Study of mitochondrial DNA ties ancient remains to living descendants

July 3 2013

University of Illinois anthropology professor Ripan S. Malhi led an analysis that links ancient and present-day Native Americans in British Columbia, Canada. Credit: L. Brian Stauffer

Researchers report that they have found a direct genetic link between the remains of Native Americans who lived thousands of years ago and their living descendants. The team used mitochondrial DNA, which children inherit only from their mothers, to track three maternal lineages from

ancient times to the present.

The findings are reported in the journal *PLOS ONE*.

The researchers compared the complete mitochondrial genomes of four ancient and three living individuals from the north coast of British Columbia, Canada. This region is home to the indigenous Tsimshian, Haida and Nisga'a people, all of whom have oral traditions and some written histories indicating that they have lived in the region for uncounted generations. Archaeological sites, some with human remains, date back several millennia. But until the current study, nothing definitively tied the current inhabitants of the area to the ancient human remains found there, some of which are 5,000 to 6,000 years old.

"Having a DNA link showing direct maternal ancestry dating back at least 5,000 years is huge as far as helping the Metlakatla prove that this territory was theirs over the millennia," said Barbara Petzelt, an author and participant in the study and liaison to the Tsimshian-speaking Metlakatla community, one of the First Nations groups that participated in the study.

Only one previous study of ancient remains found in the Americas – the hair of an Eskimo man who lived in Greenland 3,400 to 4,500 years ago – analyzed all of the ancient mitochondrial DNA sequences (the mitochondrial genome, or mitogenome). Most such studies look at only a small subset (less than 2 percent) of mitogenomic sequences.

The new analysis was made possible by technological advances that cut the cost and complexity of sequencing ancient DNA.

"This is the beginning of the golden era for ancient DNA research because we can do so much now that we couldn't do a few years ago because of advances in sequencing technologies," said University of

Illinois anthropology and Institute for Genomic Biology professor Ripan Malhi, who led the analysis. "We're just starting to get an idea of the mitogenomic diversity in the Americas, in the living individuals as well as the ancient individuals."

Focusing on the mitogenome is a good way to study the evolutionary history of these groups, Malhi said. DNA is often degraded in ancient remains, and unlike nuclear DNA, which is present in only two copies per cell, mitochondria are abundant in cells, giving researchers many DNA duplicates to sequence and compare.

Mitochondrial DNA does not recombine with paternal DNA and is passed down from mother to offspring more or less intact. This makes it easier to track unique sequences through the generations. And since the participants in the study belong to matrilineal cultures, their oral histories can be compared to evidence from the maternal lineages reflected in the mitogenome.

Another complication associated with analyzing nuclear DNA in Native Americans involves the European influence, Malhi said.

"There's a pattern of European males mixing with Native American females after European contact and so lots of the Y chromosomes in the community trace back to Europe," he said. The mitogenome offers a clearer picture of Native American lineages before European contact, he said.

Scientists, including David Archer, an anthropology professor at Northwest Community College in Prince Rupert, and biological anthropologist and co-author Jerome Cybulski, of the Canadian Museum of Civilization in Gatineau, Quebec, have worked since the 1960s to recover and analyze human remains and ancient structures in the region around Prince Rupert.

One particularly fruitful site, the Lucy Islands, yielded human remains and evidence of a house structure that is 5,300 to 6,400 years old. Careful study revealed that one of the ancient individuals found there, a young adult female, lived roughly 5,500 years ago. Her mitogenome matched that of another female found on Dodge Island, near the city of Prince Rupert. Her remains were about 2,500 years old. A living participant in the study also carried the same mitogenomic signature, the researchers found, and so is a direct maternal descendant of those two women or their mothers.

Three other living participants host a different mitogenomic sequence that matches another individual found on Dodge Island who lived about 5,000 years ago.

The archaeological sites also point to a long period of continuous occupation and stability in terms of the types of structures people used and the ways they used them, Archer said.

"Archaeology is one important source of information about the past, and oral traditions give us a lot of verifiable information about the past cultural events and patterns," he said. "But the genetic information is something that is immediately recognizable. If somebody is told that their DNA links to somebody who was present 2,500 years ago and also to someone who was present 5,500 years ago, you can summarize that in a sentence and it's very easily understood and it's exciting."

The new findings were the result of an unusual effort by the researchers to enlist the help of the indigenous people they study.

"I believe this is really a unique collaboration," said Joycelynn Mitchell, a Metakatla co-author and participant in the study. "It's very exciting to be able to have scientific proof that corroborates what our ancestors have been telling us for generations. It's very amazing how fast

technology is moving to be able to prove this kind of link with our past."

More information: "Ancient DNA Analysis of Mid-Holocene Individuals From the Northwest Coast of North America Reveals Different Evolutionary Paths for Mitogenomes," *PLOS ONE*, 2013.

Provided by University of Illinois at Urbana-Champaign