

Targeted DNA Testing



DNA testing can provide clues to solving specific genealogical mysteries but only if the right person takes the right kind of test. While you probably should test yourself, you may not be the right person to connect to the desired ancestor.

Making the right choices requires both knowledge of the different test types and of the tree of the person to whose ancestor you are trying to connect. Maybe that's you, maybe it's a family member, or friend. Regardless, to make sure you are using the best test on the best

person, let's take a step back and review the basics of DNA testing.

Types of DNA tested

- **Autosomal DNA** - Inherited from both parents, a set of 22 autosomes from each, made up of the recombined DNA inherited from their parents.
- **X-DNA** - Males inherit one X chromosome from their mothers along with a Y chromosome from their fathers. Females inherit one X chromosome from each parent. A complex inheritance pattern makes using X-DNA for genealogy more suitable for advanced users but can be useful in specific situations.
- **Y-DNA** - Males inherit from their fathers. Only sons inherit and pass to their sons.
- **Mitochondrial DNA (mtDNA)** - Both males and females inherit from their mothers. Only daughters pass to their children.

Y-DNA Facts

The first type of test available direct to consumers. Comparing Y-DNA results can confirm paternal relationships and point to a surname. Deeper analysis may reveal how closely (in generations) testers are related and how the paternal line migrated. It cannot be used as a paternity test, because it can only confirm that two men share a paternal ancestor, not positively identify which ancestor.

- **Only males inherit from father**
 - Genetic genealogy uses non-recombining portion.
 - Used to match against database to determine common paternal ancestors.
 - Does not give ethnic percentages; provides haplogroup corresponding with migratory path of paternal line.
- **Types of mutations**
 - STRs:
 - Short Tandem Repeats.
 - Counts the number of times nucleotides repeat at specific locations.
 - Traditionally used for “close” matching to determine a common paternal ancestor.
 - Number of differences between two testers known as “Genetic Distance,” a measure of how closely testers may be related.
 - SNPs:
 - Single Nucleotide Polymorphisms.
 - Change at a single location.
 - Defines haplogroups and subclades showing position on Y-DNA haplotree.
 - Corresponds to migratory paths.
- **Y-DNA tests**
 - 12-, 25-, 37-, 67-, and 111-marker STRs.
(FTDNA only offers 37- and 111- marker tests on their website. 12- and 67 - marker tests are available through Customer Service. 25 – and 67- marker tests are displayed but no longer available.)
 - Individual SNPs.
 - SNP Packs - Curated groups of SNPs from a specific haplogroup or subclade.
 - Next Generation Sequencing (NGS) tests - Exploratory tests that reveal variations in the Y chromosome unique to tester and his family lines.
 - Big Y-500 (FamilyTreeDNA - No longer sold as of 1 Nov. 2018).
 - Big Y-700 (FamilyTreeDNA - After 1 Nov 2018).

mtDNA Facts

- Cytoplasmic organelles involved in cellular energy production.



- Only 16,569 base pairs compared to more than 59 million base pairs in Y-DNA.
- Does not recombine (mix together) with DNA from other ancestors.
- Passes from mother to child so can be used to trace direct maternal line.
- Changes (mutates) slowly since many copies exist in each cell.
- Traces maternal migrations over a deeper time frame than Y-DNA traces paternal migrations, so matches may be much more distant.
- Does not give ethnic percentages but provides haplogroup corresponding with direct maternal migratory path.
- Compares testers against a reference genome.

Autosomal DNA Facts

- DNA inherited from both parents, 50% from each parent, made up of the randomly recombined DNA each parent inherited from his or her parents.
- Reveals matches sharing ancestors in approximately last 5-6 generations, gives an estimate of the tester's ancestral origins inherited from ancestors along all lines.
- Both men and women can test as everyone has autosomal DNA.

X-DNA Facts

- A man inherits one X chromosome from his mother, which is usually randomly recombined from her mother's X chromosomes but not always.
- A woman inherits one X chromosome from each of her parents.
 - Whole X chromosome from father, which he got from his mother.
 - X chromosome from mother is recombined from her two X chromosomes, not necessarily in equal proportions.
- Recombination makes tracing inheritance tricky.
- Tested as part of autosomal DNA despite being one of the sex chromosomes.

Challenges of each form of testing:

- Y-DNA reveals nothing about female ancestors.

- Y-DNA traces only a direct paternal line but cannot distinguish between brothers within the past two or three hundred years.
- mtDNA reveals nothing about male ancestors, even though men can take the test.
- mtDNA traces only the direct maternal line and changes so slowly that determining when matches share a common maternal ancestor can be difficult.
- Autosomal DNA divides in half each generation so you can only go back about five to six generations with confidence.
 - As you go back, the amount of DNA available to inherit from a specific ancestor decreases.
 - The chances of not inheriting any from a specific ancestor increases with each successive generation.
 - Recombination affects the amounts inherited.
 - Siblings inherit varying combinations of DNA from ancestors unless they're identical twins or triplets.
- X-DNA has such a specific inheritance pattern it is only useful for some of your ancestors.
 - Men only inherit X-DNA from their mothers, so it only connects to maternal ancestors.
 - Women inherit X-DNA from both parents, but there are still specific lines with which it will not connect.

Strategy: Target Testing

- To which ancestor do you want to connect?
- Is the ancestor male or female?
- How far back is that person in generations?
- Did that ancestor have children? Siblings?
- Did that ancestor's parents have siblings?
- Is there a son of a son of a son (etc.) living, accessible, and perhaps willing to test?
- Is there the son or daughter of the daughter of the daughter of the daughter (etc.) living, accessible, and perhaps willing to test?
- If you do not know any, are any of your matches descendants of that ancestor? If so, are they direct paternal descendants? Direct maternal descendants?

Summary

While it may take some research, you may be able to confirm ancestral lines by testing specific cousins, aunts, uncles, etc. Your own autosomal matches may provide you with candidates for testing, or they may have already done the testing, and all you need to do is communicate with them and share information. As more people test into the databases, you never know when that key person is going to show up in your match list. Or you may need to reach out to family members you've not contacted in years or ever.

Resources

Biology Dictionary: <https://biologydictionary.net/dna-sequencing/>

Davis, C., Sager, M., Runfeldt, G., Greenspan, E., Bormans, A., Greenspan, B., & Bormans, C.,

Big Y-700 [White paper] 2019: <https://blog.familytreedna.com/big-y-700-white-paper/>

DNAeXplained – Genetic Genealogy blog by Roberta Estes <http://dna-explained.com>

DNA and Family Tree Research blog by Maurice Gleeson, especially downloads page

<http://dnaandfamilytreeresearch.blogspot.com/p/presentations-downloads.html>

FamilyTreeDNA Learning Center: <https://learn.familytreedna.com/>

FamilyTreeDNA Public Y-DNA Haplotree: <https://www.familytreedna.com/public/y-dna-haplotree>

International Society of Genetic Genealogy (ISOGG) Wiki <https://isogg.org/wiki>

McDonald, Dr. Iain. Recent human genetic anthropology:

<http://www.jb.man.ac.uk/~mcdonald/genetics.html>

National Center for Biotechnology Information <http://www.ncbi.nlm.nih.gov/>

Vance, David – DNA Concepts for Genealogy videos:

-Y-DNA part 1 <https://www.youtube.com/watch?v=RqSN1A44IYU&t=16s>

- Y-DNA part 2 <https://www.youtube.com/watch?v=mhBYXD7Xufl&t=355s>

- Y-DNA part 3 <https://www.youtube.com/watch?v=03hRXVg9i1k&t=4s>

Vance, David The Genealogist's Guide to Y-DNA Testing for Genetic Genealogy: Self-published, 2020

Y-DNA Warehouse: <https://ydna-warehouse.org/> and <https://ydna-warehouse.org/statistics.html>