

Mitochondrial DNA and mtDNA Testing

A Primer for Family Researchers by Ancestry Bird Dog

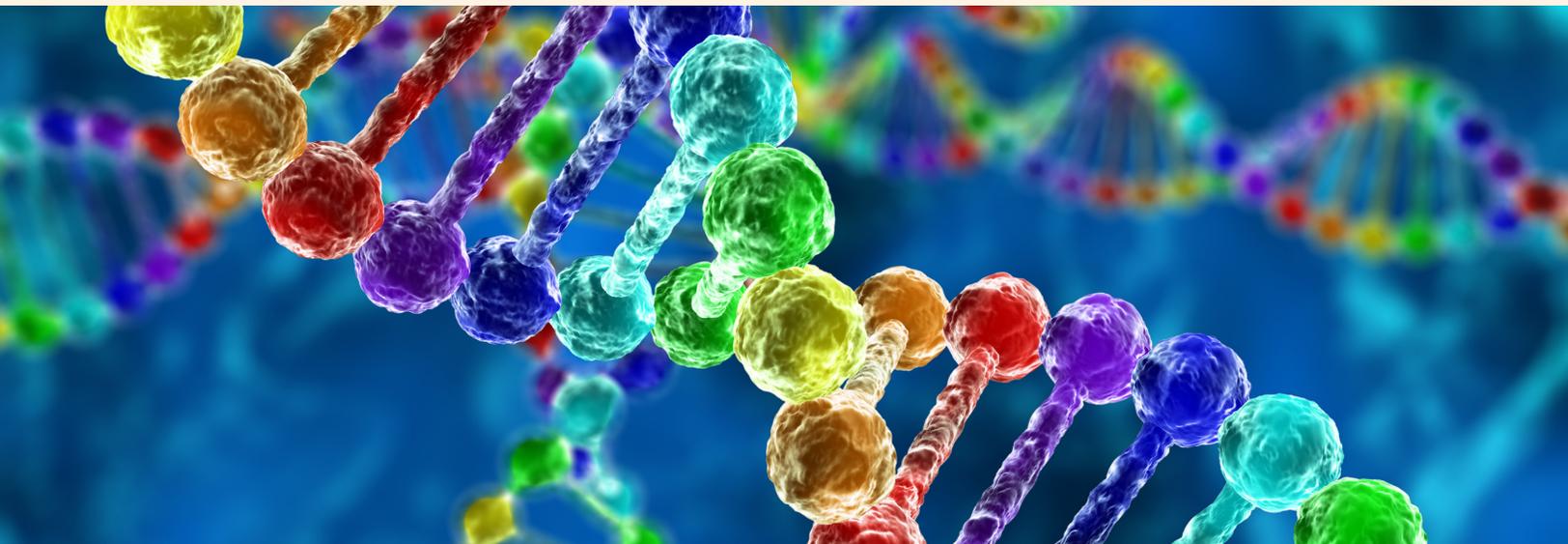


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To understand your mtDNA test results, you need to start with some basic information about human biology and genetics.

- **Human cells:** The human body is made up of millions of cells, each carrying a complete set of genetic information or instructions that guide our growth, development and health.
- **Genome:** A genome is the body's complete set of genetic instructions or DNA that are unique to each person. A genome includes all of the hereditary instructions for creating and maintaining life, as well as instructions for reproduction.
- **Humans have two genomes:** (1) The nuclear genome is found in the nucleus of each cell. (2) The mitochondrial genome is contained within the mitochondria, located in the fluid outside the nucleus.
- **Chromosomes:** The nuclear genome is stored in our chromosomes. A chromosome is a single, long molecule of DNA. Humans inherit one set of chromosomes from their mother and a second set from their father. Human cells contain 46 chromosomes, consisting of 22 pairs of autosomes (non-sex chromosomes) and two sex-determining chromosomes.

- **Sex Chromosomes:** The sex chromosomes in humans are called X and Y. Females carry two X chromosomes, while males carry one X and one Y chromosome.
- **Genes:** Within our chromosomes are sections of DNA that work together to form genes. Genes control different characteristics, such as eye color and height. Autosomal DNA has around 3.2 million base pairs.
- **Mitochondria and mitochondrial genome:** Each human cell also contains hundreds of mitochondria, located in the fluid that surrounds the nucleus. Their primary job is to provide energy to the cell. In addition, inside the mitochondria is a circular piece of DNA called mitochondrial DNA (mtDNA). It has 16,569 DNA base pairs or building blocks. Add a little bit of body text
- **Mitochondrial DNA Transmission:** Males inherit mitochondrial DNA from their mother, but do not pass it on to their children. Males inherit Y-DNA from their father, and pass it on to their sons, but not their daughters. Females inherit mitochondrial DNA from their mother; and pass it on to both their male and female children. Females do not inherit Y-DNA from their father.

- **mtDNA regions:** mtDNA is composed of three DNA regions, and mtDNA testing looks at one or more of these regions. (1) Hyper Variable Region 1; a small set of base pairs at the end of the DNA strand; (2) Hyper Variable Region 2: a small set of base pairs at the beginning of the DNA strand; and (3) Coding region: the large central portion of the DNA strand.
- **MtDNA Test Types:** There are three mtDNA tests based on the mtDNA regions. (1) Low resolution (HVR1) test; (2) High resolution (HVR1 and HVR2) test; and (3) Full genome sequence test, which looks at the entire mtDNA or all three regions.
- **Haplogroup:** An mtDNA test also provides a separate piece of information, known as a haplogroup. A haplogroup is a population of people who are all descendants of a single man or woman who lived in the distant past. Each mtDNA haplogroup has a unique set of mtDNA markers that sets them apart from being a member in a different haplogroup. There are currently 26 known mtDNA haplogroups. As science studies more populations, it is possible more mtDNA haplogroups will be added. Presently, all living persons fall into one of these 26 haplogroups. Letters of the alphabet are assigned to each mtDNA haplogroup. For instance, the L3 haplogroup is strongly associated with the out-of-Africa migration event that started nearly 70,000 years ago. It is inherited by all modern non-African populations, as well as by some populations in Africa. Research shows the population dispersal originated in East Africa and expanded to West Asia, and further to South and Southeast Asia in the course of a few millennia.

Mitochondrial DNA Test



Mitochondrial DNA Test (mtDNA): A mitochondrial DNA test traces a person's matrilineal or mother-line ancestry using the DNA in her mitochondria. Therefore, a mitochondrial DNA test can be taken by both men and women.

When two or more people share or match segments of DNA, they share a common ancestor in their past. It is from that ancestor that the shared DNA segments are inherited.

The more DNA you share with someone, the closer you are to that person. This means your shared ancestor lived in a more recent time.

Reasons to take an mtDNA Test

The most common consumer complaint about DNA testing is that the participant's results and analysis fell short of their expectations. So, to help set realistic expectations it's important to understand that each DNA test has different strengths, limitations and rationales.

The most common DNA test for genealogy is the **autosomal DNA test**. It can identify relatives between five and seven generations back, across both maternal and paternal lines.

Only men can effectively take a **Y-DNA test**, which identifies male relatives on the paternal line, reaching back 60,000 years. If you want to trace the history of your family's surname, this is the best test.

mtDNA tests can determine genetic relationships on a maternal line from 150,000 years ago, and both men and women can take this type of test. Your mtDNA test will give you a list of matches. However, this isn't particularly useful for all genealogy purposes, because even an exact match can be hundreds of years to the common ancestry and beyond any genealogical paper trail. So, mtDNA testing is generally not the first test you turn to when seeking your ancestors.

mtDNA Test, Still Valuable Research Tool

While it may not be your first choice, an mtDNA test still offers many benefits, including those listed here.

- **More generations:** Unlike *autosomal DNA testing*, mtDNA testing reliability reaches back past the fourth and fifth generation in your pedigree. But, it doesn't tell you how closely you are related to your mtDNA matches. Based on current mtDNA mutation rate data, every living person on Earth can trace their ancestry back to a single common ancestry, who lived about 150,000 years ago, the so-called "Mitochondrial Eve."
- **Break through brick walls:** mtDNA testing can help you break through brick walls, such as identifying your female ancestors who were not listed in pre-1850 census records. Or, perhaps you have an ancestor who was enslaved only listed as a woman on a Slaveholder's Census record. The test results will provide you with a list of individuals who may share direct maternal line ancestry with you, and, therefore, may be related to the woman you are seeking information about.
- **Additional documentation:** Similar to other family history records (marriage, death, birth) collected during your family research, an mtDNA test further documents your maternal line.
- **Confirm or invalidate family stories:** If there is a question about the ethnicity of your female ancestor, taking an mtDNA test will provide you with a haplogroup. Sometimes confirming your ancestor's haplogroup will confirm what you already know or dispel those multi-generational family rumors.



- **Native American Research:** For those with Native American ancestors, mtDNA testing can be especially important. Native American haplogroups are very distinct from most others. If your mtDNA haplogroup matches those found in North America or South America, you can be confident you have some Native American ancestry.
- **Research sharing:** If you get your test done by a company dedicated to genealogy, you will have the option of sharing your results with other researchers, and be able to see which have matching results. Fellow researchers may have already gathered information that will allow you to more quickly grow your family tree.
- **Tracing the geographic path of your ancestors.** Discovering your maternal haplogroup can help you determine where your ancestors likely lived over time.