

DNA Testing: In Our Blood

It is connecting lost cousins and giving families surprising glimpses into their pasts. Now scientists are using it to answer the oldest question of all: where did we come from?

By Claudia Kalb

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Feb. 6, 2006 issue - Brian Hamman had always wondered: what was up with his great-grandfather Lester? Hamman, an avid genealogist, could trace his patrilineal line back to 19th-century rural Indiana, but there was a glitch in the family records. Great-Grandpa Lester, the documents showed, was born before his parents were married. So was Lester really a Hamman? Was Brian? Three years ago DNA tests confirmed the lineage and a simple family mystery was solved: Lester's parents had hooked up before they walked down the aisle on July 25, 1898. Lester was indeed a Hamman, and so is Brian. "I'm delighted," he says.

For Debra Anne Royer, DNA unlocked a deeper mystery. Adopted at birth, Royer knew nothing about her biological parents. But certain physical traits—wide nose, dark skin—led people to guess that she was Iranian or even Cambodian. "I always wondered," she says. Two hundred dollars and a swab of her cheek gave her an answer: Royer's maternal ancestors were most likely Native American. The knowledge, she says, "makes you feel more of a person."

And then there's Prof. Henry Louis (Skip) Gates Jr., head of Harvard University's African-American Studies department. Gates always knew he wasn't 100 percent African-American. According to family legend, Gates's only white ancestor was a slave owner named Samuel Brady, who had sex with Gates's great-great-grandmother Jane on his farm in Maryland in the 1800s. But recent DNA analyses turned Gates's world upside down. There was no trace of Brady on Gates's genome. Further testing revealed that Gates, in fact, carries as much Western European blood as he does African—and that one of his white ancestors was probably an Irish servant who met Gates's sixth or seventh great-grandfather sometime before 1700. "I'm thinking I'm a Brady and maybe I'm from Nigeria, and here I am descended from some white woman," says Gates. "It's incredible."

Our blood holds the secrets to who we are, and, increasingly, individuals, families and research scientists are using genetic testing to tell us what we don't already know. Human genomes are 99.9 percent identical; we are far more similar than diverse. But that tiny 0.1 percent difference holds clues to our ancestries, the roots of all human migration and even our propensity for disease. Tens of thousands of Americans have swabbed their cheeks and mailed in their DNA to companies nationwide for testing. Far-flung cousins are finding each other; family legends are being overturned. Six years ago the term genetic genealogy was meaningless, says Bennett Greenspan, head of Family Tree DNA, which has 52,000 customers. "Now the interest is huge." So huge that celebrities like Whoopi Goldberg and Quincy Jones are signing on. This week Gates and other high-profile black Americans will tell their stories on PBS's new series "African American Lives."

As individuals track down their personal family narratives, population geneticists are seeking to tell the larger story of humankind. Our most recent common ancestors—a genetic "Adam" and "Eve"—have been traced back to Africa, and other intriguing forebears are being discovered all over the map. Last month one group of scientists found that 40 percent of the world's Ashkenazi Jews descend from just four women; another reported that one in five males in northwest Ireland may be a descendant of a legendary fifth-century warlord. The most ambitious effort by far is the National Geographic Society's \$40 million Genographic Project, which aims to collect 100,000 DNA samples from indigenous populations around the world over the next five years. The goal: to trace human roots from the present day back to the origin of our species. To create, says project director Spencer Wells, "a virtual museum of human history."

Human history lives in our genes. The DNA in each of our cells not only dictates the color of our eyes, it also contains the footprints of our ancestors. A child's genome is almost entirely a mix of genetic material created by the union of mother and father. Only two parts of the genome remain pure, untainted by the influence of a mate's DNA: the Y (passed down from father to son), and mitochondrial DNA (from mother to both sons and daughters). Occasionally, spelling mistakes or mutations arise in these regions, creating unique sequences of A's and G's and C's and T's that serve as genealogical signposts or markers—providing links backward in time,

not just to paternal and maternal ancestors but to the places they lived in the world. Scrape the inside of your cheek a few times, and for \$100 and up, a testing company will put your DNA under its microscope, map your markers into your own genetic pattern called a haplotype, then tell you which "haplogroup," or major branch of the human tree, you hail from.

Genetic genealogy has developed a cultlike following. Last fall, 175 genetic sleuths from as far away as Hawaii gathered for the second annual Family Tree DNA conference at National Geographic's headquarters in Washington, D.C., to share their haplogroups and bone up on the latest science. The genealogy garb was everywhere. Most notable: double-helix ties and pins with haplogroup logos. Forget HI, MY NAME IS JANE. Here it was R1b (Western Europe) meets J2 (Middle East). Participants who once relied only on birth records and marriage licenses to trace their family roots now listened spellbound as scientists presented arcane PowerPoints with daunting DNA lingo ("nucleotides," "autosomes," "short tandem repeats"). Over cafeteria hamburgers and hot dogs, they shared information about roadblocks they'd encountered in their ancestral paperwork—"nonpaternity events" (polite term for an affair, for example, which may have muddied the lineage) and families that had "daughtered out" (a much-bemoaned end to the family surname). Then they raved about the new frontier of DNA testing. "This is a group of geeks," Megan Smolenyak Smolenyak said with a smile during a coffee break. Her T shirt: GENEALOGY: IT'S IN MY DNA.

For Smolenyak, DNA blew up a family legend. Paper records had her believing that every Smolenyak in the world could be traced back to one of four families in Osturna, Slovakia. But after testing the DNA of individuals in each group, she discovered no genetic connection. "I got smacked upside the head," says Smolenyak, coauthor of "Trace Your Roots With DNA." "I was wrong." It wasn't just family lore that interested her: she was dating a Smolenyak at the time (now her husband), and it was nice to confirm that she could bury any fear of a kissing-cousins nightmare. "As it turns out," she says, "I could not have picked a guy more distantly related to me."

DNA testing is forcing some people to rethink their identities. Phil Goff, 42, of Naperville, Ill., thought his heritage was pure English, but a Y chromosome test matched him at least partially to Scandinavia. Now he wonders if he has any Viking blood in him. Alvy Ray Smith, 62, uncovered roots tracing back to the Puritans in 1633. "It was astonishing," says Smith, who thought his closest relatives were Irish potato farmers. "It gave me a whole different model of myself." Nick Donofrio, executive VP of innovation and technology at IBM, which is partnering with National Geographic on the Genographic Project, is a proud Italian. He was stunned when his Y test came back saying he was a member of haplogroup J2, meaning his ancestors had lived in the Middle East some 10,000 to 20,000 years ago. "You could have pushed me over with a feather," he says. After Donofrio announced his results on IBM's Web site, his in box started filling up with J2 colleagues. "A lot of Armenians have been sending me e-mails saying 'J2 rocks!' "

Armed with their haplotypes, which function as genetic blueprints, genealogists can now join Surname Projects on the Internet. These online communities bring together other Doolittles or Sanchezes or Epsteins, allowing people to compare genomes. Find a match, and you may be able to fill in branches on your family tree. Looking for relatives without your surname? You can also search within individual testing companies or in public databases like the Sorenson Molecular Genealogy Foundation, funded by Mormon philanthropist John Sorenson, which has collected 60,000 DNA samples and ancestral charts over the past 4½ years. "Eventually, you'll be able to query the database and find relatives you don't even know you have," says Sorenson's chief scientific officer Scott Woodward.

Some people think they already have. After genetic testing in 1998 revealed that Thomas Jefferson was most likely the father of at least one of Sally Hemings's children, Julia Westerinen, 71, of Staten Island, N.Y., and Shay Banks-Young, 61, of Columbus, Ohio, found each other. They look nothing alike—Westerinen's skin is white, Banks-Young's is black—but they claim one another as cousins. Neither one can prove through DNA that she is related to Thomas Jefferson himself, but that doesn't faze them. Nor does it bother Prinny Anderson, a white descendant of Thomas and Martha Jefferson's. Last week Anderson mingled with Jefferson's "unproven" black relatives at a gathering in Virginia. DNA testing isn't the end-all and be-all, she says. "I'm delighted with these additional cousins."

The science can also uncover links to ancient cultures, even religious heritage. Dr. Karl Skorecki was told from childhood that he was one of the Cohanim, descended from Moses' brother Aaron, a high Jewish priest. He was sitting in synagogue one day when he noticed that another Cohan who was called to the Torah looked nothing like him. "He was a Jewish male of African ancestry, I am a Jewish male of European ancestry,"

Skorecki, of the Technion-Israel Institute of Technology, remembers thinking. "If he has that tradition and I have that tradition, perhaps there's a greater chance that we share similar markers on the Y chromosome." Would the oral history passed down from Cohan father to Cohan son also be inscribed in their DNA? After studying DNA samples, Skorecki and geneticist Michael Hammer of the University of Arizona uncovered a genetic Cohan signature.

The research led Skorecki's team to Africa, where they tested members of the Lemba tribe, a group that believed they were descended from the Biblical land of Judea. Some of their DNA matched the Cohan signature. "We share a common paternal ancestry," says Skorecki. In 2001, Father Bill Sanchez, a Roman Catholic priest in Albuquerque, N.M., discovered he closely matched the Cohan signature, too. Sanchez's Jewish roots go back to Spain (his mother's heritage is Native American). Today he keeps pictures of his Christian and Jewish ancestors on his wall; in November he traveled to Israel. Now his niece Jessica Gonzales, 24, wants to go. Raised Catholic, she wants to learn more about her family roots. "I've been reading a lot about Judaism," she says.

DNA can now link regular people to high-profile ancestors—from Genghis Khan to the Iceman to the Irish warlord Niall of the Nine Hostages. Genghis-as-Great-Grandpa might be cool cocktail chatter, but since we don't have his DNA, proving direct descent is virtually impossible. Testing family roots through the Y chromosome and mitochondrial DNA has serious limitations, too: it tells you only about your direct paternal or maternal lineage, not the ancestor footprints hidden in the rest of your genome. Go back 10 generations, and that's 1,024 ancestors, says Stanford bioethicist Hank Greely. "Your Y might be from Japan, your mitochondrial DNA from Mexico and all other 1,022 ancestors from Sweden." Greely worries that customers may not fully understand what they're getting. One company, DNAPrint Genomics, does test markers outside of the Y and mitochondrial DNA, then maps them to four regions of the world (West Africa, Europe, East Asia and the Americas)—that's where Gates got his 50/50 ancestry. But the percentages are only estimates, not certainties. Some scientists are more than a little bit uncomfortable with the test. "I think the science of genetics is too important to become an entertainment," says Stanford geneticist Marcus Feldman, who also worries about the potential for racial stereotyping. With DNA tests, people may begin to link behaviors or characteristics with race, an idea that has been reviled in recent history. "I'm worried the more this is done, the more of that there's going to be."

The mutations in our DNA not only point to long-lost ancestors and homelands, they may also be markers for genetic disease. It's known as the founder effect: populations with marked susceptibilities to certain illnesses tend to be descended from a small group of ancestors who bred only within their own community. Sticking together meant a higher chance of inheriting a disease. The Amish, for example, are more likely to carry a genetic mutation for a condition called polydactyly, which causes extra fingers or toes. Ashkenazi Jews have an unusually high risk of certain cancers, as well as Gaucher and Tay-Sachs diseases. Men and women who inherit the mutation that causes Tay-Sachs are unaffected, but if they mate, they have a one-in-four chance of having an afflicted child. That's why Jewish parents-to-be are offered a panel of genetic tests before conceiving.

In rare instances, genetic mutations can offer medical benefits. Sickle-cell anemia is one of these double-edged swords. Patients who inherit a gene for the hereditary blood disease, which is common among people of African descent and causes red blood cells to lose oxygen, are also more likely to survive malaria. And the gene is highly prevalent in malaria-infested areas of Africa. Why? Scientists believe the gene has been naturally selected for its protective effect. Genealogical-testing companies aren't in the business of medical testing, but if you happen to discover an African ancestry you didn't know you had, should you be tested for sickle-cell? Possibly. In the brave new world of DNA testing, it would be a circuitous route to take. The express highway: submitting your genome for medical, not genealogical, analysis. In the future, this could be as routine as a physical. Already, the marketplace is eager to help. Most genealogical-testing companies stay as far from disease testing as possible, but other entrepreneurs are diving in. For \$200 and up, a company called HealthCheckUSA will test your DNA (provided by cheek swab) for eight genetic illnesses, including celiac disease (an intestinal disorder) and hemochromatosis (an overload of iron). "People call us on a daily basis and let us know we helped save their life," says company president Holt Vaughan.

The more we learn about our families, the more we learn about our beginnings. Using DNA markers and mathematical time-clock calculations, researchers have identified our ancestral Adam and Eve. Scientists say that by using Y and mitochondrial DNA, they can date the earliest female to 150,000 to 250,000 years ago and the earliest male to 60,000 to 100,000 years ago. Until DNA testing, scientists debated whether humans originated in Africa or in a number of different places around the globe. These recent findings support the theory that humans descended from a small group of people who lived in Africa tens of thousands of years

ago.

But when did groups of travelers leave that continent? Whom did they encounter and mingle with along the way? (At Arizona, Hammer is investigating the question of whether *Homo sapiens* and, say, Neanderthals mated and bore children.) Do major historical events, such as Alexander the Great's conquest of Central Asia, leave a genetic trail? These are questions National Geographic's Spencer Wells hopes to answer. The Genographic Project, launched last year, is inviting the public to test its own DNA, and already 110,000 individuals have purchased swabbing kits for \$99.95. But the project's overarching goal is to collect DNA from indigenous populations worldwide whose DNA could hold clues to our origins and global migration—and to do it fast, before whole populations die out and leave their ancestral homelands. Early testing has already started in Southern Africa, where collaborator Dr. Himla Soodyall has collected blood samples from a small group of the San tribe. Genetically the San have among the oldest roots on earth and, it is believed, they provide a direct chromosomal link to ancestral Adam and Eve. Fi Mntungwa, 28, was one of the first to donate. "We were told about genes and a huge project that is looking into the origins of people across the world. It was very interesting," says Mntungwa. "I want to revive our ancient culture."

Last fall, Wells packed up 500 blood-collection tubes, needles, alcohol wipes and cheek swabs and headed off to Chad, one of the project's first testing sites, where he took 300 DNA samples from towns and villages around the country. Thirty-five to 40 came from members of the isolated Laal community, whose population, at fewer than 750, is declining. Wells fears that this community will die out within the next 10 to 30 years, taking with it valuable DNA and cultural traditions and an ancient language—information that could provide critical insights into the first people to live in Central Africa more than 40,000 years ago. "We can use DNA to figure out some of these great mysteries, to make sense of the past," says Wells.

Not everybody supports the Genographic Project. Indigenous populations have had their share of colonialist pillaging and many, still distrustful of the dominant culture, are wary of handing over their blood and the information it contains. Debra Harry, director of the advocacy group Indigenous People's Council on Biocolonialism, has posted a petition on her Web site opposing the project, which she says has 1,000 signatures so far. But some members of the Seaconke Wampanoag Tribe in Seekonk, Mass., have already been tested. "We have our cultural story of creation, but there's another story that needs to get out, and it's right inside each and every one of us," says the tribe's chair, Michael Markley. Wells says he understands indigenous concerns, but he has found in his travels that once the details are explained, the excitement builds. "Everybody finds it fascinating that they're carrying this historical document inside their cells." In May, the project will announce plans to sponsor cultural and educational initiatives in participating indigenous communities.

The more we discover our differences, the more we find connections. Wayne Joseph grew up a black American in Louisiana and Los Angeles—even writing a My Turn for NEWSWEEK in 1994 about Black History Month. He heard about DNA testing several years ago and, seeking details about his mixed ancestry, sent away for a kit. "I figured I'd come back about 70 percent African and 30 percent something else," he says. When the results arrived in the mail "I was floored," he says. The testing company said he was 57 percent Indo-European, 39 percent Native American, 4 percent East Asian. No African blood at all. For almost a year, Joseph searched his soul, sifting in his mind the decisions he'd made based on his identity as a black man: his first marriage, his choice of high school, his interest in African-American literature. Before the test, "I was unequivocally black," he says. "Now I'm a metaphor for America." And not just for America, but for all of us.

With Karen Springen, Mary Carmichael and Karen MacGregor in Durban, South Africa

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