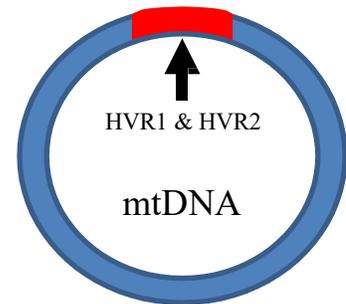


# Using Y-DNA and mtDNA to Explore Your Ancestry

Blaine T. Bettinger, Ph.D., J.D.  
[www.DNA-Central.com](http://www.DNA-Central.com)  
blainebettinger@gmail.com

## Introduction to Mitochondrial DNA

Mitochondrial DNA (mtDNA) is a small circle of DNA that is located inside our cells. Most human cells 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
T	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 [Family Tree DNA](#), 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, & Coding Region	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.





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):     ...TGTGTTGTTGTTGTTGTTGTTGTTGAC...  
Mutated (son #1)     ...TGTGTTGTTGTTGTTGTTGTTGTT**GTT**GAC  
... Mutated (son #1)     ...TGTGTTGTTGTTGTTGTTGTTGAC...

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:     CTACGTCA**G**GTTACGATTGC (denoted by “-”)  
Derived:       CTACGTCA**C**GTTACGATTGC (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:

<b>DYS#</b>	393	390	19	391	385a	385b	426	388	439
<b>Allele</b>	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.