INTERPRETING YOUR DNA RESULTS

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NOTE: There are many companies that do DNA testing and offer interpretation. Since GSMD has partnered with ftDNA, I will be following their rationale and giving mostly their online examples.

Once your have received your DNA results, you will want to interpret them which may be challenging as there are different DNA tests. If you are man you have 4 types of DNA: sex chromosomes X and Y dna (yDNA) one from each parent, mitochondrial DNA (mtDNA) solely from the mother, and autosomal DNA (atDNA) from both parents. If you are a woman you have 4 types of DNA: sex chromosomes xDNA from mother and xDNA from father, mitochondrial DNA (mtDNA), again, solely from the mother, and autosomal DNA (atDNA) from both parents. There are typically 5 tests for men: yDNA testing (father) including yDNA-STR and yDNA SNP, mtDNA testing (mother) and autosomal DNA testing (both parents). There are typically 2 tests for women: mtDNA testing and atDNA testing. X chromosome testing is not typically done but is possible having a lower yield of information as to date.

For testing purposes GSMD, at this time, recommends: atDNA for both men and women, the 111 yDNA test for men and the full sequence mtDNA for men and women. For review, there is both nuclear dna (autosomal DNA and the sex chromosomes Y and X DNA) and cytoplasmic dna (mitochondrial dna).

When you get your results back, you will have to log in online, your account will show your profile and next to it are 3 basic categories:

1. **My family Tree** – most **important**, this is your paper trail for your known family of ancestors; you should download your family tree immediately as it helps to filter and provide more information
2. **DNA TESTING**
    a. **Family Finder (autosomal dna = atDNA)**
        i. Matches---shows the name of your match and which side (Maternal, Paternal or both) they are from
        ii. Relationship Range (RR)
            1. Shared segments are based on the amount of cM or centimorgans; the higher the cM between matches, the closer those matches are longest block segment is the longest segment of atDNA measured in a cM shared by you and a genetic match

Centimorgan Chart[1] :

DNA Detectives Autosomal Statistics Chart

Created by Christa Stalcup ©THEDNADETECTIVES, 2016

cM (centiMorgans)^		Percentage (%) of Shared DNA^^		Group	Relationship	Notes
Average	Range	*Average*	Range			
3,600		50%			Parent - Child	
2650	2300 - 3900	37%	32%-54%	Group A	Full Sibling	Ancestry, FTDNA and GEDmatch (HIR only)
3600		50%				23andMe (FIR included)
1800	1300 – 2300	25%	18%-32%	Group B	Half Sibling Aunt/Uncle/Niece/Nephew Double First Cousin Grandparent/Grandchild	3/4 Siblings^^^
900	575 - 1330	12.5%	8%-18.5%	Group C	First Cousin (1C) Half Aunt/Uncle/Niece/Nephew Great-Grandparent/Great-Grandchild Great-Aunt/Uncle/Niece/Nephew	
450	215 - 650	6.25%	3%-9%	Group D	First Cousin Once Removed (1C1R) Half First Cousin (½ 1C) Half Great-Aunt/Uncle/Niece/Nephew	
224	75 - 360	3.125%	1%-5%	Group E	Second Cousin (2C) First Cousin Twice Removed (1C2R) Half First Cousin Once Removed (½1C1R)	
112	30 - 215	1.56%	0.42% - 3%	Group F	Second Cousin Once Removed (2C1R) Half Second Cousin (½ 2C) First Cousin Three Times Removed (1C3R) Half First Cousin Twice Removed (½ 1C2R)	
56	0 - 109*	0.78%	0% - 1.52%	Group G	Third Cousin (3C) Second Cousin Twice Removed (2C2R)	~10% of 3Cs will not share DNA*
30	0 - 75**	0.4%	0%-1%	Group H	Third Cousin Once Removed (3C1R) Other Distant Cousins	~50% of 4Cs will not share DNA**

^cM =Ancestry.com & FTDNA
^^Percentage of DNA = 23AndMe
^^^ 3/4 Siblings are a combination of half siblings and 1st cousins, FIRs are included.

Groups A & B: 99% within the ranges given
Groups C – I: 95% within the ranges given

2.

TYPES of RR

a. Immediate Match Range

Note: each subsequent category can contain the previous category

1. Parent/child
2. Full Sibling, (+ a.)
3. Half Siblings (+ b.)
4. Grandparent/Grandchild (+ c)
5. Aunt/niece or Uncle/Nephew(+d)

b. Distant Match Range

1. 1st cousin but also ½ Sib, GP/GC, A/N,U/N
2. 1st-2nd cousin (most likely 1)
3. 1st to 3rd cousin (most likely 2)
4. 2nd to 3rd cousin (most likely3)
5. 2nd to 4th cousin (most likely 3)
6. 3rd –to 5th cousin (most likely4)

c. Speculative Match Range

1. Means you share a significant # of Identical by Descent blocks (IBD's) but relationship extends beyond the confidence level
2. 4th and 5th to remote (up to 20th)

Note: with your matches there are links to family trees and email capability to those matches. Utilize these by communicating with these matches, which can give valuable information

Remember:

1. You will share varying degrees of DNA with your grandparents.
2. Although you get 50% of your DNA from each parent, you can inherit only 0-49% of the DNA they inherited from your grandparent (their parents) with the average being 25% due to DNA recombination

   NOTE: DNA recombination occurs during replication and division of cellular DNA where there is an imbalanced but equal exchange of DNA material (ie you could inherit 25% or 1% of grandparent DNA)

   a. We all share DNA with 100% of our immediate family members, 1st and 2nd cousins
   b. We share: 90% with 3rd cousins;
      50% with 4th cousins;
      25% with 5th cousins;
      12.5% with 6th cousins;
      6.125% with 7th cousins
      3% with 8 cousins
   c. It is nearly impossible to get accurate matches after the 8th cousin because the bits of DNA are so small.
   d. Even if known cousins do not match, don't panic…the amount of DNA transferred may have just been too small due to recombination

iii. Chromosome browser
This is a new tool that allows you to view and compare the DNA segments you share with other FF matches

iv. Linked relationships
Matches linked in your family tree

v. My origins
Gives your ethnic makeup as a percentage
Gives information shared origins mapping
Gives earliest know paternal and maternal ancestors locations

vi. Ancient origins
Compares your DNA to archeological digs within the Bronze age (metal use invaders) Neolithic age (farmers) and Mesolithic age (hunter gatherers)

vii. Wellness Report -- A new offering for the health conscious

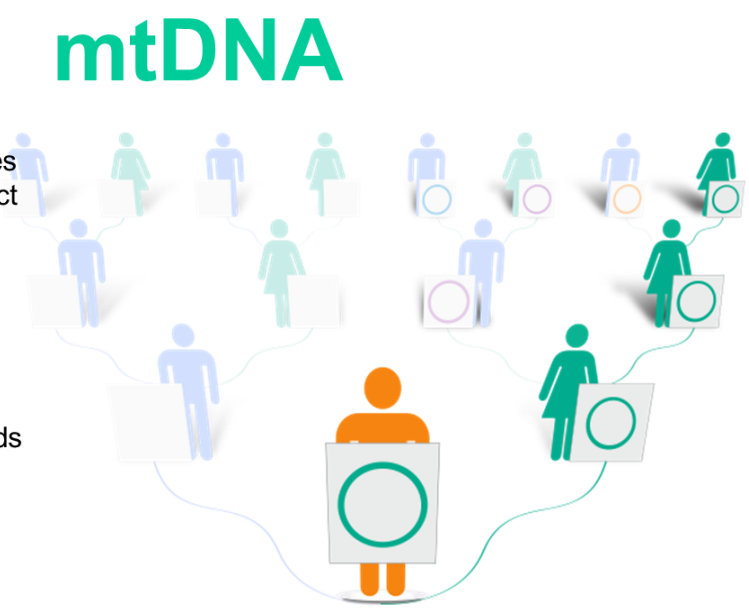
b. mtDNA (=mitochondrial dna)

i. Characteristics

1. mtDNA is passed from mother to child (male and female) but only females may pass it on (males can not pass their mitochondria because these are concentrated in the tail of the sperm for energy for motility and drop off with impregnation of the egg)
2. Allows for investigation into the maternal line of the family helping identify living relatives
3. mtDNA testing gives you your haplogroup which are distinct mutations that occurred in given groups of migrating peoples long ago which can help in identification.
4. Since mtDNA mutates slowly over time it is more helpful in giving information on the distant past (ie haplogroups) rather than more recent relatives and countries of origin
5. It will NOT give information about recent ancestors or places of origins; however, when combined with family finder test (autosomal dna), it can give you information on matches with common ancestors within 5 generation.

Example[2]

- Traces the direct maternal line (mother's mother's mother's etc. line).
- Provides a list of matches who share common direct maternal ancestry within 52 generations.
- Provides an ancestral migration route of your direct maternal line stretching back thousands of years.



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ii. Types
    1. A small chromosome consisting of only 3 regions:
        a. Hypervariable region 1 (16024-16569)
        b. Hypervariable region 2 (00001-00576)
        c. Coding region (00577-16023)
    2. Testing HVR1 and HVR2 can only reveal information on very early ancestors (more than 30 generations ago); need full sequence testing (all 3 – GSMD recs); more productive in genealogy research, can yield later matches of 5-8 generations.
    3. There is only a total of 16569 base pairs
        a. Nomenclature based on the last three numbers (ie drop 16)and first letter of base added (T= thiamine, A= adenine, C=cytosine, G=guanine)
            1. EX: Regions HVR1 – 111T, 223T, 259T, 290T
               HVR2 – 073G, 146C, 153G
    4. Strict matches in all regions indicate only a common ancestor living at least 125 years ago; match in one segment (ie HVR 1) can indicate a common ancestor of more than a 1000 years ago. Matching in 2 groups (HVR 1 and 2) means a common ancestor of about 750 years ago.

iii. Haplogroups vs Haplotypes
    1. **Haplogroups** are groups of haplotypes that are inherited together from a single parent
        a. mtDNA tests sequences of a region of the genome of the circular strand of mtDNA. These sequences are then compared to the Cambridge Reference Sequence (CRS at www.mitomap.org/mitoseq.html).
        b. The differences between your DNA and the CRS can range from 0 to many. These results can be used to estimate your Haplogroup**, which** roughly estimates the time to the most recent ancestor (MRCA).
        c. Haplogroups are only an estimate made up of similar groups of haplotypes of a common shared ancestor
        d. One can get SNP confirmation for the haplogroup (usually costs extra)
        e. There are 26 known mtDNA haplogroups
    2. **Haplotypes** are groups of alleles (or markers/mutations) that are inherited together from a single parent. They share a common ancestor by virtue of one SNP (mutation)

Example[3]:

	Sample mtDNA STR Results (= Haplotype)	
Region	HVR1	HVR2 Differences
From CRS	183C, 189C, 270T	73G, 150T, 263G, 309.1C, 315.1C

The letters represent mutations/differences between your mtDNA and the universal mtDNA from the CRS. This CRS belongs to mtDNA Haplogroup H meaning that individuals who mtDNA in Haplogroup H have fewer differences from the CRS than people in other haplogroups.

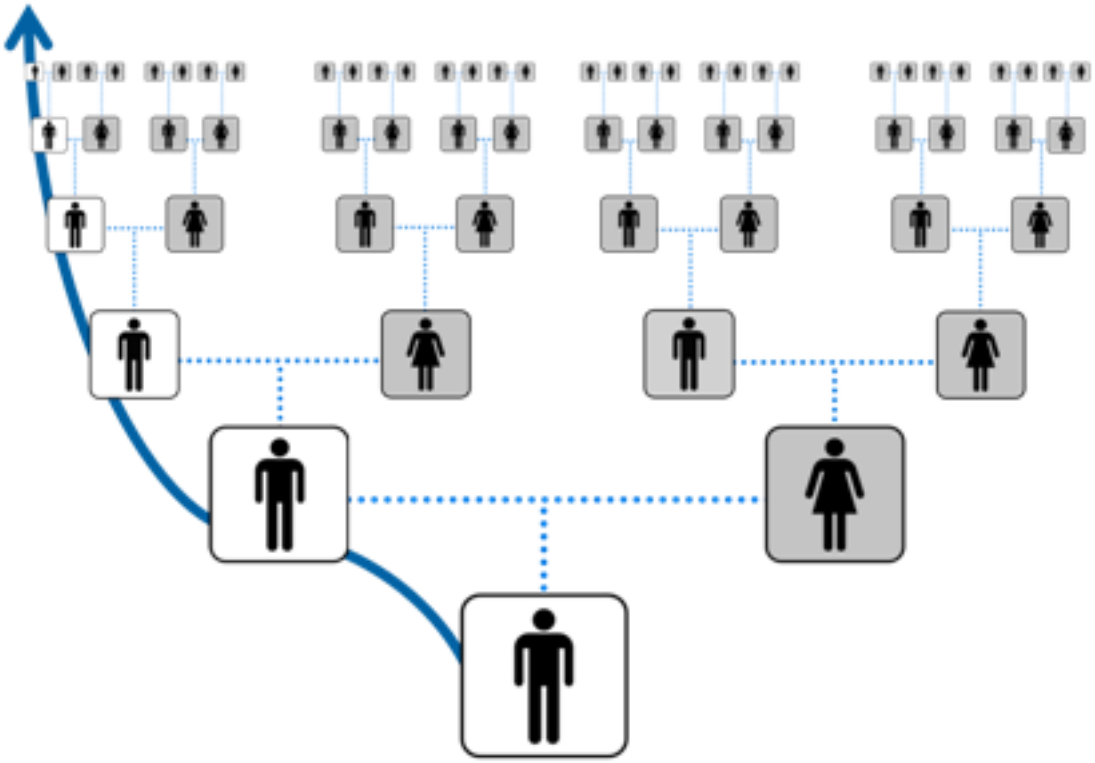
HVR1 shows 3 mutations meaning that the mtDNA sequence differs at these 3 locations. The letter represents the base change (Adenine Thymine Cytosine and Guanine). The number represents the position on the mtDNA.

HVR2 shows 5 mutations in this region of the genome. There are 2 insertion mutations of cytosine denoted 309.1 and 315.1 (deletion mutations are noted with – sign ie 255-delC)

iv. Issues with mtDNA
    1. Mt-DNA is more difficult to use genealogically because of the name changes in every generation (ie there is no surname database like in yDNA searches). There is no organized centralized data base of mitochondrial line names
    2. Only full sequence testing gives information in a genealogical relevant way.
        a. HVR1 full match indicates a common maternal ancestor living more than a 1000 years ago or 40 generations
        b. HVR1 And HVR2 full match indicates a common maternal ancestor living more than 750 years ago or 30 generations
        c. Full sequence match indicates a common maternal ancestor living 125 years ago or 5-8 generations
    3. Unlike yDNA testing, in which transmission is remarkably stable (especially in transmission of mutations), it is possible for mothers, daughters and siblings to have differences in their mtDNA due to **heteroplasmy** (presence of more than type of organelle DNA). This complicates mtDNA interpretation even more which is reason strict matches are required. (such as the remote possibility of paternal mtDNA transmission as invalidating mtDNA testing but, again, this is remote).

c. yDNA (= y sex chromosome dna)

Example[4]



i. DNA only occurs in males and is transmitted only from male to male. It traces the direct paternal line. The Y chromosome is a sex chromosome which resides in the nucleus with autosomal DNA

ii. yDNA can provide a list of matches sharing a common direct paternal ancestor and can provide an ancestral migration pattern based on haplogroup determined by the STR (= Short Tandem Repeats) or SNPs (= Single Nucleotide Polymorphisms)

   i. STR TESTING

      1. These are markers or places where your genetic code has a variable number of repeats.
      2. Unlike mtDNA, these markers change slowly from one generation to the next allowing for distinctive sets that form your signature.
      3. Comparing your results to other men in the database reveals matches that can show you have a common ancestor.

4. In terms of testing, there are standard yDNA STRs which are markers of various lengths:
   yDNA12 (1-12 markers) panel 1
   yDNA 25 (13-25 markers)panel 2
   yDNA 37 (26-37 markers)panel 3
   yDNA 67 (38-67 markers)panel 4
   yDNA 111 (68-111 markers)panel 5 (GSMD recs)
   Big Y 500 (112-561 markers)panel 6
   Big Y 700 (700+ markers)
   Note:
   a. Big Y is one of the newer marker tests where they can only guarantee for 389.
   b. You get upt to 700+ STRs and a lot more SNPs with this testing
   c. It is primarily exploratory in nature
   d. Remember! Over reliance on STRS withought additional SNP testing can be problematic, especially in those instances where there is perfect Y STR matching.

5. Unlike mtDNA, yDNA can trace both distant and recent generations

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Example[5]: Sample Y-DNA STR Results

DYS#	393	390	19	391	385a	385b	426	388	439	389-1	392	389-2
Alleles	13	24	14	10	11	14	12	12	12	13	13	29

What does this string of numbers mean?
At each of these locations on the Y-chromosome there is the potential for variation (or repeats of the same DNA sequence).

Example[4], at DYS426:
The variation consists of repeats of the DNA sequence "GTT", with 12 being the most common repeat (>60% of Y-DNA samples, according to FTDNA).
The DNA sequence would look like this, with the 12 repeats in bold:
…TGT**GTTGTTGTTGTTGTTGTTGTTGTTGTTGTTGTT**GAC…

likewise

Someone with a result of 7 at DYS426 would have the following sequence:
…TGT**GTTGTTGTTGTTGTTGTT**GAC…
Together, the particular set of alleles revealed by testing represent your personal Haplotype. In this example it falls in Haplogroup R1b. ftDNA predicts your haplogroup based on your STRs and will do further testing with SNPS if necessary.

NOTE: You may do your own haplotype prediction but this is beyond the scope of this text.

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ii. Searching for Matches
    1. This can be done in 3 ways:
        a. By Haplotype – This can be done through genetic matches via specific sequences or haplotype. If you enter the haplotype, set Show Users at minimum of 12 markers and Maximum genetic distance at 0.
            i. usually you will get overlap with ftDNA matches, if not set genetic distance to 1 or 2.
        b. By Surname group – a collaborative effort, monitored by an administrator, search for the surname and look for matching haplogroups
        c. By Geographic or Project group – a collaborative effort, monitored by an administrator

NOTE: GSMD is partnered with ftDNA for the Mayflower Society DNA Project. Mike Terry is the administrator for the yDNA studies while Susan Abanor is the administrator for the mtDNA studies accessed at https://www.familytreedna.com/groups/mayflowersociety/about

iii. SNP Testing
    1. SNP testing examines single nucleotides at specific locations on the Y chromosome (as opposed to STRs looking trinucleotides…ie GTT; SNP may only examine G)
    2. Since a mutation at a single base is very rare compared to changes in STRs, males who share a SNP usually share an ancestor who lived many generations or many hundreds of generations ago. For this reason, SNPs have been used to identify the branches in the Y-chromosome family
    3. You can see a chart of some of the most commonly tested SNPs at Family Tree DNA www.familytreedna.com/deepclade.html and www.familytreedna.com/snps-r-us.aspx
    4. SNPs tells about the ancient ancestry

Example[6]: Sample yDNA SNP results

Your SNP results: M96+ P2+, M2-, M35+, M78-, M183+, M81+, M107+, M165-, M123-
Your Y-DNA Belongs to Haplogroup E

The results give you your Haplogroup E which allows us to go directly to the Haplogroup E Tree.

These are possibilities in the haplogroup E tree:
M96+ E
P2+ E3
M2- E3a (not part of this sub-clade)
M35+ E3b1 (not part of this sub-clade)
M81+ E3b1b
M107+ E3b1b1
M123- E3b1c (not part of this sub-clade)
M281- E3b1d (not part of this sub-clade)

This yDNA sample belongs to Haplogroup E, sub-clade E3b1b1 because we tested positive for the M107 SNP. The negative results show it does not belong to E3a, E3bia, E3b1c, or E3b1d.

iv. A word on Triangulation
    1. Triangulation utilizes your DNA result, against one of your closest or best matches (atDNA or yDNA; mtDNA can be utilized but more complicated) to look for a shared match
    2. This shared match indicates a common shared ancestor; it can also indicate what the genetic makeup is of that ancestor.
        a. If you compare your DNA to a 1st cousin (say your father's brother's sons), this could indicate the actual STR marker strand in their mutual grandfather.

Remember, I liken DNA results to a small puzzle piece in a large multisegment puzzle. Looking at the piece by itself offers little help in how it fits into the big picture but comparing like characteristics to surrounding pieces will make all the difference in the world eventually giving you the big picture. DNA results will never replace a gold standard paper trail. DNA testing has become useful to make assumptions about relationships in one or two generations offering support when that paper trail is weak.

Good Luck!

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