## Overview of O'Mahony DNA Project Data - Mitochondrial DNA

As of May 2023, the O'Mahony Society DNA project includes **mtDNA** of **61 individuals**. This mtDNA data is divided into the following haplogroups:

D haplogroup: 1 individual

H haplogroup: 29 individuals (divided into 20 sub-clades)

I haplogroup: 2 individuals

J haplogroup: 5 individuals (divided into 4 sub-clades)

K haplogroup: 6 individuals (divided into 6 sub-clades)

T haplogroup: 4 individuals (divided into 3 sub-clades)

U haplogroup: 8 individuals (divided into 8 sub-clades)

W haplogroup: 4 individuals (divided into 2 sub-clades)

X haplogroup: 1 individual

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## Human mitochondrial DNA haplogroup

The **letter** names of the **haplogroups** (not just mitochondrial DNA **haplogroups**) run from A to Z. As **haplogroups** were named in the order of their discovery, the alphabetical ordering does not have any meaning in terms of actual genetic relationships.

## Your mtDNA Test Results Determine Your Matrilineal Genetic Signature

The Mutations page breaks down your raw sequence for your mtDNA test results. Your mutations determine your matrilineal genetic signature, meaning both the ancient migration path that your direct maternal ancestors took and who your closest matrilineal matches are. >

**FTDNA compares your DNA to two reference sequences**, the Reconstructed Sapiens Reference Sequence (RSRS) and the Revised Cambridge Reference Sequence (rCRS).

The **Cambridge Reference Sequence** (**CRS**) for human mitochondrial DNA was first announced in 1981.[2]

> A group led by Fred Sanger at the University of Cambridge had sequenced the mitochondrial genome of one woman of European descent[3] during the 1970s, determining it to have a length of 16,569 base pairs (0.0006% of the nuclear human genome) containing some 37 genes and published this sequence in 1981.[2]

For the RSRS, each mutation will be grouped by region of the mitochondria, and then listed with the nucleotide base (A, T, G, C) read in the DNA of the reference found at the position, followed by the position, and then the nucleotide base (A, T, G, C) read in your DNA. For example, C16222T means that at position 16222, the reference sequence has a C and your DNA has a T.

> For additional detail, you may want to take a look at this article (NATURE GENETICS): https://www.nature.com/articles/jhg2013120

For the rCRS, you will see another section that lists all of your mutations in a chart, broken down by each region of the mitochondria. The column headers in the rCRS mutations chart provide the following information:

- Position: The location of the mutation
- CRS: The nucleotide base (A, T, G, C) read in the DNA of the Cambridge Reference Sequence
- Your Result: The nucleotide base (A, T, G, C) read in your DNA at this position. Blank means your DNA matches the Cambridge Reference Sequence.

Some mutations will be defined by a letter other than A, T, G, or C. These are mutations in progress, and they are referred to as heteroplasmies. Learn more at this site:

> https://help.familytreedna.com/hc/en-us/articles/4742569461263-Understanding-mtDNA-Heteroplasmy#what-is-mitochondrial-dna--0-0

**Matching levels**

There are three sections of mtDNA used for matching: HVR1, HVR2, and the Coding Region.

- **HVR1** - Considered a low-resolution region. 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).
- **HVR2** - One of the two mtDNA hypervariable regions used in genealogical DNA testing. HVR2 is considered a high-resolution region. Matching on both 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).
- **Coding Region** - Matching at HVR1, HVR2, and the coding region 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).

The mtFull test offered by FamilyTreeDNA examines the HVR1, HVR2, and Coding Region of mitochondria and is the highest resolution test available to genealogists. The closeness of a

mitochondrial DNA (mtDNA) match depends on the matching level. Matches at higher levels are more likely to be recent.

https://help.familytreedna.com/hc/en-us/articles/4411203181711-Our-mtDNA-Test#direct-maternal-line-0-0

**"Mitochondrial DNA subject to mutation"**

In reading through the literature on the subject of mtDNA you will find mention made of mitochondrial DNA being much more subject to mutation than Y-chromosome DNA. This description may be somewhat misleading. Y-chromosome DNA is usually very slow to mutate (change), each change may take thousands of years. mtDNA may be faster to change than Y-chromosome DNA but the interval between mutations can still be multiple centuries.

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**Mutations give rise to Sub-Clades.**

The basis for a sub-clade is a mutation to a haplogroup. Establishment of a sub-clade normally requires multiple mutations to a DNA sequence.

Here is a list of some of the mutations that give rise to a mtDNA Subclade:
https://www.familytreedna.com/mtdna-haplogroup-mutations.aspx

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## What is a genetic marker

A marker (largely synonymous with the word "landmark" and often referred to as a genomic marker or a genetic marker) is **a DNA sequence, typically with a known location in a genome**. Markers can reflect **random sequences**, **genomic variants** or **genes**.

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## Haplogroup

A **haplogroup is a major branch** on either the maternal or paternal tree of humankind. Haplogroups are associated with early human migrations. Today these can be associated with a geographic region or regions.

**Note:** Though maternal and paternal haplogroups may have similar naming systems, their definitions are different. There are two kinds of haplogroups: the paternally inherited Y-chromosome DNA (Y-DNA) haplogroups, and the maternally inherited mitochondrial DNA

(mtDNA) haplogroups. They respectively indicate the agnatic (or patrilineal) and cognatic (or matrilineal) ancestry.

## Haplotype

A haplotype is a set of genetic markers inherited together from one parent.

Two individuals that match exactly on all markers have the same haplotype. *Example*: R1b-M343 is the most common Y haplogroup of men across Europe and the Americas. Example: mtDNA **haplogroup H** can be found within as much as 40% of European people, making it the most common *maternal haplogroup* in the west.

## Clade

A clade is a group of related individuals.

## Sister Clade

A sister clade is one of two haplogroups or subclades that are at the same level on a phylogenetic tree. For Y-chromosome research, this is sometimes a brother clade.

For example, on the maternal tree, H6a and H6b are sister clades.

## Subclade

A subclade is a **subgrouping in the haplogroups of the human genetic trees**. This may be either the Y-chromosome tree or the mitochondrial tree. Subclades are more specific to a location or population group than the major branches (haplogroups).

**Subclade** may be

- Subclade, any taxonomic clade which is subordinate to hierarchically higher clades, especially:
- Subclade, a subgroup of a clade
- Subclade, a subgroup of a genetic haplogroup
    - a subgroup of a human mitochondrial DNA haplogroup
    - a subgroup of a human Y-chromosome DNA haplogroup

**Yes, Your Base Haplogroup Must Match**

To even begin to look further for a common ancestor on either your Y DNA line (direct patrilineal) or direct mitochondrial matrilineal line (your mother's mother's mother's line on up the tree), your base haplogroup much match.

In other words, you and your matches must all be in the same base haplogroup. Haplogroups are defined by the presence of specific combinations of mutations which are called SNPs (single nucleotide polymorphisms) in the Y DNA.

**Are Y-DNA haplogroups the same as mtDNA haplogroups?**

No, the names in the mitochondrial DNA (mtDNA) haplogroups do not correspond with the same names in the Y-chromosome DNA (Y-DNA) haplogroups.

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Interesting mtDNA BBC video:

**Richard III: The DNA Analysis and Conclusion - Professor Turi King**

https://www.youtube.com/watch?v=-NDuzZiDWFM&t=2s

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**The ABCs of mtDNA**

*Megan Smolenyak Smolenyak*

When it comes to genetic genealogy, Y-DNA is by far the most popular type of testing–and understandably so. Since Y-DNA is passed from father to son down through the generations (just like surnames) its application is fairly obvious. But over time, mitochondrial DNA (usually shortened to mtDNA) testing has been gaining in popularity.

**How mtDNA Travels**
Many folks regard mtDNA as the equivalent of a maternal version of Y-DNA testing and while there are some parallels, there are also some differences, and that creates a lot of confusion.

Full article here: https://blogs.ancestry.com/circle/?p=479

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*Megan Smolenyak, co-author (with Ann Turner) of Trace Your Roots with DNA: Using Genetic Tests to Explore Your Family Tree (as well as In Search of Our Ancestors, Honoring Our*

*Ancestors and They Came to America), can be contacted through www.genetealogy.com, www.honoringourancestors.com, and megansrootsworld.blogspot.com.*

More on Megan Smolenyak: https://en.wikipedia.org/wiki/Megan_Smolenyak

## DNA Testing Dispels a Genealogical Myth

**It appears that Megan Smolenyak Smolenyak was interested in knowing if she had married her cousin. Here is her story:**

https://smolenyak.medium.com/dna-testing-dispels-a-genealogical-myth-7f74b851ffab

## How many genes in each mitochondria.

The human mitochondrial DNA (mtDNA) is a double-stranded, circular molecule of 16 569 bp and contains 37 genes coding for two rRNAs, 22 tRNAs and 13 polypeptides.

## Mitochondrial DNA Disease

Mitochondrial DNA Diseases – TED talk by Professor Mary Herbert, University of Newcastle

https://www.youtube.com/watch?v=pc7MyUs_ORQ

## Overview of Y-chromosome DNA project data

Background: The Y chromosome is one of the two sex chromosomes in humans (the other is the X chromosome). The sex chromosomes form one of the 23 pairs of human chromosomes in each cell. The Y chromosome spans more than 59 million building blocks of DNA (base pairs) and represents almost 2 percent of the total DNA in cells.

Each person normally has one pair of sex chromosomes in each cell. The Y chromosome is present in males, who have one X and one Y chromosome, while females have two X chromosomes.

Identifying genes on each chromosome is an active area of genetic research. Because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. The Y chromosome likely contains 70 to 200 genes that provide instructions for making proteins. Because only males have the Y chromosome, the genes on this chromosome tend to be involved in male sex determination and development.

**The present day Project report**: The number of participants continues to increase although the rate of increase has slowed in the past several years. We have a new member from the Netherlands and it is good to see participation from the continent of Europe. Hopefully we will soon be joined by O Mahony family members from Continental Europe.

The number of participants continues to grow and it is encouraging to see that we have 206 of results on the spreadsheet with others in the process at the laboratory.

The phylogenetic tree shown above has continued to develop and has been modified yet again in the past few days. These changes are based on the efforts of the "citizen scientists" (participants like ourselves) who have contributed their samples and made them available for analysis.

226 males have submitted Y-chromosome tests. 46 people have submitted sample for Big-Y testing. 10 people have submitted their GEDCOM results. Total number of Big-Y tests in the Project: 56.

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What gets tested

Y-DNA testing involves looking at Y-STR segments of DNA on the Y chromosome. The STR segments which are examined are referred to as genetic markers and occur in what is considered non-coding DNA regions.

STR markers

A chromosome contains sequences of repeating nucleotides known as short tandem repeats (STRs). The number of repetitions varies from one person to another and a particular number of repetitions is known as an allele of the marker. An STR on the Y chromosome is designated by a DYS number (DNA Y-chromosome Segment number). The example below shows the allele of Skywalker's DYS393 marker is 12, also called the marker's "value". The value 12 means the DYS393 sequence of nucleotides is repeated 12 times—with a DNA sequence of (AGAT)12.

SNP markers

*See main article: SNP testing*

A single-nucleotide polymorphism (SNP) is a change to a single nucleotide in a DNA sequence. The relative mutation rate for an SNP is extremely low. This makes them ideal for marking the history of the human genetic tree. SNPs are named with a letter code and a number. The letter indicates the lab or research team that discovered the SNP. The number indicates the order in which it was discovered. For example M173 is the 173rd SNP documented by the Human Population Genetics Laboratory at Stanford University, which uses the letter M.

Understanding test results

Y-DNA tests generally examine 10-111 STR markers on the Y chromosome, but hundreds of markers are available. STR test results provide the personal haplotype. SNP results indicate the haplogroup.

Haplotype

A Y-DNA haplotype is the numbered results of a genealogical Y-DNA test. Each allele value has a distinctive frequency within a population. For example, at DYS455, the results will show 8, 9, 10, 11 or 12 repeats, with 11 being most common. For high marker tests the allele frequencies provide a signature for a surname lineage. The test results are then compared to another project member's results to determine the time frame in which the two people shared a most recent common ancestor (MRCA). If the two tests match perfectly on 37 markers, there is a 50% probability that the MRCA was fewer than 2 to 3 generations ago, a 90% probability that the MRCA was fewer than 5 generations ago, and a 95% probability that the MRCA was fewer than 7 generations ago.

STRs results may also indicate a probable haplogroup, though this can only be confirmed by specifically testing for that haplogroup's single-nucleotide polymorphisms (SNPs).