Using Mitochondrial DNA (mtDNA) for Genealogy

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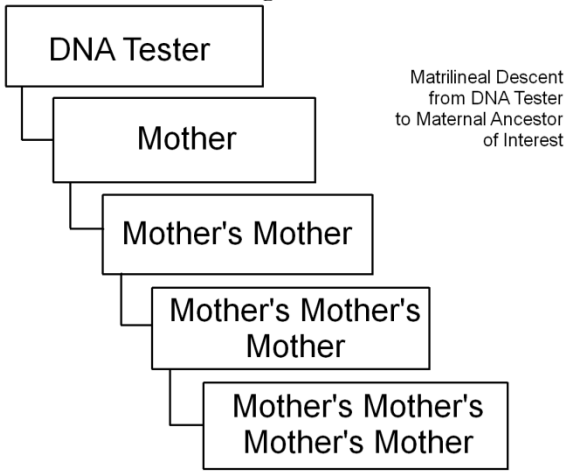
This is one article of a series on using DNA for genealogical research. There are several types of DNA tests offered for genealogical purposes. Researchers must understand that only like tests can be compared: Y-DNA to Y-DNA, mtDNA to mtDNA, and autosomal DNA to autosomal DNA. To use DNA to solve a problem, an understanding of DNA inheritance and the limits of the evidence is paramount. This article covers mitochondrial DNA.

WHAT CAN YOU DO WITH mtDNA?

Genealogists today want to know if DNA can solve a research problem. In some situations mtDNA provides conclusive evidence; in others it provides less strong evidence. The documented maternal lineage should be as deep as possible to make mtDNA most useful as genealogical evidence. The person to be tested must have a straight matrilineal descent—through women with no intervening men. A male can be tested, but the earlier ancestors must be all female back to the person of interest. See figure 1. Even without a specific problem to be solved, many are taking DNA tests to be part of this exciting technology and contribute to genealogical and scientific discoveries.

Figure 1.



As with many consumer products, the DNA tests offered have been a tradeoff between production or laboratory costs and affordability. The earliest tests offered were less comprehensive, but lower priced. Today a comprehensive test at an affordable price is available. More comprehensive tests offer a better chance of finding a common ancestor in a genealogical time frame. If the mtDNA signature is rare the test provides stronger evidence for relationships within a more recent time frame.

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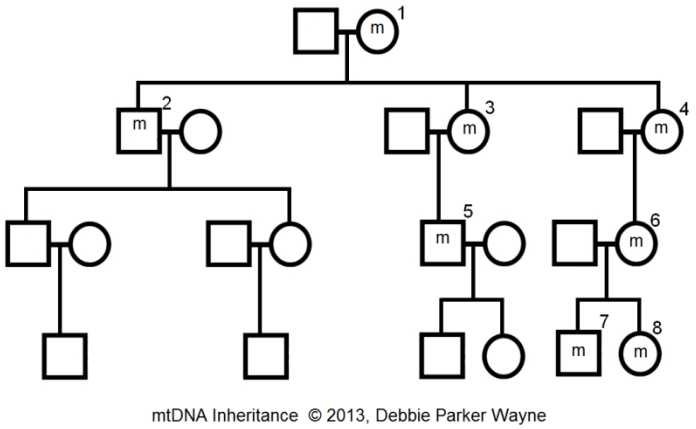
Even the low resolution mtDNA test provides strong evidence for some situations. (1) Was Native American ancestry inherited down the direct maternal line? Native American ancestry can be indicated, but DNA cannot isolate to a specific tribe. (2) Does a line descend from the first or second wife of a man? This requires that the two wives not be descended from a common matrilineal ancestor.

mtDNA INHERITANCE

Mitochondrial DNA is passed from a mother to all of her children. Daughters pass it to the next generation. This inheritance pattern is illustrated in figure 2 where men are depicted as squares and women as circles. The mother (1) passes her mtDNA to her son (2) and daughters (3, 4). The son (2) does not pass his mtDNA to his children. Daughter (3) passes her mtDNA to her son (5), but the son does not pass his mtDNA to his children. Daughter (4) passes her mtDNA to her daughter (6) who passes the mtDNA to her son (7) and daughter (8). Of the descendants shown on the bottom row, only daughter (8) will pass the mtDNA of maternal great-grandmother (1) to the next generation.

The mtDNA passes from mother to child unchanged, unless a mutation occurs. A mutation is a change caused by a copying error when the DNA is duplicated. Mutations occur at random intervals and locations. These mutations are what allow us to trace a family tree using DNA, grouping those with like changes. Mutations in mtDNA rarely occur. Two people with an exact mtDNA match may share a maternal ancestor who is one, five, ten, or more generations from the persons tested (the rate varies widely depending on which research is cited).[1]

Figure 2.



mtDNA Inheritance © 2013, Debbie Parker Wayne

All URLs accessed 13 August 2013.

[1] Brenna M. Henn, Christopher R. Gignoux, Marcus W. Feldman and Joanna L. Mountain, "Characterizing the Time Dependency of Human Mitochondrial DNA Mutation Rate Estimates," *Molecular Biology and Evolution* (2009) 26 (1): 217–230; doi: 10.1093/molbev/msn244; Open Access PDF (http://mbe.oxfordjournalsas.org/content/26/1/217.long). See mutation rate references cited in this paper.

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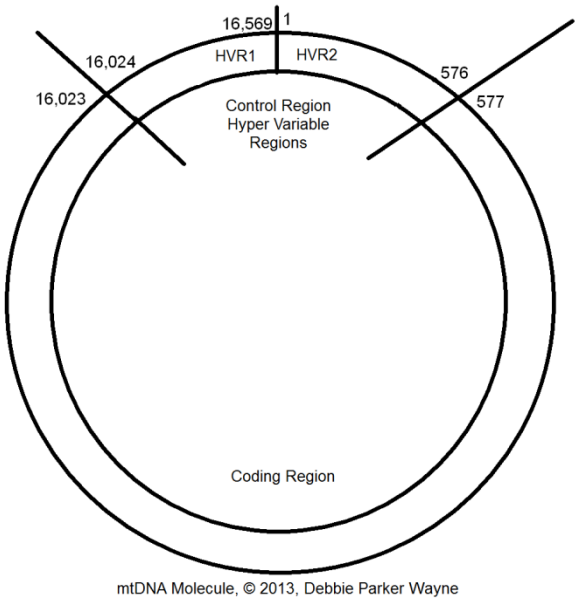
WHAT IS mtDNA?

Mitochondrial DNA is separate from the DNA in our chromosomes and is found outside of the cell nucleus. The mitochondrial molecule is depicted in figure 3. It is a small circular segment of DNA consisting of 16,569 locations (also called base pairs). The segments on either side of the start of the circle change more often than the rest of the molecule. These are called hyper-variable regions or segments (HVR, HVS). The exact start and stop points of hyper-variable regions vary between testing companies even when the same name (HVR1) is used.

The rest of the mtDNA molecule is the coding region and may contain some medically-significant information. Today the Full Mitochondrial Sequence (FMS) test is affordable for many genealogists. It is the most useful for genealogical research, but test results should be analyzed for medical significance, if any, before the results are disclosed. Because relatives have the same DNA, revealing information about your DNA could also reveal information about your descendants, ancestors, and collateral relatives. An informed decision about sharing data can be made only if you know what might be revealed.

> Note: A Full Mitochondrial Sequence should not be confused with a Full Genome Sequence. The phrase Full Genome Sequence or Whole Genome Sequence refers to the sequencing of a person's entire complement of DNA (autosomal, Y, X, and mitochondrial DNA), not just all of the mitochondrial DNA.

Figure 3.



mtDNA TEST RESULTS

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Family Tree DNA has published statements on the likelihood of a common maternal ancestor being found in a specific number of generations. These terms are easier for genealogists to understand than the numbers quoted in scientific literature. See table 1. Even when the full mitochondrial sequence is an exact match there is still a 50% chance your common ancestor is more than five generations back and a 10% chance that ancestor is more than sixteen generations back. For this reason a more complete maternal lineage makes it more likely an mtDNA connection can be identified.

Mitochondrial DNA test results consist of two parts.

1. A haplogroup with a name such as W6c, U5b1d1, or X2a1a. This represents the deep roots of the matrilineal ancestry—the location of ancestors tens of thousands of years ago. Two people in the same haplogroup share a common ancestor, but it might be thousands of years ago.

2. The location and chemical value of the mtDNA **differences** between a reference sequence and the person tested. The chemicals are Adenine, Cytosine, Guanine, Thymine, each usually represented by the first letter of the name—A, C, G, or T. This list of differences should be compared to the test results of others to find potential relatives. As shown in table 1, even with an exact match on a full mtDNA sequence there is a chance the common ancestor is many generations back in time.

   There isn't space for a complete discussion here, but be aware there is more than one reference sequence now in use. These are the Cambridge Reference Sequence (CRS) and the Reconstructed Sapiens Reference Sequence (RSRS). When comparing test results always be sure both sets of results were compared to the same reference sequence. The results will be a list of values similar to

   C146T C150T … 522.1A 522.2C … C16218T A16220D C16320T

   where
   - Numbers represent the location on the mtDNA molecule.
   - The first letter indicates the chemical that would be found in an ancient ancestral sample at the numbered location. This value is implied in a CRS comparison and not explicitly stated.
   - The second letter indicates the chemical found in the tested sample at the numbered location.
   - A deletion at location 16220 is indicated by the "D."
   - Additions are indicated by a period followed by a number and a letter representing the chemical at the location. The list above indicates an added Adenine (A) and Cytosine (C) base following location 522.

An mtDNA exact match will have the same values at the same locations. A one-step difference would have a different chemical at one location, a two-step difference would have a different chemical at two locations, and so on. The more differences there are the further back in time the common ancestor is likely to be found.

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Table 1. mtDNA Regions and Common Ancestor Matches				
Segment name	HVR1	HVR2 and HVR1	HVR3 and HVR2 and HVR1	Full mtDNA Sequence (FMS)
Resolution	Low resolution	Medium resolution	Medium resolution	High resolution
Locations tested	350 to 570 individual locations between positions 15,841 to 16,569	267 to 580 individual locations between positions 1 and 579	About 200 individual locations between positions 340 to 720 Some companies include this in HVR2	Entire mitochondrial molecule, locations 1 to 16,569
Common ancestor computations	50% chance there is a common ancestor within 52 generations for two people with exact matches	50% chance have a common ancestor within 28 generations for two people with exact matches		90% chance you have a common ancestor within 16 generations or a 50% chance you have a common ancestor within 5 generations for two people with exact matches[a]
a. "mtDNA testing (Mitochondrial DNA)," *Family Tree DNA* (http://www.familytreedna.com/mt-dna-compare.aspx). "How many generations back does mitochondrial DNA (mtDNA) testing trace?," FAQ 2139, *Family Tree DNA* "Understanding Results: mtDNA" (http://www.familytreedna.com/faq/answers.aspx?id=10).				

USING mtDNA TEST RESULTS

1. Complete your maternal lineage as far back as possible. Document this to share with mtDNA matches looking for a common ancestor.
    a. List your mitochondrial ancestral names, dates, and geographic origins. For example:
        - Minnie Josephine **McSpadden** (1874 to 1909, Independence County, Arkansas), m. Thomas A. Anderson
        - Temperance C. **Luster** (1839, Tennessee, to 1876, Independence County, Arkansas), m. Thomas A. McSpadden
        - and so on
    b. Create a privatized pedigree chart. For example, list information on your earliest known ancestor down to a great-grandparent or a recent generation that is no longer living. Include geographic locations and dates as above.
2. Join a project. Some surname (Y-DNA) projects allow mtDNA testers to join, some don't. Mitochondrial DNA haplogroup, lineage, and geographic projects welcome mtDNA testers. Ask questions of project administrators who can be very helpful in DNA analysis.
3. To find more potential matches upload data to public databases (MitoSearch.org, mtDNAcommunity.org). Investigate privacy and security policies before uploading data.
4. Search all databases and project lists for matches. Review any ancestral information shared online, and contact the match person for more information. Contact the closest matches

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first as the common ancestor is likely to be more recent. If a common ancestor cannot be identified by name, look for patterns that provide additional research clues such as geographic locales, spouses' names, etc. Matches may not have posted everything they know online. Some people don't respond to contacts, but an attempt should be made. Be patient. The person may respond months after an initial query.

5. Advanced analysis of full mtDNA sequences should be done; this does not apply when only hyper-variable regions are tested. This advanced analysis determines if any medically-relevant information is contained in the coding region so an informed decision about sharing can be made. Remember that other relatives may have this same DNA so be sensitive about what is publicly shared. Consult a medical or genetic professional as needed to fully understand the results.
6. Full mtDNA sequences can be uploaded to the Genbank database for access by academic scientists and citizen scientists. Genbank is administered by the National Center for Biotechnology Information (NCBI), part of the U.S. National Institutes of Health.[2]

RESOURCES

This article is a short introduction to mitochondrial DNA. For information on tests offered by different companies see each vendor's web site and the International Society of Genetic Genealogists (ISOGG) Wiki pages "Mitochondrial DNA tests" and "mtDNA testing comparison chart."[3] For information on the reference samples, haplogroup nomenclature, and a graphic representation of the human mtDNA phylogenetic tree see *PhyloTree*.[4] For a compendium with links to most of the known information related to mtDNA and for information to analyze medical significance see *MitoMap*.[5] Remember to consult a medical or genetic professional as needed to fully understand the medical significance.

Debbie Parker Wayne, CG, CGL, is experienced using DNA analysis, as well as more traditional techniques, for genealogical research in Texas, the South and West. She coordinates the *Practical Genetic Genealogy* course at the Genealogical Research Institute of Pittsburgh and is the Texas State Genealogical Society's DNA Project Director. See http://debbiewayne.com/ for more information.

[2] "GenBank Overview," NIH Genetic Sequence Database, National Center for Biotechnology Information, U.S. National Institutes of Health (http://www.ncbi.nlm.nih.gov/genbank/).
[3] "Mitochondrial DNA tests," Wiki, *ISOGG* (http://www.isogg.org/wiki/Mitochondrial_DNA_tests). "mtDNA testing comparison chart, " Wiki, *ISOGG* (http://www.isogg.org/wiki/MtDNA_testing_comparison_chart).
[4] Mannis van Oven, "PhyloTree.org," *PhyloTree* (http://www.phylotree.org).
[5] "MitoMap: A Human Mitochondrial Genome Database," *MitoMap* (http://mitomap.org/).

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