

I Have the Results of My Genetic Genealogy Test, Now What?

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Chapter 1: What Is (And Isn't) Genetic Genealogy?

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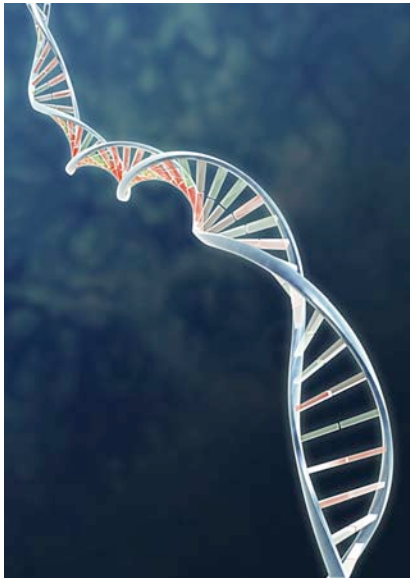
Chapter 3: How Do I Interpret My mtDNA Results?

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Chapter 1

What is Genetic Genealogy?



If you're reading this eBook, then you're probably already fully aware of genetic genealogy, the use of DNA to explore ancestral origins and relationships between individuals. It is, as I call it, "another tool for the genealogist's toolbox."

Although there are four types of genetic genealogy tests – autosomal DNA tests, X-DNA tests, Y-DNA tests, and mtDNA tests – we will only be exploring the results of Y-DNA and mtDNA testing in this eBook.

Y-DNA tests, available only to males, examine either STRs (short tandem repeats) or SNPs (single nucleotide polymorphisms) on the Y chromosome. For an STR test, short segments of DNA are measured. The number of repeats in that short sequence changes over time, and these changes are passed on from father to son. STR analysis provides a person's haplotype, which is used to predict an individual's haplogroup. SNP tests examine single nucleotide changes in the DNA sequence and are typically used to determine a person's exact haplogroup.

mtDNA tests, available to both males and females, sequence a short region of the mitochondrial genome

(although it is possible to sequence the entire mitochondrial genome). When mtDNA is tested for genealogical purposes, a region of the DNA is sequenced for mutations. The mtDNA sequence is then compared to a single mtDNA sequence, the Cambridge Reference Sequence. The differences are listed as mutations that can be compared to the thousands of other mtDNA mutation listings that are stored in the Family Tree DNA database and in publicly-available databases. The results can also be used to roughly estimate the amount of time to which individuals share a most recent common ancestor (MRCA). The results of an mtDNA test provides a person's haplotype. Family Tree DNA performs additional testing with every mtDNA test to determine an individual's haplogroup.

According to a 2006 report ([Genetic Genealogy Goes Global](#), *7 EMBO Reports* 1072-4), at least 460,000 genetic genealogy kits have been sold throughout the world. By my recent estimate, that number might be as high as 600,000 to 700,000 and increasing by as much as 80,000 to 100,000 a year. Regardless of the exact number, genetic genealogy is a quickly growing field.

What ISN'T Genetic Genealogy?

Although genetic genealogy is an informative addition to anyone's family tree, it is not without limitations. These limitations have been the focus of a great deal of media attention in recent months. Anyone who is thinking about buying a genetic genealogy test should be aware of the following:

1. The results of a genetic genealogy test do not include a family tree. DNA alone cannot tell a person who their great-grandmother was, or what Italian village their great-great grandfather came from. Genetic genealogy is an *addition* to traditional genealogical research, not a *replacement*.
2. Although Y-DNA and mtDNA can be used to determine the relatedness of individuals, it cannot directly determine the degree of relationship. For example, an mtDNA test might be used to finally determine whether two women are maternally descended from one individual, as your traditional research has suggested. However, the results will not be able to determine whether the women are first cousins, third cousins, or fifth cousins once removed.
3. Genetic genealogy testing CAN potentially reveal information about your health. Research has identified a correlation between missing DYS464 on the Y-chromosome and infertility. Out of over 85,000 testees, Family Tree DNA has identified only 11 people without a DYS464. Some metabolic and other diseases can be revealed by full mtDNA sequencing (also called FGS).
4. Finally, a genetic genealogy test will only reveal information about a small percentage of your genome. As every genealogist knows, at 10 generations there are as many as 1024 ancestors in the family tree. Thus, a Y-DNA test or mtDNA test only represents one individual out of 1024. However, almost every genealogist has spent money and a great deal of time and effort attempting to learn even the smallest bit of information about an individual in their family tree. DNA is

another way to add to that information.

Even with these limitations, genetic genealogy can be an informative and exciting addition to traditional research, and can sometimes be used to answer specific genealogical mysteries. There are many – perhaps even hundreds – of genealogical success stories thanks to the proper use of genetic genealogy. To read about some of these inspiring success stories, see the International Society of Genetic Genealogy's (ISOGG) [Success Stories](http://www.isogg.org/successstories.htm) <www.isogg.org/successstories.htm>.

Chapter 2

How Do I Interpret My Y-DNA Results?



The following is provided to anyone who might be unfamiliar with genetic genealogy. Note that it is composed of my suggestions and are not necessarily the same suggestions that someone else would make. Additionally, if you are undergoing genetic genealogy testing to answer a specific question (such as, is my surname really XYZ), your analysis will differ from the one I have provided (see question #3 under "STR Testing," below).

A. STR Testing

For the purposes of this section, let's assume that you have ordered a 12-marker STR test without SNP testing, since this is the most common type of Y-DNA testing. The results arrive as a series of markers and the results, or alleles, as in the following example:

Table 1. Sample Y-DNA STR Results

DYS#	393	390	19	391	385a	385b	426	388	439	389-1	392	389-2
Alleles	13	24	14	10	11	14	12	12	12	13	13	29

What does this string of numbers mean? At each of these locations on the Y-chromosome there is the potential for some variation (repeats of the same DNA sequence). For example, at DYS426, the variation consists of 7 to 18 repeats of the DNA sequence "GTT", with 12 being the most common (>60% of Y-DNA samples, according to FTDNA). The sequence would look like this, with the 12 repeats in bold:

...TGTGTTGTTGTTGTTGTTGTTGTTGTTGTTGTTGTTGTTGTTGAC...

Someone with a result of 7 at DYS426 would have the following sequence:

...TGTGTTGTTGTTGTTGTTGTTGTTGTTGAC...

Together, the particular alleles revealed by testing represent your personal Haplotype. Using our sample

haplotype, we will attempt to (1) identify the Haplogroup of this Y-DNA sample; (2) research the identified Haplogroup; (3) find matches in Y-DNA databases; (4) attempt to find and join a surname or geographical DNA project, and; (5) start our own DNA project.

1. Which Haplogroup Does This Y-DNA Most Closely Match?

Family Tree DNA predicts your haplogroup based on your STR results and will inform you of their prediction. Family Tree DNA customers can find their haplogroup prediction or confirmation in the Haplotree section of their myFTDNA page. If the haplogroup cannot be confidently predicted, Family Tree DNA will also provide additional testing to identify and confirm your basic haplogroup assignment.

You may also be interested in experimenting with other haplogroup predictors and analyzing your haplogroup prediction yourself. The first step in the analysis is to visit [Whit Athey's Haplogroup Predictor <www.hprg.com/hapest5>](http://www.hprg.com/hapest5), a free web-based program that allows the user to easily estimate their Haplogroup (but be sure to read the Conventions page – some testing companies report different numbers for the same alleles and it is important to enter the correct number into the program). Choose either the basic 15-Haplogroup Program, or the Beta 21-Haplogroup Program. You might want to start with the 15 to get a rough idea, and then use the 21 to potentially obtain more information. Enter your allele values into the predictor and the probability of your Haplogroup will be calculated in the right-hand field as you type.

Let's use our alleles as an example. When we input these alleles, there is a 100.0% probability that the Y-DNA

belongs to Haplogroup R1b. Don't worry if your results aren't as clear as this example; Haplogroup designation using STR results rather than SNP results is a matter of statistical probability rather than absolute certainty. Fortunately, the results in our example match the Haplogroup predicted by the testing company. If your results don't match the predicted Haplogroup, you might have to conduct more research to attempt to elucidate to which Haplogroup your DNA belongs.

If you are unable to predict a Haplogroup based upon STR testing, one possible avenue is to proceed with SNP testing, which will almost certainly be able to identify your Haplogroup. We will example SNP testing in a later section.

2. How Do I Learn More About My Y-DNA Haplogroup?

Now that we know that we (most likely) belong to Haplogroup R1b, we should attempt to learn as much as we can about the Haplogroup. Here are some links that will contain information about most, if not all, of the major human Y-DNA Haplogroups:

- Family Tree DNA provides several informative tools for learning about your haplogroup in the Haplotree section of customers' myFTDNA pages. The central Haplotree page shows the haplogroup chart and where your branch fits within the Y-DNA tree. The frequency map shows measured frequencies of major haplogroups within researched populations and displays a short description of your haplogroup. The migration map demonstrates the general path your paternal ancestors took to reach the part of the world in which they can be found today.

- Charles Kerchner's [YDNA Haplogroup Descriptions & Information Links](http://www.kerchner.com/haplogroups-ydna.htm) <www.kerchner.com/haplogroups-ydna.htm> – this great resource contains a very brief description of each Haplogroup along with a list of links (if available).
- Although Wikipedia should always be used with caution, the [Human Y-chromosome DNA Haplogroups](http://en.wikipedia.org/wiki/Human_Y-chromosome_DNA_Haplogroups) <en.wikipedia.org/wiki/Human_Y-chromosome_DNA_haplogroup> page contains links to individual pages for many of the Haplogroups and a number of subgroups.
- If all else fails, use a search engine to find pages that might contain more information.

Most Haplogroups have multiple sources of information available. Don't worry if these sources of information conflict with each other – the science is still developing and estimates change with every new discovery.

3. Does Anyone Match My Y-DNA Haplotype?

To learn more about our haplotype, and to potentially find other people who match our haplotype, we will use two resources: the Family Tree DNA database and the free public Y-DNA databases that are available, specifically [Ysearch](http://www.ysearch.org) <www.ysearch.org>.

Family Tree DNA automatically checks customers' results for matches within the company database as long as the customer has signed the release form allowing their name and email to be shared with their matches. Customers can view their list of matches in the Y-DNA Matches section of their myFTDNA page. Family Tree DNA lists the names and email addresses of any other participants whose DNA results are similar enough to

be potential genealogical matches. This list of matches can be filtered to show only matches at certain testing levels or only matches who belong to the same project as you.

You can also search for matches in public databases. Since these databases function in similar ways, we will use Ysearch as an example. Note that there are at least two ways to search each of these databases:

a. Search by Haplotype

At Ysearch.com, the user encounters a Welcome page. At the bottom of that page, click on "Search for genetic matches." Then click on "Click here to enter any sequence and search by Haplotype." This brings us to a screen where we can enter our example haplotype. Enter your haplotype and leave the other options at their defaults with the exception of "Show users that tested at least X of the markers that I did" (set this to "12"), and the "maximum genetic distance" (set this to "0" at this early stage of your research).

The next screen is a list of the profiles that match our haplotype exactly with at least 12 of our markers. For our sample haplotype, we get over 900 exact matches (as of April 2009). R1b is very common, and as a result there are many R1b haplotypes in these databases. Note that some of these profiles may overlap with your Family Tree DNA matches.

If you didn't get any matches using a genetic distance of "0", change it to "1", or "2", or more. Note that your closest matches will match by a genetic distance of "0"; the more differences between haplotypes,

the greater the amount of time to the most recent common (tMRCA) Y-DNA ancestor.

Interestingly, many of the matches in the Ysearch and Ybase databases have undergone SNP testing to show that they belong to the R1b1b2 Y-DNA Haplogroup. This might suggest that our Y-DNA sample belongs to R1b1b2, a sub-clade of Haplogroup R1b (see the SNP section, below).

Another way to use the database to search is to click on "Create a new user." This will allow you to create a profile that you can use to easily search for matches. This profile will be searchable by other users of Ysearch and will allow them to contact you for more information if necessary. Family Tree DNA customers can use an automatic upload feature which will fill their results into the "Create a new user" Ysearch page for them. This upload link is located just above the list of matches in the Y-DNA Matches section of your myFTDNA page.

b. Search by Surname

Searching by surname is yet another way to find potential matches in public DNA databases. This method of searching can even be used before a DNA test has been purchased. For instance, let's assume that we haven't yet ordered a DNA test, and that our last name is "Bettinger". Has anyone else with the last name "Bettinger" ever been tested? What were their results?

To begin, go to the Ysearch database <www.ysearch.org>. At the top of the page is a button labeled

“Search By Last Name” (there is also an alphabetical list of surnames, if you want to skip the search function). Click on that, and enter “Bettinger” in the first data field; press enter. As you’ll see, there is currently (as of January 2008) one person with the last name Bettinger in the database. Clicking on that record reveals that this Bettinger belongs to Haplogroup R1b1.

If we already have our results, we can now compare our haplotype with the Bettinger who is already in the database. There a number of ways to limit the search on the first page; I’ll let you explore these options as you become more familiar with the database.

If you are interested in maintaining a permanent presence on Ysearch with your haplotype available for searching, create a new user record. When new people go online to search for matches to their results, they will be able to compare their haplotype to yours.

4. Should I Join a Surname, Geographic, or Haplogroup DNA Project?

A DNA project is a collaborative effort to answer genealogical questions using the results of DNA testing. A surname project brings together individuals with the same (or very similar) surname, while a geographic project gathers individuals by location rather than by family or surname. Other projects bring individuals together based upon their Haplogroup designation. DNA projects are run by volunteer Administrators who are responsible for organizing results, sharing information, and recruiting new members to the group.

DNA projects can potentially provide a number of benefits to participants, including the following:

1. Confirm or reject suggested relationships between individuals in the project;
2. Identify previously unknown relationships between individuals in the project;
3. Confirm or reject the relationship of particular surname lines or surname variants (for example, do the Hoisington and Hossington surnames have a common source?);
4. Identify or learn more about the surname's country of origin;
5. Learn more about the ancient ancestry of a surname and its variants;
6. Learn more about the migration of the surname over time;
7. Learn about the genetic origins of and contributions to a geographic region;
8. Assist in genetic genealogy research projects for publication (scientists have used the results of DNA projects to learn more about human history – see the [JoGG](http://www.jogg.info) <www.jogg.info> for examples), and;
9. Join a community of exciting genetic genealogists.

There are undoubtedly many more benefits to joining surname or geographical DNA projects. One benefit that I left off the list is a financial one. Family Tree DNA offers testing discounts to project members. This is often a great motivation to join a project, and also serves as a terrific recruitment tool. By the way, don't worry if you've already taken a genetic genealogy test and later decided that you would like to join a DNA project once you are in the Family Tree DNA system. It's never too late to join!

A. How do I *FIND* a DNA Project?

Finding a DNA Project that you might be interested in joining is usually very simple. Here are three places to begin your search:

1. [Family Tree DNA](http://www.familytreedna.com/projects.aspx) <www.familytreedna.com/projects.aspx> – The search box in the sidebar lets you search by surname or by location or country. For example, typing in “Belize” leads you to two geographic DNA projects. You can also browse through the list of projects organized by name.
2. [Family Tree DNA](http://www.familytreedna.com/login.aspx) <www.familytreedna.com/login.aspx> - Family Tree DNA customers can browse and join projects using the “Join Projects” link located in the left-hand menu bar on their myFTDNA page.
3. [World Families](http://www.worldfamilies.net/search/contentsearch) <www.worldfamilies.net/search/contentsearch> – You can use the search box to search entire site.

Using traditional search engines is yet a fourth way to search for projects, with search strings such as ‘Smith DNA Project.’ For this method, as well as the methods listed above, be sure to search for variants of your surname if you are unable to find an exact match.

B. How do I *JOIN* a DNA Project?

Now that you've discovered an interesting DNA project, you're probably wondering how to become a member. Here's how to join:

After finding a Project, you will see a contact form at the bottom of the Project's introduction page. Use that form to join the project and order a test. If you've already been tested, you'll find the "Join Project" link on your myFTDNA page. This link will take you to a search page to find an appropriate Surname, Geographic, or Haplogroup Project. For more information on joining projects, see the FTDNA [FAQ page](#) <www.familytreedna.com/faq/> or the World Families "[Join a Project](#)" <www.worldfamilies.net/join>.

5. What if There is no DNA Project to Join?

The answer to this one is simple – start a new project! There is information at Family Tree DNA <www.familytreedna.com/surname.aspx?> and World Families <www.worldfamilies.net/request> about starting a new DNA Project.

B. SNP Testing – Haplogroup Determination

Single Nucleotide Polymorphism (SNP) testing examines single nucleotides at specific locations on the Y chromosome (compared to STR marker testing, which measures short regions of DNA). Advanced SNP testing is available after STR testing. Since a mutation at a single base is very rare compared to changes in STRs, males who share a SNP usually share an ancestor who lived many generations or many hundreds of

generations ago. For this reason, SNPs have been used to identify the branches in the Y-chromosome family tree and define Haplogroups, as well as very, very distantly related people. For example, males who test positive for the SNP M207 belong to Haplogroup R. You can see a chart of some of the most commonly tested SNPs at Family Tree DNA <www.familytreedna.com/deepclade.html> and <www.familytreedna.com/snps-r-us.aspx> or a full list of SNPs at ISOGG <www.isogg.org/tree/ISOGG_YDNA_SNP_Index.html>. So why would an individual order a SNP test? Here are a few of the major reasons:

1. To determine deep ancestry – some individuals might be more interested in learning about the “deep ancestry” of their Y-DNA, rather than finding close genetic relatives. For them, the results of SNP tests will typically tell them about their ancient ancestry.
2. To confirm an estimated Haplogroup – as we saw in previous sections, the results of an STR test can be used to predict or estimate an individual’s Haplogroup. Since SNPs define Haplogroups, SNP testing can confirm the estimate or re-define the Haplogroup based upon the results. Family offers both a backbone haplogroup test to confirm predictions (or to identify the haplogroup where it cannot be confidently predicted) and deep clade haplogroup testing.
3. To determine a subhaplogroup designation – some Haplogroups have multiple branches, called subhaplogroups or sub-clades. For example, Haplogroup R has branches such as R1b, which itself branches into groups like R1b1a and R1b1b (see the [Y-DNA Haplogroup Tree](#) – please note it may take several minutes to download). Deep Clade tests cover the SNPs defining these branches, or sub-clades.

The results can often (but not always, depending on how well current research has covered your haplogroup thus far) place an individual into one of these downstream branches.

1. How Do I Interpret My SNP Test Results?

To interpret results of a SNP test (other than the Haplogroup designation, which is always part of the results returned to the customer), it is helpful to have the following references: (1) the [Y-DNA Haplogroup Tree](#); and (2) the Y-DNA SNP index <www.isogg.org/tree/ISOGG_YDNA_SNP_Index.html>, which will help us interpret the numbers and letters that make up the results.

Let's use the following sample results as an example, which tell us that we belong to Haplogroup sub-clade E1b1b1b1. What exactly does that mean?

Table 2. Sample Y-DNA SNP Results

Your Y-DNA Belongs to Haplogroup E
Your SNP results are: M96+ P2+ M2- M35+ M78- M183+ M81+ M107- M165- M123-

Since the results report that our sample Y-DNA belongs to Haplogroup E, we can go directly to the [Haplogroup E Tree](#). Let's use our results, starting with the first SNP, M96. We tested positive for M96, meaning that we have that mutation. Looking at the chart, we see that M96 is characteristic of Haplogroup E. Thousands of years ago, members of Haplogroup E developed characteristic SNP mutations over time to form different branches or sub-clades such as E1 and E2. Our next SNP, P2+, is characteristic of sub-clade

E1b1. Comparing our sample SNP results to the tree, we get something like this (and it definitely helps to diagram the tree), keeping in mind that the "-" after the test indicates that we are *not* part of this sub-clade:

M96+	E		
P2+		E1b1	
M2-		E1b1a	
M35+		E1b1b1	
M78-		E1b1b1a	(<i>not</i> part of this sub-clade)
M81+		E1b1b1b	
M107+		E1b1b1b1	(<i>not</i> part of this sub-clade)
M183+		E1b1b1b2	
M165-		E1b1b1b2a	
M123-		E1b1b1c	(<i>not</i> part of this sub-clade)

Thus, our sample Y-DNA belongs to Haplogroup E, sub-clade E3b1b1 because we tested positive for the M183 SNP. The negative results also help refine our placement by showing that we do not belong to E1b1a (M2-) or E1b1b1a (M78-), for example.

The Haplotree section of Family Tree DNA customers' myFTDNA pages draws a line designating your position on the tree and notes which SNPs you have tested positive and negative for, if you have had a SNP test.

[Insert E Haplotree drawing (ex kit 101203)]

Don't be discouraged if your results are presented as clearly or in the perfect order – you can always put them in order to help make the results more clear. Also, sometimes SNPs will have different names or companies will use different SNPs to test the same thing – with a little research at the sites listed above, you'll be able to figure out what each SNP means. And lastly, now that you know your Y-DNA Haplogroup and sub-clade, you can use the steps outlined above to gather more information.

Chapter 3

How Do I Interpret My mtDNA Results?



mtDNA tests, which sequence regions of the mitochondrial genome, are available to both males and females. The customer's sequence is compared to a standard mtDNA genome called the Cambridge Reference Sequence (CRS), which is available at <www.mitomap.org/mitoseq.html>. All the differences between the customer's mtDNA and the CRS – ranging from none (i.e. a perfect match to the CRS) to many – are returned as the customer's results. These results can be used to estimate the customer's mtDNA Haplogroup and roughly estimate the amount of time to which individuals share a most recent common ancestor (MRCA).

Family Tree DNA performs Haplogroup testing for all mtDNA tests so only the subclade is predicted, when it

can be. Here is a chart of the mtDNA Haplogroups and their defining SNPs tested by Family Tree DNA:

[Insert mtDNA BB chart here]

Please note that the following is provided to anyone who might be unfamiliar with genetic genealogy. It is composed of my suggestions and is not necessarily the same suggestions that someone else would make.

1. What Is My mtDNA Haplogroup?

Family Tree DNA provides a Haplogroup along with the customer's results. Thus I will only briefly mention one method for roughly confirming Haplogroup in a separate section (see #3, below). Remember that an mtDNA Haplogroup designation based on sequencing results is just an estimate! Family Tree DNA performs SNP confirmation of basic mtDNA Haplogroups with all mtDNA tests.

To perform an example analysis of mtDNA results, we will use the sample haplotype given in the following table. As part of these results, we will assume that the testing company also informed us that the sample belongs to mtDNA Haplogroup U, and we can trace our mtDNA back to France.

Table 3. Sample mtDNA STR Results

Region	HVR1	HVR2
Differences From CRS	183C, 189C, 270T	73G, 150T, 263G, 309.1C, 315.1C

What does this string of numbers and letters mean? These letters represent mutations and are all the differences between your mtDNA and the universal mtDNA sequence known as the Cambridge Reference Sequence (CRS). The CRS belongs to mtDNA Haplogroup H, and as a result individuals who have mtDNA in Haplogroup H often have fewer differences from the CRS than people who have mtDNA in other Haplogroups.

The results in HVR1 (Hyper-Variable Region 1) are often given as three numbers followed by a letter. Note that this is just a convention – the three numbers should be preceded by “16”. In our example, the HVR1 has three mutations, 183C, 189C, and 270T. This means that our mtDNA sequence differs from the CRS at positions 16183, 16189, and 16270, with the letters given replacing the letters normally found in the CRS (this “non-mutated” letter from the CRS often precedes the numbers, such as T16189C). Remember, of course, that the “letters” represent the nucleotide bases Adenine, Thymine, Cytosine, and Guanine.

The results in HVR2 (Hyper-Variable Region 2) are also often given without the preceding “non-mutated” letter. In our example, we have five mutations in this region of the mtDNA genome. The last two mutations, 309.1C and 315.1C are both fairly common and represent an *insertion* of a new nucleotide base at that position. Thus, between position 309 and 310, we have an insertion of the nucleotide cytosine which is not found in the CRS. Another type of common mutation is the *deletion* of a nucleotide at a particular position in the mtDNA genome. This might be written as “522-523delCA”, or “522- 523-”, indicating that the individual’s genome has lost the cytosine and adenine which are normally found at position 522 and 523, respectively. That genome would go directly from the nucleotide at position 521 to the nucleotide at position 524.

2. How Do I Learn More About My mtDNA Haplogroup?

Since we have a rough idea of our Haplogroup (U), we will want to learn as much as we can about that particular Haplogroup. Here are some links that provide information about many of the major mtDNA Haplogroups (but there are many, many more):

- [Family Tree DNA <www.familytreedna.com/login.aspx>](http://www.familytreedna.com/login.aspx) - Family Tree DNA displays haplogroup descriptions in the mtDNA Results section of customers' myFTDNA page.
- Charles Kerchner's [MtDNA Haplogroup Descriptions & Information Links <www.kerchner.com/haplogroups-mtdna.htm>](http://www.kerchner.com/haplogroups-mtdna.htm) site contains a very brief description of each Haplogroup along with a list of links (if available).
- Although Wikipedia should always be used with caution, the [Human mitochondrial DNA haplogroup <en.wikipedia.org/wiki/Human_mitochondrial_DNA_haplogroups>](http://en.wikipedia.org/wiki/Human_mitochondrial_DNA_haplogroup) page contains links to individual pages for many of the Haplogroups and a number of subgroups.
- If all else fails, use a search engine to find pages that might contain more information. There are many websites devoted to sharing information about particular Haplogroups.

As I mentioned in the discussion of Y chromosome Haplogroups, the science of genetic genealogy will continue to grow and develop for many years. As a result, the information will continue to change and be refined, so don't worry if the sources of information you find conflict with each other. Scientists and genealogists are still learning about DNA, which is just one of the reasons so many people enjoy genetic genealogy!

3. Does Anyone Match My mtDNA Haplotype?

To learn more about our haplotype, and to potentially find other people who match our haplotype, we will use two resources: the Family Tree DNA database and the free public Y-DNA databases that are available, specifically Mitosearch <www.mitosearch.org>. As we find matches, we will also be gathering information about our Haplogroup designation. If we see that a person who closely matches our sequence, we can compare their Haplogroup designation to ours. This isn't an exact method when using public databases because it relies on second-hand information input, but it is an interesting exercise.

Family Tree DNA automatically checks customers' results for matches within the company database as long as the customer has signed the release form allowing their name and email to be shared with their matches. Customers can view their list of matches in the mtDNA Matches section of their myFTDNA page. Family Tree DNA lists the names and email addresses of any other participants whose DNA results are similar enough to be potential genealogical matches. This list of matches can be filtered to show only matches at certain testing levels or only matches who belong to the same project as you. You can also view the Haplogroup designations of your matches in the mtDNA Ancestral Origins section.

Let's use Mitosearch as a model for searching public mtDNA databases. Similar to Y-DNA databases, there are multiple ways to search Mitosearch. Note, however, that searching by surname is not an option for Mitosearch since mtDNA does not usually follow the surname line in a family tree.

a. Search by Haplogroup

At Mitosearch, the user first encounters a Welcome page. At the top of the page, click on "Search By Haplogroup." From the drop-down menu, pick the Haplogroup and, if known, select the country of origin. For this example we will pick 'U' and 'Western Europe'. We get a little over 30 users, some of which appear to originate in France. To compare these results to ours, click the box marked "compare" to the left of the result and then click the word "compare" at the top of that column. Hit "show" on the next page, and the results are lined up in a convenient table for us to look at. By comparing our results to the table, we see that as of March 2008, none of these results match ours exactly. It might be worthwhile to go back and compare sequences from other countries or regions, but perhaps the best approach might be the next section, search by haplotype.

b. Search by Haplotype

At the Welcome screen, click on "Search for Genetic Matches." Then proceed to the bottom of the next page where we will "Search by Mutations." Since we tested both the HVR1 and HVR2 regions in our sample results, we will search by both (but I recommend that you click the box that searches for HVR2 matches, but doesn't rule out matches based on HVR2). Enter in your results. Remember that there should be three numbers in each box, so our '73' mutation will be entered as '073'. Additionally, insertions (309.1C) and deletions (522-) must be entered carefully. Click on search.

The results show that as of March 2008, our sample mtDNA has 4 individuals who match our HVR1 sequence but did not test their own HVR2 region, and two people that match both HVR1 and HVR2

exactly. For more information about our matches – such as surname & location – click on the User ID.

So, the results of our search show that we have a number of exact matches and some other potential matches. We could then contact those individuals if we believe there might be some genealogical connection. If you are interested in maintaining a permanent presence on one of the DNA databases with your haplotype available for searching, create a new user record. When new people go online to search for matches to their results, they will be able to compare their haplotype to yours.

4. Do I Belong to a Particular “Subclade” or “Branch” of My Haplogroup?

Sometimes mtDNA results will list a Haplogroup designation (such as H) without listing a particular subclade or branch of that Haplogroup (such as H1, H2, H2a1, H2a2, etc...). This might be because (1) the sample matched the haplogroup but none of the subclades, or (2) the sample was not tested or analyzed for subclade designation. When in doubt, it might be helpful to email your testing company to determine which might be the case. Additionally, you can test your mtDNA full genomic sequence (FGS) to identify your Haplogroup subclade within the currently known tree, and you can continue to identify your placement in the tree as additional research is completed. And lastly, you can attempt to deduce a potential subclade using the mtDNA databases we’ve already discussed. Looking back at the last section, when we entered in our sample test results we obtained six potential matches, 4 who matched our HVR1 results and 2 who matched both our HVR1 and HVR2 results. Five out of those six individuals are reported to belong to Haplogroup U5b. This suggests, but does not prove, that our sample mtDNA might belong to U5b. What is even more

suggestive is that the matches have been tested by more than one company, meaning that at least two different companies interpret our mtDNA sequence to belong in U5b.

5. Should I Join an mtDNA Project?

Just like for Y-DNA, mtDNA projects bring together individuals with a similar Haplogroup designation or geographic location. DNA projects are run by Administrators who are responsible for organizing results, sharing information, and recruiting new members to the group. Joining a project may result in a number of benefits to the individual, the project, or the Haplogroup. Additionally, joining a project *before* being tested results in a testing discount, but it's never too late to join a project. Since finding, joining, and starting an mtDNA Project is very similar to the steps I described above for Y-DNA, I won't repeat them here.

Chapter 4

Monitoring The Field Of Genetic Genealogy



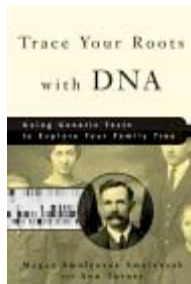
Now that we've learned a little about genetic genealogy and how to begin to interpret our testing results, how do we continue to learn more and stay up-to-date on the latest news and developments in the field? Luckily, there are many ways to follow developments in genetic genealogy. The following is a list of just some of the many valuable resources that are available:

1. Family Tree DNA Forums are a great place to share and find information about almost any genetic genealogy topic. Both newbies and experts participate in the [Family Tree DNA Forums](http://forums.familytreedna.com/) <forums.familytreedna.com/>.
2. Yahoo Groups is another method of following the field and is free to join. The [DNA-NEWBIE](http://groups.yahoo.com/group/DNA-NEWBIE) group <groups.yahoo.com/group/DNA-NEWBIE> (created in July 2005) has over 1,900 members who post an average of 350+ messages a month. The [DNA-ANTHROGENEALOGY](http://groups.yahoo.com/group/DNA-ANTHROGENEALOGY) group <groups.yahoo.com/group/DNA-ANTHROGENEALOGY> (created in October 2005) has over 1,000 members who post an average of 175/month. The [DNA-Testing](http://tech.groups.yahoo.com/group/DNA-Testing) group <tech.groups.yahoo.com/group/DNA-Testing> (created in July 2008) has 180 members. One great feature of Yahoo Groups is that you can set your email preferences to receive every message separately, or receive groups of messages, or receive no messages (and then you can independently monitor the site at your leisure).
3. The International Society of Genetic Genealogy ([ISOGG](http://www.isogg.org) <www.isogg.org>) was founded in 2005 by DNA Project administrators "to advocate for and educate about the use of genetics as a tool for genealogical research, and to promote a supportive network for genetic genealogists." Membership in the society is free.
4. The Journal of Genetic Genealogy ([JoGG](http://www.jogg.info) <www.jogg.info>) is an online science journal

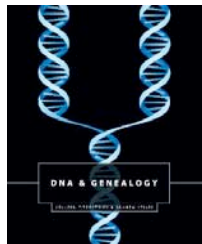
devoted entirely to genetic genealogy. The Journal is open-access, meaning that it is freely available to everyone. The JoGG welcomes article submissions about a range of topics and publishes two issues a year.

5. Family Tree DNA Newsletter is a free newsletter which announces current events at Family Tree DNA and upcoming educational opportunities, and also includes educational material on a variety of topics and questions. You can [view past issues](http://www.familytreedna.com/newsletter.aspx) <www.familytreedna.com/newsletter.aspx> of the Facts and Genes newsletter and [subscribe](http://www.familytreedna.com/facts-and-genes-register.aspx) <www.familytreedna.com/facts-and-genes-register.aspx> from the Family Tree DNA website.
6. Genetic Genealogy-Related Websites offer a wide variety content and links for more information. [Roots Television](http://www.rootstelevision.com) <www.rootstelevision.com>, created by Megan Smolenyak Smolenyak and Marcy Brown, has free online videos about many aspects of genealogy. One of Roots Television's channels, the DNA Channel, has interviews, success stories, and recordings of professional presentations. Another great website is that of Charles F. Kerchner, Jr. [Kerchner's DNA Testing & Genetic Genealogy Info and Resources Page](http://www.kerchner.com/dna-info.htm) <www.kerchner.com/dna-info.htm> contains information about genetic genealogy for both the beginner and the expert.
7. Blogs are yet another way to stay up-to-date. I can think of one great blog in particular <www.thegeneticgenealogist.com>, if I do say so myself! Another blog devoted to genetic genealogy is [Genealem's Genetic Genealogy](http://genealem-geneticgenealogy.blogspot.com) <genealem-geneticgenealogy.blogspot.com>.

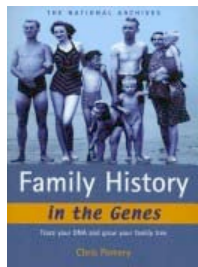
For More Information:



If you've read through this eBook and you are still looking for more information (good for you!), then I recommend "[Trace Your Roots With DNA: Use Your DNA to Complete Your Family Tree](#)" (affiliate link) by Megan Smolenyak Smolenyak and Ann Turner, M.D. This book discusses and clearly explains topics ranging from the basics of genetic genealogy to tips about creating and running your own DNA Project. I highly recommend the book for anyone who is interested in genetic genealogy, and especially for anyone thinking about starting a DNA Project.



[DNA & Genealogy](#) follows with the details on what DNA analysis is and how it can be used to reach beyond conventional documentation to find new family relationships. DNA & Genealogy has something for everyone. It includes many examples based on Family Tree DNA personal results and surname project pages.



[Thousands of family historians](#) around the world have used DNA testing to extend their family tree and the scope of their research. This clear, concise and up-to-the-minute guide shows what genetics can do for anyone in search of their ancestors, including linking people with the same surname, unraveling clan relationships and exploring the genetic profile of individuals, locations and regions. From the latest discoveries in the big surname studies to advice on starting your own DNA project, this is a fascinating introduction that will dominate future family history research.



the genetic genealogist

ADDING DNA TO THE GENEALOGIST'S TOOLBOX