



## Behind the Scenes at FamilyTreeDNA

### Janine Cloud – Group Projects Manager

#### Introduction

In April 2000, FamilyTreeDNA became the first company to offer direct-to-consumer DNA testing with a 12 marker Y-DNA STR test.

Company founder Bennett Greenspan, an entrepreneur and life-long genealogy enthusiast, leveraged the work of Dr. Michael Hammer of the University of Arizona to connect two men whom he thought should be related but could not prove.

Greenspan had read the 1999 academic paper to which Hammer contributed that reported using Y-chromosomal DNA to establish the genetic signature of the Jewish priesthood lineage. So, he contacted Hammer and eventually convinced him to do Y-DNA testing on the two men he thought should be related. Hammer convinced Greenspan to do a proof of concept to demonstrate that the technique would work in broader applications. The match was proven, and a genealogical revolution was born.

With assistance from Max Blankfeld, Greenspan's partner in a previous business endeavor, FamilyTreeDNA's headquarters were established in Houston, Texas, and for several years, testing was performed at the University of Arizona.

Over the past 21 years, the company added additional STRs, mitochondrial DNA, and autosomal DNA along with a selection of additional tests that have been phased out as their necessity waned. FamilyTreeDNA remains the only major DNA testing company to test all types of DNA along with advanced Y-DNA products.

In January 2021, FamilyTreeDNA's parent company, Gene by Gene, merged with Australian pharmacogenetics company myDNA to form MYDNA, Inc., which now oversees the operations of the various divisions and operates the in-house CAP, CLIA, and AABB accredited Genomics Research Center laboratory in Houston. Once a tester's sample arrives at the office, it never leaves the building. In addition, testers' unused samples remain in storage for at least 25 years for upgrades and additional tests.

A large portion of FamilyTreeDNA's growth, especially early growth can be attributed to Group Projects and the volunteer administrators who bought tests, recruited testers, and managed results. There are now over 10,000 projects with over 7,000 volunteers assisting with surname, geographical, haplogroup, mitochondrial, and autosomal research.



## Basics about DNA testing and FamilyTreeDNA's product catalog

### 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 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** - both males and females inherit from their mothers. Only daughters pass to their children.

### Y-DNA Facts – only genetic males can test

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, however, 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 haplotree
- Corresponds to migratory paths
- **Y-DNA Tests**
  - 12-, 37-, and 111-marker STRs
  - 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 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 – anyone can test**

- 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.
- 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 – anyone can test**

- 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.

### **X-DNA Facts – anyone can test**

- 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.
  - The amount of DNA available to inherit from a specific ancestor decreases and the chances of not inheriting any from a specific ancestor increases with each generation you go back.
  - 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: Targeted 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?

## **Resources**

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

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

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

FamilyTreeDNA Help Center: <https://help.familytreedna.com/hc/en-us>.

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

Gleeson, Dr. Maurice. DNA and Family Tree Research blog

<http://dnaandfamilytreeresearch.blogspot.com/>

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=mhBYXD7XufI&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>