Your mtDNA Full Sequence Results

My FTDNA Code: _____

Congratulations! You are one of the first to have your entire mitochondrial DNA (mtDNA) sequenced! Testing the full sequence has already become the standard practice used by researchers studying the mtDNA, and it is only a matter of time before it becomes the standard test for individuals wanting to use their mtDNA results for genealogical purposes.

For several years our clients have asked us if and how mtDNA can be used for genealogical purposes. Our best (and only) answer has been that the very slow mutation rate on the female-inherited mtDNA renders it very useful for anthropological purposes, but that this same fact has made mtDNA less useful in making solid genealogical comparisons.

Currently, scientific papers discussing mtDNA are being written based upon a scan of the entire mitochondrial sequence. This much deeper investigation of the mtDNA has shown many new mutations that will ultimately allow more precise lineages to be identified, including branches of haplogroups. The haplogroup displayed on your certificate reflects which branch we can identify based on research completed so far and your mtDNA sequence.

Because it now appears clear that most scientific papers in the future will use mtDNA full sequence tests we have decided to offer this test for genealogical and anthropological purposes ONLY. NO MEDICAL INFORMATION WILL BE DETERMINED OR DISCUSSED BY FAMILY TREE DNA FROM YOUR MTDNA FULL SEQUENCE TEST.

We offer this test for the following purposes only:

- To more accurately establish the actual mutation rate of mtDNA
- To apply statistics to better predict when two people who match likely shared a MRCA (Most Recent Common Ancestor)
- To provide the only (or final) mtDNA test that one will ever need to take
- To allow our most inquisitive clients to have results in hand when more advanced scientific papers are published in the future

Your Results: Enclosed you will find a certificate showing that you have had your mtDNA 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 and 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 the password provided on the first page of this report.

Your haplogroup, or genetic population group based on your specific ancient mutations, 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 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.

Research over the last decade regarding these mutations and haplogroups 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.

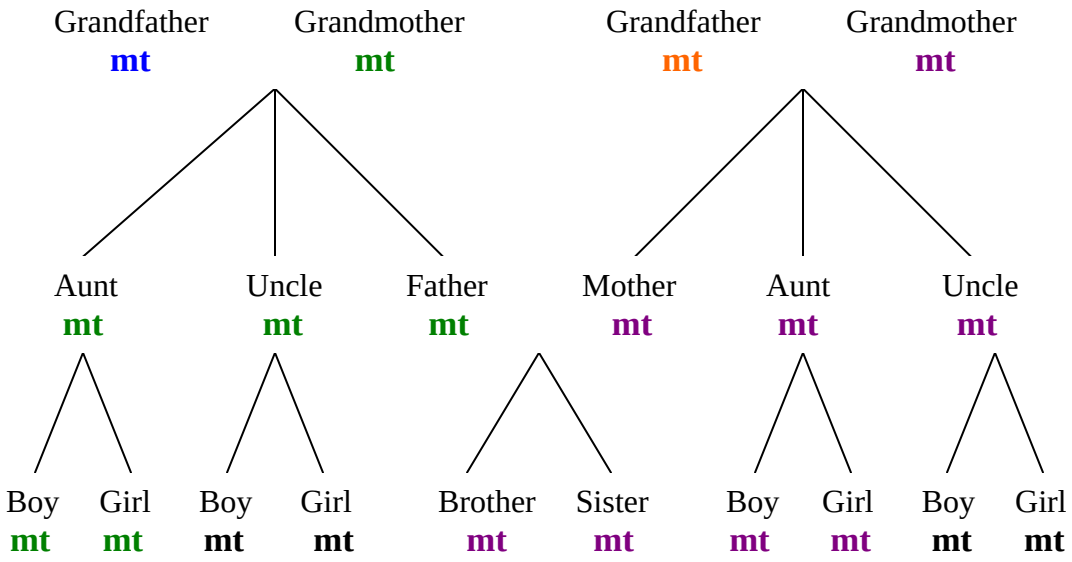
The table below indicates the likelihood that you share a common ancestor with people that you match on the full sequence if you have exactly the same results, one mutation different, etc. These data are based on what we know so far and will change as we learn more.

# differences between the results	50% probability that the MRCA lived no longer ago than this number of generations	75% probability that the MRCA was no longer than this number of generations	90% probability that the MRCA was no longer than this number of generations
0	5	9	16
1	11	18	25
2	18	26	35
3	24	34	44

Human 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 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.



MtDNA analysis is performed by looking for both similarities and differences among individuals. We test all of the 16,569 base pairs 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 mtDNA.

Mutations: 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 one base is replaced by a different base, when a base is inserted between two bases without replacing any, or when a base is removed from a position without being replaced.

These mutations are listed on your certificate. The letters represent the new code found at 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 most mutations, the base pair switches only with its partner. For example, A always partners with T, and G always partners with C. Mutations that switch in this manner (from A to T or G to C or vice versa) are indicated with a capital letter next to the number of the base pair where the mutation occurred.

You may 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 two 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 do not have a base pair at that particular place in your sequence. For example, "424 –" means that this location, 424, is not in your sequence and is represented by a minus sign at the site where the nucleotide base should have been found.

Your analysis highlights these differences and may be compared with other individuals' results. Because we are only just beginning to build our database of full sequence mtDNA test results, we only look for genetic matches in our database using the HVR1 and HVR2 sections offered through our basic mtDNA and our mtDNAPlus tests. This exact duplication of the mtDNA means two individuals shared a common female ancestor.

In the meantime, you may want to use the search available at the NCBI (National Center for Biotechnology Information) linked below. At this site, you can plug in sections of your full sequence to their "BLAST" search engine in order to search the research database for matches. This search is also linked from your personal page on our website. To access their BLAST search, go to http://www.ncbi.nlm.nih.gov/BLAST/ and click on the link which reads "Quickly search for highly similar sequences (megablast)."

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: The branches of 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.

MRCA: The most recent common ancestor shared between two individuals.

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

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

Polymorphism: A heritable mutation, or change, in the DNA sequence.

It has been a pleasure to serve you. If you have questions, please feel free to email us at info@familytreedna.com or to visit our website at www.familytreedna.com and read our ever-expanding Frequently Asked Questions page. For valuable education aids please go to www.familytreedna.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.