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Caveat Emptor?

RELATEDNESS IS FAR MORE COMPLICATED THAN THOSE TRYING TO SELL US GENETICALLY-SOUND FAMILY TREES WOULD HAVE US BELIEVE

We are related, you and I.

Darwin says so. The Bible says so. Not much controversy about it.

The question is, How related? If we're too close, there are will be restrictions on our sexual behaviour towards one another. If we're too distant – that is to say, if you're a chimpanzee – there will be restrictions as well, of a different sort.

But the middle ground is very large – about six billion people large – and we all form a network of biological kin (if not social kin). The structure of that network is the domain of human population genetics, a field newly reinvigorated by free-market genomics.

The power of molecular genetic data to address issues of identity and relatedness with scientific authority has been appreciated for decades, particularly in the domains of paternity, genealogy, and forensics. Only recently, however, has the field branched out, so to speak, into the field of family trees, and what is now often called "recreational ancestry," tapping into a universal human desire to situate ourselves within a complex social universe. The maths are simple: genomic data + folk ideology = profits, and tests have been available for several years purporting to match your Y chromosome with Genghis Khan or Moses, or your mitochondrial DNA with any of seven imaginary European "clan mothers" who lived 15,000 years ago.

The commercial success of these tests lies in how successfully they can represent biological relatedness to be the equivalent of meaningful relatedness. In fact, the two never map on to one another particularly well, as anthropologists have long appreciated. Kinship (meaningful relatedness) is constructed by human societies from a locally particular calculus combining biological ties of heredity and legal ties of marriage and adoption. Your mother's sister's child and mother's brother's child are genetically equivalent, but the first is widely considered an incestuous relationship, while the second may be a preferred spouse across diverse cultures and eras. Charles Darwin, for example, married his mother's brother's daughter, yet his face nevertheless graces the English £10 note.

The mode of transmission of mitochondrial DNA makes it particularly vexing as a surrogate for biological ancestry. Most DNA, the nuclear human genome, is transmitted probabilistically; you have a 50% chance of having inherited any particular DNA segment from any particular parent. MtDNA, however, is inherited only through the maternal line: thus, you are a mitochondrial clone of your mother and mitochondrially unrelated to your father.

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Such a fundamental discrepancy between the heredity of mtDNA and our understandings of heredity ought to raise caution about glibly confounding the two. A generation further removed, the discrepancy becomes more glaring: You are equally descended from all four grandparents, but only mitochondrially descended from one of them (your mother's mother). And of your eight great-grandparents, only one is your mitochondrial ancestor.

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In general terms, as you proceed upwards in your genealogical tree, the number of ancestors you have in every generation increases exponentially (every ancestor had two parents), while the number of mitochondrial ancestors remains constant (one – your mother's mother's… mother). From a different angle, 75% of your grandparents are invisible to a mtDNA analysis – and every generation back, that percentage increases.

Or from yet another angle, a test for relatedness derived from mtDNA carries a risk of producing a false negative result that is incalculably high. A mitochondrial match is good evidence that that the two bearers are genealogically linked, but a non-match means nothing at all. Moreover, there is a wide zone between a match and a non-match: geneticists can cluster mtDNA sequences by their degrees of similarity to one another. Thus the coalescence of the mtDNA sequences of a large population into a small number of basic groups can suggest a founder – a mother – for each of those groups.

Consider, though, what being a "member" of a 15,000-year-old mitochondrial "clan" actually implies. How many ancestors did you actually have 15,000 years ago? Conservatively assuming 25 years per generation yields 600 generations, and your 2-to-the-six-hundredth-power ancestors comprise a number with 180 zeroes, or about 173 orders of magnitude larger than the number of people alive at the time, and effectively beyond the power of language to express.

Let us call this a squijillion.

Not only do you have a squijillion ancestors 15,000 years ago, but so does everybody else. How could you have so many ancestors? Many of them are the same people – specific ancestors recur in your own tree, and many of your ancestors are other people's ancestors as well. That is to say, to some extent you are inbred, and to some extent you are related to everyone else.

And of those squijillion ancestors distributed among the 10,000,000 or so people alive back then – the ones who all contributed nuclear DNA to your genome – how many are being detected by your mtDNA? One.

Here the tenuous connection between meaningful relatedness and biological relatedness becomes helpful. There is almost nothing biological there, but the cultural associations of DNA give these data the appearance of familial association, of science, of reality. The mtDNA similarity is symbolically powerful in spite of being biologically trivial in this context.

The intersection of that symbolic power with the free market has created a hybrid nature for the science of human population genetics: partly derived from Watson and Crick, that is to say, from molecular genetics; and partly derived from P. T. Barnum, that is to say, from the fellow who said epigrammatically, "There's a sucker born every minute".

Suppose there were a scientific test that allowed you to identify all of your family members and distinguish them from people to whom you were not related? You might find distant relatives you never knew you had; you might find that you are descended from someone noteworthy; you might find something exotic, romantic, interesting, or even admirable in your DNA. You might even be able to fill in gaps in your self-identity and find out who you "really" are and where you "really" come from. That is, after all, the source of a classic dramatic arc, from Oedipus to Skywalker.

But what would such a test entail? After all, heredity is probabilistic. You have, on the average, 25% of your DNA from each of your grandparents. Or more to the point, any bit of any grandparent's DNA has a 25% chance of showing up in your genome. Consequently, you may not necessarily match any specific bit of your grandfather's DNA – since you have three other grandparents and only two sets of DNA.

Moreover, since you are related to every other human being, there is no qualitative break between your family members and non-relatives that a genetic test could detect. That is the "constructedness" of human kinship systems: some people are defined as relatives and some people are not, regardless of their biological relationships. The only kind of test that can reliably sort people into your relatives and your non-relatives would be a magic test.

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In America, hardly any social fact can be understood outside the historical context of slavery. One modern legacy is the obliteration of the pre-slavery ancestry of African-Americans. But what if your DNA matched that of an African tribe? Would that not provide a grounding in African soil and establish African kin? For a few hundred dollars, indeed that service is now provided.

One pioneering company's website "allows you to reconnect to your ancestral past—easily, accurately and profoundly" and will "connect your ancestry to a specific country in Africa and often to a specific African ethnic group." And there is no doubt that it does what promises – it connects black Americans to black Africa. But of course, that is a sloppy term – "connects" – sounding as if it has profound biological meaning, when the profound connection it provides may be more emotional than genetic. After all, of the literally thousands of genetic ancestors you had 12 generations ago – say, about the year 1700 – mtDNA is connecting you with only one. On the other hand, isn't that better than nothing?

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Well, when you consider the fact that all of these mtDNA forms are polymorphic – that is to say, varying within any population; and that the sampling of Africans is very poor, you have to begin to wonder whether a mitochondrial DNA match to a Yoruba may actually be worse than nothing. Being biologically meaningless, yet mimicking a hereditary identity, the mtDNA match might well be giving you a false identity in the name of science.

As the classic 1973 film "The Sting" showed clearly, the best scams are the ones in which the victim does most of the work. You give them the dots, and they connect them – to your advantage. In this case, the clients are paying for science and are getting it. They are getting accurate DNA results and true matches. The companies certify the match, and allow their clients to make the meaningful "connection".

Testimonials vouch for the lives thereby changed, and why shouldn't they? The only problem might be if you confuse them for scientific evidence.

Ultimately this essay is not intended as a public service or a whistle-blowing venture. Nothing illegal or even necessarily immoral is going on. Instead, this is an illustration of the way in which science has changed during our lifetimes. Science – and in particular, genetics – may never have been "pure", but until quite recently it never had to compete seriously with the profit motive for its public credibility.

In short, this isn't your grandfather's genetics.

Jonathon Marks was a Visiting Research Fellow at the Genomics Forum in October 2007.

Further Reading:

Bolnick, D. A., Fullwiley, D., Duster, T., Cooper, R. S., Fujimura, J., Kahn, J., Kaufman, J., Marks, J., Morning, A., Nelson, A., Ossorio, P., Reardon, J., Reverby, S., and Tallbear, K. (2007) The science and business of genetic ancestry testing. Science, 318:399-400.