



Understanding Your mtDNA Results

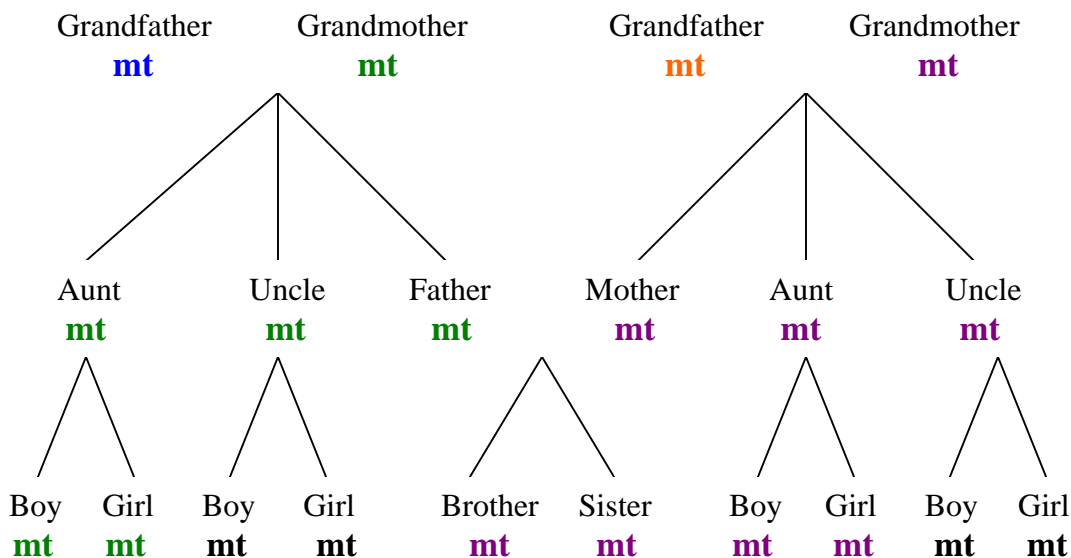
My FTDNA Code: _____

Enclosed you will find a certificate showing that you have had your mitochondrial DNA analyzed by **Family Tree DNA**. The results represent your actual DNA sequence's differences from the Cambridge Reference Sequence (CRS) and are listed on your certificate. These values are also listed on your personal page at our website. To access your personal page, please log in from the MY FTDNA box at the right side of our home page, www.familytreedna.com, with your kit number and password provided at the top of this page.

Human mitochondrial DNA (mtDNA) has distinct properties that make it an invaluable tool for genealogical and anthropological study. For example, mtDNA is inherited only from the mother. As a result, the study of mtDNA is essentially the study of female genetic lines within human populations. Mutations, when they occur, are passed down to the children. As more mutations occur over time, they accumulate in a linear or chronological manner, allowing scientists to study these changes. Using proper documentation and historical data, these mutations can also be associated with geographic areas and populations. With this information, researchers have constructed ancient migration patterns based on the presence of these mutations in human populations.

Mitochondria are present in all human cells and contain their own DNA. Both males and females have this mtDNA, but only females pass it on to their offspring. Therefore, mtDNA is passed from mother to daughter along the female line without any influence from fathers.

The chart below illustrates how mtDNA is inherited from each grandparent. The two entries in black represent mtDNA from outside the maternal lines displayed here—in this case, the uncle's spouse.





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MtDNA analysis is performed by looking for both similarities and differences among individuals. We test 569 base pairs, including the entire HVR-1 (HyperVariable Region 1) area, and use them for genealogical and genetic analysis. A base pair is a specific component of the DNA and is made of adenine (A) and guanine (G) or cytosine (C) and thymine (T). Therefore, our report will express your results as a series of letters representing the bases specifically found in your HVR-1 region. The HVR-1 area is used because mutations occur more frequently in this region and therefore they can differentiate specific lineages more easily.

MtDNA results are commonly compared to the CRS, the industry standard which was sequenced in 1981. Any place in your mtDNA where you have a difference from the CRS is characterized as a mutation. If your results show no mutations at all it means you match the CRS. A mutation happens when (a) one base is replaced by a different base, (b) a base is inserted between two bases without replacing any, or (c) a base is removed from a position without being replaced. We provide you with more details and examples on page 3.

Your analysis highlights these differences and may be compared to other individuals' results. Our database is helpful in finding other individuals with exactly the same mtDNA. This exact duplication of the mtDNA means two individuals shared a common female ancestor. Research over the last decade suggests that all the maternal lines ultimately originate from "Mitochondrial Eve" approximately 140,000 years ago in Africa. Further details may be reviewed in the journal *Nature Genetics*, November 2000; in *Science*, November 2000 or in our library at www.familytreedna.com/mtdnapapers.html.

We compare your specific sequence to other people in our database of mtDNA results. If we find someone who has exactly the same changes within their sequence as you do and you have signed the **Family Tree DNA** release form, we will inform both of you by email. In this way we are able to link you up with your "genetic cousins." However, despite the fact that the HVR-1 region experiences mutations more frequently, the mutations do not occur often enough for us to determine whether two individuals are more closely or more distantly related. As a result, we cannot tell whether you share a common ancestor with these individuals in a genealogical time frame or further back in time. Testing the HVR2 region helps to pick out the individuals with whom you most likely share a common ancestor in a genealogical time frame. You may then employ conventional genealogical techniques to see whether you can find where that person might actually fit on your family tree!

Another feature offered exclusively to our customers is our Recent Ancestral Origins (RAO) database. The database, accessible from your personal page, requires the sample code listed on your certificate and the password at the top of page one. This code should not be shared with others. It will also allow you to order additional tests or refinements using your DNA.

Mitosearch is a public database we have created to aid genealogists who want to compare their results with other individuals worldwide, regardless of the company they tested with. Now that you have your results you may wish to add them to mitosearch at www.mitosearch.org. You can also upload your GEDCOM file to www.mitosearch.org.

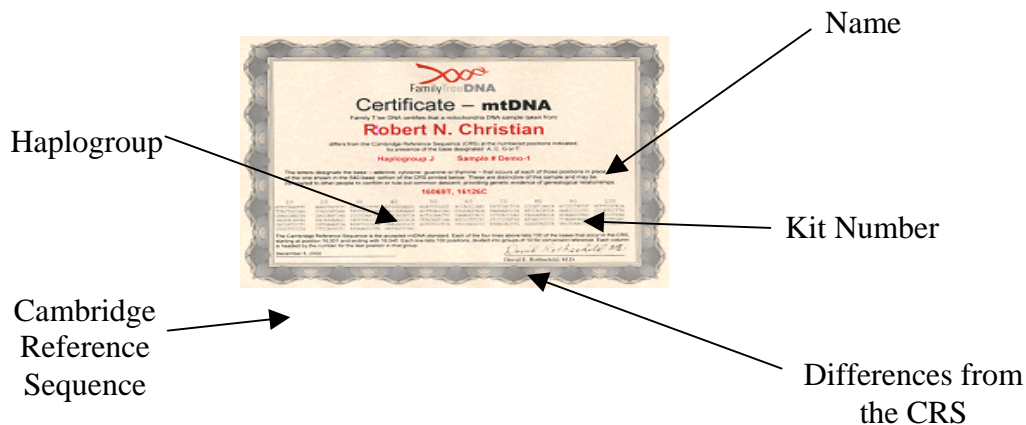
How to read your mtDNA certificate:

Your mutations from the CRS are displayed on your certificate. Below your individual mutations you will notice six rows with ten columns each. This is the HVR-1 CRS, with which all samples are compared. Begin reading at the upper left and continue to the end at the lower right.

Where you have numbers under your name, this indicates that you have a mutation from the CRS at that particular point. The letters represent the new code found that that place in the sequence. A “C” in position 16154, for example, means that at the 16154th base pair, you have a “C” in place of the “T” listed for that position in the CRS. A position like this that shows variation is called a polymorphism, or mutation.

In some cases you will see insertions in your mtDNA sequence. If you have an insertion after base pair 255, for example, the insertion will be listed as the base pair and .1C. In this case, a single base pair insertion has been found in your mtDNA string, noted by the .1. The nucleotide changed to cytosine (C) from guanine (G), therefore denoted with a C. The insertion then looks like this: 255.1C. If you have a 2 base pair insertion the results might look like this: 255.1C 255.2T

It is also possible that you have a deletion; a base pair was not copied and you just don't have a base pair at that particular place in your sequence. For example, “424 –” means that this location, 424, isn't in your sequence and is represented by a minus sign at the site where the nucleotide base should have been found.



Your haplogroup

Your haplogroup, or genetic population group based on your differences from the CRS, was determined through this testing. Haplogroups are defined by specific mutations which took place tens of thousands of years ago and have been passed down to all of today's populations. Your haplogroup identifies your deep ancestral ethnic and geographic origins on your maternal line. The world map we include with this certificate labels the general regions in which your haplogroup can be found. We also use arrows to indicate how the haplogroups are connected with one another. You can find a description of your haplogroup in the “mtDNA Results” section of your personal page on our website.



Useful terms to know:

Base pair: The DNA bases are always held together in pairs and attached to one of the strands in the DNA double helix. The order of bases is the sequence of DNA.

CRS (Cambridge Reference Sequence): The mitochondrion sequenced in 1981 became known as the Cambridge Reference Sequence (CRS) and has been used as a basis for comparison with your mtDNA. In other words, any place in your mtDNA where you have a difference from the CRS is characterized as a difference or mutation.

Deletion: A deletion takes place when a base pair is removed from the sequence.

DNA (deoxyribonucleic acid): Known by many as the structure of heredity, DNA is a chemical consisting of a sequence of hundreds of millions of nucleotides found in the nucleus of cells. It contains the genetic information about an individual and is shaped like a double-stranded helix.

Gene: the functional and physical unit of heredity passed from parent to offspring.

Haplogroup: branches in the human genetic tree (Phylogenetic tree). They are tied to deep ancestry (think 10,000 or 10s of 1000s of years).

HVR1 and HVR2 (HyperVariable Region 1 and 2): The two sections or regions of the mtDNA which are tested in the mtDNAPlus test. The results can be used to determine a person's ethnic and geographic origins, as well as to look for possible common ancestry with other individuals.

Insertion: An insertion takes place when a base pair is added to the sequence.

mtDNA (mitochondrial DNA): The genetic material found in mitochondria. mtDNA is passed down from females to both son and daughter, but sons do not pass down their mother's mtDNA.

Mutation: A heritable change that may occur in a gene or in a chromosome and may take the form of a chemical rearrangement or a partial loss or gain of genetic material.

Polymorphism: A mutation, or change, in the DNA sequence which is inheritable.

It has been a pleasure to serve you. If you have questions, please feel free to email us at info@familyreedna.com or to visit our website at www.familyreedna.com and read our ever-expanding Frequently Asked Questions page. For valuable education aids please go to www.familyreedna.com/dna101.html. Of course we encourage you to visit our web site from time to time to see what new genealogical opportunities we have developed for you.

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