



S. C. S. Computer / Genealogy Class SIG
Thursday, March 9, 2017, 10:00 a.m.
Karen Ristic

DNA: Which Test is for Me?

Part 2: mtDNA: Is This Test for Me?



Part 1
February

Y-DNA:



Part 2
March

MtDNA:



Part 3
April

AtDNA:





DNA: Which Test is for Me?

**1. What Are Your
DNA Testing Goals?**

**2. What is
Mitochondrial DNA
(mtDNA)?**

**3. How is mtDNA
Inherited?**

**4. What Does
mtDNA Test For?**

**5. What Are My
mtDNA Ancestral
Origins?**

**6. How Do I Find
my mtDNA
Matches?**

**7. Any mtDNA
Limitations?**



1. What Are Your DNA Testing Goals?

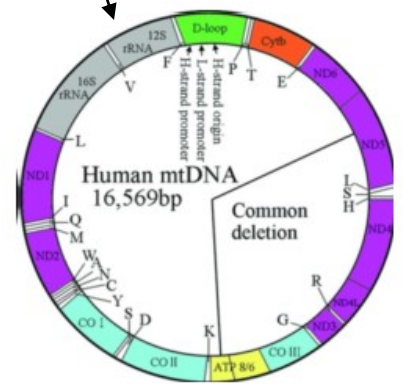
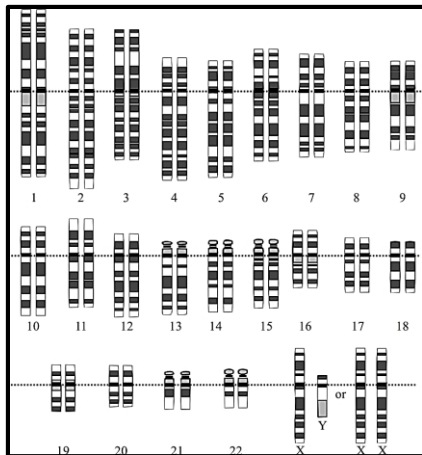
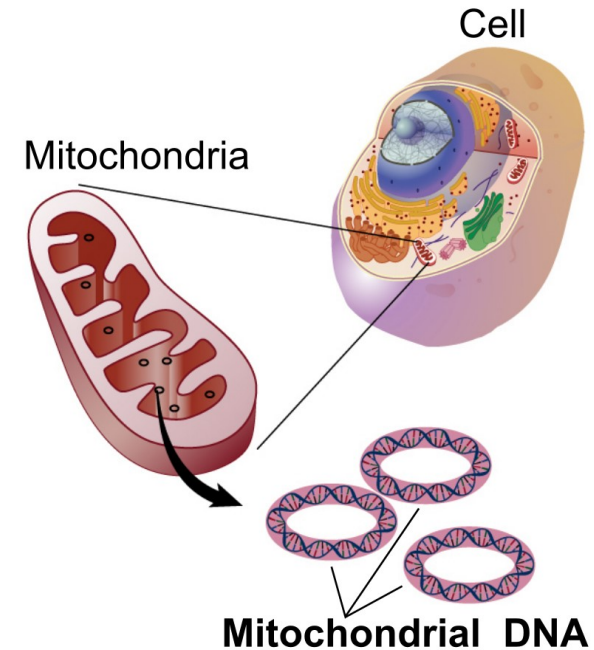
Interest in DNA testing for genealogy has reached an all-time high thanks to its increasing sophistication and the resulting visibility in the media. As a result, many family history enthusiasts have expressed their desire to venture into the fascinating world of genetic genealogy, but **don't know where to start**.

The first step is to determine your testing goals. Do you have a general curiosity about genetic genealogy or is your focus more specific? For example, please consider the following questions:

- **Are you primarily interested in researching your surname?**
- **Are there specific brick walls that you wish to target with the use of DNA testing?**
- **How far back in your family tree are these brick walls?**
- **What is the ancestral pattern back to these brick walls, i.e. mother's mother's mother or father's mother's mother's father?**
- **Are you ready for a long-term project or do you desire quick answers?**
- **Are there adoptions in your family tree that you would like to explore?**
- **Is your primary interest receiving a percentage breakdown of your overall ancestral origins or "ethnicity"?**

2. What is Mitochondrial DNA (mtDNA)?

- MtDNA are tiny energy “factories” located inside almost every cell in the body. You have hundreds or thousands of mtDNA in each cell and each one contains hundreds of copies of mtDNA. There is a lot of mtDNA in every cell.
- MtDNA is a small circular piece of DNA made up of a chain of approximately 16,569 pairs of special molecules called nucleotides.
- Although mtDNA is an unbroken loop on DNA, scientists and testing companies have given portions of the loop different names based on the DNA found within those portions.





A Detailed Example of the mtDNA Chromosome

A Video

Search Glossary

M

Marker

Meiosis

Mendel, Johann (Gregor)

Mendelian Inheritance

Messenger RNA (mRNA)

Metagenomics

Metaphase

Microarray Technology

Microbiome

Microsatellite

Missense Mutation

Mitochondria

Mitochondrial DNA

Mitosis

Monosomy

Mouse Model

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Search Glossary


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Mitochondrial DNA

Pronunciation


Mitochondrial DNA is the small circular chromosome found inside mitochondria. The mitochondria are organelles found in cells that are the sites of energy production. The mitochondria, and thus mitochondrial DNA, are passed from mother to offspring.



View Illustration




View Animation



William Gahl, M.D., Ph.D.

Dr. Gahl studies rare inborn errors of metabolism through the observation and treatment of patients in the clinic, and through biochemical, molecular biological and cell biological investigations in the laboratory. His group focuses on a number of disorders, including cystinosis, Hermansky-Pudlak syndrome, alkaptonuria and sialic acid diseases. Dr. Gahl has a long-standing research interest in cystinosis, a lysosomal storage disorder caused by a mutation in the CTNS gene. Over the past two decades, Dr. Gahl's laboratory has elucidated the pathogenesis of this disease and demonstrated the safety and efficacy of cysteamine (2-mercaptoethylamine) therapy, a treatment that depletes cells of cystine.



Listen to this genetics professional define this term

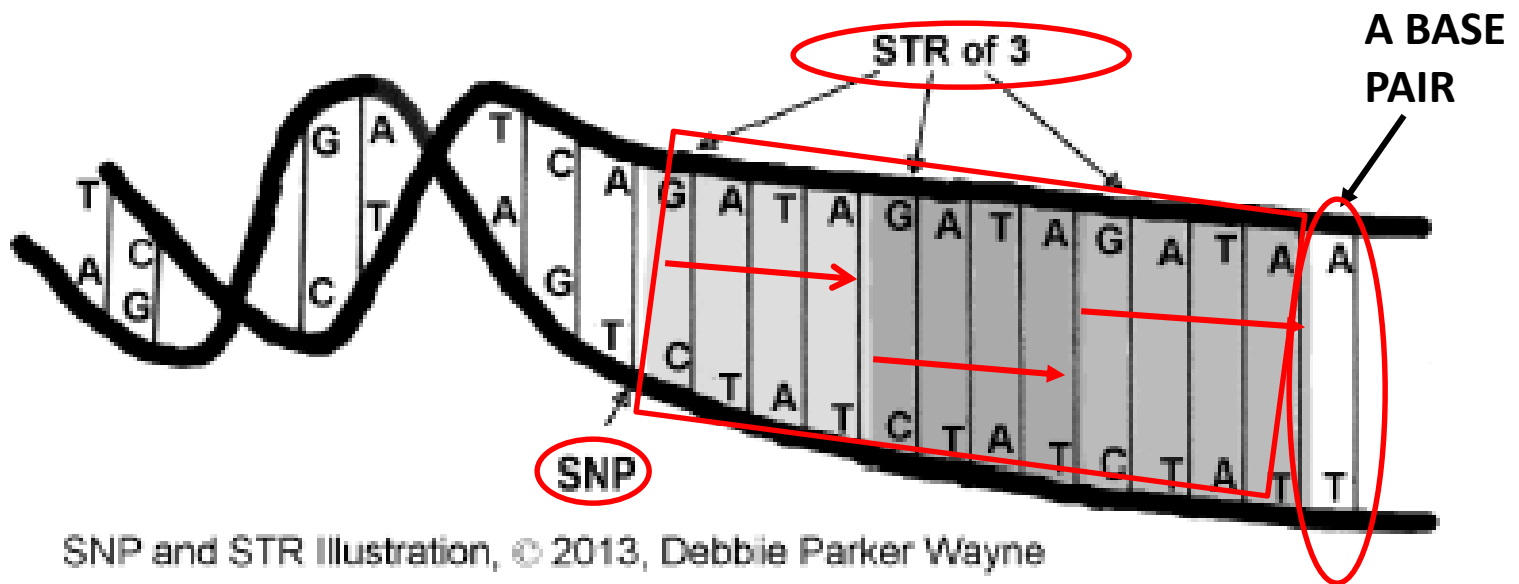
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02/09/2017

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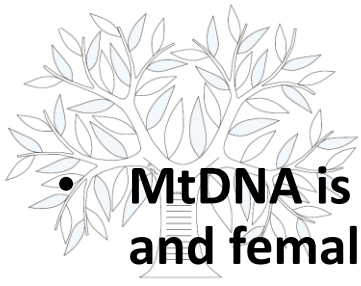


Both mtDNA and YDNA Testing Consists of Multiple Parts

- **The First Part of an mtDNA test results** depends on whether an **SNP** test is performed. When a chromosome is uncoiled it resembles a **ladder** as below. Each rung of the ladder is called a **base pair**. When the chemical at an individual ladder rung changes or mutates it is called a **single nucleotide polymorphism (SNP)**, pronounced **snip**). **SNPs may be referred to as markers** and each of these locations has a name assigned by the scientific community. This is the first part of mtDNA chromosome testing.



SNP and STR Illustration, © 2013, Debbie Parker Wayne

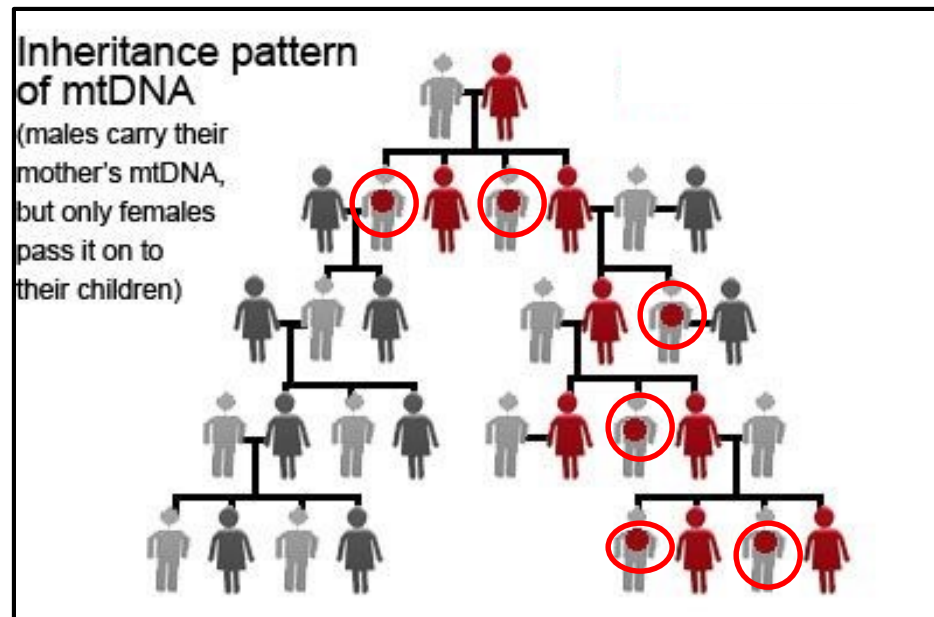


3. How is mtDNA Inherited?

- **MtDNA is always passed down from a mother to her children-both male and female.** The mother makes exact copies of her mtDNA and passes them down in her egg.
- **The image on the next page shows the path of mtDNA inheritance within a short family tree.** The bottom right level females would like to determine from whom in their family tree they inherited that particular piece of DNA. At every generation only one ancestor of hers carried the mtDNA due to this inheritance pattern. **The bottom females will know exactly which ancestor passed down her mtDNA even though she may not know that ancestor's name.**
- To find a living descendant of an ancestor who can take an mtDNA test, a genealogist must trace the mtDNA line through the generations that separates the deceased ancestors and the living descendants.
- **There is no limit as to how many generations back a genealogist can go to find an mtDNA relative, although the difficulty of researching the maternal line can be a barrier, as the surname usually changes with every generation.**

3. How Is mtDNA Inherited?

- Although mothers pass down mtDNA to both sons and daughters, only daughters will pass it on to the next generation. While every man has mtDNA he inherited from his mother and can be tested, that mtDNA ends with him. He does not pass it on to the next generation; some have labeled him a “carrier” of mtDNA.
- **For mtDNA, a male is a dead end in the testing line, although they should never be overlooked as a possible testing source.** A male may be the last living person available to take an mtDNA test for a specific ancestor.





4. What Does mtDNA Test For?

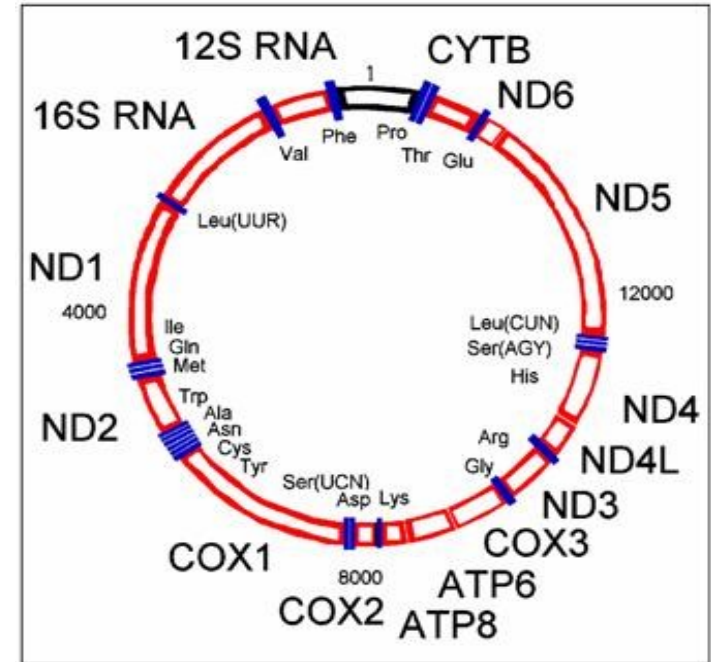
- There are two types of mtDNA tests:
 - 1) mtDNA Sequencing and
 - 2) SNP (Single Nucleotide Polymorphism) testing.
- **1) mtDNA Sequencing Testing** tests all 16,569 total base pairs. They examine short sections of mtDNA called Hypervariable Region 1 (HVR1) and Region 2 (HVR2). This test is a basic test and is useful for **analyzing ancestry and ancient origin information**
- Once the mtDNA is tested by one of these two methods, it is compared to a reference mtDNA sequence and identified.



mtDNA Sequencing Test

4.1 What Does mtDNA Test For?

- **2) The second test is SNP (Single Nucleotide Polymorphism) Testing.** This testing is a more detailed look and it looks at hundreds or thousands of locations along the circular mtDNA. **People who are very closely related should have the same SNP at every location.**
- Family Tree DNA is the primary mtDNA testing company, having tested the mtDNA of several hundred thousand customers.
- 23andMe also tests mtDNA, although it uses SNP sequencing.
- Once the mtDNA is tested by one of these two methods, it is compared to a reference mtDNA sequence and identified.



SNP (Single Nucleotide Polymorphism) Test.

4. What Does mtDNA Test For?

SNP (Single Nucleotide Polymorphism)

- A SNP is a specific location in the total DNA heredity (genome) where individuals differ in their DNA.
- As an example, below are two small sequences of DNA on one chromosome copy. **The SNP is the position where the two sequences differ and is highlighted in red.**
- SNPs are also referred to a “markers” because they refer to a specific location on the chromosome.
- The testing companies generally test over 700,000 SNPs across your genome.

↓

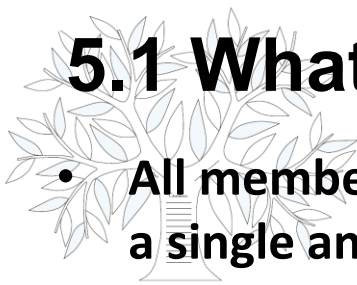
AGGAG**C**CTAT
AGGAG**A**CTAT

<p>mtDNA Plus</p> <p>ONLY \$79^{USD}</p> <p>✓ GENEALOGY</p> <ul style="list-style-type: none">• Examines two regions of mitochondrial DNA (HVR1 and HVR2)• Identifies basic haplogroup and migration paths <p>ORDER NOW</p>	<p>REFINED RESULTS</p> <p>mtFull Sequence</p> <p>ONLY \$199^{USD}</p> <p>✓ GENEALOGY + HISTORY</p> <ul style="list-style-type: none">• Examines all regions of mitochondrial DNA• Identifies basic haplogroup and migration paths• More refined results for genealogical purposes <p>ORDER NOW</p>
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5. What Are My mtDNA Ethnic Ancestral Origins

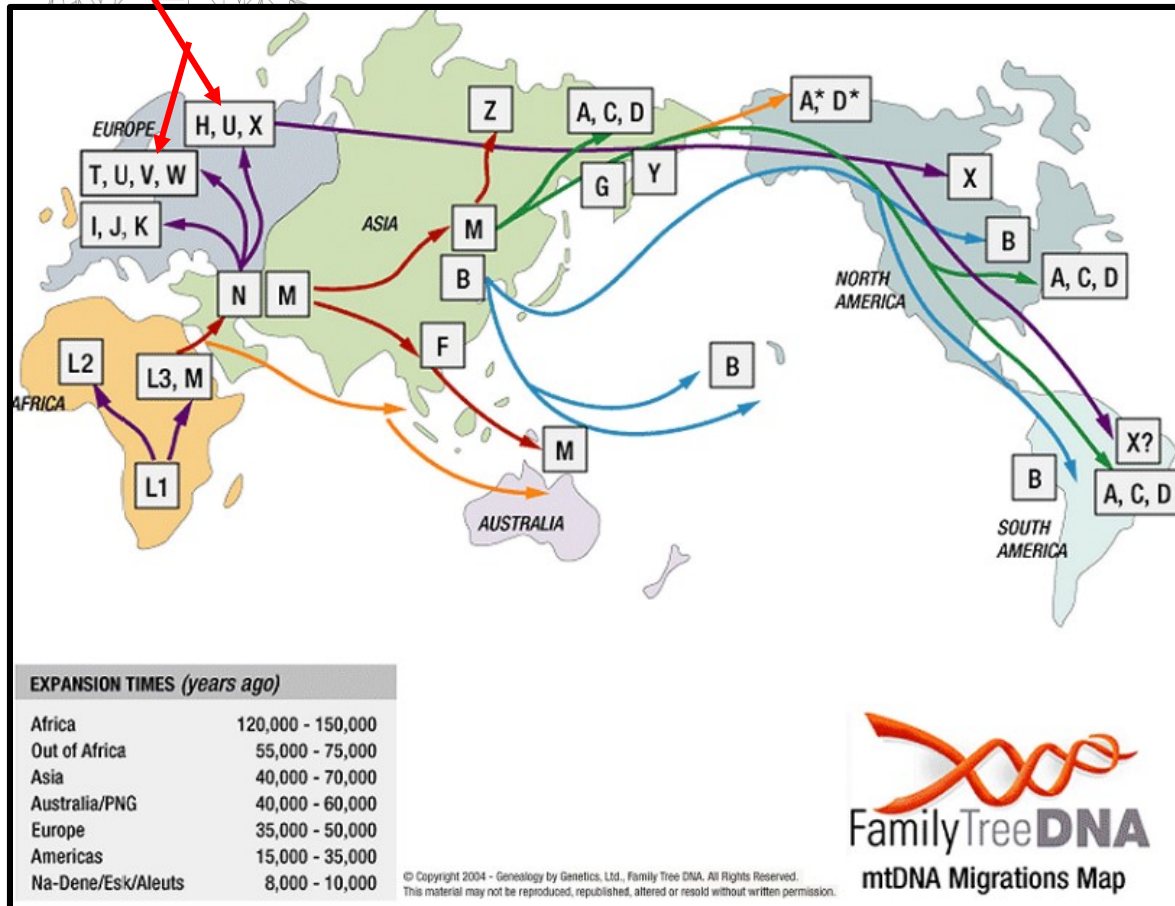
- MtDNA test results can be used to determine:
 - 1) the ancient origins of your mtDNA ancestors (haplogroup),
 - 2) whether or not two people are related on their maternal line, and
 - 3) estimate the length of time since the two tested individuals shared a **Most Recent Common Ancestor (MRCA)**.
- Regardless of the type of mtDNA test, results will reveal information about the location of your maternal line thousands of years ago: European, Asian, or Native American through the identification of a **haplogroup**
- A **haplogroup** is a group of related mtDNA results which share a common ancestor in a common place (usually several thousands of years ago). Haplogroups are named by letters of the alphabet and people in the same haplogroup will have the same or very similar list of mutations.



5.1 What Are My mtDNA Ethnic Ancestral Origins

- **All members of an mtDNA haplogroup can trace their maternal line back to a single ancestor who lived in a specific location several thousand years ago.** In most cases scientists have a good idea of the general location where mtDNA haplogroup ancestors lived. If all mtDNA sequences on earth were plotted onto a giant family tree, they would all trace back to **Mitochondrial Eve**. Each branch whether major or minor, is defined by a particular SNP mutation.
- **Each haplogroup is associated with an approximate time and place in which the founder of that haplogroup arose.** This information is based on mutation rates and modern-day distributions of the haplogroup, not on ancient samples of mtDNA although ancient DNA is being used to further study and refine information about various haplogroups.
- **Once test-takers receive their haplogroup assignment, they can seek out more information about that haplogroup and ancient origins.** For example, the mtDNA Haplogroups page at WorldFamilies (www.worldfamilies.net/mtdnahaplogroups) is a great resource with information about each of the major branches of the mtDNA family tree.

5.2 mtDNA Haplogroup Migrations Map










Haplogroups have to do with deep ancestry going back thousands of years. They tend to be associated with a geographic location. When there are mutations in the mtDNA chromosome, it leaves a signature, which is designated by letter and number combinations. With all genetic genealogy research, locations are important. This is true whether a few decades ago, or hundreds or thousands of years in the past. **With a mtDNA test you will see a visual representation of this journey, as shown here.**

6. Family Tree mtDNA Ancestor Matches

At Family Tree DNA a test-taker's mtDNA is compared to all other mtDNA in the database and the test-taker will receive a list of anyone in the database who has identical or nearly identical mtDNA. Some may have identical mtDNA, while others might differ by one or two mutations. **Generally, the fewer the differences between the two sequences, the more closely those two individuals are related.**

For example in the image below, six test-takers in the Family Tree DNA database have mtDNA similar to the test-taker's mtDNA. However, all of these individuals have a genetic distance of 1 or more, meaning the two mtDNA sequences are not identical.

HVR1, HVR2, CODING REGIONS - 6 MATCHES				
Genetic Distance	Name			mtDNA Haplogroup
1	Riley Gibson	 	FMS FF	A2w
1	John Johnson	 	FMS FF	A2w
1	Mary Roberts	 	FMS FF	A2w
2	Gwen Matthews	 	FMS FF	A2w
2	Ian Philips	  	FMS FF	A2w
3	Hiram Culpepper	 	FMS	A2w



6. Family Tree mtDNA Ancestor Matches

- In addition to the list of mtDNA matches provided by the company, a test-taker can also search MitoSearch (www.mitosearch.org) for others who share their mtDNA. MitoSearch is a free publicly available database with thousands of records from several different testing companies. Test-takers can upload their mtDNA to MitoSearch to look for matches that are already in the database, as well as compare their results to new matches that are uploaded to MitoSearch.
- Matching on mtDNA Plus (HVR1 and HVR2) means that you have a 50% chance of sharing a common maternal ancestor within the last 28 generations. That is about 700 years. Matching on the Mitochondrial DNA Full Sequence test brings your matches into more recent times. It means that you have a 50% chance of sharing a common maternal ancestor within the last 5 generations. That is about 125 years.
- If you are interested in identifying your common ancestor, you should contact your closest matches and ask them if they are interested in sharing information with you. If they are, you can review their family tree to determine whether their maternal line shares any names or locations in common with your maternal line. Sometimes your matches will list their most distant maternal ancestor, which you might be able to use to 'reverse engineer' their maternal line if they aren't interested in sharing information. For example, my most distant maternal ancestor is an Ellen Connor, born about 1789 in Grand Cayman. How far back have you traced the ancestors in your mtDNA line?



7. Some mtDNA Limitations

- **MtDNA testing can only determine whether two people are maternally related on their *direct* matrilineal line.** Further, an mtDNA test can only reveal that two people are maternally related somehow, but **it can't determine the exact nature of the relationship.** As a result, people with matching mtDNA might be sisters, mother/daughter, aunt/niece, first cousins and so on for many generations.
- These limitations must be contrasted with some of the powerful benefits of mtDNA testing. **Unlike atDNA, for example, mtDNA passes down to the next generation unchanged and therefore does not get diluted like atDNA.** A test-taker has 100% of the mtDNA of her mother's mother's mother's mother (her great-great-grandmother), but just approximately 6.25 % of her atDNA. Accordingly, even with its limitations, **mtDNA can be a powerful tool for genealogists.**
- **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 accounts.**











FAQ at FamilyTree DNA ///// Costs at FamilyTree DNA

<https://www.familytreedna.com/learn/faq/>

Some of the topics
FamilyTreeDNA answers for
you:

- Before I Buy
- Shipping
- Billing and Refunds
- After I Swab
- Website Questions
- Privacy

	Best for	Specifications	What you get	Price
FAMILY FINDER	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	Autosomal	<ul style="list-style-type: none">Family Finder MatchesEthnic Percentages	<div>\$79</div> Learn More
	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	Autosomal + 37 markers	Male specific bundle includes Family Finder plus our Y-chromosome test that analyzes 37 markers.	<div>\$248</div> <div>Males only</div>
	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	Autosomal + 67 markers	Male specific bundle with all the bells and whistles! Family Finder plus our 67 marker Y-DNA test.	<div>\$347</div> <div>Males only</div>
	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	Autosomal + mtFull Sequence	Family Finder plus a Full Mitochondrial Sequence - For both males and females, our mtDNA test traces your maternal line.	<div>\$278</div>
Father's Line Learn More				
	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	<div>37</div> markers	<ul style="list-style-type: none">Connect to matchesFree access to group projects & experts on your lineageAutomated updates to your resultsFree webinars with a professional genetic genealogistPersonalized customer supportUncover up to 340,000 years	<div>\$169</div> <div>Males only</div>
	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	<div>67</div> markers		<div>\$268</div> <div>Males only</div>
	<div>Genealogy</div> <div>History</div> <div>Ancestry</div>	<div>111</div> markers		<div>\$359</div> <div>Males only</div>
Mother's Line Learn More				
	<div>History</div>	HVR1+HVR2	<ul style="list-style-type: none">Connect to matchesFree access to group projects & experts on your lineageAutomated updates to your resultsFree webinars with a professional genetic genealogistPersonalized customer supportDiscover up to 180,000 years	<div>\$79</div>
	<div>Genealogy</div> <div>History</div>	Full Sequence		<div>\$199</div>



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ATTACHMENT 1: Family Finder, 23andMe, AncestryDNA, and MyHeritage Comparison Chart

from Tim Janzen, MD and Blaine Bettenger (as of October, 2016)

	23andMe	Family Tree DNA's Family Finder test	Ancestry.com's AncestryDNA test	MyHeritage
Website	www.23andme.com	www.familytreedna.com	http://dna.ancestry.com	www.myheritage.com
Price	\$199 with many more tests offered	\$99 =AtDNA test; \$169 Y-DNA test; \$199 for mT-DNA test	\$89 in the U.S. for At-DNA test	Levels go from free to \$119.40 per year; \$79 for At-DNA test
International product availability	56 countries	Worldwide	35 countries	48 countries
Method for collecting the DNA sample	saliva sample (about 1 cc)	cheek swab	saliva sample (about 1/2 cc)	cheek swab
Means of contacting people who share matches	Contact may be made after seeing your list of matches in DNA Relatives; the matches must be willing to share ancestry reports with you or have opted in to open sharing if you are to see what you share with your matches	email addresses of all matches are available	Contact can be made through Ancestry.com's messaging system	Messaging system
Average responsiveness of matches	Low	Medium	Medium	Unknown
Average level of genealogical knowledge of matches	Fairly low	Medium	Medium	Unknown
GEDCOM file upload allowed	No	Yes	Link is created to Ancestry.com pedigree charts	Yes



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