DNA Testing for Genealogy - What Can It Do For You??

Paper courtesy of Roberta Estes, www.dnaexplain.com, e-mail Roberta at Roberta@dnaexplain.com. Graphics courtesy of Family Tree DNA, www.familytreedna.com.

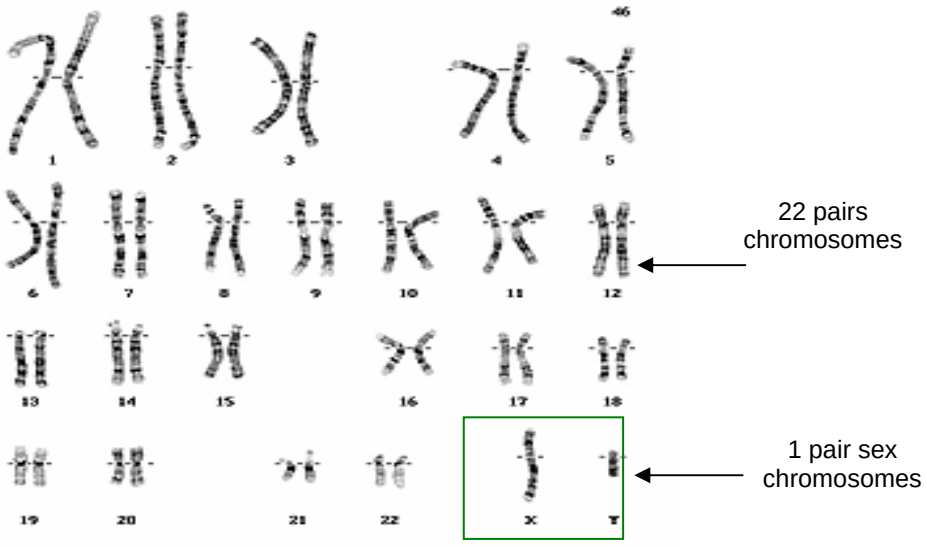
DNA testing for genealogy didn't even exist a few years ago. In 1999, the first tests were performed for genetic genealogy and this wonderful tool which would change genealogy forever was born for the consumer marketplace from the halls of academia.

Initially we had more questions than answers. If it's true that we have some amount of DNA from all of our ancestors, how can we tell which pieces are from which ancestor? How much can we learn from our DNA? Where did we come from both individually and as population subgroups? How can it help me knock down those genealogy brick walls?

In just a few short years, we have answers for some of these questions. However, in this still infant science we continue to learn every day. But before we discuss the answers, let's talk for just a minute about how DNA works.

DNA - the Basics

Every human has 23 pairs of chromosomes (think of them as recipe books), which contain most of your DNA, functional units of which are known as genes (think of them as chapters). One chromosome of each pair comes from a person's mother and the other from their father. Due to the mixing, called recombination, of DNA that occurs during meiosis prior to sperm and egg development, each chromosome in 22 of the 23 pairs, which are known as autosomes, has DNA (think of it as ingredients) from both the corresponding parent's parents (and all of their ancestors before them).

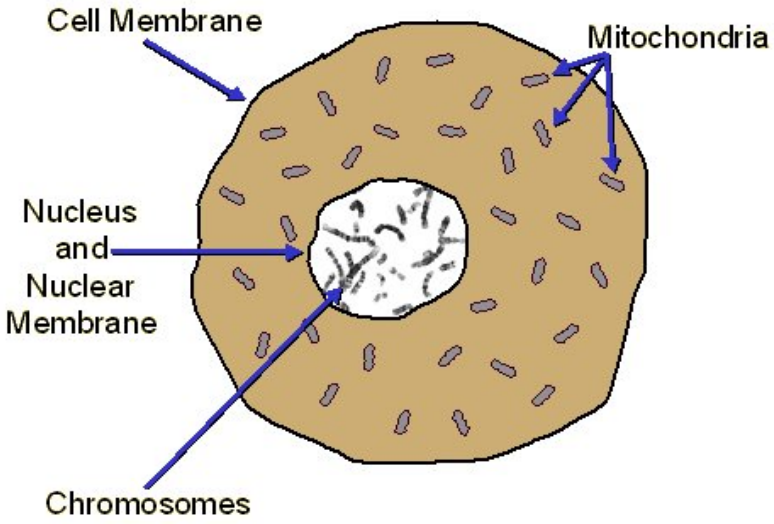


Two portions of our DNA are not combined with that of the other parent. The 23rd chromosome, in the green box above, determines the sex of the individual. Two X chromosomes produce a female and an X and a Y chromosome produce a male. Women do not have a Y chromosome (otherwise they would be males) so they cannot contribute a Y chromosome to male offspring. Given this scenario, males inherit their father's Y chromosome unmixed with the mother's DNA, and an X chromosome unmixed with their father's DNA.

This inheritance pattern is what makes it possible for us to use the Y chromosome to compare against other men of the same surname to see if they share a common ancestor, because if they do, their Y chromosome DNA will match, either exactly or nearly so.

Autosomal DNA, X chromosomal DNA and, in males, Y chromosomal DNA are all found in the nucleus of a cell. A a fourth type of DNA call mitochondrial DNA, or mtDNA for short, resides within cells but outside the cell's nucleus. Mitochondrial DNA packets are the cell's powerhouse as they provide the entire body with energy.

For both genders, mitochondria DNA is inherited only from the mother. Men have their mother's mtDNA, but do not pass it on to their offspring. Women have their mother's mtDNA and pass it to both their female and male offspring. Given this scenario, women inherit their mother's mtDNA unmixed with the father's and pass it on generation to generation from female to female. (Males carry their mother's mtDNA, but don't pass it on.) This inheritance pattern is what makes it possible for us to compare our mtDNA with that of others to determine whether we share a common female ancestor.



These animations at the Sorenson Molecular Genealogy Foundation website are an excellent visual resource for understanding how the 4 kinds of DNA are passed from the parents to a child. http://www.smgf.org/pages/animations.jspx

Autosomal DNA (not the 23rd chromosomal pair) tends to be transferred in groupings, which ultimately give us traits like Mother's blue eyes, Grandpa's chin or Dad's stocky build. Sometimes these inherited traits can be less positive, like deformities, diseases or tendencies like alcoholism. How this occurs and what genes or combinations of genes are responsible for transferring particular traits is still being deciphered.

Sometimes we inherit conflicting genes from our parents and the resolution of which trait is exhibited is called gene expression. For example, if you inherit a gene for blue eyes and brown eyes, you can't have both, so the complex process of gene expression determines which color of eyes you will have. However, this type of genetics along with medical genetics does not concern us when we are using genetics for genealogy, so we will focus initially on the unmixed Y chromosomal DNA, called Y-line for short, and mtDNA as genealogical tools.

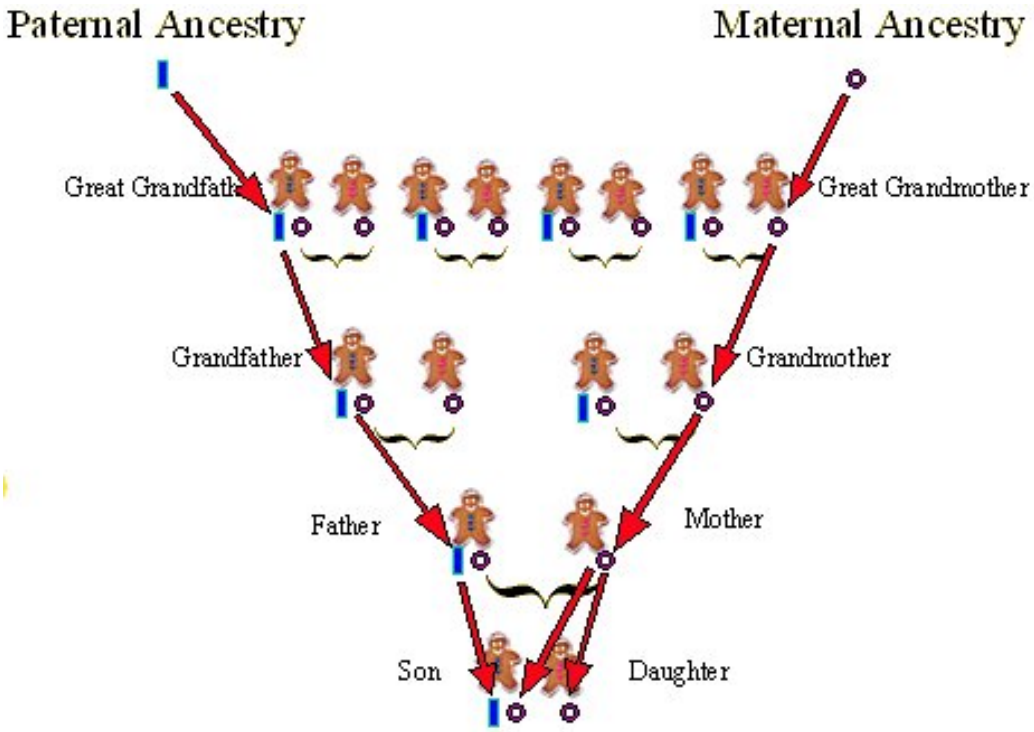
How Can Unrecombined DNA Help Us With Genealogy?

I'm so glad you asked.

During normal cell meiosis, each ancestor's autosomal DNA gets watered down by half with each generation.

However, that isn't true of the Y-line or mtDNA. In the following example of just 4 generations, we see that the Y chromosome, the blue bar marker on the left, is passed down the paternal line and the son has the exact same Y-line DNA as his paternal great-grandfather.

Similarly, the round doughnut shaped O represents the mitochondrial DNA (mtDNA) and it is passed down the maternal side, so both the daughter and the son will have the exact same mtDNA as the maternal great-grandmother (but only the females pass it on).



The good news is that you may well have noticed that the surname is passed down the same paternal path, so if this is a Jones family, the Y-line DNA travels right along with the surname. How it can help us with genealogy now becomes obvious, because if we can test different male descendents who also bear the Jones surname, if they share a common ancestor somewhere in recent time (the last several hundred years), their DNA will match, or nearly so. Surname projects have been created to facilitate coordination and comparison of individuals carrying the same or similar surnames.

Mitochondrial DNA (mtDNA) is useful as well, but not as readily useful for genealogical purposes since the surname traditionally changes with each generation.

There have been several remarkable finds using mtDNA, but they are typically more difficult to coordinate because of the challenges presented by the last name changes. Sometimes joining regional projects is more useful for finding mtDNA matches than joining surname projects. A case in point is the Cumberland Gap project which has helped many people whose families lived in close proximity of the Cumberland Gap (at the intersection of Va., Tn. and Ky.) connect with their genetic cousins. What mtDNA can easily do for us is to confirm, or put to bed forever, rumors of Native American, African or Asian ancestry.

What About Mutations?

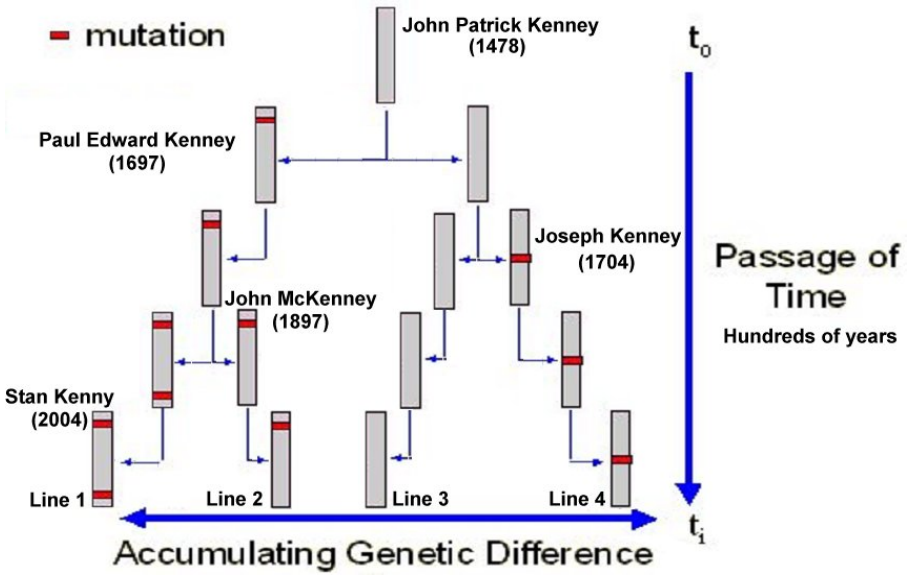
Another really good question.

Y-line DNA testing actually tests either 12, 25, 37 or 67 locations on the Y chromosome, depending on which test you select. What is actually reported at these locations is the number of exact repeats of that segment of DNA. Occasionally, either a segment is dropped or one is added. This is a normal process and typically affects nothing. These repeated segments assure that if one segment is bad, another one can take its place. However, for genealogy, they are wonderful, as the number of segments in a particular location will typically be the same from generation to generation.

When a change, called a mutation, does occur at a particular location, it is then passed from father to son and on down that line. That mutation, called a "line marker mutation" is then associated with that line of the family. If you test different individuals with the same surname, and they match except for only a couple of minor differences, you can be assured that they do in fact share a common ancestor in a genealogically relevant timeframe.

A father can potentially sire several sons, some with no mutations, and others with different mutations, as shown by the red mutation bar in the following illustration.

While mutations occur with time, individuals that share a common ancestor, should show the same markers, or markers with very few mutations.



In the above example, John Patrick Kenney had two sons, one with no mutation and Paul Edward Kenney who had one mutation. All of the male descendents of Paul Edward Kenney have his mutation and a second mutation is added to this line at a new location in the generation above Stan Kenny.

John Patrick Kenney's son who had no mutations sired a son Joseph Kenney, who had a mutation in yet a different location than either of the mutations in the Paul Edward Kenney line.

In the span of time between 1478 and 2004, this grouping of Kenney/Kenny families has accumulated 4 distinct lines as you can see across the bottom of the diagram, line 3 with no mutations, line 1 with 2 mutations, and two other lines with only one mutation each, but those mutations are not in the same location so they are easily differentiated in descendants testing today.

What Do the Results Look Like?

Results are reported in the following format at Family Tree DNA where locus means the location number, the DYS# means the name of that location, and the number of alleles means the number of repeats of DNA found in that location. This is a partial screen shot from the Family Tree DNA results page for a participant.

PANEL 1 (1-12)												
Locus	1	2	3	4	5	6	7	8	9	10	11	12
DYS#	393	390	19*	391	385a	385b	426	388	439	389-1	392	389-2
Alleles	13	25	14	12	11	14	12	12	12	13	13	29

PANEL 2 (13-25)													
Locus	13	14	15	16	17	18	19	20	21	22	23	24	25
DYS#	458	459a	459b	455	454	447	437	448	449	464a**	464b**	464c**	464d**
Alleles	17	9	10	11	11	26	15	19	29	15	15	17	17

This is interesting, but the power of DNA testing isn't in what your numbers alone look like, but in how they compare with others of similar surname.

As a DNA Surname Project Administrator of several groups, I combine the groupings of participants into logical groupings based on their DNA patterns and their genealogy.

The following table is an example from my Estes surname project which has very successfully identified the various sons of our immigrant ancestor, Abraham Estes. Based on his descendent lines' DNA, we have even successfully reconstructed what Abraham's DNA looked like so we have a firm basis for comparison. Mutations are highlighted in yellow (will show grey in black and white).

I have shown only an example of the full chart below. Moses through John R's line does have line marker mutations on markers that are not shown here. Elisha's line matches Abraham's exactly. We have had 4 descendents test from various sons of Elisha and so far we have found no mutations.

Locus	1	2	3	4	*5	*6	7	8	*9	10
Kit #	393	390	19 (394)	391	385a	385b	426	388	439	389-1
Abraham	13	25	14	**12**	11	14	12	12	12	13
English Estes Line (Reconstructs Sylvester, Abraham's father)										
16532	13	25	14	**12**	11	14	12	12	11	13
Moses through John R Line										
9993	13	25	14	**12**	11	14	12	12	12	13
11375	13	25	14	**12**	11	14	12	12	12	13
Poss John thru Elisha and Micajah line descendents										
13044	13	25	14	**12**	11	14	12	12	12	13
14107	13	25	14	11	11	14	12	12	12	13
16355	14	25	14	11	11	14	12	12	11	13
Thomas line										
12088	13	24	14	11	11	14	12	12	12	13
Sylvester line										
13805	13	25	14	**12**	11	14	12	12	12	13
17420	13	25	14	11	11	15	12	12	12	13
Robert's line thru son George son Bartlett son John Bacon										
14220	13	25	14	**12**	11	14	12	12	13	13
Elisha's line (matches Abraham exactly)										
12563	13	25	14	**12**	11	14	12	12	12	13
19696	13	25	14	**12**	11	14	12	12	12	13
Abraham's cousin Richard line - Northern Estes line										
12630	13	25	14	10	11	14	12	12	12	13
14167	13	24	15	11	11	15	11	13	10	14
This group helped us reconstruct Abraham's DNA. Please notice that participant 14167 either has unsound genealogy or an unrecorded adoption has occurred.										
Moses in SC line										
20835	13	22	14	**10**	14	14	11	14	11	13
Note that this line, even though the last name is Estes, does not match the Abraham Estes line.										
Susanna Estes line										
21235	13	24	14	11	11	15	12	12	12	13
This and the following group represent illegitimate births where the men took the mother's last name of Estes, but their DNA does not match the Estes male line.										
Nancy Estes and Jesse Mullins										
14900	13	24	14	11	11	14	13	12	13	13

What Else Can We Tell?

The results of your tests not only tell you about your genealogy, they can also tell you about your deep ancestry, known as genetic anthropology, and identify your deep ancestral clan.

Have you ever wondered where your ancestors came from before contemporary times? We know that for the most part surnames did not exist before 1066, and in some places did not exist until much later. The likelihood of us ever knowing where our ancestors were prior to 1066, unless we are extremely lucky, is very remote using conventional genealogical research methods.

However, now with the results of our DNA, we can peer through that window. Based on the results of our tests, and the relative rarity of the combined numbers, humans are grouped together in clans. We know who was a member of which clan by both the tests shown above and a different kind of test, called a SNP (pronounced snip) test.

Population geneticists are now using this information to determine how groups of people migrated, and when. We may well be able to tell if our clan is Celtic, or Viking, or related to Genghis Khan. Based on our clan type, we may be able to tell where our group resided during the last ice age, and then trace their path from there to England or America over hundreds or thousands of years. While this sounds farfetched, it certainly isn't and many people are discovering their deep ancestry. For example, we know that the Estes clan wintered the last ice age in Anatolia, and we know this because that is where other people who have this very rare combination are found in greater numbers than anyplace else on earth.

What Does This Mean For My Family?

It's easy to get started. You only need one male volunteer that carries your last name who is descended from your oldest progenitor by the same name and has a relatively firm genealogy.

To form a baseline within a family, we always eventually test two individuals from two separate lines of the common ancestor, just in case an undocumented adoption has occurred. If these two individuals match, except for minor mutations, then we know basically what the DNA of your ancestor will resemble and others can then test against that established line.

To test mitochondrial DNA, you simply need to track a direct female line forward in time from any female ancestor. To test your own mitochondrial DNA, just test, and then look back on your pedigree chart directly up your maternal branch of

the tree (your mother, her mother, her mother, etc.) to see whose mitochondrial DNA you carry.

Project Goals

If you are starting a DNA surname project, you will need to set some project goals. Here are some common examples or goals reflecting what can be discovered about genealogy from surname projects:

1. To establish the baseline DNA of a specific ancestor through his descendents.
2. To determine if all lines with that same surname descend from a common ancestor.
3. To determine the deep ancestral clan of your ancestor.
4. To determine the possible geographic origin of your ancestral family.
5. To determine whether two specific individuals with a common surname descend from the same ancestor.
6. To establish various surname groupings (as shown by DNA results) of your ancestor's surname so future individuals testing to overcome brick walls can potentially connect with an established line.

Does Testing Hurt?

Testing is done using a cheek swab that looks like a Q-tip. A test kit is shown below.

Just swab the inside of your cheek, put the swab back in the vial and mail back to the lab. I use Family Tree DNA to coordinate my projects. You can visit their web site at www.familytreedna.com and enter your various family surnames to see if there are already surname projects for your various ancestral lines. You may be surprised to find that many of your ancestors are already represented.

Family Tree DNA has been wonderful to work with, sponsors free surname and geographic projects and are infinitely patient and extremely helpful.

To see just an "average guy" collecting a sample from receiving the envelope in the mail to mailing it off again, click here http://www.davedorsey.com/dna.html.

Scholarships

If you're a female and can't test for y-line markers, you're not left out. You'll need to use traditional genealogy to find male lineal descendents of your ancestor that carry the family name. Then consider offering a scholarship for a descendent of that line to be tested.

Setting Up Your Project!!!!

I hope that this article has whetted your appetite and made you curious. You can be a pioneer with the same pioneering spirit that lived within our ancestors as they ventured into uncharted lands. The secrets of our DNA are just waiting to be unlocked.

To see a project that is already set up and underway, you can go to http://www.familytreedna.com/public/Estes/. This basic web space is provided for surname projects free by Family Tree DNA, the testing company I have selected for the surname projects I manage, and the good news is that you just type your info into a page - no programming necessary. Benefits such as this web site are some of the reasons why I selected Family Tree DNA, a decision I've never regretted. An excellent privately created web page can be seen at http://small-stuff.com/MOORE/.

To see if your family name already has a project, visit www.familytreedna.com and enter the last name into the search box located at the top of the page on the right hand side.

Receiving your Results

After you receive your results, please consider our Y-Line or Mitochondrial DNA Analysis packages at www.dnaexplain.com. Family Tree DNA customers who have tested at 37 markers for the Y-line or the mtDNAPlus for mitochondrial can order their reports directly from Family Tree DNA on their personal page. What you find in your own DNA will be priceless. Begin your adventure today!

Happy hunting!!!

Roberta Estes
Roberta@DNAexplain.com