Using Y-DNA and mtDNA to Explore Your Ancestry

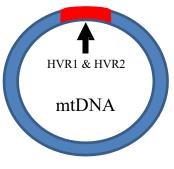
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Introduction to Mitochondrial DNA

Mitochondrial DNA (mtDNA) is a small circle of DNA that is located inside our cells. Most human cell contain hundreds or even thousands of copies of mtDNA, which is 16,569 base pairs long.

Inheritance of mtDNA

Mitochondrial DNA has a very unique inheritance pattern from one generation to the next, which makes it a great tool for genetic genealogists.



Only your mother gave mtDNA to you; your father's mtDNA was not passed down to the next generation. While male children will inherit their mother's mtDNA, they will not pass it down to their own children. This unique feature of mtDNA allows it to be used for tracing matrilineage, the inheritance of mtDNA from mother to child.

Types of mtDNA Testing

There are two types of mtDNA tests. The first type is *mtDNA sequencing*, and is performed by sequencing all or a portion of mtDNA. Most testing sequences the entire mtDNA genome. The second type of mtDNA testing, called *SNP testing*, examines single nucleotide polymorphisms ("SNPs") – or variable nucleotides (A, T, C, and G) – at many different locations along the circular mtDNA. This test is good for learning about ancient ancestry, but not quite as good at determining family relationships.

Once the mtDNA is sequenced by one of the methods above, it is compared to a reference mtDNA sequence (either the *Reconstructed Sapiens Reference Sequence* (RSRS) or the *Cambridge Reference Sequence* (CRS)). Any differences between the mtDNA sequence and the reference sequence are listed as "mutations," or changes, like this:

Haplogroup	Mutations					
Т	G709A, G1888A, A4917G, G8697A, T10463C, G13368A, G14905A, A15607G, G15928A, C16294T					

Using mtDNA Test Results

The results of an mtDNA test can be used to determine the test-taker's mtDNA *haplogroup* and ancient origins, to determine whether two people are maternally related, and if so, to estimate very roughly the amount of time since two individuals shared a most recent common ancestor (MRCA).

1. Learn About Your Ancient Ancestry

The results of mtDNA testing provide a haplogroup determination. 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.

2. Find Your mtDNA Cousins

If you test at <u>Family Tree DNA</u>, you will receive a list of people in the database that are close matches with your mtDNA sequence. These individuals are your genetic cousins and related to you through your maternal line. Some may match exactly, while others might be different from you by one or two mutations. Generally, the more mutations you share in common, the more closely related you are. However, because mtDNA mutates so slowly, you could be related very recently or several thousand years ago.

Matching Level	Generations to Common Ancestor (with <i>exact</i> match)				
	50% Confidence	95% Confidence			
HVR1 & HVR2	50-percent chance of common ancestor within about seventy generations (1,700 years)				
HVR1,HVR2,&CodingRegion	95-percent chance of common ancestor within about thirty generations (775 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.

3. Solving Family Mysteries

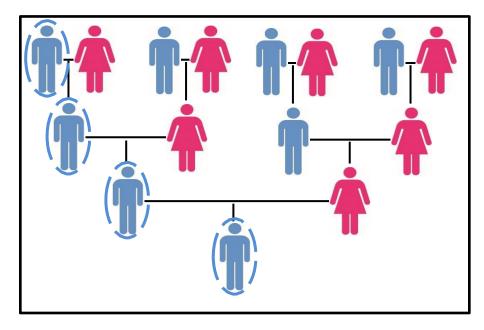
Another powerful use for mtDNA testing is to examine family mysteries and brick walls. Since mtDNA is inherited maternally, it is very good at determining whether two people are related through their maternal lines.

Introduction to Y-DNA

The Y chromosome is only found in males, who have one Y-chromosome (from his father) and one X-chromosome (from his mother). This XY pair is one of the 23 pairs of human chromosomes.

1. Inheritance Patterns of Y-DNA

The Y chromosome has a unique inheritance pattern, just like mtDNA. It is passed down from father to son without change. Over long periods of time the chromosome begins to accumulate mutations that are typically silent and have no impact on the carrier. These mutations, however, are useful for genealogical purposes – they can be used to analyze the relationships between populations and individuals. The figure below demonstrates the path that Y-DNA took from a greatgrandfather to his great-grandson:



a. Some Advantages & Limitation of Y-DNA

Important Advantages:

- Powerful tool due to unique inheritance pattern (compare to atDNA);
- Reveals ancient heritage and genetic relatives (can break through paternal brick walls).

Important Limitations:

- Can identify NPEs;
- Close paternal family members have the SAME haplotype (with rare exceptions) and are thus indistinguishable with Y-DNA alone;
- Lines daughter out.

b. Types of Y-DNA Tests

There are two types of Y-DNA testing for genealogy: *STR testing* and *SNP testing*. The STR, or "short tandem repeat" test, sequences between 12 and 111 (and sometimes more) very short segments of DNA located throughout the Y-DNA. The single nucleotide polymorphism ("SNP") test examines between a handful and hundreds of single spots throughout the Y chromosome.

i. STR Testing

Most Y chromosome tests examine between 12 and 111 STR markers, but many more are regularly being identified and used for testing. STRs are identified by their DYS number (DNA Y-chromosome Segment number), and are measured by the number of repeats of a particular DNA sequence at that location. This number of repeats can change over time at a relatively regular rate, thereby giving genealogists the ability to trace paternal lineages over time.

The results of STR testing are usually presented in a format that looks like this, with a series of STR markers and the STR results, as in the following example:

DYS#	393	390	19	391	385a	385b	426	388	439
Alleles	13	24	14	10	11	14	12	12	12

The "DYS" row are marker locations on the Y-chromosome, and the "allele" row are the number of repeats for each DYS marker location. At each of these DYS locations, there is a potential for variation of the "allele." For example, at DYS426, the variation consists of 7 to

18 repeats of the DNA sequence "**GTT**," with 12 repeats being the most common. The sequence of 12 repeats of "**GTT**" would look like this, with the 12 repeats in bold:

and with numbering to emphasize the 12 repeats:

In contrast, someone with a result of "7" at DYS426 would have the following sequence of seven "**GTT**" repeats:

...TGT**GTTGTTGTTGTTGTTGTTGTT**GAC...

Together, the group of STR markers and your allele results represent your *haplotype*, which is simply a group of DNA variations that tend to be inherited together.

When the Y-chromosome is duplicated and passed down to a son, it can accumulate an error. This typically results from mistakes introduced by the replication machinery in the nucleus of the cell, and the repetitive nature of the STRs can make them more prone to errors. Sometimes, for example, an extra repeat (or two, or more) will be introduced, and sometimes a repeat (or two, or more) will be removed:

Original (dad):	TGT GTTGTTGTTGTTGTTGTTGTT GAC
Mutated (son #1)	
	TGT GTTGTTGTTGTTGTTGTTGTTGTT GAC
Mutated (son #	1)TGT GTTGTTGTTGTTGTTGTT GAC

At this marker, each son is one "step" way from the father, although they are a step away in opposite directions.

Together, the group of STR markers and your allele results represent your *haplotype*, which is simply a group of DNA variations that tend to be inherited together.

2. SNP Testing

SNP tests examine anywhere between one and thousands of single nucleotide polymorphisms located all along the Y chromosome. SNPs are traditionally used to determine a person's haplogroup and ancient ancestry, and have been less useful for finding genetic cousins. However, new tests are identifying SNPs that may be useful on a genealogically relevant timeframe. SNP testing has several important uses, including: (i) determining deep ancestry;

(ii) confirming an estimated haplogroup; and (iii) determining a subhaplogroup designation (a "terminal SNP"). And soon, many believe that SNP testing will be used in the determination of relationships in a genealogically relevant timeframe!

A SNP is either "**ancestral**," meaning the original value of the SNP, or it is "**derived**,"

meaning that it has mutated from the original value. For example

Ancestral: CTACGTCAGGTTACGATTGC (denoted by "-") Derived: CTACGTCACGTTACGATTGC (denoted by "+")

Your testing company will usually interpret the results of a SNP test for you by placing you within the proper haplogroup or sub-clade. Use caution when analyzing SNPs, as sometimes SNPs can have different names or companies will use different SNPs to test the same thing.

a. Using Y-DNA Test Results

The results of Y-DNA test can be used to determine a person's Y-DNA *haplogroup* and ancient origins, to determine whether two people are paternally related, and if so, estimate the amount of time in which two individuals shared a most recent common ancestor (MRCA) on their direct paternal lines.

i. Ancient Ancestry

A Y-DNA *haplogroup* is a group of related Y-DNA profiles that share a common ancestor in a common place (usually several thousands of years ago). Y-DNA haplogroups are named by letters of the alphabet, and people in the same haplogroup will have the same, or very similar, list of mutations. In this table, for example, the test-taker belongs to haplogroup R1b based on an estimate of his Y-DNA:

DYS#	393	390	19	391	385a	385b	426	388	439
Allele	13	24	14	10	11	14	12	12	12
Estimated Haplogroup is R1b1b									

ii. Find Y-DNA Cousins

You will receive with your test a list of people in the database that are close matches with your Y-DNA sequence. These individuals are your genetic cousins and related to you through your paternal line, either closely or distantly. Some may match exactly, while others might be different from you by a handful of mutations. The more mutations you share in common, the more closely related you are. For example, the following chart shows the relationship between the number of markers, genetic distance, and the number of generations to the most recent common ancestor:

Number of matching STR markers	Probability that the MRCA was not more than this number of generations ago					
	50%	95%				
35 of 37	6	14				
36 of 37	4	10				
37 of 37	2	7				
110 of 111	2	7				
111 of 111	1	5				

iii. Join Y-DNA Projects

A Y-DNA project is a collaborative effort to answer genealogical questions using the results of Y-DNA testing. A surname project brings together individuals with the same (or 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.

iv. Solve Family Mysteries

Y-DNA is a great tool for examining brick walls and black sheep. Since Y-DNA is only inherited paternally, it is very good at determining whether two people are related through their paternal lines.