Y Chromosome Shows That Adam Was an African

In the beginning, there was mitochondrial Eve—a woman who lived in Africa between 100,000 and 200,000 years ago and was ancestral to all living humans. Geneticists traced her identity by analyzing DNA passed exclusively from mother to daughter in the mitochondria, energy-producing organelles in the cell. To test this view of human origins, scientists have been searching ever since for Eve's genetic consort: "Adam," the man whose Y chromosome (the male sex chromosome) was passed on to every living man and boy.

Now, after almost a decade of study, two international teams have found the genetic trail leading to Adam, and it points to the same time and place where mitochondrial Eve lived.

Described this month at a symposium on human evolution at Cold Spring Harbor Laboratory in New York, the genetic trail is so clear that it allows researchers to compare the migration patterns of men and women tens of thousands of years ago (see sidebar). It even pinpoints the living men whose Y chromosomes most resemble Adam's: a few Ethiopians, Sudanese, and Khoisan people living in southern Africa, including groups once known as Hottentots and Bushmen.

The findings, by teams based at Stanford University and the University of California, Berkeley, announced that they had found our mitochondrial ancestor. They had compared mitochondrial DNA variants found around the world and traced a common ancestor by sorting out the variants on an ancestral phylogenetic tree. But Eve, ignited fierce debate, as some scientists challenged the methods and assumptions used to place her in Africa from 100,000 to 200,000 years ago (Science, 14 August 1992, p. 873). Even if the Wilson team was right—and intense debate continues—"the mitochondrial DNA is just one gene lineage," says University of Michigan, Ann Arbor, paleoanthropologist Milford Wolpoff. Different genes might trace back to different ancestors on a different continent.

The only way to test the story of mitochondrial Eve was to trace the ancestry of other genetic lineages in the nucleus of the cell to see if separate lines of evidence also led back to African ancestors. And the obvious place to start was the Y chromosome, which is passed from fathers to sons and therefore provides an independent test of the mitochondrial Eve hypothesis. The bulk of the Y chromosome remains unchanged through generations, except for rare mutations hidden in regions that don't code for proteins. By comparing variation at the same site in different individuals' DNA (known as polymorphisms), geneticists can sort out which populations are most closely related and then build a phylogenetic tree that traces the descent of men, just as the mtDNA traces the descent of women. And by using average mutation rates for nuclear DNA, they can estimate how long ago particular mutations appeared, and thus how long ago each limb of the tree branched off.

Sketching the branches of a Y chromosome tree has been painstakingly slow, however, because few polymorphisms in the Y chromosome had been found (Science, 26 May 1995, p. 1183). But new tools to speed the detection of variation in DNA sequences have recently given a boost to research on the Y. One big break has come at Stanford University, where biochemist Peter Oefner and
The Women’s Movement

Genghis Khan, it appears, was an exception. When it comes to spreading genes around the world, scholars have often focused on the movements of men, sometimes picturing bands of males like Genghis Khan’s army sweeping across wide geographic regions and fathering more than their share of children. But with new results from the Y chromosome (see main text), researchers are finding that spreading genes into new territory may be chiefly women’s work.

The data show that variants in the Y chromosome, which sons inherit from their fathers, have a different geographic distribution from variants in mitochondrial DNA (mtDNA), which is passed from mother to daughter. Particular mtDNA markers are widespread: Women on different continents often carry the same markers, albeit at different frequencies. But most variations in the Y chromosome are restricted to small geographic areas, according to a report at a recent Cold Spring Harbor meeting by Harvard University biology graduate student Mark Seielstad and Stanford University statistician Eric Minch.

Using Y polymorphisms detected in Stanford geneticist Luca Cavalli-Sforza’s lab, they found that only 3% are distributed across continents and that most are restricted to local groups, such as Bantu-speaking males in Africa, who exhibited their own “private” polymorphism. This shows, says Cavalli-Sforza, that “women move more than men.”

This may seem counterintuitive because studies have shown that in hunter-gatherer societies, men typically travel greater distances in their lifetimes than women do. One explanation is that when it comes time to settle down and have children, men go home to their birthplaces, at least in the 70% of modern human societies that are patriarchal. This means that women move into their husbands’ homes and have their children farther from their birthplaces. Thus, over the millennia, women spread their genes farther than men do—eventually across entire continents, says Seielstad.

This doesn’t mean that Genghis Khan and other roving males didn’t leave a trail of offspring. “But from a demographic viewpoint, it hasn’t been as major a component of gene transmission as have women’s movements over their lifetimes,” says Seielstad. —A.G.

molecular biologist Peter Underhill have developed an automated system for rapidly detecting subtle differences in DNA sequences. Now they have harnessed this system to find genetic markers for evolutionary studies. The method involves mixing and heating amplified copies of Y chromosomes from two men to unzip their double-stranded DNA. Then the single strands from the two Ys are reannealed. If their sequences match, they emit a single peak of ultraviolet light. If they differ by as little as one nucleotide, they emit two or more peaks.

The Stanford researchers got the system working full-time this year. So far, they have found 93 new polymorphisms in the Y chromosomes of men from around the world. “It’s a major breakthrough—unquestionably,” exults geneticist Luca Cavalli-Sforza, head of the lab where Underhill has done this research. And when the team started sorting these polymorphisms into a phylogenetic tree, they found one particularly ancient marker, called M42. In its most ancient form, shared by other primates, this marker is an A, or adenine. Today, it is found only in Africa, in just a few of the 900 males they scanned—15% of the Khoisan, and 5% to 10% of the Ethiopians and Sudanese. These men must have inherited this ancient form from a common ancestor. “We think we have tagged Adam,” says Underhill, who reported the work at this week’s annual meeting of the American Society of Human Genetics in Baltimore.

The data further show that sometime in the past 100,000 to 200,000 years, this M42 site underwent a mutation—a change from A to T (thymine) in one of Adam’s descendants. While men with the A stayed in Africa, some of the Africans with the T left the continent and spread around the globe. Today, “all men outside of Africa, as well as most African men, carry the T,” says Underhill.

Meanwhile, another team headed by geneticist Michael Hammer at the University of Arizona, Tucson, surveyed another non-coding region of the Y chromosome in 1544 men worldwide and found the same pattern. The DNA sequences varied among individuals, but Hammer found that the variants clustered into 10 major groups, known as haplotypes, which occur in different frequencies in different populations.

Haplotype A, defined by an A at a particular site, appears to be ancestral because the A is found in chimpanzees, and Hammer’s team found that in humans, it occurs only in some Africans. “It’s at the highest frequency in the Khoisan,” he says, the same population fingered by Underhill’s team. Although the ancient form of A persisted in some groups in Africa, it also underwent a change to a G (guanine) 150,000 to 200,000 years ago in one descendant of Adam’s. Like the T in Underhill’s site, this form was carried out of Africa when men moved away and replaced other males around the globe.

Both sets of results bolster the so-called Out of Africa model of human origins. “We think that anything existing in Asian males was replaced by this,” says Hammer. Underhill agrees: “I think this speaks persuasively for an Out of Africa origin for modern humans.”

But Hammer’s group, whose findings are in press in the journal Molecular Biology and Evolution, has added a twist to the scenario. These researchers also see evidence that some of Adam’s descendants who emigrated to Asia later returned to Africa, with a new mutation on the Y that arose in Asia. In addition, Hammer says that African males could have interbred with Asian females, and traces of those women’s genes may still be in our nuclear genome. Indeed, other researchers have already reported signs of an ancient Asian origin for a β-globin gene (Science, 25 April, p. 535).

So, Hammer favors a model that shows a wave of modern people coming out of Africa, replacing most of the genes of ancient people, but interbreeding enough to add some ancient non-African genes to our genome. Paleoanthropologist Fred Smith of Northern Illinois University in De Kalb, who proposed one such intermediate model, predicts that like so much of human history, the real story “won’t be black or white. Genes and fossils are showing that population dynamics are a lot more complex than we thought.”

—Ann Gibbons

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