

Thank you for testing at Family Tree DNA.

Your mtDNA (mitochondrial DNA) Full Sequence results are complete. These results are the first step in understanding your genetic ancestry through your direct maternal line.

This guide will explain how your results help with genealogy and distant ancestry as well as how to use your myFTDNA personal account.

The Direct Maternal Line



Your direct maternal lineage is the line that follows your mother's maternal ancestry. This line consists entirely of women, although both men and women have their mother's mtDNA. This means that fathers do not pass on their mtDNA to their children. Your mtDNA can trace your mother, her mother, her mother's mother, and so forth, and offers a clear path from you to a known, or likely, direct maternal ancestor.

Note that you and your matches may share ancestors on other parts of your family tree; however, those matches are coincidence.

Matching for Genealogy

Your mtDNA may help you find genetic cousins along your direct maternal line. When we look at your mtDNA HVR1, HVR2 and Coding Region results, we look for differences in your mtDNA caused by small changes - copying errors – that we also call "mutations" or "polymorphisms." We compare those differences to the differences for other people in our database. The range of possible generations before you share a common ancestor with a match is wide. Your mtDNA HVR1, HVR2 and Coding Region exact matches may be recent, but they may also be hundreds or thousands of years in the past. We show this in the table below.

Testing Level	Matching Lough	Generations to Common Ancestor					
	Matching Level	50% Confidence Interval	95% Confidence Interval				
mtDNA	HVR1	52 (about 1,300 years)	— NA*				
mtDNAPlus	HVR1 & HVR2	28 (about 700 years)	— NA*				
mtFullSequence	HVR1, HVR2, & Coding Region	5 (about 125 years)	22 (about 550 years)				

Note: The term "mutation" in this sense does not refer to anything medical.

* The range of generations to a common ancestor at this level is too broad to calculate a 95% confidence period.

The wide range in the test results does not prevent those results from being useful. You can use this clear maternal line to provide evidence to support a relationship. You first trace two or more female lineage descendants of a single woman utilizing traditional genealogy research. The descendants then test their mtDNA. If they are exact matches, it is evidence that supports the relationship. Not matching usually disproves the relationship.



Planned comparisons are the best choice. However, you can still find your common ancestor with as little as an HVR1 or HVR2 match. To do so, use your known maternal genealogy. For each match, look for common geographic locations on the direct maternal line. Work through each of your ancestors on this line as well as their daughters, their daughters' daughters, and so forth.

Comparing genealogical records is vital when using mtDNA matching to help you in your research. You need to enter all that you know about your direct maternal line in your myFTDNA account. See the "Quick Steps" reference page for step-by-step instructions on how to set up your account and access your matching information.

The Science of Your Direct Maternal Line

We can trace our direct maternal line with our mitochondrial DNA due to a special relationship between the power supply for human cells (the mitochondria) and the cell itself.

In every human cell, there are hundreds and sometimes thousands of mitochondria. Each mitochondrion has several copies of its genetic code (represented by the letters A, C, T, and G). This genetic code is mitochondrial DNA (mtDNA) and tells the mitochondria how to function. The code also tells the mtDNA how to copy itself. Over time, the copying process can create small changes - "polymorphisms" or "mutations." If these changes are in the mother's egg cell, the child produced from that egg inherits them. If female, the child may some day pass that same change on to her own children.

Slowly, these changes build up down maternal lineages. They define and mark branches on the maternal tree. We can look at your specific mtDNA code sequence to see which changes you have in your mtDNA.

There are two scientific baseline sequences to which scientists compare these changes in mtDNA: the Reconstructed Sapiens Reference Sequence (RSRS) and the revised Cambridge Reference Sequence (rCRS). By comparing your mtDNA changes to each sequence, we can distinguish the differences between the original values in the RSRS (or the comparative values in the rCRS) and your test results. Following scientific standards, Family Tree DNA compares all mtDNA results to the rCRS, and provides you with your comparisons to the RSRS.

Your Results													
RSRS Values	rCRS Values	mt	DNA Commu	nity									
HVR1 DIFFERENCES FROM rCRS		HVR2 DIFFERENCES FROM rCRS				CODING REGION DIFFERENCES FROM rCRS							
CRS		195C	263G	315.1C	513A	522-	750G	1438G	3992T	4024G	4769G		
		523-					5004C	5773A	8269A	8860G	9123A		
							13889A	14133G	14365T	14582G	15326G		
Revised Cambridge Refere	ence Sequence												

The example above shows results for someone who has tested all three parts of their mtDNA: the HVR1, the HVR2, and the Coding Region.

In the example, each letter-number combination represents a difference between your results and the reference sequence. These individual changes do not indicate any particular ethnic or geographic origin. Their significance is in their ability to mark branches on the maternal tree, and those branches are geographically and sometimes ethnically significant.



Your Ancestral Origins

Your direct maternal ancestors have passed down their mtDNA generation after generation. The line began with our common maternal ancestor in Africa and it ultimately reached you. Our mtDNA marks the path from our ancestors in Africa to their locations in historic times. Your ancestors carried their mtDNA line on their historic migrations. Your line's present geography shows the path of this journey. Your mtDNA results are the most precise DNA tool for this line. Your mtDNA results use two strategies to explore your maternal origins.

The first method uses your main (backbone) branch on the maternal tree, known as an mtDNA haplogroup. Scientists study the history of populations across geography and time using mtDNA. They often use the frequencies of each branch in modern populations, as well as samples from ancient burial sites. With these, they are able to tell us much about the story for each haplogroup. This traces back hundreds, thousands, or even tens of thousands of years. Your branch on the tree tells you where your maternal ancestors are present today and about their likely path out of Africa. We summarize what scientists know about your mtDNA Haplogroup on the *mtDNA* – *Results* page of your **myFTDNA** account.

The second method uses your haplogroup information alongside your mtDNA Full Sequence differences. We use these to match you to the information of others in our database. These matches are more likely to share your lineage in closer genealogic time. Country frequencies thus reflect the more recent history of your ancestors. Both the *mtDNA – Haplogroup Origins* and the *mtDNA – Ancestral Origins* pages show this information. **See the "Quick Steps" reference page for step-by-step instructions on how to access this information.**

Privacy and Courtesy Standards

Privacy

Family Tree DNA cares about your privacy. When you use our internal database, we can assure your privacy. However, please use caution if anyone asks you to export your results to other databases not endorsed by Family Tree DNA. Be sure you have confidence in the privacy protections of whoever is requesting your data. You should not ever share your kit number and password outside of Family Tree DNA.

Courtesy Standards

Family Tree DNA would like genetic genealogy and DNA testing to be an enjoyable experience for all. We ask that our customers treat each other, project administrators, and Family Tree DNA staff with courtesy and respect. When your matches reach out to you, please respond to them. Even an answer of "I don't know" is better than no answer. At all times, respect the privacy of others. Everyone has his or her own comfort level for sharing genetic information. The best policy is to ask before doing.

Resources

Useful links:

myFTDNA 2.0 User Guide: mtDNA - http://www.familytreedna.com/faq/answers.aspx?id=49

Understanding Results: mtDNA - http://www.familytreedna.com/faq/answers.aspx?id=10

mtDNA Library of Scientific Papers - http://www.familytreedna.com/mtdna-papers.aspx

Glossary (complete) - <u>http://www.familytreedna.com/faq/answers.aspx?id=21</u>



Common Terms

Coding Region: The coding region is the slow changing part of the mitochondrial genome. It contains genes, causing it to have a slower change rate than the rest of the genome.

Direct Maternal Line: The direct maternal line is the line that traces your mother, her mother, her mother's mother, and so forth. With the exception of yourself if you are male, this line consists entirely of women.

GEDCOM – An internationally recognized software file (.ged) that allows you to organize genealogical information. You may export this file type from a number of software programs and upload your GEDCOM to the Family Tree DNA site. Family Tree DNA does not host or provide this software for you to create your GEDCOM.

Genetic Cousins – Individuals whose DNA test results match one another, indicating shared genetic ancestry.

Genome: A genome is a full set of genetic code. Humans have two genomes, the nuclear genome and the mitochondrial genome. The human nuclear genome is composed of 46 chromosomes (23 pairs), which contain 3 billion base pairs. The human mitochondrial genome is composed of a single circular DNA sequence that contains 16569 base pairs.

Haplogroup - A haplogroup is a major branch on either the maternal or the paternal tree of humankind. Haplogroups are associated with early human migrations. Today these can be associated with a geographic region or regions.

HVR1 (Hypervariable Region 1): HVR1 is one of two faster changing parts of the mitochondrial genome. There are two human Hypervariable Regions, HVR1 and HVR2. They do not contain genes. Therefore, they have a faster change rate than the coding part of the mitochondrial genome.

HVR2 (Hypervariable Region 2): HVR2 is one of two faster changing parts of the mitochondrial genome. There are two human Hypervariable Regions, HVR1 and HVR2. They do not contain genes. Therefore, they have a faster change rate than the coding part of the mitochondrial genome.

mtDNA (Mitochondrial DNA): Mitochondria are parts (organelles) within human cells. Our mitochondria provide cell respiration and make energy for the cell. Mitochondria were once separate organisms. Early mitochondria evolved inside primitive nuclear cells. They formed a mutually beneficial relationship with the larger nuclear cell and have their own mitochondrial DNA (mtDNA) genome. Human mothers pass their mtDNA genome to their children.

MDKA (Most Distant Known Ancestor): Your most distant known ancestor (MDKA) is the furthest person who you have documented on a specific genealogical line. In genetic genealogy, it usually refers to someone on a direct maternal line (the mother, her mother, her mother's mother, etc.) or to a direct paternal line (the father, his father, his father's father, etc.).

Polymorphism: Polymorphisms are changes to your DNA code. They are natural copying errors. One analogy is to think of a copy machine that is making many copies of a page. Occasionally it will make a mistake; an e might look more like an o, for example. This is a "polymorphism." If you then take that page with the o and copy it, it will pass on its "polymorphism" to all of its descendant copies.

RSRS: The Reconstructed Sapiens Reference Sequence (RSRS) is the sequence against which scientists compare all tested results. Researchers created it using a global sampling of modern human samples and samples from ancient hominids. It is less likely to have bias toward one population or maternal lineage.

rCRS: The historic revised Cambridge Reference Sequence (rCRS) is the sequence based on the first complete mtDNA sequence completed.



Genealogy Quick Steps

Entering Your Ancestry Information

Your **myFTDNA** account has two places for you to enter ancestry information. They are the *My Account – Most Distant Ancestors* page and the *My Account – GEDCOM/Family Tree* page. After you enter information, you will be ready to access and contact your matches.

The *My Account – Most Distant Ancestors* page is where you enter basic information about your most distant known ancestor (MDKA) on your direct maternal line. To do so:

- 1. Log in to your **myFTDNA** account. (<u>https://www.familytreedna.com/login.aspx</u>)
- 2. On the top menu bar, find the *My Account* menu.
- 3. From the *My Account* menu, select *Most Distant Ancestors*.
- 4. On the *My Account Most Distant Ancestors* page, look for the *Direct Maternal* area in the *Most Distant Ancestors* section.
- 5. In the *Direct Maternal* area, select the *Country of Origin*. If you are not sure of this, select <u>*Unknown*</u>.
- 6. In the *Direct Maternal* area, enter the name, date of birth, and date of death of your most distantly known direct maternal ancestor in the *Name* field.
- 7. Click the Save button to save your changes.
- 8. On the *My Account Most Distant* Ancestors page, find the *Direct Maternal* area in the *Ancestral Locations* section.
- 9. Click the *Add Location* button to launch the *Ancestral Locations Map* wizard.
- 10. In the *Maternal Location* section, click the blue *Edit Location* button.
- 11. Follow the steps to add your direct maternal most distantly known ancestor's location. This is usually the place where she was born. If you do not know that, it can also be where she was married or where she died.

The *My Account – GEDCOM/Family Tree* page is where you upload an electronic file of your pedigree. GEDCOM is the standard format for sharing electronic pedigree information, and almost all genealogy software packages are able to export to standard GEDCOM format (.ged). To upload your file:

- 1. First, you must export your GEDCOM file from the software in which you developed your family tree (you should be able to do this by clicking on File in that program and then Export to save this file to your computer). *Please note that Family Tree DNA does not host or offer customer support for any GEDCOM software.*
- 2. Log in to your **myFTDNA** account. (<u>https://www.familytreedna.com/login.aspx</u>)
- 3. On the top menu bar, find the *My Account* menu.
- 4. From the *My Account* menu, select *GEDCOM/Family Tree*.
- 5. On the *My Account GEDCOM/Family Tree* page, look for the *GEDCOM* section and the *Next* button.
- 6. Click on the *Next* button to start the *GEDCOM Upload* wizard.



Accessing Your Matches

You may view your mtDNA Full Sequence matches on our website by following these steps:

- 1. Log in to your **myFTDNA** account. (<u>https://www.familytreedna.com/login.aspx</u>)
- 2. On the top menu bar, find the *mtDNA* menu.
- 3. From the *mtDNA* menu, select *Matches*.

The *mtDNA* – *Matches* page has two sections. The top *Filter Matches* section is where you can change the testing level and filter for specific parameters. The bottom *Matches* section is where you can view your matches. To view your most recent mtDNA HVR1 and HVR2 matches:

- 1. In the *Filter Matches* section, change the *For:* field to HVR1, HVR2, Coding Region.
- 2. Click the orange *Run Report* button

By default, your most recent matches will be at the top of the report. You can read about additional settings for the mtDNA – Matches page by clicking on the blue page Help button at the top of the page.

Ancestral Origins Quick Steps

mtDNA Haplogroup Information

To find information about your maternal branch (haplogroup):

- 1. Log in to your **myFTDNA** account. (<u>https://www.familytreedna.com/login.aspx</u>)
- 2. On the top menu bar, find the *mtDNA* menu.
- 3. From the *mtDNA* menu, select *Results*.
- 4. On the *mtDNA Results* page, look for the *Your Origin* section.

The Your Origin section describes what we know about the origin and historic migrations of your direct maternal line.

Haplogroup and Ancestral Origins Information

To find information about your more recent ancestral origins, begin with the mtDNA – Haplogroup Origins page:

- 1. Log in to your **myFTDNA** account. (<u>https://www.familytreedna.com/login.aspx</u>)
- 2. On the top menu bar, find the *mtDNA* menu, and then select *Haplogroup Origins*.
- 3. On the *mtDNA Haplogroup Origins* page, look for blue *Help* button. The help section describes how to interpret the table of populations and frequencies on the page.

Next, check the mtDNA – Ancestral Origins page:

- 1. Log in to your **myFTDNA** account. (<u>https://www.familytreedna.com/login.aspx</u>)
- 2. On the top menu bar, find the *mtDNA* menu, and then select *Ancestral Origins*.
- 3. On the *mtDNA Ancestral Origins* page, look for blue *Help* button. The help section describes how to interpret the table of populations and frequencies on the page.