

Introduction to Genetic Genealogy

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There are two main topics that I am going to cover in this presentation. The first is how to determine which DNA test to take based on your goals. The second topic is how to choose a DNA testing company based on both the information that is provided by the company and how it is presented.

A list of genetic genealogy **terms** is available at <http://dnaadoption.com/index.php?page=glossary-of-dna-terms> and of **abbreviations** at <http://www.isogg.org/wiki/Abbreviations>.

To get started with genetic genealogy, you need to order a kit from one of the companies that offer DNA testing. The three main companies in the US are FamilyTreeDNA, 23andMe, and ancestryDNA. **Comparative information** is available at http://www.isogg.org/wiki/Autosomal_DNA_testing_comparison_chart.

Obtaining a sample for DNA testing is painless. FamilyTreeDNA uses cells obtained from the inside of your cheeks and their kit contains two “brushes” with which to collect the cells. Following cell collection, the brushes are ejected, one into each of the two vials provided with the kit which contain a preservative solution. Ancestry DNA and 23andMe each use spit as a source of DNA. Their kits come with a small vial containing preservative to which is attached a funnel into which you spit. Once you have collected a designated amount of spit, the funnel is removed and vial closed. If the testee is elderly and he/she has trouble making enough spit for the test you can request a cell collection kit similar to the one offered by FamilyTreeDNA.

When you have obtained your sample you mail it off to the testing company for analysis. Your sample is logged in, DNA is extracted from the cells in your saliva or cheek cells and copies made of your DNA in a process called amplification. This gives the company a sufficient amount of your DNA to run the test(s) that you have ordered. Your test results are then compared against those of everyone else in the company's database and those people whose DNA matches with yours are identified. Each company uses a different format for sharing match data with you.

Some **basic biology**. We all have 22 pairs of chromosomes in the nuclei of our cells, one member of each pair inherited from our mother and one member of each pair inherited from our father. These are referred to as the autosomes or autosomal DNA. We also have a pair of sex chromosomes, XX for women and XY for men. Women receive their father's X and one of their mother's two Xs; men receive one of their mother's two Xs and their father's Y. In the cytoplasm of our cells, in an organelle called the mitochondrion, which is another source of DNA. Mitochondrial DNA is present in the cells of both men and women but only women pass their mitochondrial DNA on to the next generation. A DNA test is available for each of these three types of DNA: Y DNA, mitochondrial (mt) DNA, and autosomal (at) DNA.

The **Y DNA test** is for men only as only men have a Y chromosome. Y DNA can be used to trace the direct male line, which in our culture also typically coincides with the family surname; it is passed down from father to son to his son to his son virtually unchanged. If you visualize a standard family tree with fathers on the left and mothers on the right, the direct paternal line is the far left-hand side of the family tree. There are two types of Y-DNA testing, STR offered by FamilyTreeDNA (http://www.isogg.org/wiki/Y-DNA_STR_testing_chart) and a SNP test offered by 23andMe and as the Big Y at FamilyTreeDNA (http://www.isogg.org/wiki/Y-DNA_SNP_testing_chart). The Y DNA STR markers tested generally are slow to mutate, typically mutating once every 300 or 400 years. For an explanation of the difference between a STR and a SNP see <http://dna-explained.com/2014/02/10/strs-vs-snps-multiple-dna-personalities/>.

If we look at the direct male line in terms of time, in my own family, my brothers were born in the 1950s, my father in 1919, grandfather in 1889, great grandfather in 1856, and 2x great grandfather in about 1820 and so on back through time, each generation being about 30 years. Also included in that direct male line are descendants of male siblings of your father, your grandfather, your great-grandfather, your great-great-grandfather, etc. If you are a woman and you are interested in tracing your paternal line then you can have DNA from a brother, your father, or any of the other men in the direct male line tested.

Approximately 10% of men who do Y DNA testing discover that they are not who they think they are, either due to an unknown adoption in the paternal line, a name change, for example when a woman with children from a previous marriage remarried and her children took their step-father's surname, or infidelity. When any one of these events occurs to disrupt the paternal line, it is referred to as a non-paternal event or NPE and it can be difficult to determine where in the family tree the NPE occurred.

Y DNA testing can identify patrilineal relatives many thousands of years in the past. My brothers' have an unusual haplogroup (G2a4) and terminal SNP (L-91), and if you will pardon the pun, I have unearthed a patrilineal ancestor with whom we likely share a common ancestor about 10,000 to 12,000 years ago, Ötzi the Iceman. Ötzi's 5300 year old corpse was discovered in 1991 on the mountain border between Austria and Italy. In a recent study, led by Walther Parson at Innsbruck Medical University, Ötzi was shown, based on Y chromosome markers, to be of haplogroup G and to have the rare L-91 SNP, a haplogroup and SNP he shares with my brothers. For a general article on Ötzi see <http://www.thegeneticgenealogist.com/2013/10/16/identifying-otzi-the-icemans-relatives/> and for a scientific abstract <http://www.fsigenetics.com/article/S1872-4973%2813%2900136-1/abstract>.

On that note, let's turn to **mitochondrial DNA**. Mitochondria are small organelles located in the cytoplasm of the cell and are a source of energy for the cell. Some cells have only a few mitochondria but cells such as muscle cells which require a lot of energy have many thousands of mitochondria. Mitochondria are thought to be bacterial in origin and have their own DNA. When an egg and a sperm cell fuse at fertilization, mitochondrial DNA from the egg but not the sperm cell is passed on to the resulting offspring, both male and female. So in much the same way that Y DNA can be used to trace the patrilineal line, mitochondrial DNA can be used to trace the matrilineal line. Both men and women can use this test.

Up until about five years ago, mitochondrial DNA testing was the only test available to women. However it is not particularly useful in a genealogic time frame because, unlike Y DNA which is

generally associated with the family surname, mitochondrial DNA generally is not associated with a family surname and the lineage can be difficult to trace. However the test can be useful in cases such as the recent use of mitochondrial DNA to identify bones found at Whitefriars in England as those of King Richard III. Mitochondrial DNA from a direct descendent of Richard III's sister, Anne of York was tested. Richard and his sister both inherited the same mitochondrial DNA from their mother which was then passed down through Anne's female descendants. Information on mtDNA tests is available at http://www.isogg.org/wiki/MtDNA_testing_comparison_chart. See also http://www.isogg.org/wiki/MtDNA_tools.

Mitochondrial DNA also can be useful in more specialized situations such as confirming a matrilineal relationship or inferring a particular ethnic maternal ancestry.

A new DNA test which became available about six years ago is autosomal DNA testing. http://www.isogg.org/wiki/Autosomal_DNA. This test can be used to identify any genetic relatives with a MRCA within about seven generations or back about 210 years.

To make the most of autosomal DNA testing, it is helpful to have relatives from particular family lines of interest also tested. So in addition to immediate family members, my two brothers and my son, I have had tested a female paternal first cousin (she shares both my paternal grandmother's and my paternal grandfather's ancestry plus my paternal grandmother's X chromosome), and two paternal cousins (one a second and one a third), who share only my paternal grandfather's ancestry. On my maternal side, I have had tested three cousins who share only my maternal grandfather's line, and five cousins who share only my maternal grandmother's line. At a superficial level, if someone matches me a one of my cousins, I know through which line they are related. On a more sophisticated level, using the information as to how my cousins match with me and each of my brothers I can build up a map showing which portions of each of chromosome pair have been inherited from which ancestors.

In choosing additional relatives to test and whether they likely will match with you, keep in mind the amount of DNA that you may share. As an example, you share about 50% of your DNA with your parents but the amount of DNA you share with siblings can vary widely, so it is a good idea to test as many siblings as possible. For other relatives, the amount of expected shared DNA drops by half with each generation. For the amount of DNA that you can expect to share with various relatives, see http://www.isogg.org/wiki/Autosomal_DNA_statistics.

The final topic I am going to cover is how do you decide which testing company is right for you? The three companies that I'm going to discuss are FamilyTreeDNA, 23andme, and ancestryDNA. The cost of doing autosomal DNA testing at each company is \$99. However each company varies as to what else they offer you for your \$99.

In the case of both FamilyTreeDNA (if you order the FamilyFinder test) and ancestryDNA, the only test that will be performed on your DNA sample is the autosomal DNA test. FamilyTreeDNA will also provide matching information on your X chromosome(s) for your autosomal DNA matches. The raw DNA data, including the X chromosome data, can be downloaded and then uploaded (free) to a third party website such as www.gedmatch.com where you can look for matches with people who have tested at other companies. If you wish

to do Y DNA or mitochondrial DNA testing there is an extra charge for doing that at FamilyTreeDNA. AncestryDNA no longer offers these tests.

FamilyTreeDNA has hundreds of projects run by volunteer administrators, including surname projects, geographic projects, haplogroup projects and so on, all of which are free. They also offer extensive information on not only who your matches are, but how you match them through their chromosome browser, who the matches are you have in common with another match, and the option to either upload or enter a family tree. To get the most out of testing with FamilyTreeDNA it is helpful to understand some of the science behind the tests.

Until recently, the niche that 23andme has enjoyed is not only providing genealogical information but also information about the health aspects of your genetic makeup. The latter is currently on hold pursuant to requirements by the FDA that 23andme document the accuracy of the information that they are providing. Because 23andMe provided medical information they have a rather convoluted interface where you have to invite somebody with whom you match to share your genome information before you can see how they match with you, i.e. on which chromosome(s). The person to whom you send a request to share genomes may either accept or decline the request. If you are male you receive your Y DNA test at no additional charge, and the mitochondrial DNA information for both men and women is provided at no additional charge. There is a rather nice option that allows you to see if someone you match matches with some or all of the same people that you do.

AncestryDNA capitalizes on the many thousands upon thousands of family trees in its database and if both people in a match have public trees, will identify surnames in common to the two trees and if you are really lucky, will identify the ancestors common to both trees who are your most recent common ancestors. No chromosome information is provided on matches, but you can see this if you upload your raw data to www.gedmatch.com.

Each company provides ancestral information.

Raw data from both ancestryDNA and 23andMe (V3 chip only) can be transferred to FamilyTreeDNA. Many people, especially adoptees, test first at ancestryDNA and transfer data to FamilyTreeDNA (cost is \$39) and also test at 23ndMe so that they can search for relatives in all three databases.

If you have a well documented tree, don't want to mess with understanding all the science behind the tests, and your last few generations of ancestors are in the US, you would likely have a lot of success at ancestryDNA. AncestryDNA currently is not sold outside the US..

If you are foreign born or your parents or grandparents were immigrants, you may have your best luck with 23andMe. But if you like all the bells and whistles and are not totally thrown by the more scientific interface at FamilyTreeDNA, it is the company that I would recommend as, for me, it has by far the easiest website to navigate and make contact with matches of interest.

Happy hunting!