DNA Testing – What you need to know first

This article is like the "Cliff Notes" version of several genetic genealogy classes. It is a basic general primer. The general areas include ...

Project support DNA test types – covering Y-DNA & SNP testing, mtDNA & atDNA tests Inheritance Genealogy Triangulation Ethnic Origins Helpful links

Generally speaking ...

You need to determine what you want to learn about your genetics. Then select the proper DNA test to achieve that goal. Always remember that DNA tests are tools and need to be compared to other DNA tests then correlated with paper trail genealogy to be most effective. This article should help you make a more educated decision.

Project support

There are many DNA projects (surname, geographical, haplogroup, et cetera) out there to help their related members. Some are better than others. The key is to educate your self and ask questions.

One example of a good Y-DNA Project, also known as a surname project, is the Carpenter Cousins Y-DNA Project. This project has documented over 118 Y-DNA profiles pertaining to Carpenter, Zimmerman or similar closely related variant surnames. It has a little over 400 members from several DNA testing companies. http://carpentercousins.com/carpdna.htm The critical elements of a good project include 1) having an introduction, explanation, methodology, discussion of results that are separate from the data tables which show which DNA markers are tested.

http://carpentercousins.com/carpdna.htm#toc

2) Organized groups are organized by genetic profiles or fingerprints related to the English based surname Carpenter or the Germanic version, Zimmerman. Ideally, all are related to a common genealogical ancestor within the genealogical time period. Name variants and those with adopted surnames can be included in the groups provided they match the genetic group profile and show an accepted geographic time and place of their ancestor interaction with a Carpenter or Zimmerman.

This project also has two unorganized groups for non-genetic matches. One of them is based on the common European Haplogroup R1b1b2 which has the short hand code of R-M269.

3) All groups are backed by a lineage page. These are drop down genealogical lineages showing the inter-relatedness of each member to the group ancestor. Those who match genetically, but not genealogically, are separated. Mutations to the group mean are also shown on the lineage page under the member ID number. http://carpentercousins.com/generallineage.htm

4) This project also includes general conclusions, helpful links and a FAQ or frequently asked questions section.

5) This Y-DNA project supports testing from several Y-DNA testing companies and has a genealogical research and support group called "Carpenter Cousins."

All good DNA projects should contain the basic elements above. Add in responsiveness to questions by the group administrators and one can learn a lot from such DNA projects.

DNA test types

There are different types of DNA tests. And some companies only do one type of test, while companies like FTDNA offer many types of tests.

Y-DNA Tests

The male Y Chromosome is passed from father to son virtually unchanged over the generations. This makes it ideal for surname testing.

When looking at a genealogical pedigree chart, the very top lineage is the paternal line and represents Y-DNA heritage. This is the father's fathers line.

Many companies used to have Y-DNA tests, but now only a few provide it. See comparison chart link below. And these companies test some but not all of the same DYS markers. And a few use different values (numbers) for the same DYS marker. Knowing when the test was done and by whom will allow us to convert the values into a standard format.

Y-DNA tests come in different sizes like 12, 25, 37, 67, 111. Generally speaking, the more DYS markers one uses, the higher the resolution or probability of relatedness to close matches. In general, one should consider 37 markers as the starting level.

See: <u>http://isogg.org/wiki/Y-DNA_testing_comparison_chart</u>

SNP Tests – Big Y

Single nucleotide polymorphism (SNP pronounced Snip) testing is a shotgun approach toward the Y-Chromosome. Most Y-DNA tests can estimate the basic haplogroup. SNP testing confirms the haplotyping of the Haplogroup. FTDNA calls theirs The Big Y. See the link for comparisons between the different companies who provide this type test.

http://isogg.org/wiki/Y-DNA SNP testing chart

Mitochondrial DNA tests

Mitochondrial DNA (mtDNA) is passed from the mother to her children, but only her daughters can pass it down to the next generation. Like Y-DNA this type of DNA is passed down virtually unchanged over the generations. When looking at a genealogical pedigree chart, the very bottom lineage is the maternal line and represents mtDNA heritage. This is your mother's mothers DNA. Traditionally the female assumes a married name each generation which makes it harder to track genealogically.

MtDNA is tested in Hyper Variable Regions often called HVR1, HVR2 & HVR3. A complete mtDNA test is referred to as mtFull at FTDNA. See comparison chart at:

http://isogg.org/wiki/MtDNA_testing_comparison_chart

Autosomal DNA Tests

Ancestry and 23andMe focus on autosomal DNA (atDNA) FTDNA has a similar test called Family Finder. Most people use these tests to see their ethnic heritages. Example: X% European, X% Middle Eastern, X% et cetera.

23andMe also uses atDNA type testing for medical genetic warning type tests as for Cystic Fibrosis, Sickle Cell Anemia, Hereditary Hearing Loss and et cetera.

Some use it to compare DNA fragments to others for cousin similarity up to about 5 generations.

On a genealogical pedigree chart atDNA represents all your ancestry. You share 50% of your DNA from each parent, 25% from each grandparent, then 12.5% by the next generation followed by 6.25%, 3.125%, 1.5625% and further divided numbers back into time.

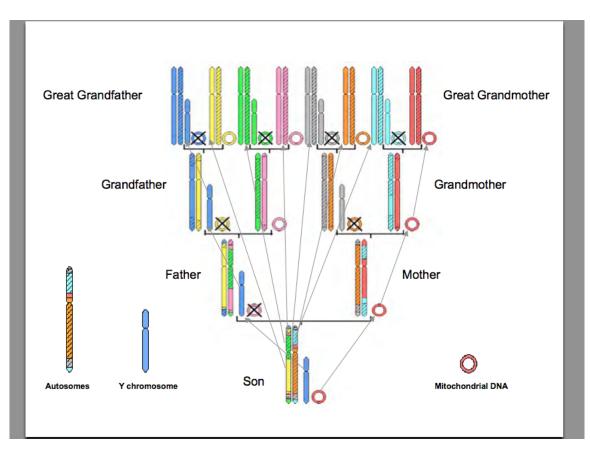
If you are surnamed Carpenter, any cousin match most likely will not be a Carpenter, but from one of your other ancestors. For example, at 5 generations the likely cousin testing match will be a Carpenter is 1/16 (one sixteenth), and more likely not surnamed Carpenter or 15/16. To see the differences between these atDNA testing companies, please go to the following link.

http://isogg.org/wiki/Autosomal DNA testing comparison chart

Are there other types of DNA tests? Yes. But the ones above are the most common ones used in genealogy. Others include **X-STR** and **paternity** tests, which also include **CODIS** markers. These tests are generally used in identification and familial matching.

Inheritance

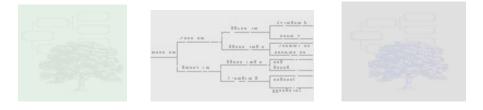
Here is a four generation inheritance chart. This shows the path of Y-DNA and mtDNA inheritance. And the atDNA segments we inherit, comes from all of our ancestors.



Please remember that those atDNA segments are reduced by 50% every generation back through time. We own 100% of our atDNA, but we get 50% from each parents during recombination (aka when a sperm joins the egg), 25% from our grandparents, then 12.5%, 6.25%, 3.125%, 1.5625%, 0.78125%, 0.390625, et cetera back in time. Simply put, after 5-6 generations the tiny amount of atDNA inherited from those generations becomes pretty much unusable.

Genealogy

Regardless of the DNA test, one also needs good genealogy for each person DNA tested. DNA tests without something to compare to is basically worthless. It is like genealogy without any documentation!



Pedigree genealogy (filling in a pedigree chart) back in time is adequate for Y-DNA and mtDNA tests. Compare person A to person B genetically then look for the genealogical most recent common ancestor. We call this triangulation. See triangulation in the next section.

But atDNA requires cousin genealogy. This is the descendants of everyone on the pedigree chart. You either compile it yourself or compile it from various genealogies from those being tested. And triangulation is much more complex because you need to trace at least one person descendant from each generation you are tracking. This means multiple atDNA tests and it really helps to have a dedicated computer program to sort out the myriad of atDNA segments that need to be tracked. GEDMATCH.com allows server time for this at a nominal cost.

Triangulation

Triangulation is a goal of genetic genealogy.

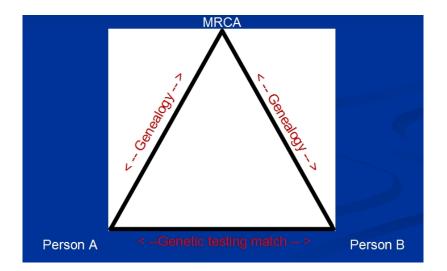
In genetic genealogy we use triangulation. Think of a triangle.

Genetic triangulation is rather simple. /_\

Person A & B match genetically and that forms the base of the triangle. _

Person A has a paper trail (genealogy) that goes back in time. / Person B has a paper trail that goes back in time. \

The top of the triangle is the MRCA or most recent common ancestor.



Person A is who you are testing. Some living biological male 2nd, 3rd or better cousin could be Person B. The most common shared ancestor is the MRCA.

If the genetics of Person A & Person B match and the paper trail goes to the MRCA, then this helps prove they are related both genealogically and genetically. This is the goal of genetic genealogy. When this is repeated several times back to a common ancestor, we then can recreate the Y-DNA markers of that ancestor. All without digging them up!

See more at: http://isogg.org/wiki/Triangulation

For many groups they have a recognizable common ancestor. For Group 2 (of the Carpenter Cousins Y-DNA Project) it is the immigrant William Carpenter b. abt 1610 in England. With triangulation we have re-created his genetic profile or fingerprint. The same goes for Group 3 and a few other groups.

Ethnic Origins

Most people use the atDNA for the recent genetic ancestry. This is usually reflects the last 300 to 500 years of your ancestry. But you need to take it with a liberal dose of caution. The estimations are just that. It is all based on mathematical modeling and reported ancestral locations.

Ethnic Makeup European	~~			and the second	Provent and
	93%				
British Isles		Aunt			
	43%	Iceland		Sweden	T S S S
Western and Central Europe				F	inland
	32%			Norway	in set prim
Eastern Europe	15%			Sara Es	stonia
Finland and Northern Siberia	1.3.70		66		itvia
Finand and Northern Siberia	3%		A GR	Denmark Lithu	Jania
Middle Eastern	0.0		IE	Germany Poland	Belarus
Middle Eastern	6%			BE CZ	man himmen buch she
	0 /0		Fra	AT AT	Ukraine Kazakhstan
Asia Minor	5%			Switzerland HR Ron	MD ania
Eastern Middle East	570			BA RS	BG Gaating and the set
Eastern Middle East	1%		Spain	AL	Azerbaijan
collapse all			P1	Gree	ece Turkey
conapse an				Tunisia	Syria Lebanon Afghanistan
			Morocco		Iran Iran
red Origins My Ancestral History				•	Jordan Kuwait Pakistan
All Matches 🔹	Rel	lation 💧	European	Middle Eastern 🖕	Egypt Saudi Qatar
			European	midule Lustern	Arabia Oman Indi
					Oman
Ruth Carpenter	Full S	Sihlinas	93%	7%	
	Full S	Siblings	93%	7%	Eritrea Yemen
	Half	Siblings,	93%	7%	Sudan Fritrea Yemen
Ruth Carpenter	Half : Gran	Siblings, adparent/			Sudan Eritrea Yemen Sudan Ethiopia Sri La
	Half Gran Gran Aunti	Siblings, adparent/ adchild, / Uncle,	93% 87%	13%	Sudan Ethiopia South Ethiopia Sudan Somalia Sri La
Ruth Carpenter	Half Gran Gran Aunt Niece	Siblings, Idparent/ Idchild, / Uncle, e/		13%	Sudan Eritrea Yemen Sudan Ethiopia Sri La
Ruth Carpenter	Half Gran Gran Aunt Niece Neph	Siblings, Idparent/ Idchild, / Uncle, e/ hew		13%	Sudan Ethippia South Ethippia Sudan Somalia Sri La Tatic Kenya
Ruth Carpenter	Half I Gran Gran Aunt Niece Neph Half I Gran	Siblings, Idparent/ Idchild, / Uncle, e/ hew Siblings, Idparent/		13%	Sudan Eritrea Yemen South Ethiopia sudan Somalia artic Kenya
Ruth Carpenter	Half J Gran Auntu Niece Neph Half J Gran Gran	Siblings, Idparent/ Idchild, / Uncle, e/ hew Siblings,		13%	Sudan South Ethiopia Sudan Somalia Aratic Nic Kenya Io

Even worse in mathematical modeling is what we call Deep Ancestry. This is before the genealogical time period. This is based on estimations of when DNA (Y-DNA & mtDNA) Haplogroups and haplotypes developed back in time. And those estimations (educated guesses) are measured in thousands and tens of thousands of years. Some haplotypes can be compared to archeological DNA found in ancient human remains which gives the impression of close relatedness, when it is really very distant fragments of relatedness.

Every living thing on this earth and what has lived on this earth through out time, is related to you and me. The food we eat is related to us genetically. Otherwise we could not digest or use it. We are all mutts, composites from our genetic past. We have a little bit of dinosaurs in us and when we go to the Zoo, we really are visiting our genetic cousins!

Helpful Links

Here are a few helpful links regarding DNA and common DNA terms.

A glossary of basic DNA terms can be found at: http://www.kerchner.com/books/glossary.pdf

The FTDNA version is at: https://www.familytreedna.com/learn/glossary/

General stuff about DNA https://www.genome.gov/25520880/deoxyribonucleic-acid-dna-factsheet/deoxyribonucleic-acid-dna-fact-sheet/ See also: http://www.nature.com/scitable/topicpage/introductionwhat-is-dna-6579978

FTDNA info on Y-DNA testing: https://www.familytreedna.com/learn/y-dna-testing/

Prepared by John R. Carpenter - 23 May 2017