Introduction to Genetic Genealogy

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"Put a scientist to work on your genealogy"

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 DNA from 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 cells in your saliva as a source of DNA. Their kits come with a small vial, containing preservative, to which you attach a funnel (provided) into which you spit. Once you have collected a designated amount of spit, the funnel is removed and vial closed.

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 millions of 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 algorithm for determining a match and they each use 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, two X chromosomes (XX) for women and an X and a Y (XY) for men. Women receive their father's single X chromosome and a recombined version of their mother's two Xs; men receive a recombined version of their mother's two Xs and their father's Y chromosome.

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 as the Big Y at FamilyTreeDNA (http://www.isogg.org/wiki/Y-DNA_SNP_testing_chart) and used by 23andMe to determine the the Y haplogroup. 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/.

The direct male line includes brothers, father, paternal grandfather, paternal great grandfather and so on, each generation being about 25-30 years. Also included in that direct male line are all the men who share your family surname: male descendants of male siblings, descendants of father's male siblings, and those of your grandfather, your great-grandfather, your great-great-grandfather, etc. If you are a woman tracing your paternal line you can have a brother, your father, or any of the other men in the direct male line tested.

When testing it is important to test a sufficient number of markers to obtain reliable match results. As shown in the table below, at 12 markers, even an identical match is not useful unless it is with someone with the same surname. You need to test at least 37 markers and then the match would need have no more than one difference (genetic distance = 1) with the man who has tested and preferably only 1 to 2 differences at 67 or 111 markers.

The expected relationship between you and your Y-chromosome DNA (Y-DNA) match is dependent on both the number of markers you have tested and the genetic distance. The chart below shows the interpretation of your relationship at each testing level (Y-DNA12, Y-DNA37, etc.) for relevant genetic distances.

For example, if you and your match have both tested at the Y-DNA37 level and are a 36/37 match this is a genetic distance of one. You are then considered tightly related.

	Y-DNA12	Y-DNA25	Y-DNA37	Y-DNA67	Y-DNA111	Interpretation
Very Tightly Related	N/A	N/A	0	0	0	Your exact match means your relatedness is extremely close. Few people achieve this close level of a match. All confidence levels are well within the time frame that surnames were adopted in Western Europe.
Tightly Related	N/A	N/A	1	1-2	1-2	Few people achieve this close level of a match. All confidence levels are well within the time frame that surnames were adopted in Western Europe.
Related	0	0-1	2-3	3-4	3-5	Your degree of matching is within the range of most well-established surname lineages in Western Europe. If you have tested with the Y-DNA12 or Y-DNA25 test, you should consider upgrading to additional STR markers. Doing so will improve your time to common ancestor calculations.
Probably Related	1	2	4	5-6	6-7	Without additional evidence, it is unlikely that you share a common ancestor in recent genealogical times (1 to 6 generations). You may have a connection in more distant genealogical times (less than 15 generations). If you have traditional genealogy records that indicate a relationship, then by testing additional individuals you will either prove or disprove the connection.
Only Possibly Related	2	3	5	7	8-10	It is unlikely that you share a common ancestor in genealogical times (1 to 15 generations). Should you have traditional genealogy records that indicate a relationship, then by testing additional individuals you will either prove or disprove the connection. A careful review of your genealogical records is also recommended.
Not Related	3	4	6	>7	>10	You are not related on your Y-chromosome lineage within recent or distant genealogical times (1 to 15 generations).

A percentage of men who do Y DNA testing discover that they are not who they think they are: the men with whom they match have a different surname, 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.

Y-DNA haplogroups can be used to infer ancestry. Broadly speaking, a haplogroup is a collection of mutations common to people of shared deep ancestry. The seven Asian branches are derivations of the African branches and further branch off into Oceania, Europe and America. All European lineages are derivations of African and Asian branches. All American Indian lineages are derivations of Asian branches. You will receive your Y DNA haplogroup when you do a Y DNA test at FamilyTreeDNA and when you o an autosomal test at 23andMe.

M**itochondrial DNA (mtDNA)**. Mitochondria are small membrane bound organelles located in the cytoplasm of the cell and are a source of energy for the cell. The number of mitochondria per cell varies widely. In humans, erythrocytes (red blood cells) do not contain any mitochondria, whereas liver cells and muscle cells may contain hundreds or even thousands. Mitochondria, thought to be bacterial in origin, have their own DNA. When an egg and a sperm cell fuse at fertilization, mtDNA 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, mtDNA can be used to trace the matrilineal line. Both men and women can use this test.

Up until about eight years ago, mtDNA 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, mtDNA generally is not associated with a family surname and the lineage can be difficult to trace. Unlike nuclear DNA, mtDNA is not specific to one individual. It therefore is used in combination with other evidence, for example anthropological information, to establish identification.

The greater number of copies of mtDNA per cell increases the chance of obtaining a useful sample, and a match with a living relative is possible even down a great many generations. Outlaw Jesse James's remains were identified using a comparison between mtDNA extracted from his remains and the mtDNA of the son of a female-line great-granddaughter of his sister. Stone AC, Starrs JE, Stoneking M (January 2001). "Mitochondrial DNA analysis of the presumptive remains of Jesse James". J. Forensic Sci. 46 (1): 173–6. mtDNA was recently used to identify bones found at Whitefriars in England as those of King Richard III: Richard and his sister both inherited the same mtDNA from their mother which was then passed down through 17 generations of Anne's female descendants to a living descendant who was an identical match. 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.

MtDNA can be useful not only in specialized situations such as confirming a matrilineal relationship but also for inferring a particular ethnic maternal ancestry based on the mtDNA haplogroup. See https://en.wikipedia.org/wiki/MtDna_haplogroups_by_populations and https://en.wikipedia.org/wiki/Human_mitochondrial_DNA_haplogroup. You receive your mtDNA haplogroup as part of your mtDNA test at FamilyTreeDNA and also when you do an autosomal DNA test at 23andMe.

The European mtDNA haplogroups consist of H, I, J, K, T, U, V, W, and X. Asian mtDNA haplogroups are A, B, C, D, F, G, M, and Z. African mtDNA haplogroups are L1, L2, and L3. Other haplogroups are as noted on the map above.

Autosomal DNA. A DNA test which became available about eight 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 most recent common ancestor (MRCA) within about seven generations or back about 210 years.

To make the most of autosomal DNA testing, it is helpful also to have relatives from family lines of interest tested. So as funds allow, test all of the oldest (in generations) relatives you have in addition to your parents if they are alive, all your siblings, and as many first, second and third cousins who will indulge you and provide a sample. At a superficial level, if someone matches you and one of your cousins, you know through which line your match is related to you. On a more sophisticated level, using the information as to how your cousins match with you and each of your relatives, you can build up a map showing which portions of each of your chromosomes has 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.

Which testing company? 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 is $99 at AncestryDNA and FamilyTreeDNA (but check for sales). 23andMe is now charging $199. However each company varies as to what else they offer you for your testing fee.

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 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.

The niche that 23andme enjoys is that they not only provide genealogical information but also some information about the health aspects of your genetic makeup. Because 23andMe provides 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.

There is a new option to see chromosome information on your matches: if you opt in to sharing with all your matches, then you may see chromosome information on other 23andMe folks with whom you match who have also opted in without going through the invitation process.

If you are male you receive your Y DNA haplogroup at no additional charge, and the mtDNA haplogrouop 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 also 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 now see how much autosomal DNA you share. In order to see chromosome information you need to upload your raw data to www.gedmatch.com (free) or have your matches transfer their raw data to FamilyTreeDNA ($39)..

Each company provides ancestral information in the form of an estimate of ethnic composition which is continually being updated.

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. 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, in my opinion, it has by far the easiest website to navigate and make contact with matches of interest.

For online introduction classes to navigating the test company websites go to www.DNAadoption.com.