# 10 DNA Testing Myths Busted, and Other Favorite Posts

*By Blaine T. Bettinger*

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## 10 DNA Testing Myths Busted

(Originally posted October 25, 2007)


1. **Genetic genealogy is only for hardcore genealogists.**

Wrong! If you've ever wondered about the origins of your DNA, or about your direct paternal or maternal ancestral line, then genetic genealogy might be an interesting way to learn more. Although DNA testing of a single line, such as through an mtDNA test, will only examine one ancestor out of 1024 potential ancestors at 10 generations ago, this is a 100% improvement over 0 ancestors out of 1024. If you add your father's Y-DNA, this is a 200% improvement. Now add your mother's mtDNA, and so on. However, please note the next myth:

**2. I'm going to send in my DNA sample and get back my entire family tree**.

Sorry. DNA alone cannot tell a person who their great-grandmother was, or what Italian village their great-great grandfather came from. Genetic genealogy can be an informative and exciting addition to traditional research, and can sometimes be used to answer specific genealogical mysteries.

**3. I would like to try genetic genealogy, but I'm terrified of needles.**

Good news! Genetic genealogy firms don't use blood samples to collect cells for DNA testing. Instead, these companies send swabs or other means to gently obtain cells from the cheek and saliva.

**4. I would like to test my ancestor's DNA, but they died years ago**.

You don't always need your ancestor's DNA to get useful information from a genetic genealogy test. If you are male, you contain the Y-chromosome (Y-DNA) that was given to you by your father, who received it from his father, and so on. Both males and females have mitochondrial DNA (mtDNA), which was passed on to them by their mother, who

received it from her mother, and so on. Everyone of us contains DNA (Y-DNA and/or mtDNA) from our ancestors that can be studied by genetic genealogy.

**5. I want to test my mother's father's Y-DNA, but since he didn't pass on his Y-chromosome to my mother, I'm out of luck.**

Wrong! There is a very good chance that there is another source of that same Y-DNA. For instance, does your mother have a brother (your uncle) who inherited the Y-DNA from his father? Or does your mother's father have a brother (your great-uncle) who would be willing to submit DNA for the test? Sometimes there might not be an obvious source of "lost" Y-DNA, or no one in the family is willing to take a DNA test. The secret to solving this problem is to do what every good genealogist does – use traditional genealogical research (paper records, census information, etc) to "trace the DNA". Follow the line back while tracing descendants in order to find someone who is interested in learning more about their Y-DNA. This applies to finding a source of mtDNA as well.

**6. Only men can submit DNA for genetic genealogy tests, since women do not have the Y-chromosome.**

Wrong! Most genetic genealogy testing companies also offer mtDNA testing. Both men and women have mtDNA in their cells and can submit that DNA for testing. In addition, women can test their father's or some other male relative's Y-DNA to learn more about their paternal ancestral line, even though they did not inherit the Y-chromosome.

**7. My genetic genealogy test will also reveal my propensity for diseases associated with the Y-chromosome and mtDNA.**

Wrong, thank goodness. Most of the information obtained by genetic genealogy tests has no known medical relevancy, and these firms are not actively looking for medical information. It is important to note, however, that some medical information (such as infertility detected by DYS464 testing or other diseases detectable by a full mtDNA sequence) might inadvertently be revealed by a genetic genealogy test.

**8. I don't like the thought of a company having my DNA on file or my losing control over my DNA sample.**

This is, of course, an understandable concern. However, most testing firms give a client two options: the DNA is either immediately destroyed once the tests are run, or it is securely stored for future testing. If the DNA is stored, the firm

will typically destroy the DNA upon request. If the long-term storage of DNA is a concern, be sure to research the company's policy before sending in a sample.

**9. If my test reveals Native American ancestry, I plan to join a particular Native American affiliation group.**

Although genetic genealogy can potentially reveal Native American ancestry (for instance, my mtDNA belongs to the Native American haplogroup A2), it is incredibly unlikely that this information will be sufficient to positively identify the specific source of the lineage (such as a tribe) or allow membership in a particular Native American affiliation.

**10. My DNA is so boring that genetic genealogy would be a waste of time and money.**

Very wrong! A person's DNA is a very special possession – although everyone has DNA, everyone's DNA is different (okay, except identical twins – if your identical twin has been tested, you should think twice about buying the same test!). As humans settled the world, Y-DNA and mtDNA spread and mixed randomly. As a result, it is impossible to guess with 100% assurance that a person's Y-DNA or mtDNA belongs to a particular haplogroup (a related family of DNA sequences) without DNA testing.

**BONUS MYTH: My genetic genealogy test says that my mtDNA belongs to Haplogroup A2. Juanita the Ice Maiden, a frozen mummy discovered in the Andes Mountains in Peru, also has Haplogroup A2 mtDNA. Therefore, she must be my ancestor!**

Unfortunately, although genetic genealogy can reveal that a person is RELATED to an ancient DNA source, it cannot prove that a person is a DESCENDANT of an ancient DNA source. For instance, perhaps you are descended from Juanita's sister, or her 5th cousin. Thus, although Juanita might be your great-great-great-great…great-grandmother, she might instead be your great-great-great-great…great-aunt. And since Juanita died when she was just 12 to 14, it is unlikely she has any descendants.

If you understand the risks associated with genetic genealogy (such as the detection of non-paternal events) and other risks) and are ready and willing to embrace the results to learn more about your genetic ancestry, then genetic genealogy might be for you. I recommend that you read archived posts here at The Genetic Genealogist, and do some online research through one of the many companies that offer genetic genealogy testing

## To Sequence or Not to Sequence - That is the Question

(Originally posted October 15, 2007)


An article appearing Sunday at Bloomberg.com, "Cheap, Detailed Genetic Testing Might Soon Be Ready for Market", highlights some of the recent developments in DNA sequencing. The article is a response to three studies published at Nature Methods which reportedly "explore cheap technologies to decipher and analyze individual patients' DNA by allowing researchers to quickly find the small portions of the human genome that make protein and describe them, while discarding irrelevant data."

According to the author of the Bloomberg article, "complete" DNA sequencing for as little as $300 could be ready within months. Although it is unclear what the author means by "complete", it is entirely foreseeable that SNP testing will soon be available for a reasonable price.

### All this leads to the question which is so hotly debated in the blogosphere - if inefficient sequencing becomes available to the average consumer, should they get their genome sequenced?

As the article points out, there are already around 1,000 different DNA sequencing tests which range in price from $200 to $3,000. However, Cathy Wicklund, the president of the National Society of Genetic Counselors, believes that people should "think hard before asking for complete genome testing":

"Just because we have the technology doesn't necessarily mean that we should jump to offer it," she said. "Consumers should ask themselves, `What is this going to tell me, is it going to give me information that's helpful right now?' "

There are a number of strong voices in this arena, others who believe that genomic sequencing without further extensive studies that link genotype and phenotype is useless and potentially harmful to any consumer who does not have a strong

genetics background. Although I respect this position, I believe that attempting to ward people away from genomic sequencing will prove to be ineffective. Genetics is about to leave the hands of the medical professional, and there's nothing we can (or perhaps should) do about it.

Fortunately or unfortunately, the wave is coming. In just months or a few short years, anyone will be able to open an envelope or log into a website and see their entire genomic sequence, from the very first nucleotide to the very last. **Thus the question is not whether people should get their genome sequenced - because they invariably will - but rather what can be done to educate consumers**. With a background in genetics, I know better than many consumers all the dangers that my genetic sequence will reveal. But I'll still be ready to swab my cheeks the instant I can afford a complete genomic sequence.

Is there really no proper place for the average non-geneticist, non-physician-assisted consumer in the whole genome market? What if I can't afford a genetic specialist - should I be denied the opportunity to sequence my genome? What if my health insurer refuses to pay for genetic sequencing? Should only the knowledgeable or the rich be allowed to learn more about their genes?

I would argue that there is a place for the "early consumer." Early consumers are the pioneers, the curious who do something because it is new and exciting and they want to learn more about the technology and about themselves. For instance, there are so many people that get into genetic genealogy even though they don't know the first thing about genetics. When they get their results back, they do what the human mind was designed to do - they go out and attempt to learn more (and helping educate them is exactly why I started The Genetic Genealogist in the first place). Luckily, there are already others who are leading the consumer education front.

All new technology comes with risks. Even genetic genealogy, the sequencing of a few SNPs or a few 100 base pairs, can reveal unexpected or unwanted results. But should the risks really cause so much fear and caution? We are who we are regardless of whether or not we get sequenced. Sequencing just arms us with information that could, now or in the future, be useful. For me, the benefits far outweigh the risks.

In my opinion, the answer is to educate, educate, educate. Convincing people that their genome is scary or useless will dissuade very few from sequencing and will likely only alienate the pioneers.

## Top 5 Reasons to Save Your Grandmother's DNA

(Originally posted May 10, 2007)

**1. You got those big blue eyes from your grandmother, but chances are you inherited less desirable genes as well.** We inherit our DNA from our parents, who inherited it from their parents. Since we all possess genes that can cause or contribute to disease, knowing one's DNA and family medical history can be a great resource for someone who learns they have a genetic disorder.

**2. Full genome sequencing is right around the corner!** The X-prize quest for the $1000 genome will lead to efficient and affordable whole-genome sequencing. As commercial companies crop up and compete for customer's business, leading to even lower prices.

**3. Your grandmother's DNA contains clues to her ancestry.** X-chromosome, mtDNA, and autosomal genealogy tests contain clues to a person's ancestry, both recent and ancient.

**4. Even if you aren't interested in this whole genetic genealogy craze, somebody you know will be**! Genealogy is one of the most popular hobbies in America, and the use of DNA to augment traditional genealogical research is growing faster than ever. Chances are that someone you know will someday be interested in your grandmother's DNA!

**5. All the undiscovered possibilities.** No one knows what uses will be discovered for DNA in the future. Save that DNA just in case!

Disclaimer: Some people are very uncomfortable with the thought of gathering and storing a loved one's DNA, and those beliefs should be honored and respected. It is ALWAYS best to obtain your grandmother's permission before you gather her DNA. So don't delay, call her now!

## Famous DNA Review – Genghis Khan

(Originally posted May 21, 2007)

In 2003, researchers from around the world released a paper that suggested that 8% of all Mongolian males have a common Y chromosome because they are the descendants of Genghis Khan (See "The Genetic Legacy of the Mongols," 2003, Zerjal, et. al., *American Journal of Human Genetics*, 72: 717-721). The researchers examined the Y chromosome variability of over 2000 people from different regions in Asia and discovered a grouping of closely related lines. The cluster is believed to have originated about 1,000 years ago in Mongolia and its distribution coincides with the boundaries of the Mongol Empire.

Genghis Khan's empire (he ruled from 1206 – 1227) stretched across Asia from the Pacific Ocean to the Caspian Sea and was reportedly extremely prolific. Khan's son Tushi had as many as 40 sons. His grandson Kublai Khan is reported to have had as many as 22 sons, and perhaps many more. Together this family may have as many as 16 million descendants alive in Asia today. **It is extremely important to note that until DNA can be extracted from Khan's bones (which have never been found), there is no definitive proof that this Y chromosome cluster is actually descended from Genghis Khan**.

When Family Tree DNA compared the markers in the paper to their database, they determined that the Y chromosome cluster belongs to Haplogroup C3 (M217+). Forty-seven samples in their database at that time exactly matched the markers identified in the paper. The company has summarized the marker results from the paper and have made that information freely available.

A newly released study from Russian scientists examined the Y chromosomes of 1,437 men from 18 Asian ethnic groups (Altai Kazakhs, Altai-Khizhis, Teleuts, Khakasses, Shor, Tuvinians, Todjins, Tofalars, Soyotes, Buryats, Khamnigans, Evenks, Mongolians, Kalmyks, Tajiks, Kurds, Persians and Russians). The researchers discovered that approximately 35% of Mongolians possess the "Khan" Y chromosome. Surprisingly, the results of the study suggest that although the Mongol Empire held eastern Russia for 250 years, there are few "Khan" Y chromosome carriers in that region.

You can read more about the 2007 study at UK Channel 4.

## You and the $1000 Genome - Part I: The Archon X PRIZE for Genomics

(Originally posted May 22, 2007)

The Archon X PRIZE is a challenge from the Archon X PRIZE Foundation to foster the development of efficient and inexpensive genomic sequencing. Not only will the X PRIZE for Genomics change the face of medicine, but it will also have an ENORMOUS impact on the field of genetic genealogy, which we'll discuss in Part IV of this series.

**History of the Archon X PRIZE for Genomics:**
In 2003 the J. Craig Venter Science Foundation announced a $500,000 Genomic Technology Prize that would be awarded to an the group whose technology significantly enhanced "the field of high throughput DNA sequencing by enabling a human genome to be sequenced for $1,000 or less." The Foundation believed that crossing this threshold would enable the majority of individuals to afford genomic sequencing as part of medical treatment.

By 2006, Dr. Ventor's $1000 genome challenge was picked up by the X PRIZE Foundation to create the Archon X PRIZE for Genomics, a $10 million dollar incentive for the first successful team. **To win the prize purse, the registered group must build a device and use it to sequence 100 human genomes within 10 days or less, with an accuracy of no more than one error in every 100,000 bases sequenced (that's just 0.001%!!) for no more than $10,000 per genome**. As of May 2007 there are three teams registered for the competition; VisiGen, 454 Life Sciences, The Foundation for Applied Molecular Evolution (FfAME), and Reveo, Inc. If you're curious, Genomics & Proteomics Magazine has summarized a number of the leading technologies that are being developed in pursuit of the X PRIZE (very technical information).

In August 2005, the National Human Genome Research Institute announced that it had awarded grants in excess of $32 million to promote the development of sequencing technologies that would significantly lower the cost of whole-genome sequencing. At the time, it cost roughly $10 million to sequence a human genome (a 50-fold decrease from the previous decade), and the NHGRI set a final goal of $1000 or less for an entire genome. As the NHGRI pointed out, "the ability to sequence an individual genome cost-effectively could enable health care professionals to tailor diagnosis, treatment, and prevention to each person's unique genetic profile."

**Four years later, has there been progress?**
454 Life Sciences, for example, has just announced in March that they have essentially completed sequencing of James

Watson's genome, arguably the first time a single person's genome has been sequenced (the Human Genome Project's source of DNA was reportedly an amalgam of different sources). For those that don't know (can there be anyone?), James Watson is famous for having discovered the structure of DNA over 50 years ago. **Interestingly, Watson has asked 454 to withhold his results for the *apoE* gene - associated with Alzheimer's disease - as well as a number of other results, citing privacy concerns**. Watson, after all, has a son who received 50% of his genetic makeup from Watson's genome. In light of this, 454 has decided to hand over the results to Watson, who will then decided what to release to the public. (See Marshall, Eliot, "Sequencers of a Famous Genome Confront Privacy Issues" *Science* 30 March 2007:Vol. 315. no. 5820, p. 1780DOI: 10.1126/science.315.5820.1780). 454 estimates that the six-fold coverage of Watson's genome cost an estimated $1 million. Still a long way to go to reach the $1000 goal.

Meanwhile, Reveo, Inc. just joined the competition on April 30th of this year, but Reveo's founder, Dr. Sadeg M. Faris, believes that their technology will eventually be able to read an entire human genome "in minutes for pennies per genome."

The X PRIZE Foundation has released a video that explains the aims of the project. In the next post I will be examining whether or not the $1000 genome is really necessary considering recent developments in a related field.

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