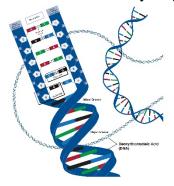
Introduction to Genetic Genealogy Shannon Combs-Bennett @tntfamhist | tntfamhist@kandsbennett.net

Why should I do this? This the question I am frequently asked when people are considering DNA testing for genealogy. Genetic genealogy has taken off in the last decade as access to testing has become easier and more affordable for genealogy. As with all new technical advances it has had its growing pains but has become a mainstay in the savvy genealogist's toolbox. As with everything we do in genealogy, DNA testing is considered a tool. It may not



DNA Advanced, from the National Institute of Health, image by Darryl Leja, NHGRI

give you definitive answers to your questions. However, coupled with paper genealogical research it can help validate possibilities. DNA has led to more than one person validating a hunch and proving a connection that was only theoretical on paper.

Many people are afraid of taking a DNA test when approached by an eager relative. In fact, many of you may have the same concerns. Here are three common concerns that I am asked regularly.

<u>Common Myth #1:</u> Will an insurance company or my employer be able to use my DNA test against me? No. In 2008 the US government passed the Genetic Information and Non-Discrimination Act (GINA) so that this will not happen. Right now the only people who have access to your DNA results at the genetic genealogy testing centers are you and those you give permission to. Your employers and your insurance companies cannot access this

information. For further details you can read about GINA here: <u>http://www.genome.gov/10002328</u>.

<u>Common Myth #2:</u> I don't have to exhume my ancestors to obtain their DNA do I? No. Their DNA is your DNA in most instances. If you don't have the type of DNA (yDNA or mtDNA) that needs to be tested you can always recruit living relatives to help.

<u>Common Myth #3:</u> These tests are done by blood draws and are really expensive because of it. No. The DNA testing companies that help genealogists use saliva samples and cheek scrapes to collect DNA samples. No needles needed!

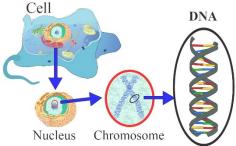
I hope you can see that there is really nothing to be worried about as far as modern world concerns. In fact, many people are more often shocked and concerned about the information a test brings to light. A topic we will cover in detail.

Useful Terminology:

As with all genealogy niches genetic genealogy has terms that you should be familiar with. Below are the more common words you will come across. It is by no means a complete list. I recommend people create a lexicon of their own of words they are unfamiliar with when

starting in the field. This way you will always have a definition (with a citation of where you got it) at your fingertips.

- Allele: specific form of a gene, one of multiple possibilities. In genetic genealogy used as the reference for a marker.
- Chromosomes: structure in the cell where genes are located.
- DNA (Deoxyribonucleic acid): a molecule that contains the genetic instructions for living organisms.
- Gene: a unit of heredity in living organisms. One gene may have multiple forms, or alleles, defined by different DNA sequences.
- Genome: the collection of genes in an organism
- Haplogroup: genetic population who share a common ancestor on the paternal or maternal line with similar SNP mutations.
- Haplotype: the set of numbers that make up your Y-Chromosome or mtDNA results.
- Locus: physical location of an allele on the chromosome.



Eukaryote_DNA.svg:(http://creativecommons.org/licen ses/by-sa/3.0) via Wikimedia Commons

- Marker: the specific place on a chromosome with two or more forms. The inheritance of it can be followed through each generation.
- MRCA: Most Recent Common Ancestor
- Phylo-tree: Phylogenetic Tree is the reference for diagrams that show all the haplogroups.
- Proxy: the contact person for the DNA test if not the person taking the test.
- SNP: Single Nucleotide Polymorphism, this test confirms you haplogroup by identifying if it has mutated from the ancestral state.
- STR: Short Tandem Repeat, the number of repeats for a base pair sequence which then determine the marker value.
- Sub-clade: an offshoot branch of the phylo-tree or a reference to deep clade testing.

Types of DNA Testing

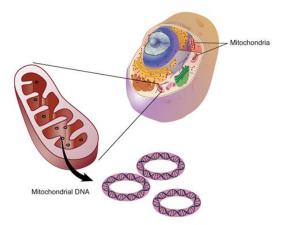
There are 3 tests for genetic genealogy: autosomal (atDNA), Y-chromosome (yDNA) and mitochondrial (mtDNA). In addition, you get a bonus result with some companies when testing your atDNA, which are results for your X-Chromosome. Each will tell you something different about your ancestry and when used together can help break down brick walls.

Autosomal DNA, also known as admixture DNA, it is the non-sex-determining chromosomes in a person's cell. Anyone may take this test and will give you ancestral results for about 7 generations back from you. Each generation loses genetic material from the generation before leading to a more difficult time in determining some cousin relationships after several generations.

A chromosomally normal human has 22 pairs of autosomal chromosomes. The 23^{rd} pair is a person's X or Y sex determining chromosome. Each of us inherited one chromosome from our father and one chromosome from our mother for each pair.

atDNA is best used in conjunction with paper genealogy. It can help confirm possible genealogical relationships. Testing more than one sibling, or more than one cousin, can help in this endeavor. Each person may have inherited a different segment of DNA from their parents or grandparents due to recombination.

yDNA testing is taken only men as it tests the Y-chromosome found in males. It is the sex chromosome that determines maleness and is passed from father to son. What will Y-DNA Testing tell you? Besides showing you a direct paternal line of inheritance, it will also give you the probable haplotype that you belong to giving you an idea of the origins of your ancient male relatives. After your test is finished you may choose to enter a surname study for men of your



National Institutes of Health. National Human Genome Research Institute. 'Talking Glossary of Genetic Terms.' http://www.genome.gov/glossary/

last name. This is a good way to find other men who share your ancestry and possibly make headway into genealogical brick walls.

An mtDNA test can be taken by anyone, male or female, and will trace direct maternal ancestors. It is passed down from a mother to all of her children. A child may trace their mother's line through the mitochondria in their cells. Mitochondria (mitochondrion singularly) are organelles found in most eukaryotic cells. That is to say they are found in cells that have internal structures enclosed by a cell membrane. Mitochondria are unique in that they have their own DNA. mtDNA Testing is a tool for Deep Ancestry. Research has lead scientist to believe that everyone is descended originally from "mitochondrial Eve" who lived approximately

140,000 years ago in Africa. The genetic population you are descended from is determined from the mutations identified in your mtDNA. These changes happened thousands of years ago helping the scientists to determine your ancestral ethnicity and geographic origins.

Everyone has an X-Chromosome, women 2 (one inherited from each parent) and men 1 (inherited from their mother). Analysis of xDNA can open up the branches to hidden lines that otherwise would be difficult to find DNA results for in other tests. It can also help you narrow down lines from different sides of the family when used in conjunction with atDNA. Analysis of xDNA is a relative new comer to genetic genealogy and there are new tools and tips made available every day.

Testing Companies

There are 3 main companies for genetic genealogy testing in the United States. The National Geographic Geneographic Project also has some interesting results for genealogy if you are interested in participating in it. Listed below are brief descriptions of the companies. For a nice comparison of all the companies, and their features, I suggest you see Tim Janzen's Autosomal DNA Test Comparison Chart found on the International Society of Genetic Genealogy Website www.isogg.org/wiki/Autosomal_DNA_testing_comparison_chart.

<u>FTDNA</u>: Family Tree DNA was founded by Bennett Greenspan in 2000 because he needed to solve a family mystery and DNA was the answer to his problem. Based out of Houston, Texas it offers complete genetic testing for genealogical purposes. https://www.familytreedna.com

<u>23and Me</u>: Based in Mountain View, CA this is a privately held personal genomics and biotechnology company. Testing began in 2007 and quickly became known for their medical genetics testing. https://you.23andme.com

<u>Ancestry DNA:</u> a segment of ancestry.com that handles all of their genetic testing and databases. They are primarily an autosomal testing facility and using software algorithms match your genealogical information to others who are potential relatives. There is a process where you can upload information on Y-DNA and mtDNA tests from other companies to your tree also. http://dna.ancestry.com/

National Geographic Genographic Project: launched 13 April 2005 by the National Geographic Society and IBM. It is a multiyear study in genetic anthropology that has the aim of creating a map of human migration patterns. They are collecting thousands of DNA samples from people all over the world to aid in the endeavor. https://genographic.nationalgeographic.com/

<u>My Heritage DNA:</u> genetic testing by MyHeritage began in November 2016. As the newcomer to the scene information on its testing is still coming out. I have recently taken a test but have not received my results back to give you an honest opinion. https://www.myheritage.com/dna

Ethics

I cannot stress enough how important it is to think about others, and not just your need for information, when you delve into genetic genealogy. While it would be nice and wonderful for all living relatives to willingly submit a DNA test not everyone will. Some don't care. Some are afraid. Still others don't want to know. You, as a researcher, must respect everyone's right to do with their body (and a DNA sample is part of person's body) what they want to.

Please remember this, if you administer someone else's results these results are not your results. You have no rights to them, you are not the owner of the DNA, the donor is. There are some researchers out there who feel that if they paid for it the results are theirs to do with what they will. This is far from the case. As the administrator (or purchaser) of the DNA kit you may ask for permission to see the results but you cannot keep someone from seeing their results.

In January 2015 Judy Russell author of The Legal Genealogist blog wrote a post about this topic. Titled "Whose DNA it is anyway?" http://www.legalgenealogist.com/blog/2015/01/18/whose-dna-it-is-anyway. This is a must read blog for continued discussion of genetics and ethics within the genealogy community.

On 10 January 2015 the Genetic Genealogy Standards Committee released the first ever guide for how genealogists should use results from genetic genealogy. The 3 page document, simply called "Genetic Genealogy Standards was designed to be a guide on how to be a good genetic genealogy researcher. For regular ole paper genealogy most people use the Board for Certification of Genealogists (BCG) standards that are published in their standards manual. This is the same type of guidelines but for genetics.

Additional Sources

Websites

- DNA Interactive http://www.dnai.org/
- Genetics Home Reference http://ghr.nlm.nih.gov/
- Guild of One Name Studies http://www.one-name.org/
- International Society of Genetic Genealogists (ISOGG) http://www.isogg.org/
- ISOGG Wiki http://www.isogg.org/wiki/Wiki_Welcome_Page

Blogs

- DNAeXplained by Roberta Estes http://dna-explained.com/
- On-line Journal of Genetics and Genealogy http://jgg-online.blogspot.co.uk/
- Musings on Genealogy, Genetics, and Gardening by Kitty Cooper http://blog.kittycooper.com/
- The Genetic Genealogist by Blaine Bettinger http://www.thegeneticgenealogist.com/13/
- Through the Trees by Shannon Christmas http://throughthetreesblog.tumblr.com/
- Your Genetic Genealogist by Cece Moore http://www.yourgeneticgenealogist.com/

Books

- Colleen Fitzpatrick and Andrew Yeiser. DNA and Genealogy. Rice Book Press, 2005
- David Dowell. *NextGen Genealogy: The DNA Connection*. ABC-Clio/Libraries Unlimited, November 2014.
- Debbie Kennett. DNA and Social Networking: A Guide to Genealogy in the Twenty-First Century. The History Press, November 2011.
- George Redmonds, Turi King and David Hey. Surnames, DNA and Family History
- Richard Hill. *Guide to DNA Testing: How to Identify Ancestors, Confirm Relationships, and Measure Ethnic Ancestry through DNA Testing,* (ebook) Atrax LLC, 2014.
- Megan Smolenyak and Ann Turner. *Trace Your Roots with DNA: Using Genetic Tests to Explore Your Family Tree.* Rodale Books, 2004.
- Terrence Carmichael and Alexander Kuklin. *How to DNA Test Our Family Relationships*. DNA Press, 2000