Using Autosomal DNA Testing for Family History

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March 2015

What is Autosomal DNA testing? How is it different from other popular types of DNA testing? How can Autosomal DNA testing help you tear down brick walls in your search for ancestors? What are its strengths and limitations? These are a few questions that this article hopes to answer.

In each human cell, the nucleus contains 23 pairs of chromosomes for a total of 46. See Figure 1 for a photo of the 23 pairs. These 23 pairs contain the nuclear DNA of a person. Two chromosomes are special – X and Y – the sex chromosomes. You receive one sex chromosome from your father and one from your mother. What you receive determines your sex – XX for females and XY for males.

The non-sex chromosomes or pairs 1-22 in Figure 1 are the *autosomal chromosomes*. Autosomal DNA testing uses carefully selected markers on these 22 pairs of chromosomes.

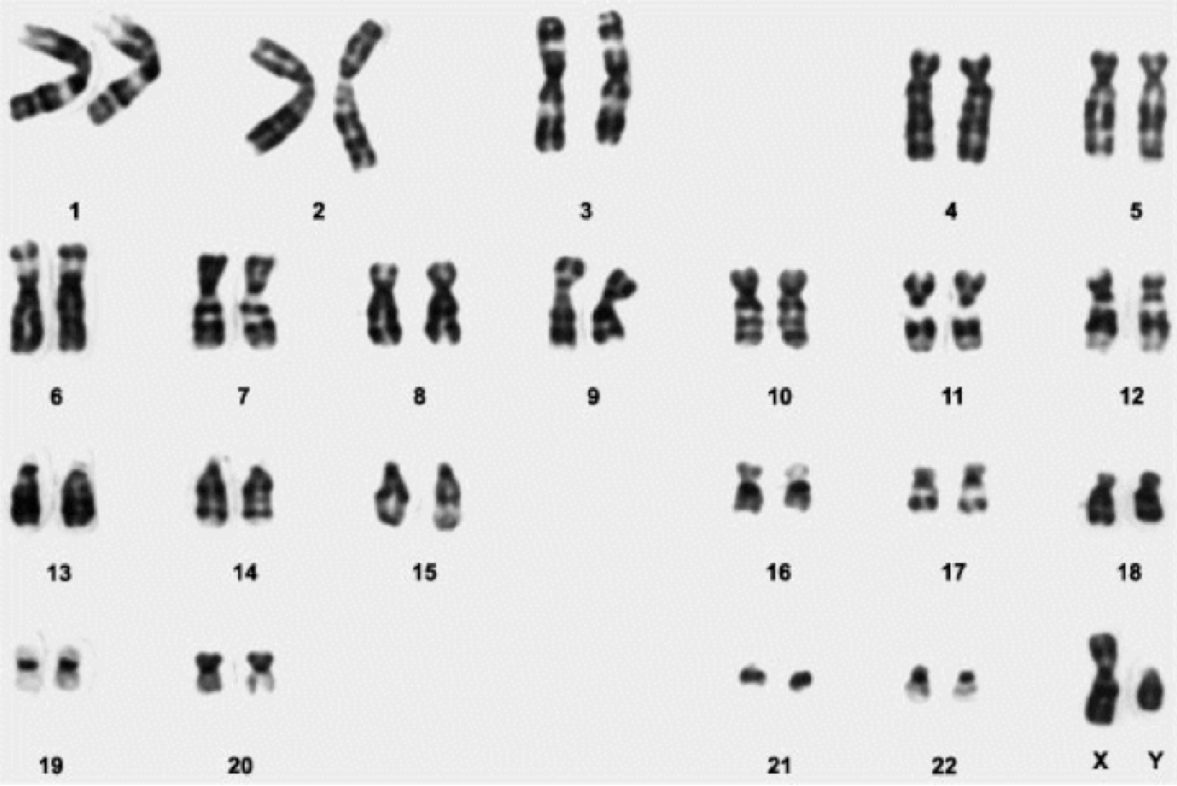

Figure 1: Photo of Human Chromosomes – 23 pairs for a total of 46 in a cell's nucleus.

Three Popular DNA Tests – Autosomal DNA (atDNA), Y-DNA, and Mitochondria DNA (mtDNA

There are three popular DNA tests that are of interest to genealogists. The Autosomal DNA (atDNA) test compares markers on the autosomal chromosomes (pairs 1-22). This article explores this test in detail.

The Y-DNA test compares markers on a man's Y chromosome. Since a father passes his Y chromosome on only to his sons, Y-DNA follows surname in most cultures. Since only males have a Y chromosome, only males can take a Y-DNA test. A man's Y-DNA results on its own reveal little.

Matches with others are the key. For example, Y-DNA testing may confirm paper trails of two lines of males with the same surname and indicate they have a recent common male ancestor.

The third DNA test uses markers in the DNA of a cell's mitochondria (mtDNA). As shown in Figure 2, mitochondria are rowboat shaped bodies in a cell's cytoplasm but outside of the cell's nucleus. These mitochondria provide energy for the cell. Mitochondria DNA (mtDNA) is genetic material that is passed from a mother to her children, but only her daughters pass it on. Therefore, mtDNA is useful for tracing direct maternal lines. Since a cell may contain hundreds of mitochondria and mtDNA is much more stable than nuclear DNA (the 23 pairs of chromosomes), mtDNA is valuable for identification of degraded remains, e.g., thousands of years old teeth.

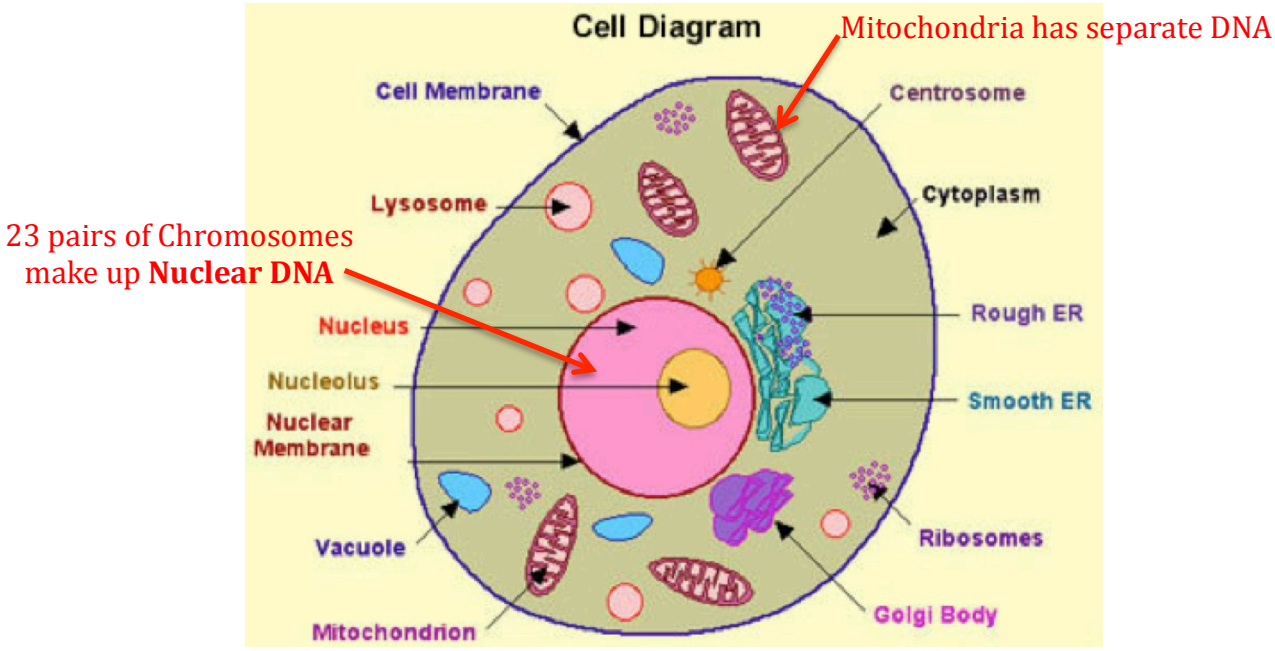

Figure 2: Cell diagram showing the Mitochondria in the Cytoplasm.

Autosomal DNA (atDNA) Test Companies

As mentioned above, the Autosomal DNA (atDNA) test compares markers on the autosomal chromosomes (pairs 1-22). The DNA testing company Family Tree DNA uses about 700,000 markers and calls their autosomal DNA test "Family Finder." Ancestry.com currently has only one DNA test "AncestryDNA" which is an autosomal one, and also uses about 700,000 markers but not the same ones as Family Tree DNA. The company 23andMe calls their autosomal DNA test "Relative Finder" and they state on their website it uses tens of thousands of markers. The current cost for atDNA testing at all three companies is $99 per test.

How Autosomal DNA (atDNA) is passed on to next generation

Whereas a man's Y chromosome is passed on to his sons essentially unchanged and a woman's mitochondria (mtDNA) is passed on essentially unchanged, we inherit a mix of atDNA from both parents (about 50% each) but it is shuffled and diluted with each new generation. An egg or a sperm only has 23 chromosomes and at conception the 23 from both parents combine to form the 46 of a normal cell. atDNA is scrambled when each woman's egg and each man's sperm are formed. When an

egg or sperm is formed, each atDNA chromosome (1-22) is made by using bits and pieces of the two chromosomes in the pair (recombination process). Note that the sex chromosome X and Y are handled differently.

As a result of this shuffling, you share about 50% atDNA with a sister or brother but a different 50% with each sibling! This is because each egg and sperm has its own shuffling for each of the 1-22 chromosomes.

Because of this shuffling and dilution of atDNA at each generation, genetic scientists steered clear of using atDNA for testing for a long time. Only recently has the technology with 700,000 markers allowed comparisons of individuals' atDNA for genealogy purposes.

How much atDNA is shared across generations?

You share about 50% atDNA with a parent. You share about 25% atDNA with a grandparent; about 12.5% with great-grandparents, etc. Back more than 5 generations the atDNA is too diluted to be useful.

The percentages of shared atDNA for cousins are halved when you come down the ancestral tree. For example, you share about 12.5% atDNA with a 1st cousin; about 3.125% with a 2nd cousin; and about 0.0488% with a 5th cousin. See Figure 3 for other percentages.

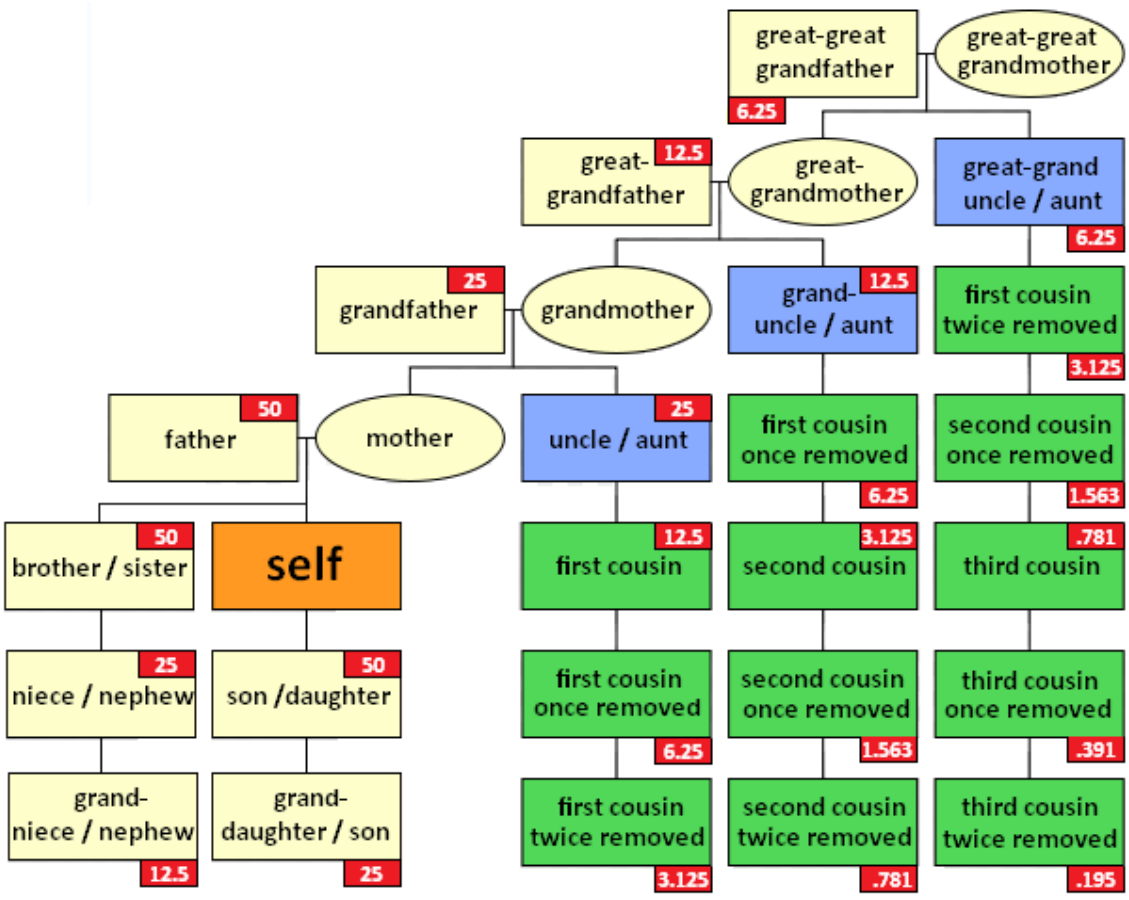

Figure 3: Average amount of autosomal DNA shared by you and your close relatives.

Figure 3 (courtesy Dimario, Wikimedia Commons) shows the average amount of autosomal DNA shared by you and your close relations up to the third cousin level.
Source: http://www.isogg.org/wiki/Autosomal_DNA_statistics

Note that these percentages are mathematical averages. The actual values can vary quite a bit. For example, I share 18% atDNA with my first cousin where the mathematical average predicts 12.5%.

How is atDNA test useful?

The atDNA test is useful for finding matches with relatives up to 5th cousins. When you test with a major DNA testing company such as Family Tree DNA, Ancestry.com or 23andMe, they supply you with the names of matches, estimates on the relations, and ways to contact the matches, usually via email. Both males and females can take the test and identify cousins from both their paternal and maternal lines. Unfortunately, since it is a relatively new DNA test, few people have tested with it and the people you are really seeking have not tested.

Besides identifying unknown cousins, an atDNA test can aid you in determining if a close relative, e.g., g-aunt, is a full or half relative. It could help you determine if a g-grandpa and g-grandma were first cousins.

The atDNA test "Problem"

Many family history researchers were excited about testing for atDNA. However, they were quickly disappointed. Though they received plenty of matches, they were frustrated when they discovered it was difficult to verify how a match may be related. Let explore this "problem" and why this can be so frustrating.

The atDNA test can identify relatives up to and including 5th cousins. Your 5th cousin and you share a common ancestor in a gggg-grandparent. To effectively use the atDNA test results, you need to determine as many descendants from your 64 gggg-grandparents as possible. Now most family history researchers have a tendency to search back in time by identifying parents of great-grandparents, etc. and not filling in all the collateral lines. Therefore, one source of frustration is that our research strategies of the past have not produced the information we need to verify our atDNA matches.

How many descendants would these 64 gggg-grandparents produce? Let's make a hypothetical guess. Say, on average, each union has five children that live to marry and have children. Assuming no cross marriages (pedigree collapse**)** as when two cousins marry, that's 19,530 possible descendants to research in the resulting six generations! Yipes! We have identified a second major source of frustration. There are potentially thousands of people you need to research.

In the next section we will see a third source of frustration. atDNA testing is an inexact science.

Details on how to compare atDNA.

The degree of atDNA sharing between two individuals is measured by the DNA testing companies in units of genetic distance known as *centiMorgans* although in practice it is not the total number of centiMorgans that is more significant but the length and number of *shared segments*. The percentages and the number of centiMorgans can vary. For example, a brother might share 53% of his DNA with

one sibling and 47% with another sibling. Because of the random way that autosomal DNA is inherited third, fourth and more distant cousins will not necessarily match you with the currently available autosomal DNA tests. According to Family Tree DNA's figures there is a 90% chance that third cousins will share enough DNA for the relationship to be detected, but there is only a 50% chance that you will share enough DNA with a fourth cousin for the relationship to be identified. These "estimates" and the resulting false matches are another source of possible frustration in determining if a match is a relative. (Source: http://www.isogg.org/wiki/Autosomal_DNA_statistics)

CentiMorgans (cM) are used to denote the size of matching DNA segments in atDNA tests. Segments which share a large number of centiMorgans in common are more likely to be of significance and to indicate a common ancestor within a genealogical time frame. 23andMe provides information on both the percentage of DNA shared and shared cM. Family Tree DNA does not provide percentages and only provides information on the shared cM. In order to get an approximate percentage of shared DNA from a Family Tree DNA Family Finder test, take all of the segments above 5 cM, add them together and then divide by 68. The way the calculation works is that your total genome in cM with the Family Finder test is 6770 cM. So dividing by 68 is a close approximation.

For example, the shared cM between you and a first cousin might be 850 cM. Dividing 850 by 68 gives 12.5% of shared atDNA which is what we would expect with a first cousin. Note determining that you share 12.5% atDNA with an individual does not necessarily imply he or she is a first cousin. Great-grandparents, first cousins, great-uncles, great-aunts, half-aunts/uncles, half-nephews/nieces all share 12.5% atDNA with you. Other information such as age must be used to sort out the relationship.

Note that AncestryDNA does not provide information on the shared centiMorgans or the percentage of shared atDNA. However, AncestryDNA customers can upload their raw data to the free GedMatch website in order to extract the necessary cM data for making comparisons and to check the relationship predictions. For GedMatch, see http://v2.gedmatch.com/login1.php and for an article about GedMatch see http://www.legalgenealogist.com/blog/2012/08/12/gedmatch-a-dna-geeks-dream-site/ David Pike's tools available at http://www.math.mun.ca/~dapike/FF23utils/ can also be used.

Poeple can download their raw atDNA files from Family Tree DNA, AncestryDNA, or 23andME and upload the results to GedMatch for free. This is a great way to expand your potential matches for cousins.

Use of Family Finder's Chromosome Browser

Comparing the percentage of shared atDNA of two individual is useful to predict their relations, such as 2nd or 3rd cousin. However, further detailed information can be obtained by using Family Finder's Chromosome Browser. 23andMe has a similar tool. The Chromosome Browser allows you to view on each chromosome where you share atDNA with another person (or up to five persons).

In Figure 4, a Family Finder's graphic shows the shared segments in orange where my first cousin and I share the same atDNA. Note that the 22 chromosomes are numbered down the left side with the X chromosome at the bottom. I match my first cousin on 58 shared segments (for a total of 1224.10 shared cM or 18%). The long orange segment on chromosome 4 is 83 cM wide. The gray areas are SNP poor areas and not tested by Family Finder.

The graphic of Figure 4 is nice to look at but how can you use the information? First, you can compare up to five individuals in the Chromosome Browser at the same time. Comparing one person with known

relation to you, e.g., first cousin on mother's side, with individuals of unknown relation, one looks for overlaps. For example, if an unknown person overlaps segments with the cousin on your mother's side, this might be an indicator that this unknown person is related to one of your mother's parents. This will narrow your paper-based search.

Figure 4: A graphic from Family Finder's Chromosome Browser

Figure 5 shows a graphic from Family Finder's Chromosome Browser of the shared atDNA between myself and the same first cousin (blue), his daughter (orange), and his son (green).

To help determine which DNA segments come from which parents, genetic genealogists use both *chromosome mapping* and *phasing*. Both techniques are helpful in determining how new genetic cousins are related to you. However, these two techniques are beyond the scope of this article. People who are interested should consult a good reference such as Chapter 12 in Emily D. Aulicino's book *Genetic Genealogy: The Basics and Beyond*, published by AuthorHouse LLC, 2014.

However, to use the Chromosome Browser effectively in this way, you need to test many close relatives, e.g., parents, grandparents, great aunts and great uncles from your several lines. Many family researchers do not have access to the appropriate relatives because they are not living or unavailable for a variety of other reasons.

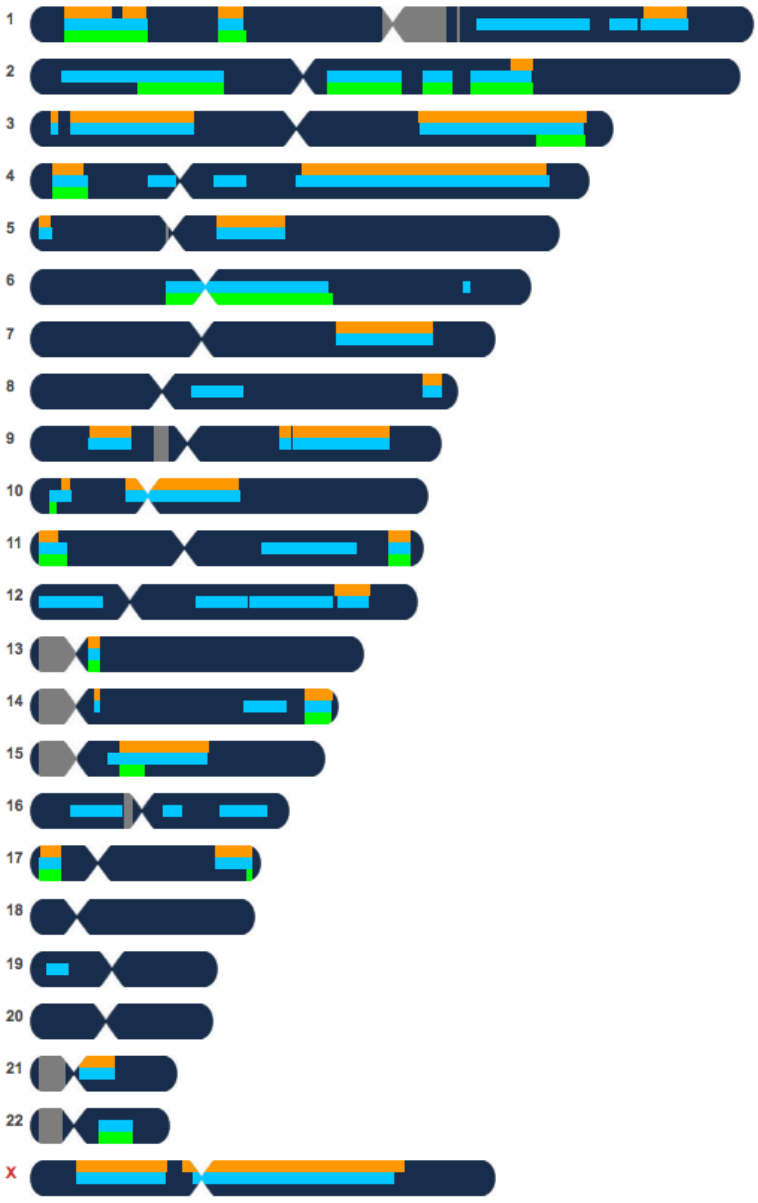

Figure 5: Graphic from Family Finder's Chromosome Browser showing shared atDNA between myself and my first cousin (blue), his daughter (orange), and his son (green).

Summary

Autosomal DNA testing provides another tool for the family history researcher's toolbox. Its strength is the ability to discover relatives up to 5th cousins. However, determining if a match is a true relative and how you are connected can be tricky.

DNA Testing Companies

23andMe – https://www.23andme.com
AncestryDNA - http://dna.ancestry.com
Family Tree DNA - https://www.familytreedna.com