

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

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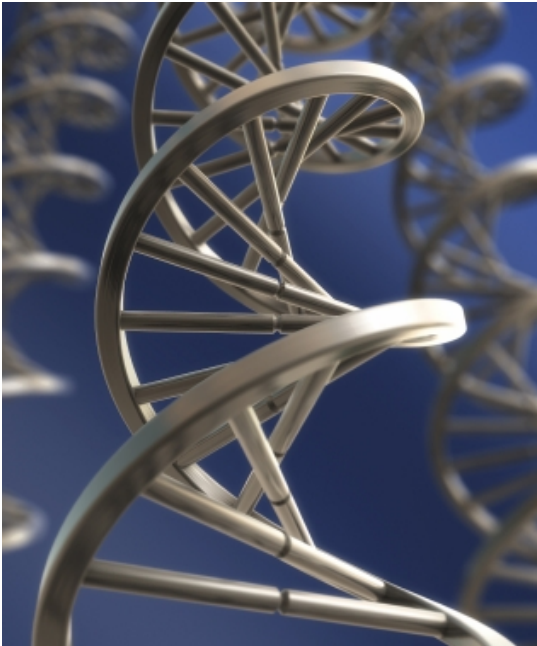
Chapter 5: Monitoring the Field of Genetic Genealogy

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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>.

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_haplogroup) <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](http://www.familytreedna.com/faq/) <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-
M123-

E1b1b1c

E1b1b1b2a
(*not* 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 Haplotype 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.

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

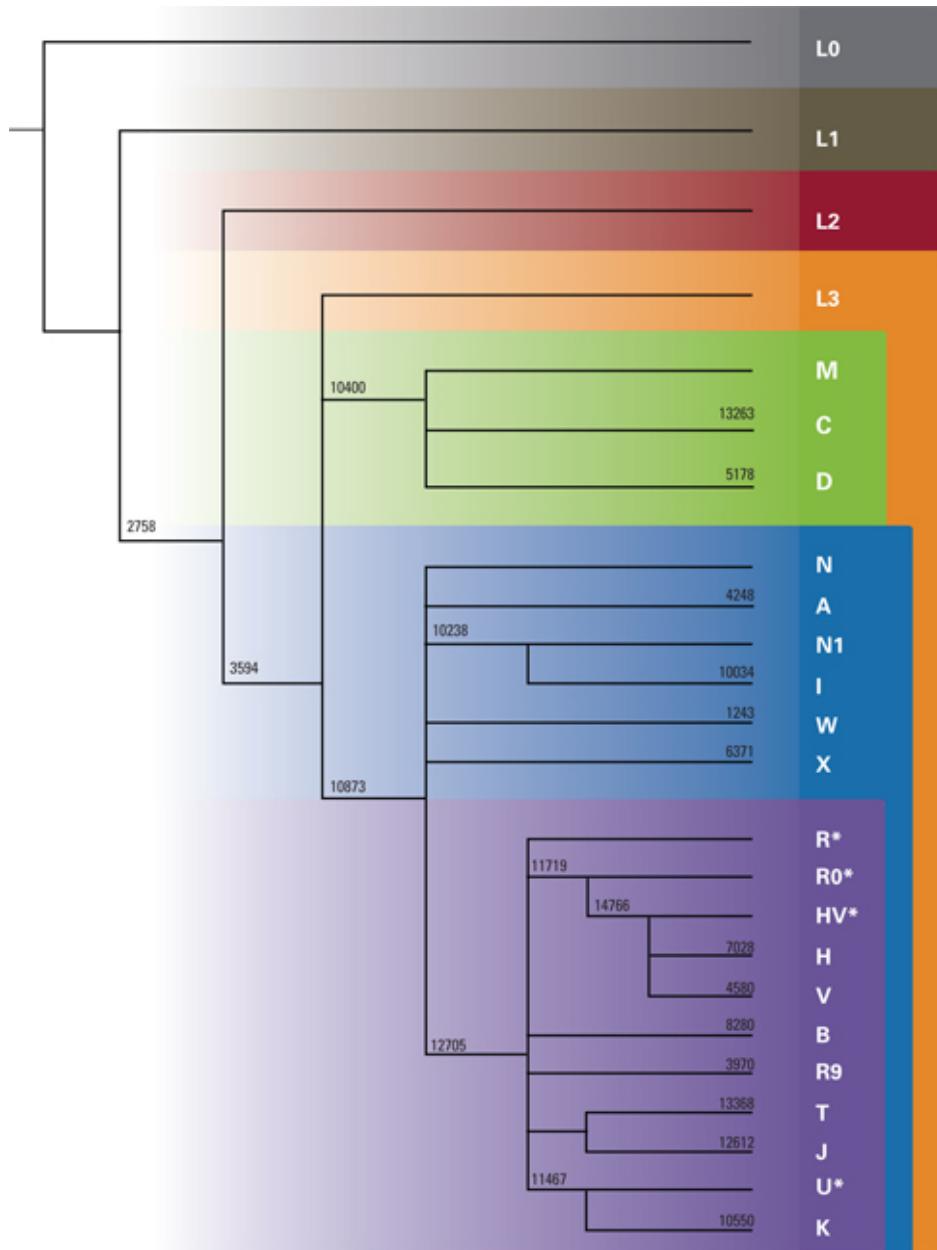
How Do I Interpret My mtDNA Results?



mtDNA tests, which sequences 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:

(Chart is on the next page)



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 **A**denine, **T**hymine, **C**ytosine, and **G**uanine.

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](http://www.familytreedna.com/login.aspx) <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](http://www.kerchner.com/haplogroups-mtdna.htm) <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](http://en.wikipedia.org/wiki/Human_mitochondrial_DNA_haplogroup) <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?

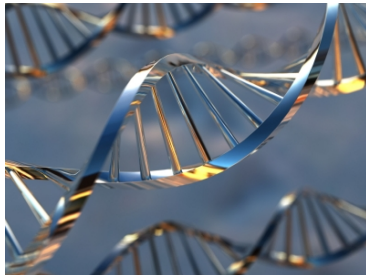
Sometimes mtDNA results will list a Haplogroup designation (such as H) without listing a particular subclade or branch of that Haplogroup (such as H1, H2, H2a1, H2a2, etc...). This might be because (1) the sample matched the haplogroup but none of the subclades, or (2) the sample was not tested or analyzed 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

How Do I Interpret My Family Finder Results?



In this new chapter added by Family Tree DNA, we explore new autosomal testing that has become available since the eBook's initial publication: the Family Finder test.

This autosomal test, available for both men and women, examines SNPs (single nucleotide polymorphisms) across the 22 autosomal chromosome pairs. These chromosomes contain DNA from all sides of your family. The Family Finder test examines hundreds of thousands of SNP positions and uses the results to find recent relatives and geographic origins.

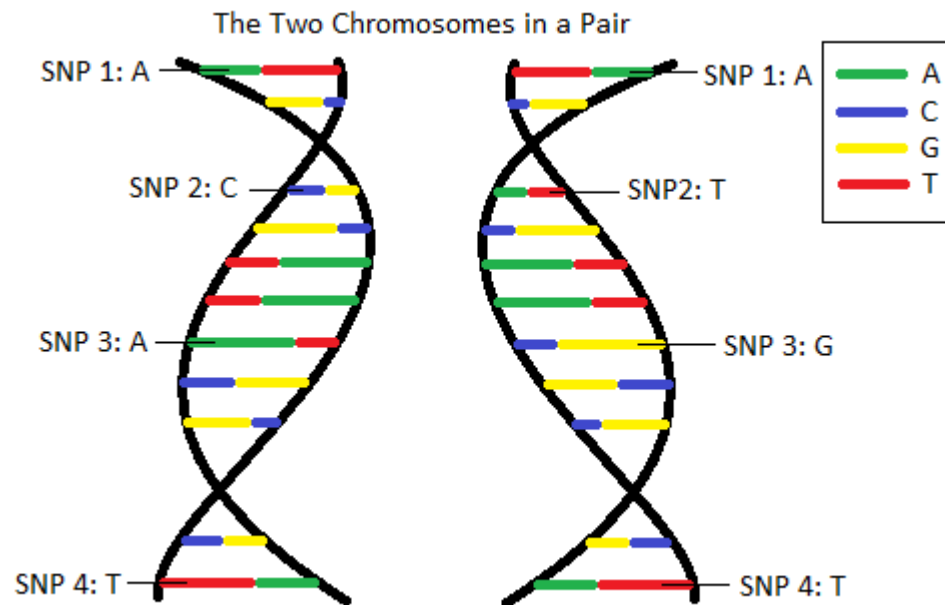
A. Family Finder Testing

For the purpose of understanding how the results are used, we will look briefly at the raw data generated by the Family Finder test. Please keep in mind that you will probably never need to view your own raw data.

Autosomal chromosomes, or autosomal DNA, are inherited from both parents and mixed from one generation to the next. Every person has 46 chromosomes in total, 23 from each parent. 2 of these 46 chromosomes are the sex chromosomes. The other 44 are autosomal chromosomes.

The autosomal chromosomes come in pairs, with one chromosome from each parent in every pair. The 22 pairs are numbered 1 through 22. Testing autosomal SNPs yields two results for each SNP because the same position in the DNA sequence exists in both chromosomes in a pair. The two results may not be the same, though, depending on what DNA you received from your parent.

Here is a brief example of what these results look like:



SNP	AA
1	
SNP	CT

2	
SNP	AG
3	
SNP	TT
4	

What do the letters mean? Each letter refers to a base in the DNA sequence (similar to a letter in a sentence). The four bases are represented by the letters A, C, G, and T. Two A's mean that the SNP result on both chromosomes is an A. One C and one T mean that one of the chromosomes has a C for this SNP and the other has a T.

A **homozygous (homo = same)** result is when both letters are the same, like AA. A **heterozygous (hetero = different)** result is when the two letters are different, like CT.

We cannot tell which result is from which chromosome; in other words, we cannot tell whether it was your mother or your father who gave you the C in SNP 2.

When used by the thousands, SNP results from two different people can be compared to find if they are related in a recent time frame and, if so, how closely related they are. We can also compare the results with populations that have been tested around the world to find out the regions of the world where your ancestors most likely originated.

B. Relationship Testing (Family Finding)

How does it work? Just as you have two of each chromosome, your parents do, too. The chromosome 1 you received from your mother is a mix of the DNA from both of her chromosome 1s, and the chromosome 1 you received from your father is a mix of the DNA from both of his chromosome 1s.

Let us call your father's chromosome 1 pair Chromosome 1A and Chromosome 1B. Chromosome 1A came from your father's father, and Chromosome 1B came from your father's mother. During sperm creation, Chromosomes 1A and 1B trade large sections of their sequence to build a unique new Chromosome 1 that is ultimately passed down to you. Your chromosome 1 from your father contains a chunk from Chromosome 1A, then a chunk from Chromosome 1B, etc. This process is called **recombination**.

For example, in the picture of a chromosome below, imagine this is a chromosome you inherited from your father. The sections in blue are from your father's father, and the sections in orange are from your father's mother.



Recombination takes place in all the autosomal chromosomes you inherit from both of your parents.

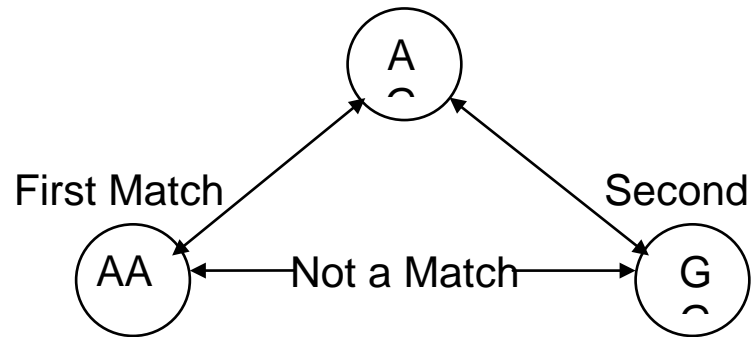
Because DNA is exchanged in sections, SNPs that are found close together on a chromosome are likely to belong to the same section and to be inherited together. When we search for matches, we're not just looking for what %

of SNPs match between two people. After all, we humans share well over 99% of our genome with all other humans. Instead, we are looking for sections of many SNPs in a row that match. The segments' size is a key factor in determining whether and how closely two people are related.

Family Finder is a $\frac{1}{2}$ identity by descent (IBD) test

When two people share the same segment of DNA, they are **identical by state (IBS)**. In other words, the state or sequence of their DNA is identical. This could either be because they share a common ancestor and inherited this same DNA from that common ancestor, or it could be by coincidence. If they did inherit the matching DNA from a common ancestor, then they are **identical by descent (IBD)** meaning the reason they are identical in this segment is that they descend from a common ancestor.

$\frac{1}{2}$ IBD means that only half of the result needs to match between two people in order for them to be related. For example, if your result for a particular SNP is AG, the A from one of your chromosome pair and the G from the other, then at that SNP you match anyone else who has at least one A or at least one G. You are also a match to anyone else who has both (AG matches AG), but it is not necessary to share both letters in order to match at the SNP. This is because you get one of these from each side of the family; someone related to you on your father's side would not be expected to match the DNA you received from your mother's side, too.



Generally when two people share a large segment in common, that segment is IBD. On the other hand, it is very common for people to have small segments of DNA that are IBS but not IBD, especially among members of the same population group, because the same DNA has been circulating in the population for many generations. If two people inherit a segment of DNA from their common ancestor but the segment of DNA is too small, we cannot tell whether it is IBD. In those cases, a connection between the two people is either speculative or not suggested at all.

Don't worry if your distant relatives test and do not show up as a match to you. The more generations there have been since two people share a common ancestor, the smaller and fewer the segments of DNA they share. After enough generations have passed, even though both could have inherited some DNA from a common ancestor, they may not have inherited the same segments of DNA as one another and the relationship cannot be detected.

Relationships can be detected up to 3rd cousin. Sometimes more distant relationships can be detected if the two people inherited enough of the same DNA from their shared ancestor.




1. Does Anyone Match My Family Finder Results?

To find other people who match your Family Finder results, we will take a look at the Family Finder's Matches and Chromosome Browser sections.

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 address to be shared with their matches.







Customers can view their list of matches in the Family Finder Matches section of their myFTDNA page.

Customers can visually compare their matches to see what portions of their chromosomes they share in common using the Family Finder Chromosome Browser tool.

What does the Family Finder Matches section tell me about my potential relatives? Here you can find the names and likely relationships of the people you match. Under each person's name is an envelope icon () you can click to email your match. You can also click the paper and pencil () to write notes to yourself about this match, such as whether you have contacted him, what actual relationship you think you share, or something similar about your ancestry that you think might lead you to find your family connection. Finally, if your match has uploaded a GEDCOM file into the system, you will see a third icon () you can click to view your match's family tree.

The suggested relationship tells you the likeliest relationship you have with your match, while the relationship range tells you the possible relationships you have with your match. Occasionally, especially for speculative matches, a person will be related to you more recently or more distantly than the relationship range shows, depending on the amount of DNA you have in common.

Next come two columns showing cM (centiMorgan) values: Shared cM and Longest Block. A centiMorgan (cM) is a measurement of how likely a segment of DNA is to recombine from one generation to the next. A single cM has a 1% chance of being broken apart and recombined in one generation. The shared cM number is the total size of all of your shared blocks of DNA. The longest block number tells you how big the largest segment of DNA you share is. People who are more closely related tend to have higher values for both, but the actual values vary from match to match depending on the size of the segments you happened to inherit in common.

Filter Matches By:		Relations:	<input type="checkbox"/> Exact Spelling for Matches <input type="checkbox"/> Hide 3rd Party Matches			
		Name:	<input type="text"/>	Ancestral Surnames:	<input type="text"/>	<input type="button" value="Filter"/>
Name	GENEALOGICAL MATCHES BASED ON DNA				KNOWN GENEALOGY	
	Relationship Range	Suggested Relationship	Shared cM	Longest Block	Known Relationship	Ancestral Surnames <small>(Bolded names match your surnames)</small>
Harold Weimar   	1st Cousin - 3rd Cousin	2nd Cousin	384.11	98.54	<u>2nd Cousin</u>	Babior Podolsky Weisblat
Anthony Bolden   	1st Cousin, Half Siblings, Grandparent/ Grandchild, Aunt/ Uncle, Niece/ Nephew	1st Cousin	1052.63	98.64	(Pending) <input type="button" value="Assign"/>	Becky (Rebecca) Braun Harris Klein

The last two columns are pieces of information you and your matches can enter to share with one another. The known relationship column is a place where you can enter the actual relationship when you know exactly how you are related to your match. Your match will have to confirm the relationship is correct before it is listed in this column. The last column shows the ancestral surnames entered by your match. You can enter your ancestral surnames in the User Preferences section of your myFTDNA page or by uploading a GEDCOM, or family tree file, and then importing the surnames. See the Family Finder FAQ for more details about this chart and instructions

on how to enter known relationships and ancestral surnames:

<http://www.familytreedna.com/faq/answers.aspx?id=33>

You can filter your Family Finder matches based on how closely related they are to you, whether they share a surname in their list of ancestral surnames, whether you have entered a known relationship, and whether they are new matches since the last time you logged in. Here is a legend to how the filter divides matches by relationship:

Immediate Relative	Parent/Child, Sibling, Half Sibling, Grandparent/Grandchild, Aunt/Uncle, Niece/Nephew
Close Relative	1 st Cousin to 2 nd Cousin
Distant Relative	3 rd Cousin to 4 th Cousin
Speculative Relative	May or may not be related, probably more distant than 4 th Cousin

If you have also tested Y-DNA or mtDNA, you may be interested in comparing your list of Family Finder matches with your closest Y-DNA and mtDNA matches to see if any are on both lists. An exact match at the highest level of Y-DNA or mtDNA testing may or may not be close enough to be detected as a Family Finder match, but you can certainly compare your matches sections to find out. For example, if someone matches you in Family Finder

and is a close match on Y-DNA, then the relationship found by the Family Finder test is almost certainly the same common ancestor you share on the Y-DNA line.

How can I see what portions of my DNA I inherited in common with my matches? The Chromosome Browser tool is an excellent way to visualize how much of your DNA and what sections of your chromosomes you share with your matches.

In the Family Finder Chromosome Browser section of your myFTDNA page, you can choose up to 5 of your Family Finder matches to compare at once. If you compare matches and find that you share one or more of the same segments with multiple people, that segment may have been inherited from a single ancestor that all of you share. It can be helpful to compare a known cousin with other unknown relatives to see if some of them share the same segments. If so, they may be related on the same side of the family as your known cousin. When you are comparing matches, you can click on their name to view more details about the match, including a space to enter notes.

Here is a sample comparison in the Chromosome Browser:

(Chart is on the next page)

Compare Genes		5+ cM	Remove
No Photo	Anthony Bolden Shared Segments:65		
No Photo	Harold Weimar Shared Segments:42		

Personal Details

Harold Weimar

Total CentiMorgans (cM): 384.109988
Shared DNA Segments: 42

Predicted Relationship: 2nd Cousin

Variance Range: 1st Cousin - 3rd Cousin

Surnames: (Matches Appear Bold)
Babior (), Podolsky (), Weisblat ()

Enter Notes about this relative.

The blocks in orange and light blue represent segments of DNA shared with this person's two selected matches. All 22 chromosomes are represented in descending order. Some sections overlap, but many segments do not. This person is probably related to these two individuals through different family lines.

Don't worry if you find that two of your matches share the same significant segment of DNA with you but, when you communicate with these matches, you discover they do not match one another. At first, it can seem that there must be an *error* and that they should match one another. However, this is not necessarily the case. Almost always in this situation, one of your matches is related to you on your father's side and one on your mother's side, and they are not related to one another. They can match on the same segment of the chromosome and not be related to one another because you have two of each chromosome. Here is a simplistic example:

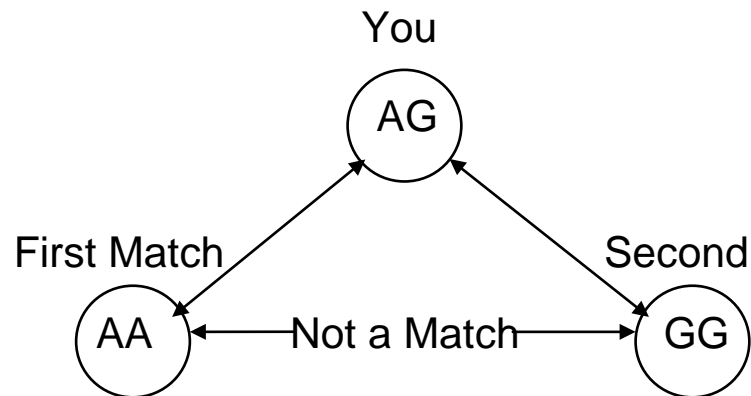
At a SNP, your result is AG, meaning one of your chromosomes has an A for this marker and the other has a G. Remember one of these came from your mother and one from your father.

At the same SNP, your first match's result is AA. Both of his chromosomes have the A. His result is not exactly the same as yours, but since most of your relatives are related on only one side of the family and not both, it only takes one matching letter for the SNP to match. You match him at this SNP because you both have an A.

Also at this SNP, your second match's result is GG. Both of her chromosomes have the G, and you match her at this SNP because you both have at least one G.

First Match	You	Second Match
AA	AG	GG

However, your first match and your second match do not match one another: AA and GG do not share anything in common. Further, since you inherited your A from one parent and G from the other, the one who matches your A is related through the first parent and the one who matches your G is related through your other parent.



Two people are a match when a very large number of SNPs in a row match. If there are many places where you have a result like AG and your two matches are AA and GG, then your two matches differ from one another at too many places for this to be a segment of DNA they share in common. Because of this, they do not show up as a match to one another even though they are both related to you and coincidentally match you in the same or similar segment of DNA.

2. Should I Join a Project?

A DNA project is a collaborative effort to answer genealogical questions using the results of DNA testing. Surname Projects bring together individuals with the same or very similar surname; Geographic Projects gather individuals by location. Some projects are focused exclusively on one type of testing, such as Y-DNA testing. Other projects focus on one type of test but include others as additional information. If you are unsure whether a project accepts or uses Family Finder test results, contact the project's volunteer administrator to find out.

There is a type of project that is specifically for Family Finder results: Family Finder Projects. These are very specialized projects and tend to focus on known descendents of a particular ancestor in an effort to learn more about that ancestor's DNA. Because of their specialized nature, these projects operate by invitation only.

C. Population Testing (Searching for Origins)

Autosomal results can also be compared with populations from around the world to see which ones your DNA resembles the most. Only some populations in the world have been tested, but we can still use those populations to represent the regions of the world. With this comparison, it is possible to estimate what % of your DNA is inherited from ancestors of indigenous European, American, African, Middle Eastern, and Asian origin. Sometimes it is even possible to point to subcontinents, depending on how closely your DNA fits the reference populations.

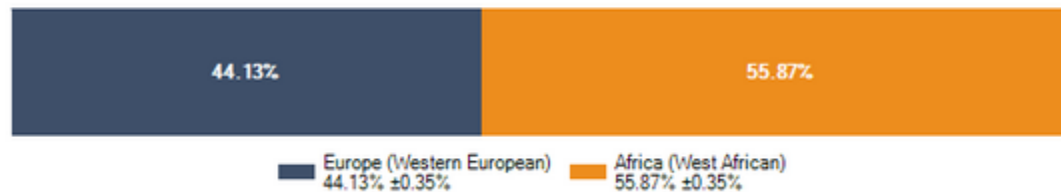
You can find the results from this analysis by checking the Population Finder section of your myFTDNA page.

1. How Do I Read My Population Finder Results?

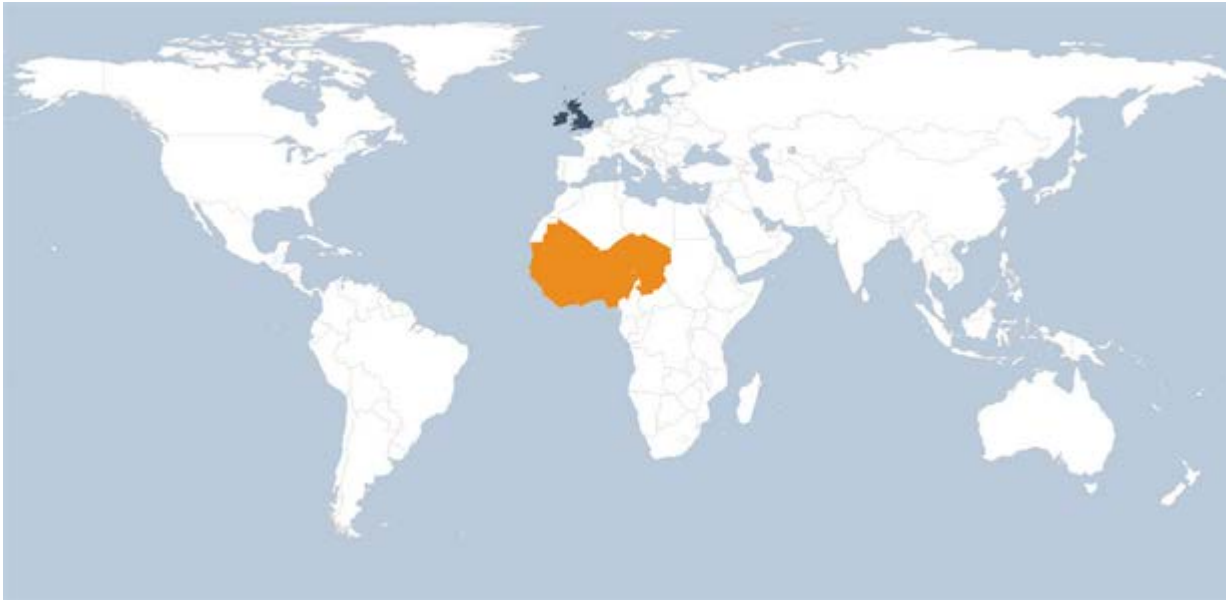
In your Population Finder section, you can find the breakdown of your origins based on how well your DNA matches the populations in the database. Over time more world populations may be tested to compare you with, which would have a minor effect on the breakdown of your origins shown in this section.

You can use the chart in either the default bar graph or in the pie chart option to see how much of your DNA fits with what region of the world. The world regions are broken down into continents and major subcontinent areas, such as Europe (Western Europe). If your DNA does not clearly fit into a subcontinent, you will just see the continent listed. Here is a sample from a person with European and African origins:

Chart Type: Stacked Bar Pie



The map gives you a general idea of where your ancestors came from based on the populations you did and did not match. Your ancestry is not absolutely restricted to these colored areas, but is most likely from these places.



Also provided are the world populations you matched from each region, such as:

Continent (Subcontinent)	Population	Percentage	Margin of Error
Europe (Western European)	Orcadian	44.13%	±0.35%
Africa (West African)	Yoruba, Mandenka	55.87%	±0.35%

Remember that not all populations in the world have been tested, so these populations are similar to but not necessarily the same as your own ancestral populations. For example, if your matching populations include

Finnish, French, and Tuscan, but you know at least some of your ancestry is from Germany, these results do not conflict with what you know about your ancestry; all of these populations are from Europe.

D. What Can Family Finder NOT Do?

1. The results of this testing still does 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. Autosomal testing cannot detect a relationship if the two individuals did not inherit any of the same segments of DNA from their common ancestor. Family Finder does not control what you inherited from your parents, or what your parents inherited from your grandparents. In other words, if the relationship is too distant or you just did not happen to inherit the same segments of DNA from your common ancestor, the DNA test cannot detect the relationship. This only tends to happen when the relationship is more distant than 3rd cousin.
3. Autosomal testing also cannot confidently detect a relationship if two individuals inherited only very small segments of DNA in common from their shared ancestor. That is because humans already have extremely similar results to one another, so two random people can also match on very small segments of DNA without having shared a recent common ancestor.

4. This test still only surveys a small portion of your genome. We select positions in the DNA that are known to sometimes differ between unrelated people, which gives us the best opportunity to find variety and distinguish between related and unrelated people. However this is not the same thing as a full genomic sequence.

5. The Family Finder test also avoids DNA positions that have known medical implications as this test is designed to be for genealogical and anthropological purposes only.

E. Additional Resources

1. Chromosome Browser Tutorial. You can find this tutorial in the top right corner of the Chromosome Browser section of your myFTDNA page

2. Frequently Asked Questions – Family Finder: <http://www.familytreedna.com/faq/answers.aspx?id=33>

3. Frequently Asked Questions – Population Finder:
<http://www.familytreedna.com/faq/answers.aspx?id=22>

Chapter 5

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

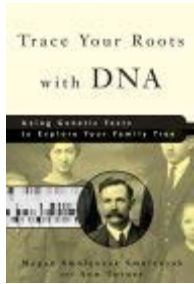
message separately, or receive groups of messages, or receive no messages (and then you can independently monitor the site at your leisure).

3. **The International Society of Genetic Genealogy (ISOGG <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 <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 <www.familytreedna.com/newsletter.aspx>](http://www.familytreedna.com/newsletter.aspx) of the Facts and Genes newsletter and [subscribe <www.familytreedna.com/facts-and-genes-register.aspx>](http://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 <www.rootstelevision.com>](http://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.

[Insert DNA & Genealogy book ref here]

[Insert new Chris Pomery book ref here]



the genetic genealogist

ADDING DNA TO THE GENEALOGIST'S TOOLBOX