

Ancient Siberian genome reveals genetic origins of Native Americans

November 20 2013



Burial of Mal'ta child redrawn from Gerasimov (1935), with photos of the plaque and swan from the burial and a representative Venus figurine from the excavation. Credit: Kelly E Graf

The genome sequence of a 24,000-year-old Siberian individual has

provided a key piece of the puzzle in the quest for Native American origins. The ancient Siberian demonstrates genomic signatures that are basal to present-day western Eurasians and close to modern Native Americans. This surprising finding has great consequences for our understanding of how and from where ancestral Native Americans descended, and also of the genetic landscape of Eurasia 24,000 years ago. The breakthrough is reported in this week's *Nature* (Advance Online Publication) by an international team of scientists, led by the Centre for GeoGenetics at the Natural History Museum of Denmark (University of Copenhagen).

The search for Native American ancestors has been focused in northeastern Eurasia. In late 2009, researchers sampled at the Hermitage Museum, St. Petersburg the remains of a juvenile individual (MA-1) from the Upper Palaeolithic site of Mal'ta in south-central Siberia. The MA-1 individual dated to approximately 24,000 years ago. Now, the team reports genomic results from the MA-1 individual which unravel the origins of the First Americans – ancestors of modern-day Native Americans.

"Representing the oldest anatomically modern human [genome](#) reported thus far, the MA-1 individual has provided us with a unique window into the genetic landscape of Siberia some 24,000 years ago", says Dr. Maanasa Raghavan from the Centre for GeoGenetics and one of the lead authors of the study. "Interestingly, the MA-1 individual shows little to no genetic affinity to modern populations from the region from where he originated - south Siberia."

Instead, both the mitochondrial and nuclear genomes of MA-1 indicate that he was related to modern-day western Eurasians. This result paints a picture of Eurasia 24,000 years ago which is quite different from the present-day context. The genome of MA-1 indicates that prehistoric populations related to modern western Eurasians occupied a wider

geographical range into northeast Eurasia than they do today.

Dual ancestry of Native Americans

The most significant finding that the MA-1 genome reveals is its relation to modern Native Americans. This relative of present-day western Eurasians shows close affinity to modern Native Americans, but surprisingly not to East Asians who are regarded as being genetically closely related to Native Americans.



Cross section through the Mal'ta humerus. The central void is the medullary cavity. Credit: Thomas W Stafford, Jr

Furthermore, the team finds evidence that this genetic affinity between MA-1 and Native Americans is mediated by a gene flow event from MA-1 into the First Americans, which can explain between 14-38% of

the ancestry of modern Native Americans, with the remainder of the ancestry being derived from East Asians. Supported by numerous reasons against these signatures being caused by contamination from modern DNA sources or from post-Columbian admixture (post 1492 AD), the study concludes that two distinct Old World populations led to the formation of the First American gene pool: one related to modern-day East Asians, and the other a Siberian Upper Palaeolithic population related to modern-day western Eurasians.

"The result came as a complete surprise to us. Who would have thought that present-day Native Americans, who we learned in school derive from East Asians, share recent evolutionary history with contemporary western Eurasians? Even more intriguingly, this happened by gene flow from an ancient population that is so far represented only by the MA-1 individual living some 24,000 years ago", says Professor Eske Willerslev from the Centre for GeoGenetics who led the study.

Early cosmopolitans

Additionally, results from a second south-central Siberian from Afontova Gora-2 site are presented in order to address human occupation of the region during and after the Last Glacial Maximum (LGM; ca. 26,000 to 19,000 years ago), a climatically cold period when glacial ice sheets extended to their maximum range. At approximately 17,000 years ago, this post-LGM individual demonstrates similar genomic signatures as MA-1, with close affinity to modern western Eurasians and Native Americans and none to present-day East Asians. This result indicates that genetic continuity persisted in south-central Siberia throughout this climatically harsh period, which is a significant consideration for the peopling of Beringia, and eventually the Americas some 15,000 years ago.



These are the remains of the 24,000-year-old Mal'ta boy. Credit: State Hermitage Museum in Russia

Dr. Pontus Skoglund from Uppsala University, and one of the lead authors of the study, explains, "Most scientists have believed that Native American lineages go back about 14,000 years ago, when the first people crossed Beringia into the New World. Our results provide direct evidence that some of the ancestry that characterizes Native Americans is at least 10,000 years older than that, and was already present in Siberia before the last Ice Age."

Professor Kelly Graf from the Center for the Study of the First Americans (Texas A&M University), who together with Professor Willerslev did the sampling, adds, "Our findings are significant at two levels. First, it shows that Upper Paleolithic Siberians came from a cosmopolitan population of early modern humans that spread out of Africa to Europe and Central and South Asia. Second, Paleoindian

skeletons with phenotypic traits atypical of modern-day Native Americans can be explained as having a direct historical connection to Upper Paleolithic Siberia."

As such, results from this study contribute a major leap forward for resolving the peopling of the Americas.

Take-home messages:

- First Americans descended from the meeting and admixture of at least two populations, of which one is related to contemporary East Asians and the other to present-day western Eurasians.
- These findings may explain the presence of mitochondrial lineage X in Native Americans.
- The presence of a population related to western Eurasians further into northeast Eurasia provides a more likely explanation for the presence of non-East Asian cranial characteristics in the First Americans, rather than the Solutrean hypothesis that proposes an Atlantic route from Iberia.
- Genetic continuity in south-central Siberia before and after the LGM provides evidence for the presence of humans in the region throughout this cold phase, which is of consequence to population movements into Beringia and ultimately the Americas around 15,000 years ago.

More information: [dx.doi.org/10.1038/nature12736](https://doi.org/10.1038/nature12736)

Provided by University of Copenhagen

Citation: Ancient Siberian genome reveals genetic origins of Native Americans (2013, November 20) retrieved 28 March 2023 from <https://phys.org/news/2013-11-ancient-siberian-genome->

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