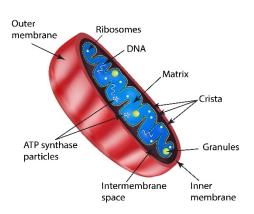


Mitochondrial DNA Testing

Mitochondrion



FamilyTreeDNA is the only commercial co mpany that tests the full mitochondrial genomic sequence with a matching database. Some companies offer a haplogroup with their autosomal test, but it may not be completely accurate since it is from a selection of mtDNA SNPs (markers) rather than the entire genome.

The haplogroup designation consists of a sequence of letters and numbers and corresponds with migratory paths over time, giving insight into the geographic origins of one's direct maternal line over centuries and even millennia.

DNA Matches

Your mtDNA values are compared against the other mtDNA testers in the database. When your values match another tester's values within the threshold, it means that you both share a common direct maternal ancestor at some point in time. We also report the Genetic Distance (number of mismatches) you have.

To be considered a match at the HVR1 level and the HVR2 level, you and another tester must be exact matches (0 mismatches). At the Coding Region level, which you get with the Full Sequence test, you may have a Genetic Distance of up to 3 (3 mismatches).

When a person's cells contain more than one base pair at the same position, it's called a **heteroplasmy.** In other words, some copies of a person's DNA have one result and other copies have another. This is most common in mtDNA because there are hundreds, and sometimes thousands, of copies of mtDNA in each cell. As of January 2021, each heteroplasmy shows as a mismatch, artificially adding a Genetic Distance of 1 for each heteroplasmy. To see whether or not you have a heteroplasmy, look at your mtDNA mutations and compare to the chart <u>here</u>.

Matching on HVR1 means that you have a 50% chance of sharing a common maternal ancestor within the last 52 generations or about 1,300 years. Matching on HVR1 and HVR2 means that you have a 50% chance of sharing a common maternal ancestor within the last 28 generations



or about 700 years. **Matching exactly on the mtDNA 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 5-16 generations or about 125-400 years.

mtDNA Projects and Your Research

To help with understanding and analyzing your mtDNA results, you may want to join a Group Project. Run by volunteer administrators, there are several categories of Group Projects, including:

- o Y-DNA Geographical, mtDNA Geographical, Dual Geographical
- Y-DNA or mtDNA Haplogroup
- mtDNA Lineage
- Family Finder (autosomal DNA)

Group Project Administrators have varying levels of experience and expertise, but in general, they can provide data comparison for grouping in the project, data analysis within matches and within the project, and data integration with paper trail research. They may also recruit test candidates and, in some cases, solicit donations to crowdfund tests for those who might not be able to afford them.

Resources

FamilyTreeDNA Learning Center, https://learn.familytreedna.com/

Heteroplasmy nomenclature,

https://www.familytreedna.com/learn/mtdna-testing/heteroplasmy-nomenclature/

FamilyTreeDNA Public mtDNA Haplotree,

https://www.familytreedna.com/public/mt-dna- haplotree/L

Behar, et al, A "Copernican" Reassessment of the Human Mitochondrial DNA Tree from its Root, https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3322232/

Mannis van Oven's Phylotree.org website, http://phylotree.org/

Estes, Roberta. DNAeXplained: Mitochondrial DNA Resources – Everything You Need to Know, https://dna-explained.com/2019/09/04/mitochondrial-dna-resources-everything-you-need-to-know/