Genetic genealogy goes global

Although useful in investigating ancestry, the application of genetics to traditional genealogy could be abused

Like millions of Americans, Thomas Robinson wanted to investigate the roots of his family tree before the point at which his ancestors emigrated to the USA. "I did paper-based genealogy research on my family and could not get very far, especially on my father's side since I had never met my paternal grandfather and knew little about him," said Robinson, a professor of accounting at the University of Miami, FL, USA. "I also read quite a bit on the origins of man and how DNA testing was used to trace migration patterns." Robinson, who also studied biology at university, thus sent a cheque with a cheek scraping to Oxford Ancestors (UK), one of many companies that use molecular genetics to unravel family histories. In early 2003, he received his results: a signature from his Y chromosome and detailed information on his mitochondrial DNA. The latter indicated that his family might have originated on what is now the Spanish/French coast, whereas the Y-line information was not specific.

But the story did not end there. In March 2006, a representative from Oxford Ancestors called Robinson to tell him that he was apparently descended from Genghis Khan, the thirteenth-century Mongol emperor. Robinson's Y-signature matched seven out of nine markers in Khan's signature, which are found in unusually high numbers—about 8%—of men living in a wide swathe of Asia from the Pacific Ocean

to the Caspian Sea (Zerjal *et al.*, 2003). "It was exciting to hear and nice to put a 'name' to an ancestor," said Robinson, a rather unlikely-looking member of the Mongol horde. The story was picked up in newspapers

TX, USA) concluded that he and Genghis Kahn shared no common ancestry within the past 30,000–40,000 years, let alone the past 800 years. Robinson was booted out of the yurt. The Mongolian ambassador never followed through with the reception and the newspapers ran retractions. It was "a bit disappointing", Robinson admitted, "but I learned quite a bit more [about DNA and Genghis] in the process."

"The Y-STR [short tandem repeats] data from Oxford Ancestors and Family Tree DNA matched perfectly, but the interpretation differed," said Chris Tyler-Smith from the Sanger Institute (Cambridge, UK), who is one of the authors of the study on Khan's signature. "In particular, Family Tree DNA typed some additional Y-SNPs [single nucleotide polymorphisms] that showed that the two lineages were not as closely related as appeared from the Y-STRs."

Although this particular case is unusual, Robinson is just one of many who rely on molecular biology to find their ancestors. Companies such as Oxford Ancestors, Family Tree DNA and DNAPrint Genomics (Sarasota, FL, USA) have attracted more than 300,000 customers in the past six years. Most cases address more modest objectives, such as confirming relationships between people with similar surnames or investigating family stories about a Native

around the world, and the Mongolian ambassador to the USA promised to hold a reception in Washington, DC, to toast the new-found descendant of the Great Khan.

However, at the suggestion of a filmmaker who had contacted him about a documentary on Mongolia, Robinson pursued more detailed testing. Family Tree DNA (Houston,

American ancestor. Jill Servian Whitehead, a 55-year-old social researcher from London, UK, is such a customer. Through traditional paper-based searches, she found that her Jewish ancestry could be traced back to eighteenth-century Poland and Lithuania.

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As she had a longstanding interest in genealogy, history and biology, Whitehead had her DNA tested by Family Tree DNA. Her results showed that she had a relatively rare haplotype, and she used the company's online database to find matches to people with shared Jewish roots from the Baltics. At the same time, Whitehead was contacted by Mike Moseley through the Jewish Genealogical Society of Great Britain, who was searching for people named Servian. Testing confirmed that the two, who had previously never met, shared common great-grandparents. "Even without paper trails, genetic testing has enabled us to find fairly close relations going back over the last 300 years or so. This has been very exciting and enriching," Whitehead said.

However, as the Robinson case highlights, customers should take their results with a pinch of salt. Scott Woodward, a molecular genetics researcher and President of the Sorenson Molecular Genealogy Foundation (Salt Lake City, UT, USA), a non-profit organization that collects DNA samples to build up database-integrated family trees, worries that overselling can backfire. "The public is getting the impression that DNA is the answer to everything. That's not necessarily so. I think we have to be careful about emphasizing what the realities are, what you really can do with DNA," he said.

As a commercial service, genealogical DNA testing holds the same promises and pitfalls as any business, but the potential drawbacks for customers are not normally dramatic. "As far as mixing science and capitalism goes as in fee-for-services testing, it is a 'let the buyer beware' marketplace," said Peter Underhill, a senior research scientist at Stanford University (CA, USA). "How much responsibility a company has in educating a potential client prior to sample submission

and credit card number is a value judgment matter. Also mistakes do occur, during either the actual data collection or its interpretation. Fortunately errors in paternal ancestry assignment are not life-threatening, as would be the case in blood typing prior to transfusion, for instance."

"[A]s usual, there are those who simply want to make money out of it, those for whom it is part of serious science, and those who are extremely desperate to look for their famous ancestors and who will do anything to reach their target," commented Peter de Knijff, a geneticist at Leiden University, the Netherlands, who was involved in a highly publicized study showing that a male from the bloodline of US President Thomas Jefferson fathered a child with his slave Sally Hemings (Foster *et al*, 1998). There is "a lot of total rubbish on many of the Y-STR project sites [online], but it seems that those involved are perfectly happy with what they do," de Knijff said. "People are entitled to have their dreams, and if they are willing to pay for that, well, fine with me."

Not surprisingly, the commercial use of genetic tools in genealogy initially was rather off-putting for both genealogists and molecular biologists. Megan Smolenyak, a genealogist who conducts studies for the US military to identify remains of soldiers from the Vietnam and Korean wars, said many traditional genealogists are reluctant to leave the archives and take a dip in the genetic pool. "It's very much the same sort of life-cycle that we went through when the Internet came along," she said. "[S]ome of the pros were saying, 'Oh, this is cheating. This is just people looking to take shortcuts.'" But things changed, she said, as genealogists realized that the genetic tools were complementary to methods such as digging out paper records that can be used to prove or disprove family links.

Bryan Sykes, a professor of human genetics at Oxford University and founder of Oxford Ancestors, said some of his colleagues have indeed looked down on his commercial enterprise. "There are always some academics who believe it is entirely wrong to almost corrupt pure academic investigation by turning these into commercial services." Sykes, whose own research involved genotyping Ötzi the Iceman and the Romanovs, said, "The only way to open up a field to the general public [...] is by offering a proper commercial service. It can't and shouldn't be done in

research laboratories because they are there to do other things."

For Woodward and the Sorenson Molecular Genealogy Foundation, it is also a matter of people partaking in research that they supported in the first place. "To find disease genes, we used pedigrees to identify people in those families who've had a particular disease. It was through genealogy and family trees that we were able to identify those pieces of DNA," he said. "And then we just sort of turned it around and brought some of those tools back to the people who would now be able to use them to find their families."

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Underhill also sees another positive aspect. "On the one hand, such enterprises open the door for the public to get a better understanding of science, genetics, human evolution and the important fact of the African ancestry of our species, as well as the very high degree of genetic similarity between people of the world," he said. "On the other hand, these activities potentially reinforce a culture of personality [spotlighting famous people] [...] as well as potentially trivializing science. I don't think it is a bad thing if neighbours at a weekend BBQ stand around and discuss what haplogroups they belong to, as long as such activities do not cause the public to mistrust or view the underlying science of deciphering human evolution and human history as trivial and shallow."

Spencer Wells, a senior explorer with the US National Geographic Society (Washington, DC) and leader of its Genographic Project, believes that commercial genealogists can contribute knowledge, just as amateur astronomers and ornithologists have made important contributions to their respective fields. "It's probably similar to the astronomy community thinking that somebody standing out in their backyard with their telescope looking at the sky isn't going to find anything," Wells said. "In come cases, amateur astronomers actually do discover things. They discover comets, nebulae and so on. I think there is something to be gained from tapping into the interest in genealogy."

This is what the Genographic Project aims to do. Launched by Wells, the US National Geographic Society, IBM (Armonk, NY, USA) and the Waitt Family Foundation (La Jolla, CA, USA), it is an anthropological rather than a genealogical exploration. It hopes to collect DNA samples from 100,000 indigenous people, along with a minimum of 100,000 public participants who pay US\$99 to get a glimpse of their ancestry while helping to fund both the research and a payback project for the indigenous groups. During the first 15 months of the five-year project, 160,000 people signed on, far more than had been anticipated. "I think it is part of people's general interest [...], particularly people who live in countries with large immigrant populations like the United States, where everybody, unless you're a Native American, came from somewhere else relatively recently," Wells said. "They have the desire to connect to the old country. There is a tremendous interest in genealogy. And this application of a new tool tells us a little more about our ancestry."

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However, not everyone is as pleased with the project. Debra Harry, a Paiute Indian, and Executive Director of the Indigenous Peoples Council on Biocolonialism (Pyramid Lake, NV, USA), and Le'a Malia Kanehe, a Native Hawaiian attorney for the group, noted that the study consent form states that the research "may contradict an oral, written, or other tradition held by you or by members of your group" and criticized that research could undermine these beliefs (Harry & Kanehe, 2006). "The discounting of indigenous historical knowledge goes beyond just a difference of opinion. A claim that challenges the 'indigeness' or 'aboriginality' of certain indigenous peoples could pose serious political threats," they wrote.

"The Genographic Project is an anthropological and historical research initiative, and some indigenous people might find value in that very same knowledge gained by the project scientists," Wells commented. "In addition, the Genographic Project has established the Genographic Legacy Fund [...] to provide tangible benefits to indigenous communities in support of their aspirations to promote and

protect their cultures." He added, "ultimately, the Genographic Project has the potential to highlight human unity and connectedness while celebrating cultural diversity."

However, Edwin Black, author of *War Against the Weak: Eugenics and America's Campaign to Create a Master Race*, warned that the data could be abused, for example, to discriminate against indigenous people. He also worries that the Genographic Project and others could sell their databases to governments, which could then use the information to persecute minorities. Wells dismissed Black's worries. "Technologies are powerful and they can be misused. People do find reasons to discriminate that have nothing to do with genetics." He also discounted Black's concerns about the Genographic Project database being sold. "Our database, once we publish the data, will be in the public domain. For that reason alone, I don't think anybody would be willing to spend any money on it because it's going to be freely available," Wells said. "We are simply testing genetic markers that tell us about ancestry. We're not doing anything that is medically or clinically relevant. [...] It's only information about deep ancestry with no information tying it to a particular person."

The same worries apply to commercial companies. For example, at Family Tree DNA, consumers can consent to having their DNA stored for future testing. "We offer storage and allow people to have the DNA destroyed if they wish," said Bennett Greenspan, the company's founder. "To date, we've had 70,000 test takers and only two people have asked that their DNA be tossed out, so we accommodate both sides." He added that the company's database is not for sale and that samples are bar-coded to protect their consumers' identity.

Nevertheless, concerns remain. Howard Sachs, an 80-year-old federal judge from Kansas City, MO, USA, who used Oxford Ancestors and Family Tree DNA to trace his Jewish roots, worries that genetic information could be abused by people with racist agendas. "This would have been a great tool for the Nazis," he said. Sykes agrees, "Some companies do so-called ethnicity testing. They will use any range of autosomal markers [to determine] the percentage of [ethnicity]," and declare whether an individual is 25% Native American or 10% Asian, for example.

The question then is whether any interested group could use molecular genealogy to discriminate against other people. Matt Thomas, a senior scientist at DNAPrint Genomics, which performs autosomal testing for genealogical purposes, said that this is possible, but his company opposes testing to determine "racial purity" and he is unaware of it being done. "It has been our policy as a company and as individuals who work for the company to resist that kind of use of the technology," he commented, but conceded that DNAPrint Genomics would not be able to exclude clients by questioning their motives. Ultimately, the idea of a genetically pure race has no biological basis, as it implies that people once existed in racially pure groupings. "They never did," Sykes said. "We're just a temporary state of a huge swirling cloud of genes coming from all over the place."

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The increasing fascination with ancestry—not only in the USA but also worldwide—suggests that genealogical DNA testing is here to stay. But as the Robinson case so aptly illustrated, "interpretation of the results can be difficult, and surprising conclusions need to be checked particularly carefully," said Tyler-Smith. By and large, the results will prove very useful to the studies of anthropology and ancestry, for both scientists and society. More nefarious uses will be subject to the principles of the individual. As Thomas pointed out, "The results and the sample belong to our customers. Whatever they do with it is their business."

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