Mitochondrial DNA Results - What Do They Mean and What Do I Do With Them?

By Roberta Estes, www.dnaexplain.com, robertajestes@att.net

DNA testing for genealogy celebrated its 10th anniversary this year. Most of us, by now, are familiar with Y-line DNA testing. These families are relatively easy to follow because the Y chromosome also follows the paternal surname. If a male Estes tests, for example, then his Y chromosome, and therefore his test results, should be the same as his father, his grandfather and so on, with maybe a mutation or two on upstream. The Y chromosome follows the surname so the genealogy is easy - or as easy as genealogy ever gets.

Mitochondrial DNA is different and many people make the mistake with both Y-line and mitochondrial DNA testing of thinking that you take the DNA test, and voila, your genealogy is magically set forth before you. That's not the case with any type of DNA testing.

Working on the female lines of our family is always more challenging due to the name changes. If marriage records are not available, there are a few ways to determine surnames of wives, but these are not always successful. Bibles are wonderful tools of course, but many times they don't survive. Wills of the wife's father are another excellent way to determine a surname, but this only works if there was a will, it still exists, and the records of that particular county have an "every name" index. Sometimes the will is in another location and the name in that other location remains unconnected to the woman and her husband in a different county. Sometimes when there was no will, there are still estate administrative papers that are loaded with information, but these types of records are rarely indexed or published.

In later years, cemetery records and some church records will contain maiden names, and of course, in the 1900s and in some locations somewhat earlier, death certificates will sometimes reveal maiden names. Many death certificates say "unknown" for the mother's name or her surname, and sometimes the information is incorrect. When you're looking for surnames, don't forget to check the information of siblings as well who shared the same mother.

DNA is another tool in the genealogists toolbox, but it's not an absolute answer. Let's look at the information we received from Family Tree DNA relative to mitochondrial DNA testing and results.

The first thing most people want to know is if they are Indian, African or European. Generally, this is quite straightforward and easy to determine. The haplogroup gives us that answer. Haplogroups A, B, C, D and X2 are Native American. If you happen to be X2, some additional analysis needs to be done as some X2 is European, but X2 is quite rare. Haplogroup L is African. Haplogroups H, I, J, K, T, U and V are European. If you are assigned a different haplogroup not listed above, it's rare and needs special

analysis. Of course, you maybe a subgroup of any of the above haplogroups, such as H1a for example.

Haplogroups are assigned based on the mutations found in your mitochondrial DNA. There are 16569 locations in your mitochondrial DNA. Think of this as a clock. The HVR1 test tests the DNA from 11:55 to noon on the clock. The HVR2 test then tests the DNA between noon and 12:05. The full sequence test tests all of your mitochondrial DNA. Some haplogroup subgroups cannot be assigned without the full sequence test, and some haplogroup subgroups have very unique migration patterns.

Mutations are reported by comparing your results with the CRS, or Cambridge Reference Sequence. That is simply the reference model that is considered the "norm", and everyone else is compared to it and differences are reported as mutations. This makes it easy to compare our results to those of others.

Most people have many mutations. Typically 2 or 3, or sometimes more in the HVR1 and HVR2 regions, respectively, and several in the coding region, which is what the area that is not the HVR1 or HVR2 regions is called. If you recall, this would be the time on the clock between 5 after and 5 till the hour.

Let's say you have a mutation in the HVR1 region of 16519C and a mutation in the HVR2 region of 522C. If you match someone at in the HVR1 region, but not the HVR2 region, then you're probably related, but not for several thousand years. So genealogically, it's not relevant. If you match someone on both the HVR1 and HVR2 levels, you may be related in a genealogical timeframe, especially if you have lots of mutations and match on all of them. At Family Tree DNA, you are only matched with people who are 100% matches because mutations occur infrequently in mitochondrial DNA. Of course, a mutation can happen in any generation at any time, but in general, if you don't match exactly, you're not related in a genealogical time frame.

If you match at the HVR1 level and the other person has not tested at the HVR2 level, perhaps they would upgrade. If you match at both the HVR1 and HVR2 level and you want to know for sure if you are a match, you should both upgrade to the full sequence level. If you match exactly at the full sequence level, then you may well be related in a genealogical timeframe. Now, of course, the key is to find your common ancestor.

If your common ancestor is in a dead end line - you may have to rely on geography and not genealogy to make the connection. I always suggest that people compose a one page introduction to their matches and to contact everyone with whom they share an exact match. Make your e-mail as easy and straightforward as possible. The shorter and simpler, the more likely it will be read and you'll get a response. Here is the table I recommend using following a short introductory paragraph:

Name of Ancestor	Birth Date	Birth Location	Death Date	Death Location	Spouses Name
Self					

Name of Ancestor	Birth Date	Birth Location	Death Date	Death Location	Spouses Name
Mother - living					
Edith Barbara Lore	1888	Indianapolis, Indiana	1960	Warsaw, Indiana	John Ferverda - couple lived in Silver Lake, Indiana
Elnora Kirsch	1866	Aurora, Indiana	1949	Lockport, NY	Curtis Benjamin Lore - couple lived in Rushville, Indiana
Barbara Drechsel	1848	Goppmannsbuhl, Germany	1930	Wabash, Indiana	Jacob Kirsch - couple lived in Aurora Indiana
Barbara Mehlheimer	1823	Goppmannsbuhl, Germany	1906	Aurora, Indiana	George Drechsel - couple immigrated to Aurora, Indiana
Elisabetha Mehlheimer	Abt 1800		Before 1851	Probably Goppmannsbuhl, Germany	Unknown

Locations are extremely important when tracking mitochondrial DNA because if you match someone who is in the same area as your ancestor, then you're close to finding your common ancestor. The records that may well prove the connection may be located in that geography as well.

Some people are lucky enough to connect to a surname. Since they change every generation, the surname will likely be buried in the information of the other individual. You should ask them for their info as well, along with the areas where their ancestor lived.

Don't neglect sisters and who they married. Your ancestor's sister may hold the key to your ancestry as well. I generally take my matches' ancestor's names and compare them to names in my Gedcom file to see what I find. It's amazing how often I find something close geographically or sometimes I find their ancestor already listed as a sibling or niece or cousin of one of my ancestors. That's powerful information.

Other data bases hold clues and possible matches as well. You can enter your DNA information at Sorenson at www.smgf.org. Sorenson does not facilitate matches with

individuals, but they do provide a genealogy along with the DNA information. If you find a line you believe you connect to, you can then peruse the various forums such as www.rootsweb.com lists and boards and www.genforum.com boards to find information about that line or someone who connects.

Another resource is www.mitosearch.org. You can upload your information directly from your Family Tree DNA matches page to Mitosearch. The key here is that you are looking for people who did not test at Family Tree DNA, as your results are already being compared to those who did. Sometimes the information found here for those who you match at Family Tree DNA is important as well, as participants can enter their oldest ancestor and some additional information not available through Family Tree DNA directly.

A final resource is www.ancestry.com. Click on the DNA tab at the top and enter your information. Unfortunately, Ancestry does a very poor job of both haplogroup assignment and matching. They show matches as everyone you match at any mutation. Once your matches are displayed, click on "table view" and then click on "download". A spreadsheet will open, and you can manually sort the match results. Eliminate all matches that do not match your results exactly. You will have a small subset remaining, if any. Use that subset to initiate contacts with those individuals. To do that at Ancestry, click on the name of the individual, and then click on "contact" and a message form will appear. You cannot contact them directly, but a message will be sent to them through Ancestry and they can choose to respond or not. I always put my own e-mail address in the message hoping they will contact me directly.

The great thing about DNA is that even if you don't have any matches today, your DNA is out there "fishing" for you every day, 24X7. One day you'll receive a match notification from Family Tree DNA, and you never know what tidbit of information your match may have that will help one of your brick walls fall. Be sure to check the alternate data bases regularly. While Family Tree DNA notifies participants of matches, the others don't, so check your matches when you change your smoke detector batteries. Your ancestor may be waiting for you!