even, and Ulchi: Contribution to Prehistory of Native Siberians and Their Affinities to Native Americans

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ABSTRACT To fill remaining gaps in mitochondrial DNA diversity in the least surveyed eastern and western flanks of Siberia, 391 mtDNA samples (144 Tubalar from Altai, 87 Even from northeastern Siberia, and 160 Ulchi from the Russian Far East) were characterized via highresolution restriction fragment length polymorphism/single nucleotide polymorphisms analysis. The subhaplogroup structure was extended through complete sequencing of 67 mtDNA samples selected from these and other related native Siberians. Specifically, we have focused on the evolutionary histories of the derivatives of M and N haplogroups, putatively reflecting different phases of settling Siberia by early modern humans. Population history and phylogeography of the resulting mtDNA genomes, combined with those from previously

Studies on present day world populations, especially those based on maternally inherited mitochondrial DNA (mtDNA), suggest that modern humans expanded from East Africa around 70 kya (1000 years ago). They dispersed along the Southwest-Southeast Asia shore and reached the Siberian Pacific \sim 38-37 kya (reviewed by Pope and Terrel, 2007). In the other direction, archaic cultures, including Neanderthal and previously unknown hominin, who lived in Denisova Cave in Altai, southwestern Siberia, around 40 kya, were replaced by modern humans no later than 30 kya (Krause et al., 2007, 2010b; Derevianko, 2011). During the last Ice Age (43-12 kya), a special feature of Siberia has been its relative isolation from the rest of Eurasia and the New World, making it a case study for understanding the main pattern of cultural and biological adaptation and subsequent dispersions to the New World. At the height of last glacial maximum (LGM), the earliest Siberians were largely confined to their strongholds, south of the 56°N parallel, which were areas of continuous occupation (Finlanson and Cartion, 2007; Kuzmin, 2008; Graf, 2009).

Current distribution of maternal lineages across the world indicates that Siberia lacks autochthonous M and N lineages, the Eurasian descendants of African L3 that are abundant along the proposed southern coastal migration route (reviewed by Forster and Matsumura, 2005; Mellars, 2006). Many phylogenies across Siberia/ Beringia based on complete mtDNA sequences were reconstructed and sequence-divergence estimates were obtained (Derbeneva et al., 2002a; Starikovskaya et al., 2005; Derenko et al., 2007; Perego et al., 2010; Volodko published data sets, revealed a wide range of tribal- and region-specific mtDNA haplotypes that emerged or diversified in Siberia before or after the last glacial maximum, ~18 kya. Spatial distribution and ages of the "east" and "west" Eurasian mtDNA haploclusters suggest that anatomically modern humans that originally colonized Altai derived from macrohaplogroup N and came from Southwest Asia around 38,000 years ago. The derivatives of macrohaplogroup M, which largely emerged or diversified within the Russian Far East, came along with subsequent migrations to West Siberia millennia later. The last glacial maximum played a critical role in the timing and character of the settlement of the Siberian subcontinent. Am J Phys Anthropol 148:123–138, 2012. \odot 2012 Wiley Periodicals, Inc.

et al., 2008). However, it is still not quite clear where in the southern extent of Siberia the M and N offshoots arose, when and how they spread over the higher latitudes, setting the stage for colonization of the Americas. As these and related studies have progressed, the Altai-Sayan Mountain system, represented by the Tubalar, immediate descendants of autochthonous hunters and gatherers, emerged as a strategic area of very high relevance to these issues. However, lack of entirely sequenced mtDNA data precluded elucidation of Altaic prehistory, potentially preserved in the intrinsic diversity of mtDNA lineages. This is also true for the genetic history of the Tungusic world (Pakendorf et al., 2007), in East Siberia represented largely by the Evenki and Even, the sparse groups of nomadic reindeer herders and hunters, and the Ulchi, in recent traditional times a

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