

**DNA Science:**  
**Combining Genetic and Traditional Genealogy**

By  
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**FORWARD:**

This article provides a basic tutorial on the three main types of DNA testing that are used to aid genealogical inquiries. It is especially tailored to individuals who are curious about their ancestral lines and are considering DNA testing to support their traditional, document-based research. This paper describes the three current major genetic genealogy testing approaches, their differences, and their usefulness in breaking through some of the brick walls met in conventional genealogy.

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Perhaps because the United States is for the most part “A Nation of Immigrants,” the American public is eager to learn about their family histories and is fascinated to be able to trace their ancestral roots to other places in the world. Genealogy has become a wildly popular pastime, second only to gardening among American hobbies. Genealogy comes in second place among the most visited categories of internet websites. Several television programs trace the ancestry of actors, singers, athletes, and other public figures. This billion-dollar industry is supported by millions of volunteers who now back their traditional research with genetic studies based on data derived from internet-ordered, mailed-in genetic test samples.

DNA testing has several uses. Various types of DNA assessment look at different sources of DNA molecules, and no single test serves all purposes well. In questions of legal parentage, courts generally accept DNA evidence above all else. Medical research is heavily invested in seeking genetic ties to various conditions. Criminals are often identified through DNA evidence. DNA has been used to solve historical mysteries including identifying the remains of the fallen in past and long-dead royalty. As for DNA testing specifically related to genealogy, there are three basic types, Y-Chromosome DNA, mitochondrial DNA, and autosomal DNA.

Y-Chromosome DNA (Y-DNA) tests trace only a male line – from a male to his biological father to his paternal grandfather, his paternal great-grandfather, etc., through hundreds and thousands of years. In many cultures a surname follows the paternal line, and because Y-DNA passes virtually unchanged from father to son, Y-chromosome tests are very useful in tracing a surname back through time, possibly to times before surnames even came into use, over forty generations or more ago. Y-DNA usually passes unchanged from father to son, but in any specific father-to-son transfer of Y-DNA, one or two very small but measurable changes may occur. Between any two men who share nearly identical Y-DNA, the number and locations of even a few small changes, that appear at specific sites of a Y-DNA marker string,<sup>1</sup> can lead to an estimate of the time when their Most Recent Common Ancestor (MRCA) was alive. It is important to note consistencies within surnames are frequently disrupted by adoptions, name changes, and

other non-paternity events. Test participants should be cautioned that some men have been surprised and even shocked to discover their Y-DNA points to a very different genetic surname from their legal surname.

The second basic type of genealogy-related DNA testing is the mitochondrial DNA (mtDNA) test, which traces the female line from either a male or female to his/her mother to the maternal grandmother, then to the maternal great-grandmother, etc. MtDNA changes only very slowly over thousands of years, and while the results are of great interest to anthropologists and geneticists, they usually are of little use to genealogy hobbyists. In addition, because females often change their surnames upon marriage, the surname associated with the mtDNA result generally changes each generation, complicating the process of tracing the maternal line through traditional pedigrees.

The third, and most recent, significant type of genealogy-related DNA test is the autosomal test, which looks for close relatives along not only male and female lines but also all ancestors in between, such as the mother's father or the father's mother's mother. Humans have 22 pairs of autosomes and one pair of sex chromosomes (the X and, for males, the Y chromosomes).<sup>2</sup> Each person receives on average half of each of their biological parent's DNA, an average of a quarter of each grandparent's DNA, about an eighth of each great-grandparent's DNA, and about a sixteenth of each great-great-grandparent's DNA. Autosomal tests are excellent for identifying first, second, and third cousins, but beyond about fifth cousins, genetic connections, regardless of surname, become too weak to be greatly useful. Nonetheless, if an individual has many dozens or hundreds of distant cousins who have contributed DNA to a database, some of them will likely be identified as sharing unique DNA segments with one another. It is also helpful for an individual to encourage living relatives such as parents, siblings and known close relatives including first and second cousins to participate in genetic testing because it is likely that each has inherited some autosomal DNA, which is unique to that particular individual but still inherited from the same ancestors. Thus, siblings or close cousins may help identify overlapping yet distinct groups of third, fourth and distant cousins that may aid in substantiating particular ancestral lines. This is known as 'casting a wide net.'<sup>3</sup> It is important to note that autosomal tests, like Y-DNA tests, only compare results with other results already in that company's database.

A potential fourth test involves the X-Chromosome, but this will be a far more complex process with a unique pattern of inheritance.<sup>4</sup>

The first three types of tests provide some indication of ethnicity, but the results vary widely by test and by the company reporting them. In particular, mtDNA can only give a rough indication of the place of origin of the female line with low resolution, and often over several thousands of years. All three types of tests have some transferability to websites such as GEDMatch that analyze data for free<sup>5</sup>.

## WHICH DNA TEST SHOULD I TAKE?

Men can be tested directly using any or all of the three approaches, but since women do not carry the Y-Chromosome, they can only directly participate in mtDNA and autosomal DNA tests. However, if a genetic link can be verified to a male relative in the direct paternal line to an ancestral surname group of interest (e.g., a father, brother, nephew, or male cousin bearing the surname of interest), then that male's Y-DNA can be used to trace the genetic ancestry of their mutual male ancestors. In all cases, the results are useful only in relation to the results of previous test samples already in the company's database.

A quick internet search reveals many companies offer DNA testing. When a test is conducted for genealogy and not for legal or medical purposes, the three most popular options in the United States are 23andMe, Ancestry.com, and Family Tree DNA. The companies have slightly different procedures, but all three companies accept payment from new test participants on line, and then send the individual a kit to collect saliva samples. After the test samples are returned to the companies, results and matches to likely genetic relatives are available via the internet some weeks later. To protect privacy, each company posts an individual's results to a private web site that can be accessed with a user name and password. Each company provides suggestions about how the results match those of previous test participants, and may send a message when future matches appear in their databases. The companies provide suggestions as to the strength of each match, which give hints at how far back the shared Most Recent Common Ancestor (MRCA) lived. Both participants must authorize the release of their personal data before the companies will enable the newly identified relatives to contact each other.

The major genetic genealogy testing companies have interesting and instructive online tutorials regarding the DNA testing process – what their tests can and cannot do, and how the results can be used in conjunction with traditional family trees. Errors involved in the testing process are minimal – probably less than 1% - and the companies are very good at answering customer questions. Autosomal “cousin finding” tests cost about the same at all three major genealogical DNA testing companies. The DNA testing company 23andMe<sup>6</sup> formerly provided health-related genetic reports to individual customers, but in November 2013 the U.S. Food and Drug Administration forbade them from further reporting such results. More recently the company has shifted emphasis to ancestry-related genetic reports.<sup>7</sup>

In contrast, for decades Ancestry.com<sup>8</sup> has focused on traditional genealogy research and the creation, documentation, and sharing of online family trees. In the last few years the company has also offered genealogy-related DNA testing. Ancestry.com offers an extremely useful feature: When a test participant is linked genetically to another person, and both participants have posted traditional family trees on the ancestry.com website, then the company's computer algorithms search automatically for parallel trees and common ancestors. Over sixty million overlapping “trees” containing billions of people are already posted on ancestry.com. This is an amazing method to find both living relatives and deceased ancestors. Even adults who were adopted as young children and

who have no idea of the identity of their biological relatives or their genetic heritage might discover their genetic third or fourth cousins. Then they might conduct interviews to find first cousins and perhaps even siblings and parents. It is emphasized that traditional pedigree “trees” must be publicly posted on the Ancestry website for such genetic contacts to be made and both parties must agree to share information. While ancestry.com charges a yearly membership fee to access most of the record collections on Ancestry, creating and posting your online family tree is completely free. Even without an Ancestry subscription, AncestryDNA results include a dynamic list of DNA member matches to help find new relatives to about fifth cousins and the test participant’s genetic ethnicity report. Occasionally, AncestryDNA may find seventh and eighth cousins that agree with well-documented pedigree lines.

As noted above, both autosomal and Y-DNA analysis, the two types of DNA testing that are most useful in genetic genealogy, compare results with other results that are already in that company’s database. Consequently, the larger the database, the greater the likelihood of finding genetic relatives. The largest database in the field of genetic genealogy, with almost three quarters of a million records, is that of Family Tree DNA (FTDNA).<sup>9</sup> FTDNA is especially noted for their special interest groups that include those based on surnames, haplogroups, location, and Y-DNA. Men looking for their father’s father’s father’s paternal line should first check the FTDNA website to see if their paternal surname is represented. If so, the next step would be to join that surname group, and submit a DNA sample through them. Not only do these special interest groups have volunteer administrators who can provide advice, but also by purchasing tests through a surname group, new test participants receive a price discount. It must be noted, however, that not all of these volunteer administrators are uniformly knowledgeable and helpful. At FTDNA, participants may choose the number of Y-DNA markers to be tested, usually 37, 67, or 111 markers. A higher level of testing provides more precise results, but costs more. Since many samples have only been tested at 37 or fewer markers, the results of those potential matches can only be compared at those lower levels. For additional fees, the original sample can usually be tested for additional markers and types of tests<sup>10</sup>, and at FTDNA autosomal and mtDNA analysis can also be performed on the same sample. When FTDNA conducts autosomal testing, both men and women can, at no charge, join additional surname groups that represent other lines of known ancestors. When the regions or groups from which ancestors came are known, many surname project participants also join those FTDNA regional projects, again at no additional cost. While many surname project participants later choose to pay for more in-depth testing of their original sample, FTDNA only charges a one-time fee for each type of test, with no additional subscription fee.

FTDNA encourages free transfers of raw autosomal DNA results from ancestry.com and 23andMe. The twenty closest autosomal matches are provided at no cost, with full release of all matches provided at low cost. To build their own databases, other websites including Y-Search and GEDMatch<sup>11</sup> also offer free transfer of DNA results from FTDNA, ancestry.com, and 23andMe.

## FINDING DISTANT COUSINS CAN BREAK THROUGH BRICK WALLS

As noted above, for genealogical purposes DNA test results are meaningless until they are compared with other people's results. Just as genetic siblings share biological parents, so also first cousins share a set of grandparents, second cousins share a set of great grandparents, and third cousins share a set of great-great grandparents. For example, once you find a genetic second cousin, comparison of family trees and DNA results should reveal which of their great-grand parents are also your direct, genetic ancestors. Further research along these lines, combined with discussions and other records, might reveal a first cousin or even a parent or sibling. Participation in genetic testing by these suspected close relatives would confirm the relationships. Y-DNA testing is even easier to interpret because genetic matches usually quickly reveal surnames of paternal lines, and newly tested males have a good chance that their Y-DNA will match an existing pedigree line.

This is one of the primary features of genetic DNA testing: to help people break through "brick walls" in traditional document-based family trees. In addition, a growing application for genetic DNA testing is to help adoptees and descendants of adoptees discover their actual genetic parentage.

Each of the three major testing companies mentioned in this paper indicates the degree of each genetic match and provides a method for these apparent genetic relatives to make contact, but only if both parties have chosen to be able to communicate with each other. Because of their different purposes, and because matches may differ, many participants test with more than one testing company. New cousins can be discovered. Once close genetic matches are discovered, participants can choose to share their own family trees with others who have been tested, a process that may reveal a wealth of traditional ancestral documentation.

Genetic genealogy can support, but can neither replace nor prove traditional ancestral research. DNA results cannot prove that the information in traditional trees is accurate; they can merely suggest a common ancestor, but they might disprove pedigree lines that are suspect, or even lines that have long been accepted as true. Potential test participants should be prepared to discover previously unsuspected family secrets.

The probability of discovering both ancestors and living relatives is rapidly increasing as more people choose to be tested. The presentations of results are becoming more user-friendly and the databases are growing exponentially. Whether people have long and well-documented pedigrees, are only beginning to document their family trees, or even have no idea who their biological parents were, everyone can benefit from genealogical DNA testing. The ability to compare traditional pedigrees with DNA results is a powerful tool that provides an additional dimension to genealogy.

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#### END NOTES:

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<sup>1</sup> The results of a Y-DNA test are reported as a series of numbers at specific DNA locations. Different companies test different locations; the resultant “markers” overlap but do not completely match the results of other companies. The number of reported markers ranges from 12 to 111 depending on the company and specific test selected.

<sup>2</sup> See the description of autosomal DNA at the following web site of the International Society of Genetic Genealogy: [http://www.isogg.org/wiki/Autosomal\\_DNA](http://www.isogg.org/wiki/Autosomal_DNA).

<sup>3</sup> <http://www.legalgenealogist.com/blog/2013/04/07/widen-the-net/>

<sup>4</sup> See <https://www.familytreedna.com/learn/autosomal-ancestry/universal-dna-matching/use-x-dna-test-results/>.

<sup>5</sup> See <http://www.pallasweb.com/blog/compare-ancestry-com-results-to-gedmatch.html>  
See also <http://www.gedmatch.com> and <http://www.ysearch.org>.

<sup>6</sup> <https://www.23andme.com>.

<sup>7</sup> Results from a testing agency can be uploaded to a third-party tool like Promethease (<http://www.snpedia.com/index.php/Promethease>) to obtain health and traits reports.

<sup>8</sup> <http://dna.ancestry.com>.

<sup>9</sup> <https://www.familytreedna.com>.

<sup>10</sup> Description of additional types of Y-DNA tests including FTDNA’s Big-Y test for Single Nucleotide Polymorphisms can be found at the FTDNA web site and at <http://www.isogg.org>, where definitions, age estimates, and importance of haplogroups, Short Tandem Repeats (STRs) and Single Nucleotide Polymorphisms (SNPs) are given.

<sup>11</sup> <http://www.ysearch.org>; <http://www.gedmatch.com>.