

How Can I Use mtDNA?

Testing your mitochondrial DNA (mtDNA) may help you find answers to any of these genealogical questions:

- Who is my ancestor's mother?
- Where did my female ancestor come from?
- How can I fully document my direct maternal line?

How Does mtDNA Work?

Mitochondrial DNA has a unique inheritance pattern. It is passed from a mother to all of her children, regardless of gender. However, only the females can pass it on to the next generation. Therefore, you

Getting Started

Take the mtDNA test from FTDNA and set up your FTDNA account at www.familytreedna.com.

Link Your Family Tree This is good genealogical etiquette and shows matches what you already know. In the top menu bar, click Family Tree. Enter tree data or upload an existing tree file.

Enter Earliest Known Maternal Ancestor Under your profile (upper right corner) > Account Settings > Genealogy > Earliest Known Ancestor, enter your earliest known maternal ancestor's name.

Enter Beneficiary Information Designate someone to manage your account (should something happen to you) under your profile > Account Settings > Account Information > Beneficiary Information.

Check Privacy/Sharing Settings Under Account Settings > Privacy and Sharing, choose among several ways to ensure you are comfortable with the ways others can view your data.

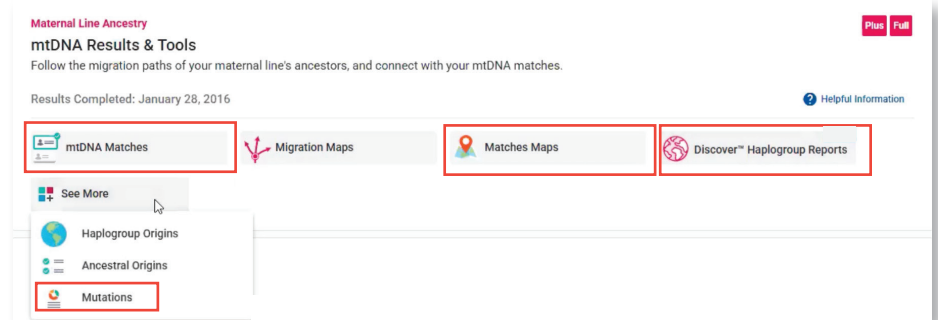
have your mom's mtDNA, as do all of your siblings, and it is very likely to be exactly the same mtDNA your 5th great grandmother carried around with her all those years ago. This means that any two living people today who share mtDNA should share a common direct maternal line ancestor. However, because of the way mtDNA is inherited, that shared ancestor may be many, many generations ago.

Which Test Should I Take?

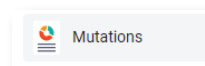
The Full Mitochondrial DNA Sequence (FMS) test evaluates every single mitochondrial DNA location, making it the most genealogically valuable kind of mtDNA test. Currently FamilyTreeDNA (FTDNA; www.familytreedna.com) is the only commercial genetic genealogy company providing this FMS testing. Two other DNA testing companies, 23andMe and Living DNA, also provide you with mtDNA origins information but they do not provide cousin matching.

What Do I Get?

Logging into FTDNA brings you to the *Dashboard*. Under the mtDNA header you will see several menu items that highlight different aspects of your test. We will cover the four most important sections in this guide.



Each mtDNA test will produce two kinds of information: an mtDNA profile, which is just a list of numbers, and a haplogroup, or deep ancestral group. In general, your profile is useful for genealogy as it helps you find genetic cousins, while the haplogroup can give you clues to the origin of your maternal line. See an example of an mtDNA profile on page 2, and learn about haplogroups on page 3.



Mutations

Your mtDNA data is reported as your differences (or mutations) from a standard reference sequence at the locations evaluated. Taken all together, your list of differences is called your mtDNA profile. Understanding the numbers in your mtDNA profile can provide insight into how mtDNA matching works and



Reference Sequences

While a version of the rCRS has been used in academia and beyond since its inception in 1981, in 2012 a new standard was introduced, the RSRS. When you view your mtDNA results at FTDNA by choosing *Results* from the *Dashboard*, you will be initially presented with your values as compared to the RSRS. However, the rCRS is used by most of the academic community.

HVR1 DIFFERENCES FROM rCRS					HVR2 DIFFERENCES FROM rCRS					CODING REGION DIFFERENCES FROM rCRS				
★16224C	16311C	16319A	16463G	16519C	73G	152C	199C	263G	309.1C	750G	1189C	1438G	1811G	2706G
					315.1C					3480G	★4646C	4769G	5913A	7028T
										8860G	9055A	9698C	9962A	10289G

To learn why two positions in this mtDNA profile are starred, see page 4.





therefore impact the way you use your results.

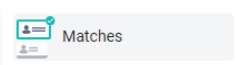
The image above is an mtDNA Profile. Each value in the profile consists of a position on the mtDNA (indicated by the number) where this person's mtDNA differs from the reference sequence, followed by a letter. The letter is their value at that location. Because only differences are reported, the number of values different people have in their profile will vary.

The table is divided into three sections: HVR1, HVR2, and the Coding Region. HVR1 and HVR2 are the most variable regions, and the Coding Region is where most of the instructions for the day-to-day business of the mitochondria are held.

Taken all together, your mtDNA profile represents you and everyone in your direct maternal line. It is like having a primary genealogical record right there in your DNA.

Coding Region (115) ⓘ Hypervariable Region 2 (327) ⓘ Hypervariable Region 1 (2564) ⓘ [Filter](#) [Export CSV](#)

Name ⓘ ↑	Genetic Distance ⓘ	mtDNA Haplogroup ⓘ	Mitotree Haplogroup ⓘ	Maternal Country of Origin ⓘ	Maternal Earliest Ancestor ⓘ	Match Date ⓘ	Actions ⓘ
Emily Yates	Exact Match Link on...Tree	U5a1b	<input checked="" type="checkbox"/> U5a1b <input checked="" type="checkbox"/> F5697126	England	Carrie Payne b. 1862 London	Aug 8, 2024	 
Alex Stephen	1 step Link on...Tree	U5a1b	<input type="checkbox"/> U5a1b <input type="checkbox"/> F2549637	Unknown Origin	-	Jun 3, 2021	 



Match Page

You will spend a majority of your genetic genealogy efforts on the mtDNA Match Page. Outlined here are the sections you need to explore in detail.

1 Regions

This page will initially show you matches at the highest level of testing you have had completed. If you have completed the FMS test, you can click the tabs to change the view to show those who have had just HVR1+HVR2 tested, or only HVR1 tested. The FMS test is shown as Coding Region.

Keep in mind that as you move down in testing resolution that the match table is cumulative, meaning that it includes everyone who was tested at that level and above. This brings up the second section of results, Genetic Distance.

2 Genetic Distance

This column is only present when you are viewing Full Mitochondrial DNA Sequence results. That is because FTDNA allows you to see individuals with up to **three differences** from your profile at the FMS level. However, at lower levels of testing only **exact** matches are shown. Please keep in mind that those on the list may be matching exactly at the lower level of testing, but they may not be matching exactly at the FMS level. Be sure to check the level of testing

they have had completed in the third column of this table (see the Tools section below) to be sure you are making the correct assumptions about the match strength. **Only exact mtDNA matches should be evaluated for a possible relationship.** There are exceptions to this rule, as a mutation can occur at any generation, but in general you will want to stick to your exact matches, as even with an exact match, you are not guaranteed a shared recent common ancestor.

3 Name

Click on the name to bring up a snapshot of your match's profile page, complete with contact information, when provided.

4 Actions

Family Tree Icon: When filled in, the dark blue icon indicates you may view your match's family tree.

Notes Icon: This is where you want to record anything you have learned about this DNA match.

5 Maternal Country of Origin and Maternal Earliest Known Ancestor

Review the genealogical information posted by your matches, looking for locations familiar to your maternal line. Uploading your family tree will not automatically fill in these fields. See *Getting Started* on page 1 for more details.

6 mtDNA Haplogroup and Mitotree

Haplogroup

A haplogroup is a deep ancestral group helping to define where your direct maternal line ancestors were anciently. There are two “trees” in circulation describing these haplogroups and their relationships to each other. Mitotree is the proprietary haplogroup tree being built by FTDNA. So, over time you may see slightly different values in these two columns as the general academic tree and Mitotree diverge in the way they are talking about things. For now, under the Mitotree column you may see two values - your general haplogroup, and a specific mutation that is, so far, relatively unique to your line and does not yet have a place in the larger Mitotree structure (but it may one day). Keep reading for more on your haplogroup.

7 Total Number of Matches


An often overlooked bit of information, the number of matches to your profile can actually reveal some interesting information about your probability of success using mtDNA in your family history research. If you have very few matches, especially at the HVR1 or HVR1+HVR2 level, your profile is overall very rare. Sometimes having a very rare profile makes the matches you do have even more valuable and more likely to indicate a shared recent common ancestor.

On the other hand, if you have hundreds of matches, it is likely that your mtDNA profile is not doing a very good job isolating your own maternal line. This means that an exact match could be your close cousin, or your 20th cousin, and it is really difficult to determine which one it is. Plus, with so many matches it can be overwhelming to try to find the most valuable matches. In that case, you need to be especially aware of any genealogical clues, like names and locations, that might set one match apart from another.

If you do have an overwhelming number of matches, consider using the *Matches Maps* tool to help you find your most valuable matches.

8 Filters

Clicking this button will bring up an additional menu that will let you sort your mtDNA matches according to desirable features. While many are available, one of the most valuable is the Family Finder filter. This lets you view your mtDNA matches who have also taken the Family Finder (FF, or autosomal DNA test) at FTDNA. Investigating their FF relationship, or lack thereof, can often be helpful when trying to determine your relationship to another DNA mtDNA match.

 Matches Maps

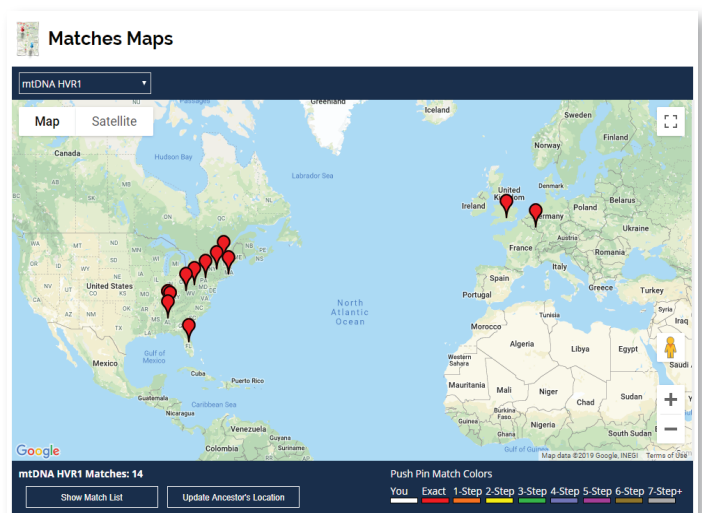
Matches Maps

Access this tool by selecting *Matches Maps* from your Dashboard. This tool can be a great resource to help you sort through your mtDNA matches, as it takes the information reported by your match in the *Earliest Known Ancestor* column and places it on an interactive map. There you can easily zoom into the locations of interest for your maternal line and locate matches of particular interest.

The tool defaults to showing you matches at the HVR1 level, so **be sure** to double check your match table, or use the dropdown menu in the upper left corner of the map tool, to determine if the individual is still matching you at higher levels of testing.

mtDNA Haplogroups

A haplogroup is a deep ancestral group usually associated with a general geographic location. It is named with an alternating letter/number combination, like K1b1a. You can think of these letter-number combinations like descendency charts, where the first letter is the earliest known ancestor, and the numbers and letters that follow are other ancestors on that line. Just like your ancestors, each of the characters in your haplogroup name has a time and a place. Just like your own genealogy, we don't always know all of those times and places, but we can get a general idea.



You are assigned to a haplogroup based on the values in your mtDNA profile. For example, let's take a closer look at the values in the table at the top of page 2. This individual is in haplogroup K1b1a1a. In a tree like Mitotree you will see that the presence of the *16224C* value in your mtDNA designates you as a member of haplogroup K. Find the value *16224C* in the chart. It is under the first section, *HVR1*, and marked with a star. There are other values that correspond to other subgroups of K including K1, K1b and so on, all the way to K1b1a1a. This final haplogroup assignment is defined by *4646C*, which you see starred under the third section.

Each character in your haplogroup name represents a tier of heritage. For example, values corresponding to the K portion of your haplogroup are likely upwards of 35,000 years old and were developed in Asia. So you will share these values with lots and lots of people. The K1 and K1b levels bring us 10,000 years into the future with movement west, toward Europe. And so it goes, on down to K1b1a1a that is found in France, Switzerland, and Ireland. So the values defining this more recent haplogroup will be shared with fewer people, but still quite a few. This is why you may find your mtDNA profile matching hundreds of people.



While most haplogroup origins are too far back to be helpful in determining where your maternal line ancestor is from, you can see that by understanding that not all of your mtDNA values are created equally, you are in a better position to understand your match list. If your profile is very common, and therefore you are in a very old haplogroup, then your matches are less likely to produce an identifiable recent common maternal ancestor, simply because that common ancestor could be thousands of years ago.

mtDNA and Your Health

In the early days of genetic genealogy, much effort was put forth to reassure those tested that no health information could be gathered from YDNA and mtDNA testing and that the results would not be able to uniquely identify you. While it is still true that your mtDNA and YDNA test results cannot identify you uniquely, with the onset of the Full Mitochondrial DNA Sequence (FMS) test there is a chance that a limited amount of health information can be revealed.

Your mitochondria provide essential functions to your cell. Therefore, it is possible that a change in your mtDNA code could cause a disruption in the normal cellular process. Because the FMS test evaluates every position on the mtDNA it has the potential to reveal if your mtDNA has undergone some adverse changes. The list of mtDNA associated diseases isn't that long.

Reports

This section contains several sections you will want to investigate to learn more about your own maternal origins. In this guide we will highlight the most powerful for your family history: the mtDNA Match Time Tree (mtMTT). The mtMTT is a powerful way to view your mtDNA full sequence matches, allowing you to view your connections on a timeline that helps you see the connections between the tested maternal lines.



Note specifically the dates across the top, showing the connection of these matches to an ancestor named V7e before 500 CE. Now review the four circled matches on the right and their F5930709 label. This label is the name of an mtDNA mutation that these four share, but that is not yet a part of the mtDNA tree, and therefore does not officially define a haplogroup. The fact that they all share this value tells us that these four individuals may share a common direct maternal ancestor with each other, before they share a common ancestor with the rest of the descendants of V7e.

In fact, some are even helpful, like recognizing you may have the mtDNA value that indicates exercise intolerance!

If you do have concerns about mtDNA and your health, you can visit mitomap.org. Partway down on the homepage, see the header *Disease Mutations (reports of possible disease-associations)*. You can look through the list for values present on your mtDNA profile to see if any of your mtDNA mutations have disease associations. Pay special attention to the status column which will tell you if a particular mtDNA value is simply reported as an association, or is actually confirmed. Discuss any concerns with your healthcare provider.

Read [mtDNA-related tips and stories](#) on our blog!

