

# Genetic Genealogy: DNA Testing for Family Researchers

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## INTRODUCTION:

Genetic genealogy – *DNA testing for family history research* – has become popular. DNA is found in every cell of the body. As parents' donate genes to their children, most parental DNA is shuffled, halved and diluted with each generation; but some portions of DNA escape scrambling and remain robustly constant over many generations. Different tests of both stable and ever-changing DNA can help prove ancient kinship between individuals. DNA won't solve all genealogy mysteries, but can solve some types – as well as determine ethnicity & find new cousins.

## DNA Testing Basics:

- DNA collection is painless & self-administered – no needles, just spit or swab your inner cheek & mail to the lab.
- Costs range from \$59 to over \$700 depending on the type of DNA test and number of DNA markers tested.
- Results are returned in weeks and include numerical values (alleles) for each DNA marker and an interpretation.
- Your DNA results or "Genotype" are posted anonymously online at the lab's website & you're told of matches.
- Additional tests can sometimes be ordered later from your original sample stored at the DNA testing lab.
- Except for ethnicity estimates, DNA results must be compared to tests of others who are suspected relatives.
- The closer that two people's results are to one another, the more closely related are the two individuals.
- There are 3 types of DNA tests: Y-Chromosome DNA, Mitochondrial DNA and Autosomal (non-XY Chromosome) DNA; each test type reveals different ancestral insights and should be chosen to meet a specific research need.
- DNA testing can have privacy implications: despite labs' best efforts, it is possible for law enforcement or others to gain access to your DNA test results. Think carefully about testing if you (or a family member) are a criminal.

## Things Genealogical DNA Testing Cannot Do:

- Most DNA tests are not a personal DNA fingerprint; close kin can have identical results (family genotype).
- One DNA test type is nearly a personal DNA fingerprint; autosomal-DNA helped identify the Golden State Killer.
- DNA test labs do not reveal genetic disorders or foretell disease for technical and legal reasons, although
- Third party services linked DNA results to thousands of medical studies by web sites for as little as \$5.
- DNA is contributory evidence; it tells whether 2 people share an ancestor, but **not** exactly who or when.
- DNA can disprove a genetic relationship; vast DNA test differences almost always mean no relationship.

## THREE BASIC TYPES OF DNA TESTING:

- **Stable DNA** Y-Chromosome DNA (Y-DNA) reveals a man's father's father's line through the Y (male) chromosome. Only men have Y-DNA and can contribute cells for Y-DNA testing. Female genealogists can ask direct line male relatives (brother, father, or paternal uncle or nephews) to contribute DNA on their behalf. Y-DNA tests typically look at dozens to hundreds of marker values. Y-DNA is useful in the genealogical time frame (the last 800 years). When two living test subjects' Y-DNA tests differ by 2 or fewer marker values, then the comparison is considered genealogically relevant. Labs maintain confidential masked results and build free, online Y-DNA test result databases that can be searched to compare your paternal line's Y-DNA results to other families.
- **Very Stable DNA** Mitochondrial DNA (mt-DNA) reveals any person's mother's mother's line through the mother's egg without any paternal contribution. Men and women both receive their mother's mt-DNA, and may donate cells for mt-DNA testing. But only women pass their mt-DNA to later generations. Like Y-DNA, mt-DNA tests typically look at dozens to hundreds of marker values. Unlike Y-DNA, only exact mt-DNA matches are considered relevant – and even then, may only point to a general relationship. mt-DNA is useful in an ancient time frame (10,000-plus years). A few online mt-DNA databases exist for comparing your family's results to others'. Because of the extremely slow mutation of mt-DNA, it is considered an anthropologic, rather than genealogic test.
- **Dynamic DNA** Autosomal DNA (at-DNA) tests hundreds of thousands of markers from chromosomes other than X or Y. An at-DNA test costs as little as \$59 on sale. at-DNA tests broadly reflect your entire pedigree. As a stand-alone test, at-DNA can indicate the proportion of different ethnicities and unique regional origins of a person's collective ancestry – for instance, 75% Irish or 5% Native American. at-DNA most accurately reflects the last 100 to 150 years (4 to 6 generations). This is because the at-DNA of each successive generation is diluted by half, as parents each randomly donate half of their genetic heritage to each child, while the other half is lost. Your ethnicity estimates rarely agree with your records research – or your siblings' ethnicity estimates - because of random dilution effects. Moreover, your ethnicity estimate often varies greatly between labs, because each lab uses DNA of its own proprietary "reference populations" to define each of its "pure" ethnic sub-types (e.g., "the Irish").

at-DNA is mostly useful in determining if two living people are related. Testing labs like AncestryDNA, 23andMe and FamilyTreeDNA each compare results among test subjects and let you know whenever an at-DNA overlap meets the lab's threshold to notify you by email. Overlap is typically reported as a percentage and/or as the number of centimorgans (cM) of shared DNA. The amount of at-DNA overlap of two test subjects can reveal a "recent" shared relative, but can't tell the exact relationship (see chart). More precise relationship estimates can be achieved by "triangulation": testing more of your family members whose unique genetic inheritance (their halving outcomes) may better reveal relationship insights. Additional services are offered by Ancestry.com and MyHeritage.com who attempt to link family trees to DNA test results, though a subscription may be required.

### **Medical Insights of at-DNA Testing**

Even though genes are analyzed, at-DNA testing labs are limited by the FDA from providing extensive medical interpretations of their findings. However, you can download your DNA results from some labs and then upload to third party web sites like Promethease.com who offer possible health implications based on your at-DNA test data.

### **Criminal Insights into at-DNA Testing**

US law enforcement already uses Familial DNA searches of criminal DNA databanks (sometimes commercial labs) to identify "near-match" leads. New European laws allow law enforcement officials to analyze trace DNA samples for genetic markers that help determine hair color, eye color, skin color, age, and "biogeographical ancestry."

### **Advice about Autosomal DNA Testing**

- Population size matters: use one of the "Big 3" DNA testing labs: Ancestry, FamilyTreeDNA, or 23andMe.
- Autosomal DNA is a good test to begin with – it will answer many questions and open doors to new relatives.
- Use AncestryDNA \$59- \$99 for autosomal DNA testing, then upload your results to FamilyTreeDNA & others.
- FamilyTreeDNA is the only company to broadly test mt-DNA and Y-DNA to answers specific questions.
- Test oldest relatives first. Results of grandparents, parents, cousins, uncles & aunts will help sort later matches.
- Ethnicity estimates are interesting, but are often perplexing; they randomly emphasize only recent ancestry.
- Third party tools can help you. These include GEDMatch.com, DNAGedcom.com and Promethease.com.

### **Limitations and Restrictions Inherent in all DNA testing:**

- The deep history tests only reveal the maternal (mt-DNA) & paternal (Y-DNA) "edges" of a pedigree.
- Newer at-DNA tests like AncestryDNA work best in the recent era – before too much "gene halving" & dilution.
- All DNA tests are contributory evidence that can only suggest a relationship, not absolutely prove one.
- DNA won't substitute for traditional research; you should consider DNA as just another piece of evidence.
- DNA statistics can calculate a likely *Most Recent Common Ancestor* (MRCA) of two DNA test subjects, but this relatedness estimate only tells you if enough DNA overlap exists to further explore a possible relationship.
- DNA tells "**whether**" a relationship exists, but it cannot reveal the exact "**who**" or "**when**" of the relationship;

### **STRATEGIES FOR DNA TESTING**

DNA cannot solve every research question. The key to DNA testing is to pick a test to answer a specific answer:

- **First**, does your research question involve a branch of the pedigree that can be revealed by DNA testing. For example, does the question deal with your father's father's line (Y-DNA), your mother's mother's line (mt-DNA) or a question of ethnic origin or finding lost branches by comparing test results (at-DNA)?
- **Second**, do you have sufficient traditional research to propose a theory that DNA evidence can help prove?
- **Third**, do living suspected relatives exist to test/compare results to the proposed answer to your question?
- **Fourth**, will these suspected relative agree to the inconvenience, cost and disclosure of confidential results?

### **CONCLUSION**

DNA testing is a powerful tool capable of contributing to family history research when your question is either:

- **Very general** – Autosomal DNA test to answer, "what's my ethnicity" or "to whom might I be related", or
- **Very specific** - Y-DNA or mt-DNA tests to answer certain questions of maternal or paternal lineage.

Because of the cost, it is helpful to develop a DNA testing strategy. Even at their best, DNA test results are shrouded in "probability" rather than "certainty". So, DNA test results are usually contributory evidence and not absolute proof. As such, DNA test results should be considered just another fact to be considered, weighed and evaluated in the light of all evidence available on any question of kinship in your research.

Because DNA test results are on the verge of producing personally identifying "fingerprint-level" identification, consider the security issues and read the privacy policies of your DNA lab and any other site to which you upload your test results.

# DNA Research Case Studies

## mt-DNA Case Study - Identifying the Remains of the Russian Royal Family

**Question:** In 1990, Russian scientists unearthed the charred remains of what were believed to be the Russian Royal Family who were executed in 1917 at the time of the Russian Revolution.

**Answer:** With the help of sample mt-DNA provided by Prince Philip, who shares the same maternal line from Queen Victoria with the Russian Royal family, the DNA tests identified the bodies of the Romanovs, the Russian imperial family. The mt-DNA results of all but two of the bodies were identical. By other means, these two were found to be the family doctor and a maid. In a related test, tissue from an old New York hospital biopsy of a deceased woman who had claimed to be the escaped Princess Anastasia Romanov was also compared. Despite years of claims to the contrary, she was proven to be unrelated to the Romanovs.

## mt-DNA Case Study - To Which Wife do these Daughters Belong?

**Question:** A male ancestor had more than one wife. Can we match several daughters to their correct mothers?

**Answer:** Find and test mt-DNA of maternal descendants of all the daughters; exact matches establish a maternal family lineage. If no direct female descendants survive, try “climbing the maternal pedigree” until you find an ancestor with a living direct female descendant.

## at-DNA Ethnicity Estimate Case Study – An AncestryDNA email says we’re related. Is the overlap meaningful?

**Question:** An email from AncestryDNA test “cousin” with 75 cM shared at-DNA with me asks if/how we’re related?

**Answer:** Overlapping DNA greater than 30 cM (0.4%) makes a shared recent ancestor very probable. Use the web site [www.DNAPainter.com/tools/sharedcmv3](http://www.DNAPainter.com/tools/sharedcmv3) to determine that 75 cM (1.0%) shared DNA has 39 possible relationships with two most likely: **3<sup>rd</sup> Cousin** or **2<sup>nd</sup> Cousin Twice Removed**. Search for shared surnames and compare online family trees to find we’re actually: **3<sup>rd</sup> Cousins-Once Removed** (which statistically averages 48 cM).

## Autosomal DNA Ethnicity Estimate Case Study - Proving Native American Ancestry

**Question:** You have always heard that your family has a Native American ancestor “somewhere”.

**Answer:** If it’s your father’s father’s line OR the mother’s mother’s line uses a test from: [FamilyTreeDNA](http://FamilyTreeDNA). If it you want % mix of primal ancestral groups (including Native American), use: [AncestryDNA](http://AncestryDNA).

## Y-DNA Case Study - Thomas Jefferson & Sally Hemming

According to a Nov. 5, 1998 article in Nature Magazine, DNA tests “proved” that Thomas Jefferson had a liaison with his slave, Sally Hemming. The article stated that “Jefferson sired the last son, Eston through that union, and probably 6 others.” Conclusions were based on Y-DNA samples of direct male descendants of Sally’s eldest son and descendants of a Jefferson relative.

**Lesson One:** since there were no surviving male descendants from Thomas Jefferson's marriage to be tested, climb up the paternal pedigree until you find an even older ancestor who has a living male descendant. In this case the researchers climbed-up Jefferson’s pedigree to his paternal grandfather and then tested a living male descendant of Jefferson’s paternal uncle, Field Jefferson.

**Lesson Two:** Don’t leap to conclusions. These results were reported by Pulitzer Prize winning author Phillip Ellis, but he jumped to his conclusion, because DNA proof is not definitive. A later issue of Nature retracted the original conclusions because at least 25 male Jeffersons (presumably with near-identical Y-DNA) were living at the time of Sally’s conception of Eston, including eight adult males who frequently lived at Monticello. Each of them also is also a potential father of Sally’s child, along with, then, 64 year old Thomas Jefferson. Additionally, there is always a possibility of later DNA mingling from the Jefferson line with the Hemming line during the intervening 200 years.

**Lesson Three:** Better traditional documentation might have allowed this DNA test to become a final confirmatory proof of this “Thomas Jefferson/Sally Hemming Liaison” hypothesis. So, know what DNA can and cannot do. As a stand-alone test (even when combined with family tales), DNA is merely suggestive of proof. On the other hand, badly mismatched DNA would be definite disproof – as was the case for the 6 other children of Sally Hemming, once the results were more carefully examined.

# MAJOR GENEOLOGICAL DNA TEST TYPES

DESCRIPTION	mt-DNA	Y-DNA	at-DNA
DNA Source	Mitochondria	Y Chromosome	Autosomal Chromosomes (44 of 46 chromosomes; all except X & Y)
Location	Cell Cytoplasm	Cell Nucleus	Cell Nucleus
<b>TEST SUBJECTS</b>			
Who can be tested?	Women or Men	Men only	Women or Men
<b>TIME FRAME</b>			
Does DNA get Shuffled?	No	No	Yes, each generation
DNA Stability	Extremely Stable	Stable	Unstable (shuffling and halving each generation)
Looks how far back?	1000s of generations	100s of generations	5-6 generations
Ave. Mutation Rate?	Once every 10,000 years	Once every 800 years	Inconsequential (minimal mutation is overshadowed by shuffling)
Test Type	Anthropological	Genealogical	Genealogical
<b>DNA USES</b>			
Primary Use	Matching to others with a shared ancestor	Matching to others with a shared ancestor	Matching to others with a shared ancestor
Secondary Uses	Migratory Insights	Migratory Insights	Ethnicity Estimates (%) Migratory Insights
<b>LABS &amp; COSTS</b>			
Best Lab	FamilyTreeDNA	FamilyTreeDNA	AncestryDNA
Cost (# of Markers)	\$89 (2 regions) to \$149 (all regions)	\$169 (37 markers) to \$649 (500 markers)	\$59-\$99 (> 700,000 markers)
<b>ACCURACY &amp; CERTAINTY</b>			
Overlap required to demonstrate Kinship	100% (no mismatched DNA markers)	98-99% (2 or fewer mismatched DNA markers)	Anything > 0.5% (as little as 16 cM)
Match Certainty	MRCA Estimated	MRCA Estimated	MRCA Estimated
Ethnicity Accuracy	Moderate	Moderate	Slightly Better

Ancestry.com "potential DNA match"

Home Trees Search **DNA** Help Extras

AncestryDNA Home > Member Matches for David Bradford **← 1**

Member since 2016, last logged in Aug 4, 2017

**Name Obscured for Privacy** **Send Message**

Member since 2016, last logged in Aug 4, 2017

**Predicted relationship: 4th Cousins**

Possible range: 4th - 6th cousins (What does this mean?)

Confidence: Extremely High **1** **← 2**

**Amount of Shared DNA**

75 centimorgans shared across 5 DNA segments

What does this mean?

4 **←** 3 **←** 2 **←** 1

Confidence Score	Approximate amount of shared centimorgans	Likelihood of a single recent common ancestor
Extremely High	More than 60	Virtually 100%
High	45—60	About 99%
Very High	30—45	About 95%
High	16—30	Above 50%
Good	6—16	15—50%
Moderate		

