

Past and present genomes tell the story of Native American biological origins

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The Lucy Islands in British Columbia, Canada were the discovery site for skeletal remains over 6,000 years old . Pictured here is an ancient shell midden found on the islands.

The first human inhabitants of the Americas lived in a time thousands of years before the first written records, and the story of their transcontinental migration is the subject of ongoing debate and active

research. A study by multi-institutional, international collaboration of researchers, published this week in *Science* presents strong evidence, gleaned from ancient and modern DNA samples, that the ancestry of all Native Americans can be traced back to a single migration event, with subsequent gene flow between some groups and populations that are currently located in East Asia and Australia.

The study was led by the Centre for GeoGenetics at the University of Copenhagen; more than 80 researchers contributed sequence data and analyses of key ancient individuals, and from living individuals in the Americas and possible ancestral regions, including Siberia and Oceania. This breadth of sampling increased the power of the study to distinguish between alternative hypotheses for the timing and pattern of migration events. Ripan Malhi, an associate professor of anthropology at the University of Illinois and one of the senior coauthors, focused on genome sequence obtained from 6,000-year old skeletal remains found on Lucy Islands in British Columbia, Canada, and modern descendants of those individuals.

"There were multiple reasons why certain ancient individuals were selected," said Malhi, who is also an affiliate of the Regenerative Biology and Tissue Engineering research theme at the Carl R. Woese Institute for Genomic Biology (IGB). "We wanted a variety of locations as well as ages . . . what we were able to show overall, other than the late Pleistocene Anzick boy that was published last year, all of the individuals ranging from 6,000 years ago to more recent times show closer genetic affinity to the modern-day Native Americans in that same geographic region than anywhere else."

Migration to the Americas: how early, how often?

This study, like previous work, identified two major groups of Native Americans; Athabascans and northern Amerindians (comprising mainly

northern populations) and other Amerindians (comprising mainly southern populations). Comparing genome sequence within and between these groups and ancestral individuals led the researchers to estimate that both of these groups diverged from their shared ancestors at about the same time, no earlier than 23,000 years ago. This finding supported the idea that a single group migrated to the Americas, and subsequently split into two distinct populations.

Ancestral Native Americans traveled into the Americas via the Bering Land Bridge; during the Pleistocene epoch, this grassland region connected what are now Alaska and Siberia. The current study suggests that the migrating population remained isolated in this area for about 8,000 years, before moving further into the Americas.

These findings challenge an alternative model that proposes a migration from East Asia and Oceania that gave rise to the earliest Americans, called Paleoamericans, who were then replaced after a second migration via the Bering Land Bridge.

This alternative hypothesis has received support in the past from structural similarity of skull shapes in putative Palaeoamerican remains to those of their possible Australo-Melanesian relatives. However, the extensive genomic comparisons made in this study found no evidence of a close relationship between these groups. Instead, the Central and South American remains examined were more closely related to other Native American groups. There were traces of genetic similarity to East Asian and Australo-Melanesian populations found in some of the Native American groups examined; analyses suggested this similarity was produced through sporadic contact between Native American and Eurasian populations, rather than two independent migration events.

Challenges and opportunities for future research

Highlighting the difficulty of obtaining and analyzing data that can help answer these and related questions, a study published simultaneously in *Nature* arrived at a few conflicting conclusions. Most notably, authors of the *Nature* study found support in their analyses for two separate migration events. Further research in this area will benefit from more data, and continued improvement in genomic technology and analysis.

Malhi noted that the *Science* study that he contributed to had several strengths compared with prior work. Past studies have relied more heavily on anatomical measurements, which can be misleading, and on mitochondrial or Y-chromosome DNA sequence, which, because they are inherited from a single parent, are more vulnerable to bias. The present study used biparental genomic data, and examined an unusually large and diverse set of individuals. Malhi also emphasized that the partnerships formed with modern Native American populations during his career have strengthened and informed his research program.

"A lot of my new thoughts and ideas, perspectives on genomic analysis of indigenous people in the Americas, come from interacting with Native Americans," said Malhi. Malhi is co-organizer of an annual workshop, the Summer Internship for Native Americans in Genomics (SING) held this year at the IGB, that provides a forum to discuss the uses, misuses and limitations of genomics as a tool for Native American communities. The workshop also assists in training Native Americans in the concepts and methods currently used in genomics.

More information: Genomic evidence for the Pleistocene and recent population history of Native Americans, *Science*, [DOI: 10.1126/science.aab3884](https://doi.org/10.1126/science.aab3884)

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