Genetic Genealogy Demystified:
Reading and Understanding Your Family Tree DNA Results
Part 2: mtDNA

Presented By
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> **You've taken the plunge and ordered a genetic genealogy DNA test from Family Tree DNA, but now that you have your results, you don't understand them? This webinar is for you! Join us to learn how to read and understand your mtDNA results.**

DNA Tests for Genealogy

- Y Chromosome (Y-DNA)
  - Direct paternal line (father's father's father's father, etc)
  - Only men can test for it
- Mitochondrial DNA (mtDNA)
  - Direct maternal line (mother's mother's mother's mother, etc)
  - Both men and women can test for it
- Autosomal DNA (Family Finder)
  - Identify cousins via any ancestral line
  - Both men and women can test for it

This webinar focused on mtDNA tests.

Review of DNA Basics

Our DNA consists of:

- 46 chromosomes (23 pair)
  - 22 pair = autosomal chromosomes, or autosomes
  - 1 pair = gender chromosomes (X,Y) (women have XX, men have XY)
- Mitochondrial DNA (mtDNA)

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1

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DNA Sequences & Mutations

DNA is made up of four (4) bases (nucleotides):

- Adenine
- Cytosine
- Guanine
- Thymine
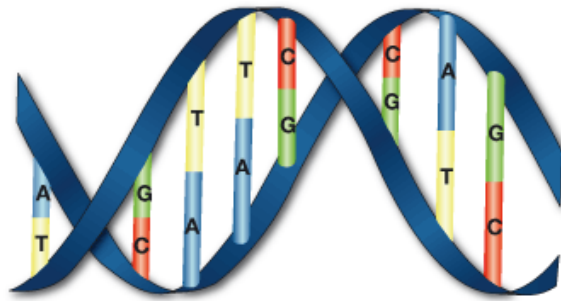
Thymine (Yellow) = T Guanine (Green) = G

Adenine (Blue) = A Cytosine (Red) = C

Mutations (changes) in the DNA happen over time. Mutations happen randomly, but always between a parent/child pair. Mutations are what enable us to do genealogy and ancestry studies. They differentiate us from people who we're not closely related to, while matching us with people who we are closely related to. Genetic genealogy uses two types of mutations: STR and SNP. For mtDNA testing, we only use SNPs.

DNA Sequence – a string of consecutive bases

> Example: TAGCAAAGTCCCATGCCCCAGTCAGTCGGGA

Single Nucleotide Polymorphism (SNP) – Base change at a single location in the DNA

> Example: C → G, or A → T

> Rare occurrences in Y-DNA and mtDNA, so great for deep ancestry studies.

> Great for autosomal studies when used in large quantities.

Haplogroup – Broad division of the human population, defined by mtDNA and Y-DNA SNPs.

> Represents deep/ancient ancestry (thousands to tens of thousands of years).

> Separate mtDNA and Y-DNA haplogroups.

> Can be divided into subgroups, known as subclades.

> Haplogroups are hierarchical, similar to an outline.

> Haplogroup names start with a capital letter, then alternate numbers and lower case letters.

> Example: K1a

>> K is the main haplogroup name

>>> K1 is a subclade of K

>>>> K1a is a subclade of K1.

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2

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Reading & Understanding mtDNA Results

Family Tree DNA offers three (3) levels of mtDNA tests:

- mtDNA
  - Hyper Variable Region 1 (HVR1)
  - Bases 16001-16569 (Total of 569 bases)
  - Backbone haplogroup determined

- mtDNAPlus
  - Hyper Variable Regions 1 & 2 (HVR1+HVR2)
  - Bases 16001-16569 plus Bases 1-574 (Total of 1143 bases)
  - Backbone haplogroup determined

- mtFullSequence or Full Mitochondrial Sequence
  (also known as FGS, Full Genomic Sequence)
  - Entire mtDNA genome
  - Bases 1-16569 (Total of 16,569 bases)
  - Full haplogroup determined

Cambridge Reference Sequence (CRS)

First full mtDNA test ever completed. Family Tree DNA reports results as differences from the CRS. The CRS is in haplogroup H2a2a. However, the ancestress of all human maternal lines living today is in haplogroup L0.

Control Region
- HVR1+HVR2

Coding Region
- Everything outside of HVR1+HVR2

Matches

Family Tree DNA provides names and email addresses of other customers who your results match. You can contact your matches to share your family trees and work together to identify your common ancestor. FTDNA displays only exact matches for mtDNA:

- Exact mtDNA Matches (Low resolution, HVR1)

- Exact mtDNAPlus Matches (High resolution, HVR1+HVR2)

- Exact Full Mitochondrial Sequence Matches (FGS/FMS)

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Reading and Understanding Your Family Tree DNA Results, Part 2: mtDNA

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3

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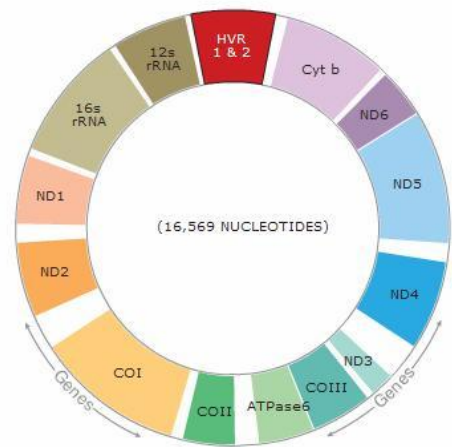
Results

The following chart shows results for a sample Full Mitochondrial Sequence test: HVR1, HVR2 and the Coding Region (CR). Each value shown is a difference from the CRS.

Haplogroup - K1a1b1a

HVR1 differences from CRS		
16224C	16234T	16311C
16519C		

HVR2 differences from CRS		
73G	114T	263G
315.1C	497T	

CR differences from CRS		
750G	1189C	1438G
1811G	2706G	3480G
4769G	7028T	8860G
9055A	9698C	10398G
10550G	10978G	11299C
11467G	11470G	11719A
11914A	12308G	12372A
12954C	14167T	14766T
14798C	15326G	15924G

How to Read the Results

16224C = At location 16,224 on the mtDNA genome, this sample has a C instead of the reference value.

The chart to the right shows the reference (CRS) value at position 16224 as T, while this sample has a C.

HVR1 REFERENCE SEQUENCE

Show All Positions

Position	CRS	Your Result
16224	T	C
16234	C	T
16311	T	C
16519	T	C

Comparing Results

The differences from the CRS should be compared. In most cases, mtDNA matches should be exact across all tested areas.

Haplogroup	HVR1 differences from CRS	HVR2 differences from CRS
K	16224C, 16234T, 16311C, 16519C	
K	16224C, 16234T, 16311C, 16519C	73G, 114T, 263G, 315.1C, 497T
K	16224C, 16234T, 16311C, 16519C	73G, 114T, 263G, 315.1C, 497T
K1a1b1a	16224C, 16234T, 16311C, 16519C	73G, 114T, 263G, 315.1C, 497T
K1a1b1a	16224C, 16234T, 16311C, 16519C	73G, 114T, 195C, 263G, 309.1C, 315.1C, 497T
K2a2a	16224C, 16311C, 16519C	73G, 146C, 152C, 263G, 315.1C, 512C

In the sample comparison above, the first 5 share the same HVR1 results, and several share the same HVR2 results as well. However, the 5th person has two additional mutations in HVR2, so they are likely not related to the others within a genealogical timeframe (hundreds of years). They do share the same deep ancestry (thousands of years), since they are in the same subclade of haplogroup K.

The 6th person has differences in both HVR1 and HVR2, and is also in a different subclade of haplogroup K. So while they still share extremely deep ancestry that goes back to the founding of K (25,000 years ago), they don't share more recent deep ancestry and certainly don't share recent genealogical ancestry (on the direct maternal line).

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4

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Time to Most Recent Common Ancestor

mtDNA (HVR1) Match
  50% chance of sharing a common ancestor within 52 generations (approx 1300 years).

mtDNAPlus (HVR2+HVR2) Match
  50% chance of sharing a common ancestor within 28 generations (approx 700 years).

mtFullSequence (FMS/FGS) Match
  50% chance of sharing a common ancestor within 5 generations (approx 125 years).

Ancestral Origins

Anonymous listing of your matches' reported ancestral origins on their direct maternal lines. This is not a breakdown of your own ancestry – rather, it's a hint as to where you could potentially have direct maternal line ancestry from, based on your matches' known ancestry.

HIGH RESOLUTION (HVR1+HVR2)					
Country	Your Matches	Comment	Match Total	Country Total	Percentage
Austria	5	-	7	207	3.4%
	2	Ashkenazi			
Belarus	7	-	9	156	5.8%
	2	Ashkenazi			
France	1	-	2	973	0.2%
	1	Ashkenazi			

How to Read

You match 7 out of 207 people who tested HVR1+HVR2 and reported direct maternal line ancestry from Austria. Similarly, you match 3.4% of the people who tested HVR1+HVR2 and reported direct maternal line ancestry from Austria.

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mtDNA Haplogroups

- Broad groupings of the human population.
- Represent deep ancestry (thousands to tens of thousands of years)
- Hierarchical – can be broken down into subclades (subgroups)
- Generally determined by coding region mutations.
  - With the mtDNA and mtDNAPlus tests, Family Tree DNA also tests a panel of 22 SNPs to determine the backbone haplogroup.
  - The Full Mitochondrial Sequence is required to obtain a full haplogroup assignment.

See also:

http://www.familytreedna.com/faq/answers/default.aspx?faqid=10#475

Phylotree:

- http://www.phylotree.org
- Similar to the Y-DNA Haplotree, but for mtDNA and managed by an independent group.
- Shows the mutations (differences) that define each branch of the mtDNA haplogroup tree.
- Phylotree starts at haplogroup L0, colloquially known as "Mitochondrial Eve"
- Remember, the CRS is in haplogroup H2a2a!

Haplogroup Origins

Anonymous listing of your matches' mtDNA haplogroups and reported country of origins. Matches who have only HVR1 or HVR1+HVR2 results will only have a backbone haplogroup shown. Matches who have Full Mitochondrial Sequence results will have a complete haplogroup shown.

HIGH RESOLUTION (HVR1+HVR2)			
Haplo	**Country**	**Comment**	**Match Total**
K	Austria	-	1
K	Austria	Ashkenazi	1
K	Belarus	-	3
K1a1b1a	Belarus	-	4
K1a1b1a	Belarus	Ashkenazi	2

Migration Maps

Provides two maps to help you understand the migration and distribution of your haplogroup:

Migration Map

Migration path for each haplogroup, traced back to Africa.
Downloadable version at http://www.familytreedna.com/pdf-docs/mt_migrationmap.pdf

Frequency Map

Charts the distribution of each haplogroup in 10 world regions.

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6

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Making the Most of Your Genetic Genealogy Experience

Receiving, reading and understanding your results shouldn't be the end of your genetic genealogy journey. Here are some ways you can make the most of this experience:

- **Share.** Your myFTDNA account has fields to enter your Most Distant Known Ancestor's (MDKA) name and location, as well as all the surnames in your ancestry. You can also upload a GEDCOM of your family tree. Entering all this information will enable your matches to learn something about your genealogy before they even contact you.

- **Communicate.** Don't be afraid to email your matches, and definitely respond to emails that you receive from your matches! All customers can choose whether to sign the green release form that Family Tree DNA provided with your swab kit, which gives FTDNA permission to provide their name and email address to matches. So everyone who you see on your Matches page had signed this form and given permission.

- **Participate.** Family Tree DNA has over 6,000 different projects which focus on surname, geography, heritage and haplogroups. Projects are all run by volunteer administrators and are free to join. Joining projects is the best way to stay informed about news related to the topic of the project, and you have the added benefit of getting support and assistance from your project administrator. For mtDNA, consider joining appropriate haplogroup, heritage and geographical projects.

- **Be Proactive.** No project for your mtDNA haplogroup or direct maternal country of origin yet? Considering starting one and recruit other people to get tested. You don't have to be an expert on genetic genealogy to start a project.

- **Learn.** Keep learning about genetic genealogy by attending Relative Roots Webinars, consulting the Family Tree DNA FAQ, visiting the International Society of Genetic Genealogy website, and reading discussion forums and email lists that discuss genetic genealogy. Project administrators are also invited once a year to attend a conference hosted by Family Tree DNA in Houston.

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7

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Websites

- Relative Roots
  - http://www.relativeroots.net
  - http://www.facebook.com/RelativeRoots
  - http://www.twitter.com/RelativeRoots
- Family Tree DNA
  - http://www.familytreedna.com/
  - http://www.familytreedna.com/faq/
  - http://www.facebook.com/FamilyTreeDNA
- International Society of Genetic Genealogy (ISOGG) – it's free to join!
  - http://www.isogg.org
- Genetic genealogy mailing lists and discussion forums:
  - http://www.yahoogroups.com/group/ISOGG
  - http://www.yahoogroups.com/group/DNA-Newbie
  - http://forums.familytreedna.com
  - http://www.dna-forums.org
  - http://lyris.jewishgen.org/ListManager (JewishGen)
  - http://lists.rootsweb.ancestry.com/index/other/DNA/GENEALOGY-DNA.html

Public Searchable Databases

- Mitosearch   http://www.mitosearch.org
- SMGF   http://www.smgf.org

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8

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