MOLECULAR ANTHROPOLOGY

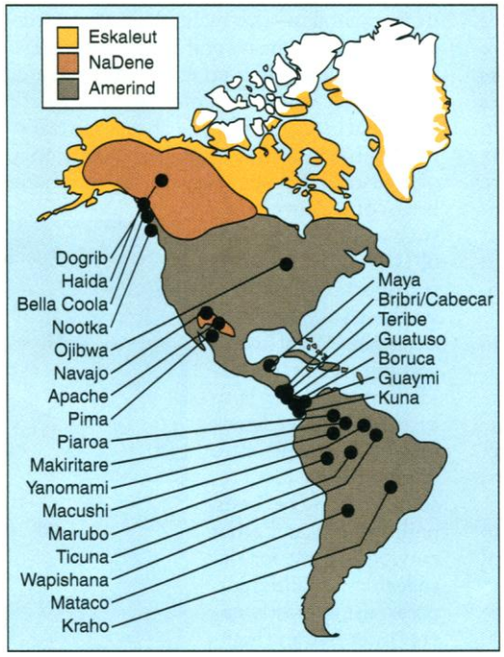
Geneticists Trace the DNA Trail of the First Americans

Who were the first Americans? If ever there were a controversial question in American anthropology, this is it. Ever since anthropologists discovered an 11,000-year-old fluted stone blade lodged in a mammoth bone in Clovis, New Mexico, in 1932, there has been a heated debate over whether the so-called Clovis people who made those tools were the first to cross a landbridge from Asia, or whether there were other groups who entered long before the Clovis ancestors came, some 12,000 years ago. That debate is hardly settled (*Science*, 17 August 1990, p.738). In fact, says Southern Methodist University archeologist David Meltzer, who has written extensively on the debate, there is "absolutely no agreement" in the research community on when the first Americans arrived on these shores.

But while that debate has been raging—based largely on analyses of such things as stone tools and fire pits—another area of investigation on the early Americans has quietly been making progress. That line of work depends not on stone, bone, and cinder, but on DNA. Yet it also has a lot to say about just how the Americas were populated. Indeed, at last month's meeting of the American Anthropological Association in San Francisco, much attention was given to the findings by several geneticists on the ancestry of American Indians. The researchers have found that all present-day American Indians can trace their descent to one of four maternal lineages originating in Asia—an important confirmation of anthropologists' longstanding suspicions that the first Americans came from Asia. The finding also suggests that the first settlers were ancestors of the Amerind-speaking people, whose modern-day descendants include the Pima of Arizona, the Mayans of Mexico, and the Yanomami of Venezuela—tribes spread out in North, Central, and South America. And while the geneticists' views of the past are no more unanimous than those of their stone-and-bone colleagues, at least a few think their data suggest that the first Americans set foot in Alaska well before the Clovis people did.

Most of these conclusions are based on the population geneticists' analyses of genes from both living native Americans—ranging from the Aleut Eskimos of Alaska to the Kraho tribe who live in Brazil—and from the well-preserved remains of some paleoindians, including 3000-year-old mummies from Chile and 8300-year-old skulls from a Florida bog. In particular, the researchers are comparing the sequences of genes from mitochondria (the cell's energy factories) from people of various tribes as a way of tracing their origins and determining how closely related they are.

For this kind of work, mitochondrial DNA (mtDNA) offers some major advantages over the bulk of the cell's DNA, found in the nucleus. It's inherited exclusively from the mother, without undergoing the gene shuffling and mixing that can obscure the evolutionary trail of nuclear genes. And because mtDNA accumulates mutations much faster than nuclear DNA, it provides more points of comparison and therefore allows ancestral relationships to be traced more accurately.



Gene map. The DNA analysis of 24 tribes of Amerind and NaDene speakers shows they settled in America in at least two waves.

SOURCE: ANTONIO TORRONI/EMORY UNIVERSITY ILLUSTRATION: D. DEFRANCESCO

One of the first researchers to apply mitochondrial analysis to sort out the lineages of American Indians was Emory University geneticist Douglas Wallace. In the mid-1980s, Wallace found that the Pima Indians of Arizona have a marker, or rare variant mtDNA sequence, found in Asians but not in Europeans or Africans. That was consistent with the long-held general view that native Americans originated in Asia. But it indicated something else as well. Since the variant is 20 times more frequent in the Pima than in Asians, it suggested the Pima were descended from a small number of female ancestors who brought the variant with them in their genetic baggage as they migrated from their Asian homeland.

Having established that mtDNA analysis can provide information about the origins of native Americans, Wallace's group, which includes geneticist Antonio Torroni and anthropologist Theodore Schurr, also at Emory, went on to analyze the mtDNA of 547 American Indians from 24 tribes, "from Alaska to Argentina," Torroni says. (The DNAs were part of a collection assembled by University of Michigan population geneticist James Neel.) For comparison purposes, the researchers analyzed mtDNA from 404 Siberians from 10 tribes.

The results: Every fullblooded American Indian tested carries one of four different rare mtDNA variants that is also found in Asians but not in Europeans or Africans. (Wallace's group published a preliminary account of this conclusion, based on a smaller sample size, last year in *Genetics*.) What's more, the frequency of the variants is much higher in Native American populations than in Asians, indicating that all American Indians, not just the Pimas, are descended from a small number of "founding mothers" from Asia. The number could be as low as four lucky mothers who traveled in a group in which the other women's mtDNA lineages later went extinct—or it could be that there were four groups of closely related women, each of which carried one of the four mtDNA variants. Either way, says Torroni, "there was a clear reduction in genetic variation from Asia to America, possibly due to a bottleneck."

Other researchers also are coming to agree with Wallace's conclusion about the four maternal lineages. At the anthropological association meeting, Pennsylvania State University graduate student Anne Stone presented data from her analysis of the mtDNA from the remains of 50 Oneonta Amerindians who lived in Illinois before contact with Europeans.

Working with Penn State molecular anthropologist Mark Stoneking, Stone found that 96% of the individuals had mtDNA that fell in one of the four genetic lineages described by Wallace and Torroni. Further confirmation came from Pittsburgh University geneticists Robert Ferrell and Andrew Merriwether, who reported preliminary results from studies of mummified remains of people who lived in Chile from 3000 to 500 years ago. They found high frequencies of two of the four mtDNA lineages, while a study of 40 living Mayans found all four. And a separate study of the mtDNA of Hohokam mummies in a collection at the University of Arizona found similar results: These mummies from Ventana Cave in Arizona also carried the same four clusters of mtDNA, says University

of Munich geneticist Oliva Handt. "It looks like there were a small number of maternal lineages, and that the women were all very closely related," says Merriwether. "After that there was a rapid population explosion."

Based on these findings, Torroni proposes the following scenario: A band of settlers—with women carrying the four lineages—enters the New World. The founders find no other people to compete with for space or resources, so they thrive in the New World and quickly spread out, radiating throughout North, South, and Central America, where they multiply their numbers rapidly.

After they had been in the New World for a while—perhaps thousands of years—another wave of immigrants lands on the shores of Alaska. But the mtDNA data indicate that these people were different from the initial settlers, because the women carry only one of the four mitochondrial lineages. And that one lineage traces back to a population in Siberia that also carries only that mtDNA variant—a population that is distinct from those Asian populations that carry all four lineages.

The evidence for those two waves of migration is found in the DNA of their descendants: When the Wallace group compared different tribes, an interesting correlation emerged. Individuals in tribes that are Amerind-speakers (including the Maya, Yanomami, and other tribes from the United States to South America) inherited one of the four lineages, but overall, four lineages are present in the Amerinds. But only a single lineage was found in the NaDene-speakers (including the Dogrib, Tlingit, and other tribes of the Pacific Northwest). That implies that the Amerind and NaDene had different ancestors—who must have entered the New World in two migrations.

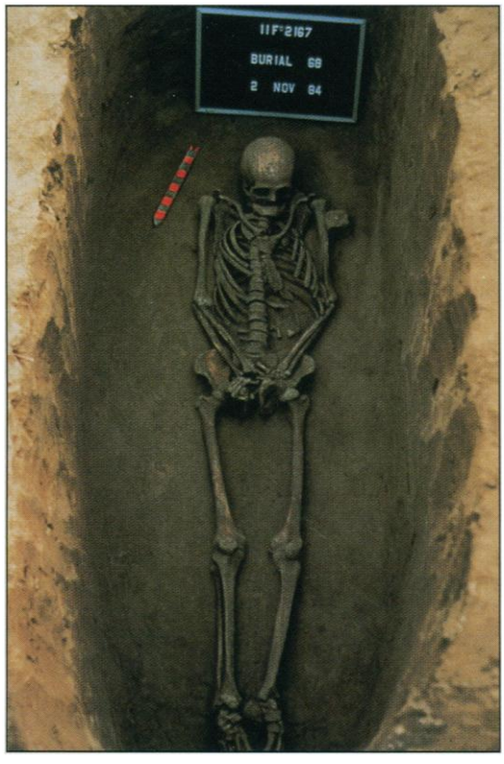
As it happens, the Wallace group's scenario dovetails nicely with a controversial theory proposed in the mid-1980s by Stanford University linguist Joseph Greenberg, Christy Turner of Arizona State University, and University of Arizona anthropologist Stephen Zegura (*Science*, 27 April 1990, p. 439). Based on their analysis of languages, genes, and teeth, they concluded that the ancestors of all American Indians arrived in three distinct waves—with the Amerinds migrating first, and the NaDene and Aleut-Eskimos coming in separate migrations more recently.

The Wallace group findings differ in at least one significant regard from those of the Greenberg group, however. Greenberg put the date of the Amerind migration at 15,000 years ago. But based on their analysis of the accumulation of variation in the mtDNA lineages, which can be used as a sort of clock to time the split of two individuals from a common ancestor, the Wallace group, in collaboration with Neel, proposes that the first Amerinds migrated to America between 42,000 and 21,000 years ago. Says Torroni: "The Amerind are very old. We say this confirms they were pre-Clovis."

JAMES NEEL/UNIV. OF MICHIGAN

Direct descendants. The Makiritare who live in the Amazon trace their ancestry to the founding mothers of the Americas.

The second group, which was ancestral to the NaDene, came between 12,500 and 6000 years ago, as did the Aleut-Eskimos. Torroni and Wallace arrive at these dates by analyzing all the variation that has arisen within the four mtDNA lineages in the American Indians over the millennia, and calculating how long it would take the DNA to mutate enough times to account for that variation. And the Amerinds show far more variation—by a factor of at least three—than the NaDene. That means they had far more time for their mtDNA to accumulate mutations, and must be of more ancient ancestry.



MARK STONEKING/PENN STATE UNIV.

Ancient DNA. The mtDNA from this extinct Oneonta paleoindian offers clues to his ancestry.

Still not everyone agrees that the variation the Wallace group sees in the four mtDNA lineages had to have arisen after the migration from Asia. Take, for example, University of Utah geneticist Ryk Ward. In a paper published in 1991, he argued that the mtDNA of 63 individuals from an Amerind tribe in the Northwest—the Nuu-Chah-Nulth—can be grouped into four clusters that, he now says, correspond to Wallace and Torroni's four genetic groupings. But he also finds substantial diversity between those clusters, so much so, Ward says, that it may be too much to have arisen only after the tribe was in the New World. He points out that different molecular lineages usually arise long before populations split. Ward argues that Wallace's four lineages aren't necessarily those of the first American women—but might be four maternal lineages that instead arose in Asia and began mutating there. If that's the case, extrapolating backward using the four maternal lineages wouldn't help to pinpoint the date when various ancestral groups entered the New World. In fact, says Ward, if he's right, the founding populations could have come over in a continuous trickle and "we will still have to rely on archeological data to show us where the humans were at the time (when their genes diverged)."

Archeologist Meltzer agrees that if the geneticists want to convince anthropologists to take their dates seriously, they will have to "get their house in order" by addressing Ward's concerns. Torroni says new studies of Asians find they have far less variation within the four lineages, indicating it originated in America. But he's also looking for gene markers on the Y (male) chromosome to see if paternal DNA lineages display the same kind of diversity as the maternal lineages, or to trace the ancestry of more nuclear and mitochondrial genes in American and Asians. Others geneticists are hoping to study more samples of ancient DNA, including those from 91 skulls found in a Florida bog that date back 8300 years.

In the end, however, it appears that no one discipline on its own will reach a definitive account of the peopling of the Americas. While anthropologists can place an early human at a specific site and time in a landscape, it will take an analysis of the languages, teeth, and genes of their descendants to portray the first Americans in detail. Says University of Michigan's Neel: "Genetic tools are increasingly shedding light on these old stories."

–Ann Gibbons