Family Tree DNA and Genetic Genealogy

by Susan C. Meates

The Beginning

The new discipline of DNA testing for genealogy, called Genetic Genealogy, was pioneered by Family Tree DNA, of Houston, Texas. In 2000, a genealogist, Bennett Greenspan, wondered if some Nitz's located in Argentina were related to his family tree. Having read about DNA testing for scientific endeavors, he approached Dr. Michael Hammer at the University of Arizona. Dr. Hammer is a molecular biologist who utilizes DNA testing for research and population studies. Intrigued with Bennett's genealogical problem, the two men designed a proof of concept to test the application of these scientific tests to genealogical problems. The pilot project was successful, and Family Tree DNA was launched.

Family Tree DNA provides DNA testing for genealogists. A test kit is sent in the mail consisting of three small brushes and three vials of a preservative fluid. To properly prepare a sample to send to the lab, you take each brush, one at a time, and rub the brush repeatedly against the inside of your mouth, and then put the tip of the brush into a vial. This process is easy, painless, and takes hardly any time. The vials are placed in the return envelope provided and mailed back to Houston.

Each test kit has a unique serial number, which is assigned when a test kit is ordered. This serial number appears on each vial. When the test kit arrives in Houston, an email is sent to notify the participant that their kit has arrived. These test kits are grouped together, hundreds arriving each day, and then sent to the University of Arizona where the lab will perform the tests ordered.

An important part of the process is the separation of the person's name and the DNA sample. The test vials with the sample have only a serial number, and this is all the information that is provided to the lab. At Family Tree DNA, their database connects the person's name to the serial number, but they do not have access to the sample. This extra level of security protects the DNA samples.

At the lab, the sample goes through a variety of steps depending on the test ordered, and the test results are electronically returned to Family Tree DNA who then matches the result by serial number and notifies the participant that their results are ready. A web page, called a Personal Page, is created at the Family Tree DNA web site for the participant who logs into this page with their serial number and a password. Once at their Personal Page, their journey of discovery begins with a variety of selections to click to learn more about their ancestry, origins, and to find others to whom they match.

From the beginning just 7 years ago in 2000, Family Tree DNA has grown dramatically and has provided testing services to people in 179 countries. Every day, hundreds of genealogists around the world are discovering the power of DNA testing and how the results can help them with their genealogy research.

The Benefits

DNA testing for genealogy can be used by anyone interested in family history research. You do not need a background in science or any special knowledge. The knowledge you have acquired working on your family history research, combined with learning about this new source of information, will enable you to effectively use DNA testing for your family history research.

It is important to point out that DNA testing is not a substitute for family history research. DNA testing is another tool available for the genealogist and is used in conjunction with your family history research.

This new tool provides information that can not be uncovered from the paper records. The information from DNA testing can also provide new information and clues to help with your research.

Here are just a few of the benefits that can be achieved:

- Validate research
- Find any mistaken connections in your research
- Determine which family trees are related
- Bridge gaps in the paper records
- Confirm variants
- Find unknown variants
- Sort out multiple families found in the same location
- Discover information which may solve research problems, and/or resolve brick walls
- Get clues regarding migrations
- Confirm suspected events, such as illegitimacy and adoption
- Discover information to define the major branches of the tree going back to the origin of the surname
- Discover information about the evolution of the surname
- Discover clues regarding the origin of the surname
- Combine results with research in early records to determine the number of points of origin for the surname
- Get clues to help your research

How Does It Work?

Scientists have discovered two areas of DNA that are passed on to children, typically unchanged. For males, a section of the Y chromosome is passed from father to son. For females, mtDNA is passed from the mother to both the sons and daughters, though only the daughters pass on the mtDNA.

These areas of DNA that are passed on to the each generation unchanged are unique. At conception, most of our DNA is a mix of our two parents, and our parents are a mix of their parents.

Scientists call this process recombination. These unique areas, on the Y chromosome for men, and mtDNA for both women and men, do not go through the shuffling at conception. Therefore, these small sections of DNA provide us with a window to the past. This small portion of DNA is passed down to each succeeding generation. For a male, his father, his grandfather, and his great grandfather would all have an identical or very close result if this area of the Y chromosome was tested for each of them. For mtDNA, a son or daughter, the mother and the mother's mother would all have the same result.

Types of DNA Tests for genealogy research:

Y-DNA	A section of the Y chromosome is passed from father to son, typically unchanged. By testing this section, you would discover information about the direct male line, which is the man's father, his father, and back in time. You must be male to take this test.
mtDNA	A mtDNA test provides information about the direct female line, which is your mother, her mother, and back in time. Both males and females inherit mtDNA, though only females pass on mtDNA. Both males and females can take this test.

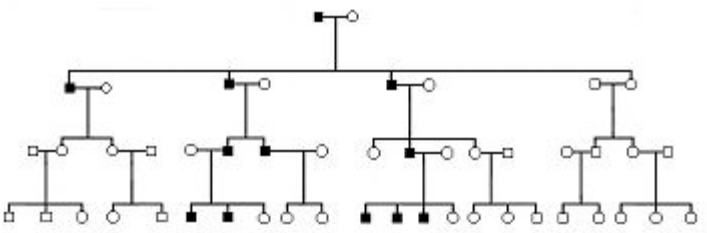
DNA, deoxyribonucleic acid, is the chemical inside the nucleus of all cells that carries the genetic instructions for making living organisms. Much of the DNA is termed 'Junk DNA' and has no known function. Within the nucleus of each cell, each of us has 23 pairs of chromosomes. Chromosomes are long segments of DNA, which contain genes and Junk DNA. A gene is the basic unit of heredity.

One set of 23 chromosomes is inherited from the father, and one set is inherited from the mother. The 23rd chromosome is also known as the sex chromosome. The child receives an X chromosome from the mother, and either an X or a Y chromosome

from the father. A child with XX is a girl, and a child with XY is a boy.

A section of the Y chromosome is passed from father to son, typically unchanged. While the Y chromosome carries some genes, there is also a significant amount of DNA located between these genes. This section of the Y chromosome is in the area scientists call Junk DNA, since it has no known function. In the chart below, the squares are males, and the circles are females. All the squares which are black show the path of the Y chromosome down the male line of the family tree.

Y Chromosome Inheritance



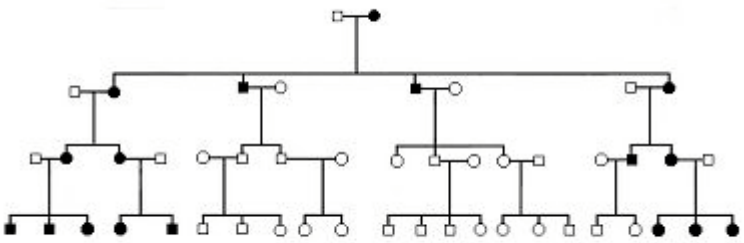
The black squares show all the males who inherited the same section of the Y chromosome from the male ancestor. From a male at the bottom, whose square is black, looking back up his family tree, following the Y chromosome, is called the "direct male line". The direct male line starts with a male, and is his father, his father, and on up the family tree.

mtDNA is inherited by both males and females, though only females pass on mtDNA. mtDNA are the circular DNA contained inside the mitochondria, which are small organelles which reside in all our cells and provide the power to the cell.

The chart below shows the path of mtDNA through the generations. The males are squares and the females are circles. A

solid square or circle shows that the mtDNA of the ancestor was inherited by this person.

mtDNA Inheritance



mtDNA follows the direct female line. Your mtDNA was inherited from your mother, who inherited the mtDNA from her mother, who inherited from her mother, and so forth. Both males and females inherit mtDNA, and only females pass on mtDNA.

Testing Y-DNA from a male would provide information about the direct male line. Testing your mtDNA would tell you about your direct female line.

Example Results

The result from a Y-DNA test is a string of numbers, which is a count of short repeats of DNA at locations on the Y chromosome called markers. The markers tested are those in the section of the Y chromosome which is passed from father to son, typically unchanged. By comparing the result of two men, you can determine if they had a common ancestor, and approximately when the common ancestor occurred.

Below are two results from a Y-DNA test:

12 22 15 10 13 15 11 14 11 12 11 28 15 8 9 8 11 24 16

20 29 12 14 15 15 12 22 15 10 13 15 11 14 11 12 11 28 15 8 9 8 11 24 16 20 29 12 14 15 15

In the example above, two men took a 25 marker Y-DNA test at Family Tree DNA. Each man belongs to a different family tree, and they both share the same surname. Their 25 marker Y-DNA test result is an exact match, which is also called a 25/25 match. The Y-DNA evidence tells us that the men had a common ancestor. Sharing the same surname tells us that these two men share a common ancestor since the adoption of surnames. These two family trees connect at some point in time, since the adoption of surnames. Each man, on his web page at Family Tree DNA has a small symbol they can click, that will bring up a screen that estimates the time frame of the common ancestor.

The test results also provide the major population group for the direct male line. These population groups are known as haplogroups, and date back thousands of years. All men in a haplogroup would share a common ancestor. The haplogroup for the above result is I1a, and the description from the vendor's web site about this haplogroup is the following:

"The I1a lineage likely has its roots in northern France. Today it is found most frequently within Viking / Scandinavian populations in northwest Europe and has since spread down into Central and Eastern Europe, where it is found at low frequencies." (Description copyright Family Tree DNA.)

Although the haplogroup is not relevant to genealogy research, often the results are interesting, and tell you about the distant origins of the direct male line.

Testing mtDNA does not have as many genealogical applications as testing Y-DNA. The surname for married females changes with each generation, and mtDNA mutates at a slower rate than Y-DNA, therefore providing a longer time frame for the common ancestor.

A mtDNA test will provide information about your direct female line. The test result also provides information about the distant origin of your direct female line. The test result is compared to a standard, called the Cambridge Reference Sequence. This reference standard is the first mtDNA sequenced. Sample mtDNA results are shown below**.**

Haplogroup	**HVR1 Mutations**	**HVR2 Mutations**
K	16145A 16224C 16311C 16519C	73G 150T 152C 195C 263G 315.1C 497T 523- 524-
U5a1*	16192T 16256T 16270T 16399G	73G 263G 309.1C 315.1C
H	16304C	152C 263G 315.1C 456T

Since mtDNA mutates at a slower rate than Y-DNA, only exact matches should be investigated. In addition, both Hyper-Variable Region 1 (HVR1) and Hyper-Variable Region 2 (HRV2) should be tested and match for genealogy applications.

The haplogroup is the major population group of your direct female ancestor. This is also known as the Daughter of Eve, or Clan Mother. For each haplogroup, Family Tree DNA supplies information about the origin of that haplogroup.

For example, the description for haplogroup K is as follows: "Haplogroup K is found through Europe, and contains multiple closely related lineages indicating a recent population expansion. The origin of haplogroup K dates to approximately 16,000 years ago, and it has been suggested that individuals with this haplogroup took part in the pre-Neolithic expansion following the Last Glacial Maximum."

(Description copyright Family Tree DNA.)

Matches on HVR1 are those where the time frame of the common ancestor could be thousands of years ago. For genealogy applications, both HVR1 and HVR2 should be tested.

There are genealogical applications for mtDNA testing. For example, your family tree may have a male ancestor who had two wives, and you can't find the documents to tell you who was the mother of the 3rd child, who was a girl. By following the female line of the various female children from the first and second wife, and testing the mtDNA of descendents and comparing the result to the mtDNA result of a descendent of the 3rd child, you would be able to determine which wife was the mother of the 3rd child.

DNA Testing for Genealogy

Genetic genealogy is the application of DNA testing for family history research. Who is tested, and which test is selected, depends on the information you want to uncover. The application of Genetic genealogy to your family history research is very similar to your approach to your research. For example, for your family history research you may decide to look for a census entry for your grandparents as your next step. In applying DNA testing to your family history research, you would also select an objective, which is the information you want to uncover.

DNA testing has a wide variety of applications or objectives, depending on the genealogy research problem or the information to uncover.

It is also important to understand that DNA testing will **NOT** provide all the answers. If DNA testing shows that two people are related, the results will not tell you exactly when they were related, or the name of the common ancestor. A DNA test wouldn't tell you exactly where an ancestor lived, though the test results might provide clues for a geographic area. For this reason, DNA testing

is combined with your family history research to get further information from the test results.

One of the most exciting elements of DNA testing for genealogy is that often the test results will provide information that cannot be uncovered from other sources. For example, you may not be able to find any paper records to determine if two males in two different households with the same surnames in the census are related. To find out if these two males were related, you would test one or more direct male descendents from each of these households.

DNA testing for genealogy has a wide variety of applications, depending on the genealogy research problem or the information to uncover. The objective could be to determine if two people with the same surname are related, or the objective could be a more complex application to determine the surname of an adopted ancestor where no paper records could be found, and it is suspected that he is the son of the widow next door who died.

How Many Y-DNA Markers to Test

The first choice for a Y-DNA test is the number of markers to test. Twelve to 67 markers are offered. At Family Tree DNA, you can upgrade your test to additional markers. For example, you can start at 12 markers, and then later upgrade to either 25 or 37 or 67 markers.

Each test kit includes 25 years of storage. This makes upgrades easy, as well as ordering other tests, such as mtDNA or advanced Y-DNA tests. In addition, your sample is available for future scientific advances. This is very beneficial when there are limited living males for your family tree, especially elderly males, so they can leave a legacy for future genealogists.

Twelve markers are best at identifying those to whom you are **not** related. When you have a match at 12 markers, you usually want to upgrade to reduce the time frame to the common ancestor, and determine if you are related in a genealogical time frame.

A simple guideline is: More markers provide more information, as well as reduce the estimated time frame to the common ancestor.

Tests of 12 markers or below are considered low resolution. Tests for 25 markers or above are considered high resolution.

It is usually more cost effective to order a high resolution test initially, instead of starting with a low resolution test and upgrading later.

To receive genealogically useful information, a high resolution test should be conducted.

For Y-DNA, Family Tree DNA also offers a variety of advanced tests, ranging from a SNP test to confirm your haplogroup or major population group, to a test that will determine if you could be immune from the plague, if it occurred today.

Y-DNA Surname Projects

Y-DNA testing is organized based on the surname. If two people have the same or variant surname, and their Y-DNA is a match or a close match at 25 or more markers, then without any documentation, you would know that these two people are related since the adoption of surnames. A more precise time frame can be estimated based on the closeness of the match.

The adoption of heredity surnames occurred in different locations at different times. The process took centuries, and during this time the form of the surnames often changed, and variants arose.

Y-DNA testing will identify which family trees for a surname are related. This information is quite valuable. For example, you may be trying to make a connection to the ancestral country. If your surname has two different Y-DNA results, and all the trees with your result go back to one location, then you would want to focus your research in that geographic area.

Identifying the related variant surnames can also be very beneficial. For example, previously unidentified variants may be found, which, when you review the census records with these variants, you are able to find your ancestors, and overcome your brick wall.

To get started with Y-DNA testing, you would want to determine if there is an existing Surname Project for the surname. Testing with an existing project simplifies the comparison of results. To determine if a Surname Project exists, go to the Family Tree DNA home page, shown at the end of this article, and enter your surname in the Project search facility in the upper right.

As a participant, your results include a certificate and report in the mail, and a Personal Page is created at the Family Tree DNA website. On your Personal Page, you can click selections, and view results and information. One selection is Y-DNA Matches. Clicking this selection will show those whom you match or are a close match. The person's name and email are shown. In addition, if they up loaded a Gedcom of their family tree, a symbol appears to the right of their name. The FTDNATiP symbol is also shown for matches. Clicking this symbol generates a report comparing you to the match using a proprietary algorithm. A report is generated that shows the probability of when the common ancestor occurred. This tool is very valuable to determine whether to search for documentation supporting the relationship.

Another selection is titled Haplogroup. This selection will tell you the major population group, known as Haplogroup, and provide a description of the Haplogroup as well as showing the matches in the Haplogroup database and where they are located.

The selection Recent Ancestral Origins provides information about the origin and migration of the ancestors of those whom you match.

Family Tree DNA customers also have an opportunity to join the Genographic Project, by clicking on a selection on their Personal

Page. This project is sponsored by National Geographic and has two components. One component is to test indigenous peoples around the world to trace the journey of human kind out of Africa. The second component is public participation, where you join and participate in a real time scientific project. There is a small fee to join the Genographic Project. Once you join, a Personal Page is created at the Genographic site filled with maps and educational material about the distant history and migration of your ancestor.

If a Surname Project has not been established for your surname of interest then you can easily start a Surname Project. Testing under the umbrella of a Surname Project provides discount pricing. Therefore, starting a Surname Project would reduce the cost of testing. If you do not want to start a Surname Project, perhaps one of your fellow researchers would be interested. Otherwise, you can test outside a Surname Project, and could later join a project when one is established for your surname.

If you are considering starting a Surname Project, Family Tree DNA supplies assistance, educational materials, help interpreting results, and guidance. Included with a Surname Project are a set of administrator tools and reports that make managing the project and interpreting results easy.

Application: Confirm Your Family Tree

One application for DNA testing is to confirm your family tree. This step involves confirming your research utilizing Y-DNA. Two distant direct line males would be selected to test. Assuming that your family history research is correct, you would expect the two results to match or be a close match. If you have any suspicions or areas of weak documentation, you would want to make sure at least one participant was a descendent from the weak branch. If your family tree has a lot of breadth, or goes back several centuries, you would probably want to test three or four males representing the major branches to achieve a comprehensive DNA review of your family history research.

There is one weakness in your family history research that DNA testing to confirm your family tree may not catch. If there are other families in a geographic area with the same surname, and you connect your family tree to the wrong family, the DNA testing may not catch this situation, if the males are related by a prior ancestor. For example, 3 brothers immigrate to the US in the 1700's. They each have sons called William. You family tree shows the son William of the 3rd brother as your ancestor, when the correct ancestor is the son William of the 1st brother. Since the brothers were related, they would all have the same or close Y-DNA results, and so would their male descendents. Therefore, your DNA testing to validate your family tree would still have matches, even though from the research you selected the wrong William. If you test at 37 or 67 markers this problem of a mistaken connection can often be identified.

Application: Determine If Two People Are Related

DNA testing can be used to determine if two people are related through their direct male line, or related through their direct female line. To determine if two people are related through the direct male line, one or more males from each family tree would take a Y-DNA test. Typically, you would want to test two males in each family tree. The step of testing the second male is to validate the result of the first male, and establish the Ancestral Result for the family tree. Assuming the two results match or are a close match, then any extramarital event or unknown adoption in the family tree has been eliminated for the branches tested.

Testing two males in your family tree provides a result that can be used for multiple applications.

It is always possible that hidden in a branch you do not test is an extramarital event or unknown adoption. If you do not have any clues in the paper records that any of these types of events could have occurred, such as being unable to find a birth, or a married couple living apart, then it probably is not worth further testing. The only way to confirm for 100% certainty that there was no

extramarital or unknown adoption in the living male line of your tree is to test every living male descendent. This is usually not done.

To compare two family trees to see if they are related along the direct male line, you would select two males from each family tree to test. For example, consider the situation where in a census entry you have two males living next door to each other with the same surname, and you have been wondering if they are related. Determining with DNA testing whether these two families are related might be a clue to help sift through the immigration records or a clue to help find the ancestral homeland.

If either of the family trees only have one living direct descent male today, then you would only be able to test one male from that family tree.

A close match can occur because a mutation occurs for one of the markers, which are the locations tested on the Y chromosome. A mutation simply indicates a change. Family Tree DNA estimates that a mutation for a marker occurs about once every 250 generations. A mutation can occur at any time. You may find a living descendent who has a mutation, or the mutation might have occurred several generations ago. There is a methodology that can be used to determine exactly where the mutation occurred.

Below are the Ancestral Results for the two family trees being compared:

15 23 15 10 15 16 13 13 11 14 12 30 16 8 9 11 11 26 15 20 29 11 11 14 16
13 24 14 11 11 14 12 12 12 14 13 30 16 9 10 11 11 25 15 18 30 15 15 17 17

When comparing the result from each family tree to each other – they are clearly not related. Only 5 of the numbers match between the two results, and to be related, we are looking for an exact, 25/25 match or close match, 23/25, 24/25.

If you had no further information than two families with the same surname living next to each other in the 1880 census, it probably would not be surprising that the results do not match.

On the other hand, if you had strong documentary evidence that the two families were related, and then the two results don't match, it is time to review the family history research, as well as review the two family trees to make sure that the two persons who were tested are distantly related, so that one extramarital event or adoption would not impact both of their results. For example, if the founder of a family tree had multiple sons then each participant tested would ideally be from a different son. If you test two participants from one son, and that son was adopted, then the results would match for the two participants, but would not reflect the founders result.

Today

As the pioneer who brought DNA testing for genealogy to the mainstream, Family Tree DNA provides a variety of tests to help with your genealogy research, as well as tests to explore your distant origin or to learn more about your ancestry. As new scientific discoveries occur, Family Tree DNA brings new tests to market.

The easiest way to learn more about DNA testing for genealogy is to take a test. The first step is to decide whether you want to test your direct male line or your direct female line. If you are female, and want to learn about your direct male line, you would have your father, brother, uncle or other male relative in the direct male line take the test. Both males and females can take the mtDNA test to learn about your direct female line. Family Tree DNA provides education material, consultation and email support to understand your results.

To learn more about DNA testing for genealogy, be sure to subscribe to the free educational newsletter provided by Family Tree DNA. The link to subscribe or read past issues is shown below.

DNA testing for genealogy is a very powerful tool, and we are just at the beginning of this emerging discipline. It was only a few years ago that DNA testing for genealogy was first introduced. Each year since then, new and expanded tests have been introduced as the scientists make more discoveries. Each new test provides additional information for genealogists.

Through DNA testing, people have learned information that could not be uncovered from other sources. A DNA test could supply the answer to break through a brick wall. You can begin your journey of discovery by ordering a test kit today.

Family Tree DNA
http://www.FamilyTreeDNA.com

Free monthly educational newsletter from Family Tree DNA
http://www.familytreeDNA.com/fgregister.asp

Past Issues of the Family Tree DNA newsletter
http://www.familytreeDNA.com/facts_genes.asp?act=past

Genographic Project:
http://www5.nationalgeographic.com/genographic/index.html

FTDNATiP™ Report

Family Tree DNA Time Predictor*
Version 1.1 - Patent Pending

In comparing 37 markers, the probability that L7: Greg Meates and L1: Richard Meates shared a common ancestor within the last...

100 years is	200 years is	300 years is	400 years is	500 years is	600 years is
83.49%	97.28%	99.55%	99.93%	99.99%	100.00%

The above numbers are based exclusively on the comparison of their Y-DNA results, which show no mismatches.

* The FTDNATiP™ results are based on the mutation rate study presented during the 1st International Conference on Genetic Genealogy, on Oct. 30, 2004. The above probabilities take into consideration the mutation rates for each individual marker being compared.