Paleontologists use genomics to delve into the lives of ancient humans

July 7 2015, by Kyle Frischkorn, Earth Institute, Columbia University

The ancient Siberian femur bone on the lab bench. Credit: Bence Viola, MPI EVA

Like an immutable passport, birth certificate and social security number, our DNA is a marker of who we are and where we've come from.

Recently, a spate of studies by paleontologists have used genomics to delve into the lives of ancient humans. These studies have capitalized on futuristic next generation techniques to reveal the genealogy, travel plans and sex lives of our ancestors.

Unlocking the invisible messages in the code of DNA, this research has unveiled otherwise unattainable secrets of human origins.

The Case of the Kennewick Man

The story of the Kennewick Man opens like a scene out of a crime drama. It's the summer of 1996 and two teenagers skulk through the Columbia River in Kennewick, Washington. While attempting to break into a race track, they glance down and find the hollow eyes of a skull staring back.

These human remains were not a murder victim, but rather the bones of a man who died during the Holocene, approximately 11,000 years ago. The recovery of the so-named Kennewick Man incited a decades-long legal battle. On one side were Native American tribes who lived in the region and sought to perform burial rites on the remains. On the other side, researchers vied to keep the skeleton as a specimen used for scientific study.

The dispute was complicated by the Kennewick Man's bone structure. Forensic anthropologists asserted that though it was geographically confounding, his anatomy did not resemble Native Americans, but ancient Polynesians instead.

How could a case like this be solved with no witnesses and no evidence other than the bones of a man dead for thousands of years? The only hope was that some DNA remained in the Kennewick Man's bones.

The researchers were in luck. "We got a very, very tiny sample," said Eske Willerslev, a paleontologist based at the Natural History Museum of Copenhagen. "We sampled it to complete exhaustion—there's nothing left." That faint dusting of bone was enough, however. Willerslev is corresponding author on a recent *Nature* publication that showcases the Kennewick Man's genome. The genomic analyses present in the paper put to rest the dispute over this ancient man's heritage.

Based on sequence comparison to databases of human genomes, Polynesian ancestry has been ruled out. David Meltzer, a researcher at Southern Methodist University and co-author on the paper said, "I'm quite certain we've made the case that Kennewick was Native American." In fact, using the DNA sequence like a molecular clock, the researchers posit the Kennewick Man's genome has continuity with Native American lineages for eight millennia.

For now, these results do not undermine a court decision in 2004 to let the remains stay with scientists, rather than being returned to the region's tribes. Douglas Owsley, a researcher at the Smithsonian Institution in Washington DC, cautions about reading too much into the conclusions of DNA testing. While the results are an important advancement and underscore that morphological classification alone is insufficient, they are far from definitive. In an interview with the Guardian, he explained how certain aspects of an individual are not reflected in the sequence of their genome. He said, "We don't know anything about his culture. We don't know who his people are."

Naia's Bones and the Origin of Paleoamericans

The incongruity between anatomy and genome-enabled heredity seen in the Kennewick Man calls into question what scientists know about how the American continents were peopled. Deep in an underwater cave in the Yucatan lie the 12,000 year old remains of a teenage girl that could help solve this conundrum.

Paleontologists have nicknamed this skeleton Naia. Like the Kennewick Man, Naia's bone structure is inconsistent with patterns seen in Native Americans. Even though Naia had been in a submerged cave for millennia, scientists were still able to recover a DNA sample. Using part of her genome, they have formulated a hypothesis for why the bones of Paleoamericans seem out of place. These results were published in

Science last May.

Previous genetic research supported the theory that ancient humans trekked across the Bering land bridge to populate what would become present day America. Morphometric analyses of the oldest Paleoamerican remains like Naia and the Kennewick Man cast doubt on this theory, as they do not fit with modern Native Americans or Siberians.

The genome showed that though Naia looks different, key genetic signatures indicate she is related to the group who once crossed the land over the Bering Strait.

Naia's DNA goes a long way towards bolstering confidence in the origins of Native Americans. "It helps support the consensus view, from archaeological, genetic and linguistic evidence, that the Americas were initially peopled 15,000–20,000 years ago from Siberia," said the Wellcome Trust Sanger Institute's Chris Tyler-Smith.

In the publication, the authors argue that the confounding bone structures seen in ancient remains are likely due to changes brought about by isolated evolutionary events rather than different ancestry.

Unzipping Our Genes: The Illicit History of Neanderthals and Humans

Genomes are an intrinsic family tree, passing down genealogical information from generation to generation. When this genomic evidence comes into play, there are few skeletons that can be kept in the closet. The results of a *Nature* study published in October of last year provide compelling new evidence about illicit intermingling between ancient humans and Neanderthals.

The corresponding author on the *Nature* study, Svante Paabo of the Max Planck Institute for Evolutionary Anthropology, recently discussed the findings on NPR Science Friday.

The story begins in 2008 when a human femur bone was found sticking out of alluvial deposits on the banks of the Irtysh River in Siberia. Isotopic dating and anatomy indicated the femur belonged to a man who died 45,000 years ago. The ancient owner of this femur was not given a catchy name like the Kennewick Man or Naia, perhaps because the rest of him was never recovered. Miraculously, however, the DNA was so well-preserved that just shy of a gram of his bone yielded enough DNA to assemble his genome.

Approximately 2% of the modern human genome is Neanderthal in origin. Like lingering evidence of ancient affairs between species, the Neanderthal-like DNA is distributed in small chunks throughout human chromosomes. Previously, what remained unclear was when this interbreeding took place. Existing evidence was only able to narrow the window to between approximately 30,000 and 80,000 years ago.

Within the a genome sequence from the 45,000 year old bone were found long stretches of Neanderthal-like regions–about 4 times longer than those found in modern human genomes.

Based on this data, Paabo and colleagues estimate that the admixture between Neanderthals and humans occurred 300 generations before the owner of the femur bone was born. Based on that, they narrow down the time window of interbreeding to between 50,000 and 60,000 years ago.

This corresponds to the era when humans were radiating out of Africa and the Middle East, a time when Homo sapiens were beating out Neanderthals as the dominant hominids.

A seemingly futuristic technique unattainable until just a few years ago, DNA sequencing on ancestral genomes offers a window into our past. Future applications of this technology to anthropology are limited only by the imaginations of paleontologists, and of course the ancient remains lying in wait to be discovered.

Provided by Earth Institute, Columbia University

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