DNA links Native Americans with Europeans

A Danish-led international research team has mapped the hitherto oldest genome of an anatomically modern human: the genome of a boy buried at Mal’ta near Lake Baikal in south-central Siberia some 24,000 years ago.

Surprisingly, the genetic material reveals that the boy was European, which means that a European culture reached all the way east to Lake Baikal.

The really sensational news, however, is that a large proportion (about a third) of all living Native Americans are descendants of the Mal’ta people. In other words, Native Americans have partly European ancestry.

"This is incredibly surprising. At first I didn’t believe it," says team leader Professor Eske Willerslev of the University of Copenhagen.

‘Jaw-dropping’ discovery

The results reveal that Native Americans are a mixture between Western Europeans who reached Siberia and an East Asian population. This paints a new picture of Native Americans and at the same time solves a number of puzzles regarding the colonisation of America.

"For American archaeology, this is a really, really big thing,” says Willerslev.

The study has just been published in the journal Nature; however, the results started to leak out about a month ago, after Willerslev spoke about the discovery at a conference in the US.

Here, geneticist Connie Muligan of the University of Gainesville described the discovery as ‘jaw-dropping’.

The colonisation of America has for decades been a hotly debated topic among researchers, with one of the big questions being who the first Americans were and where they came from.

Very few skeletons from the right time and location

Genetic analyses can help find the answers in two ways:

- Either by mapping the genetic traces of living people and reconstructing how our geographical distribution may have taken place.
- Or by going directly to the genetic material from prehistoric remains to identify their relations.
The first method has the advantage that it is easy to get hold of samples, but the analysis is more complex. The second method provides definitive answers, but very few skeletons from the right place and location are available.

It was one of these skeletons that in 2009 prompted Willerslev to go to Saint Petersburg in Russia together with American archaeologist Kelly Graf. They wanted to find out who the first Americans were, and they were familiar with the find in Mal’ta. If they were lucky, the boy skeleton (named MA-1) could be an individual from the tribal community that wandered into America across the Bering Strait some 16,000 years ago.

"It was a bit of a long shot, but the age was just right," says Willerslev.

**Sequencing the Siberian genome**

He remembers how a Russian archaeologist, Svetlana Demeshchenko, opened up the door to a huge building complex, which was originally the Tzar’s palace but which today is the famous Hermitage Museum.

Demeshchenko's office was located far away from the polished floors of the Tzar’s stately halls in a tumbledown part of the building where the walls were covered with faded posters from old archaeological expeditions. She found a small wooden box containing bones from the boy, and after a few days Willerslev was allowed to take samples from the upper arm bone.

He also took samples from the femur of another, roughly 17,000-year-old skeleton excavated in Afonova Gora in the same region.

Back in the US and in Denmark, the researchers confirmed the dating using modern technology and started sequencing the genetic material.

The professor was disappointed at first because the preliminary examination revealed that the mitochondrial DNA, which is only inherited in the female line, had a distinctively European profile known as haplotype U.

"I thought, 'This can’t be right. Surely there must have been some contamination by archaeologists who have been in contact with the bones';" he says.

They had expected to find an east-Asian haplotype, as studies have shown that 97 percent of living Native Americans have one of four mitochondrial haplotypes called A, B, C and D, which outside of America are found in eastern Asia. (The remaining 3 percent is the mysterious exception known as haplotype X, which we shall get back to).

**The project was resumed instantly**

To make a long story short, the project was put on low speed for more than a year, until the study’s first author, Maanasa Raghavan, also from the University of Copenhagen, sequenced more genetic material and suddenly could see details of the boy’s sex chromosome Y. These details revealed a very old and basal lineage dating back to before the Y chromosomes of living Europeans and West Asians.

This prompted the researchers to take another look at the mitochondrial haplotype, and it soon became clear that they had found a special haplotype U, which is closest to what is found in the first hunter-gatherers in Europe.

Having sequenced the Siberian genome, the oldest human genome sequenced to date, the researchers now had enough material to start analysing kinship.
It is clear that the boy is of the same lineage as living Europeans, and the archaeological finds, which include Venus figurines, thus represent a culture that has been far more extensive than previously assumed.

**Europeans and East Asian start to mingle**

However, the big breakthrough didn’t come until Pontus Skoglund, a bioinformatician from Uppsala University in Sweden, revealed in his analyses a close connection to American Indians, but none to East Asians.

According to the researchers’ calculations, 14-39 percent of the Native American genetic material comes from Mal’ta.

"That really is a lot,” says Willerslev. “It shows us that Europeans and East Asians met and had lots of sex, and that’s what created the Native Americans.”

**Two branches of modern man**

The analyses show that Native Americans carry about one-third European genes and two-thirds East Asian. This reveals a meeting between two branches of modern man: one branch that followed the east coast of Asia, and one that travelled east from Europe to the steppes of Asia.

The researchers cannot at this point say with any certainty exactly where the two branches covered, but they estimate that they met after the East-Asian lineage split into distinct groups in the high northeast at the gate to America in the vast land area between Siberia and Alaska, known as Beringia. Here, it is conceivable that there has been some sort of a nesting box from which various genetic lineages of Native Americans originate.

The discovery also shows that the European traces that have so far been explained as a mixture between Indians and Europeans after Columbus discovered America in 1492 goes much further back in history. It also provides a logical explanations to many archaeological finds that have puzzled the researchers.

**Head shape more similar to Eastern Europeans than East Asians**

Many skulls from the earliest American Indians, such as the 9,500-year old Kennewick Man, have a head shape that is more similar to that of Eastern Europeans than East Asians.

And then there is the so-called haplotype X mystery, where some tribes of Native Americans today carry a large proportion of the mitochondrial lineage X, which is otherwise only known from Europe. This means that there is a big hole in Asia with no haplotype X, which has made it difficult to reconcile it with the idea that the Native Americans’ ancestors wandered in from Asia.

Together with the discovery of some stone points that resemble spikes that are only known from the Solutré region of France, these puzzles have given rise to some pretty wild theories that the first Americans actually reached America by crossing the Atlantic Ocean.

The Mal’ta find now puts an end to such speculation, and it appears that the researchers can now say with certainty that man entered America through Asia.

**Primitive hunter-gatherers may have survived the Ice Age**

Up to now, most researchers have agreed that the road to America was closed, so to speak, during the Ice
Age, and that the Beringia gate did not open up until the huge ice sheets started to retreat.

However, the other 17,000-year-old skeleton from Afontova Gora may overturn this view. It turns out that it has the exact same genetic lineage as the MA-1. In other words, this area is very likely to have been inhabited by the same people throughout the period.

This may not sound like a big deal, but some 20,000 years ago the Earth underwent the harshest period of the last Ice Age, and no-one has thought that primitive hunter-gatherers could have survived the cold Siberian temperatures that far north. The Beringia is situated even further north than this, but in 2004 researchers found 30,000-year-old spears and stone tools from hunter-gatherers up by the Yana Rivers at a latitude of 65 degrees north.

So although the evidence is inconclusive, there is increasing evidence that man arrived in America much earlier than 16,000 years ago, as previously thought.

"There are no particularly good arguments to prove that the first humans should not have reached America earlier, even much earlier," says Eske Willerslev.

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Read the Danish version of this article at videnskab.dk [8]

Lake Baikal in south-central Siberia, where the village of Mal’ta is situated. The genome of the MA-1 revealed that an Upper Palaeolithic population from this region admixed with ancestors of present-day East Asians, giving rise to the First American gene pool. (Photo: Niobe Thompson) [9]

Burial of the MA-1 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. (Photo: Kelly E. Graf) [10]

A cross section through the MA-1 individual’s humerus. The central void is the medullary cavity. (Photo: Thomas W. Stafford, Jr.) [11]

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Eske Willerslev's profile [13]

Rasmus Kragh Jakobsen [14]

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