DNA Testing and the Melungeons

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DNA testing has become an integral part of any genealogical endeavor, generally as part of a surname project. However, genealogy testing for a larger group purpose, such as the Melungeons, poses some unique challenges.

As the DNA advisor for the Melungeon DNA projects, I would like to take this opportunity to discuss DNA testing, the various types of tests, how they work and why they are important to the Melungeon Historical Society and research process as well as to the personal genealogical research of each participant.

Before discussing how DNA works, let's first talk about the kinds of questions we would like to attempt to answer by using various types of DNA testing.

1. Are the individuals of the same last name living in the primary Melungeon "home area" of Hancock and Hawkins County (and other very nearby locations) related on their paternal ancestral line? This means, for example, are all of the various Collins families (by way of example) from a common male ancestor?
2. Are the various individuals in this area related to the same maternal lines? This means are there "founding mothers" of this group? This is an important question because in both African and Native American cultures, families were matrilineal in nature, meaning that the surnames could be the same, from the mother, but the fathers in that social culture could be different.
3. How much truth is there to the various reports of African, Native American and Portuguese ancestry within the descendant families? Which families are admixed and can we determine the source of that admixture?
4. Can we tell, using DNA, the source of the Melungeons as a population group? Is the source of the entire group the same, or are there different subgroups? Can we eliminate or lend support to any of the proposed theories, such as shipwrecked sailors, descendants of the Lost Colony, "white Indians", and others?
5. Can we connect the genealogy of the individuals, the documented historical records of the families, the recorded history of the areas where they are found in the earliest records and along the migration path to Hawkins/Hancock and the DNA to create an answer to the question, "who were the Melungeons and where did they come from"?
6. Are other groups, such as the Redbones, Brass Ankles, Carmel Indians, Salyersville Indians, Lumbee, Saponi and others, specifically other similar tri-racial isolate groups, related to the Melungeons, and if so, how? Perhaps in some cases the proper question is "are the Melungeons and these groups descended from common ancestors", and of so, who, when and where?

Other questions may arise from the answers, such as "if your surname matches a core Melungeon surname, and your dna matches as well, but your genealogy does not take you back to Hawking/Hancock, are you a Melungeon"? These

kinds of social and identity questions are not DNA-related questions and it is not my goal to address these kinds of issues.

Let's take a look at how DNA testing works and the various types of DNA testing available in the market today and how they can address the various Melungeon scientific questions we have set forth above.

The company we have selected to be our partner for DNA testing is Family Tree DNA, www.familytreedna.com. This discussion will reference their tests and products. Family Tree DNA provides us with surname projects, geographical projects like the Melungeon project and related projects like the Cumberland Gap and Lumbee projects, as well as haplogroup projects for research. For participants, they provide a personal web page, e-mail notifications of matches, customer support, the benefits of project administrators and more. Some of the graphics below are courtesy of Family Tree DNA as well.

In the Beginning….

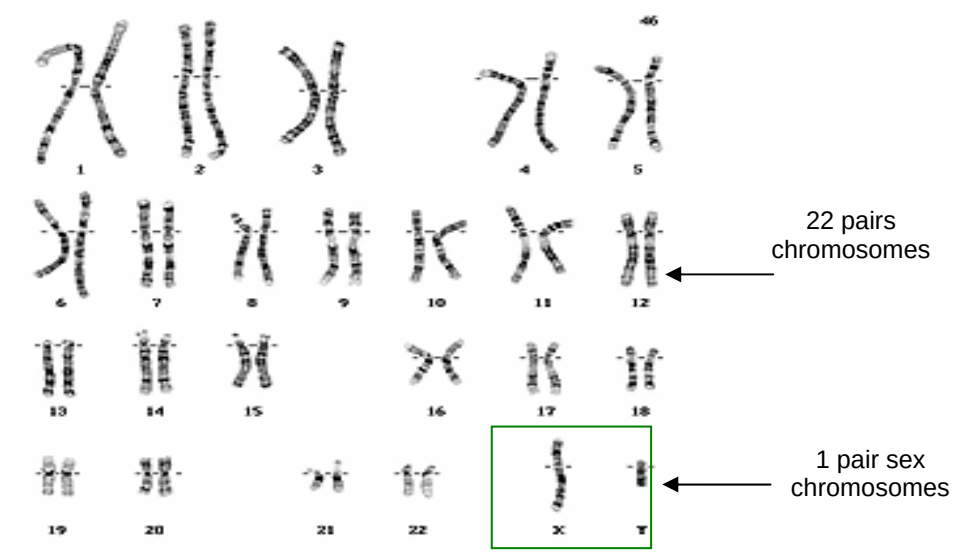
DNA testing for genealogy didn't even exist a few years ago. In the year 2000, the human genome was finally deciphered. We each have enough DNA to go from the earth to the moon and back several times.

It would follow shortly that that someone would wonder how our DNA, which holds the story of our history as humans, could assist us in our search for our genealogy. 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?

In just a few short years, we have answers for some of these questions, but before I give you the answer, 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 recipes). 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 (think of this as mixing ingredients), each chromosome in 22 of the 23 pairs, which are known as autosomes, has DNA 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 chromosomal pair, 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.

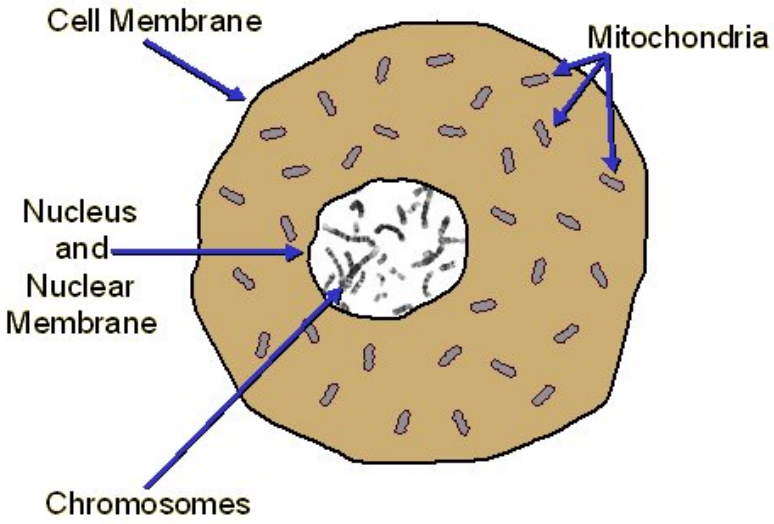
This inheritance pattern is what makes it possible for us to use the Y chromosome to compare against other men of the same surname to see if they share a common ancestor, because if they do, their Y chromosome DNA will match, either exactly or nearly so.

On the average, offspring receive equal amounts of DNA from all four of their grandparents, but due to the recombination which occurs during meiosis, any given individual may receive more DNA from some grandparents than from others -- more about this later in the autosomal section.

In addition to autosomal DNA, X chromosomal DNA and, in males, Y chromosomal DNA, all found in the nucleus of a cell, there a fourth type of DNA call mitochondrial DNA, or mtDNA for short, which resides within the cell but outside the cell's nucleus. Mitochondrial DNA can be thought of as the cell's powerhouses as their job is to provide energy for the entire body.

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 tends to be transferred in groupings, which ultimately give us traits like Mother's blue eyes, Grandpas 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.

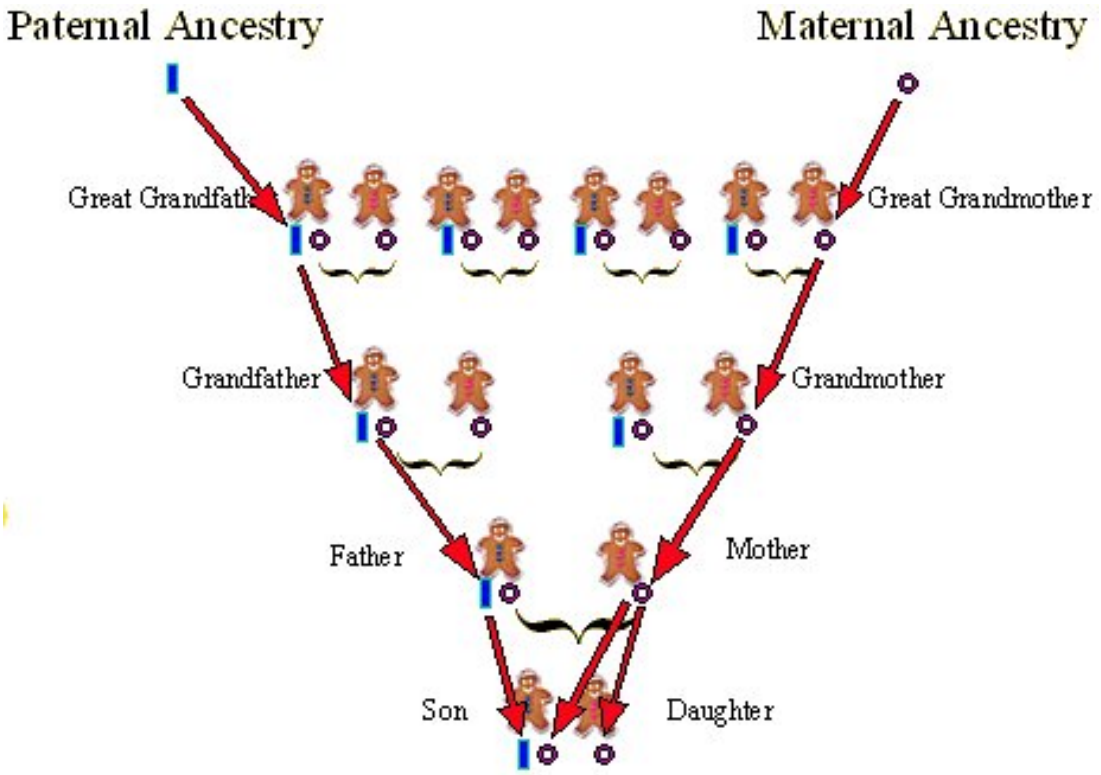
Sometimes we inherit conflicting genes from our parents and the resolution of which trait is exhibited is called gene expression. For example, if you inherit a gene for blue eyes and brown eyes, you can't have both, so the complex process of gene expression determines which color of eyes you will have. 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 Yline for short, and mtDNA as genealogical tools.

How Can Unrecombined DNA Help Us With Genealogy?

I'm so glad you asked.

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 as the Y chromosome, 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, their DNA will match, or nearly so.

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 recently been several remarkable finds using mtDNA,

but they are typically more difficult to coordinate because of the challenges presented by the last name changes. However, as more people test, individuals are increasingly finding relatives and solving genealogical puzzles using mtdna.

What Yline and mtdna can readily do for us, aside from genealogy, is to confirm, or put to bed forever, rumors of Native American, African or Asian ancestry. Our ancient mutations segment each of us into a clan, called a haplogroup, and each of these ancestral haplogroups is distinct from other haplogroups. Identifying our haplogroup allows us to track our ancestors' migrations patterns.

What About Mutations?

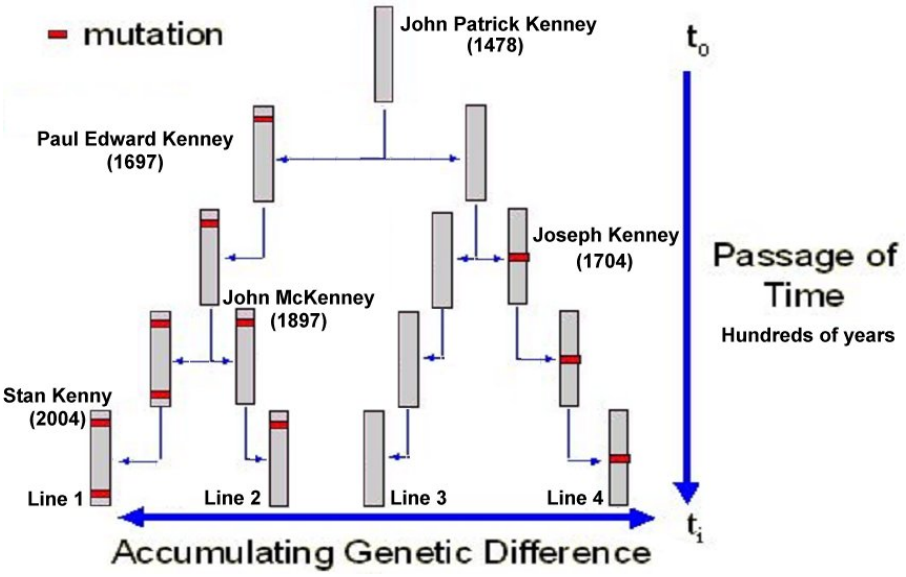
Both Yline and mtdna testing and matching depends on mutations that occur over time, typically hundreds, thousands and tens of thousands of years. Some locations mutate rapidly (over hundreds of years) and some very slowly (tens of thousands of years).

Yline DNA testing actually tests either 12, 25, 37 or 67 locations on the Y chromosome, depending on which test you choose. 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 within a family.

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, typically 500 years.

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, compliments of Family Tree DNA, 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 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 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.

What Do the Results Look Like?

Results are reported in the following format 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.

FTDNA DYS markers

We provide the actual scientific Allele values and DYS #'s for your results unless the markers were discovered at the University of Arizona and do not have a publication schedule. When that situation occurs we provide your results in "scores" to allow us to use the marker without compromising the discoverer until publication dates have been established.

We are pleased to report your results below:
Understanding your results.

Locus	DYS#	Alleles
1	393	13
2	390	25
3	19*	16
4	391	10
5	385a	10
6	385b	14
7	426	12
8	388	12
9	439	10
10	389-1	13

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 DNA tests not only tell us about genealogy, they also tell us about deep ancestry, known as genetic anthropology or population genetics, and identify an individual's deep ancestral clan. This is how we determine whether a participant tested is of African, Asian, Native American or Indo-European descent for the particular surname tested.

Genealogy ends in fairly recent 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 and related to royalty, is very remote using conventional genealogical research methods.

Deep Ancestry

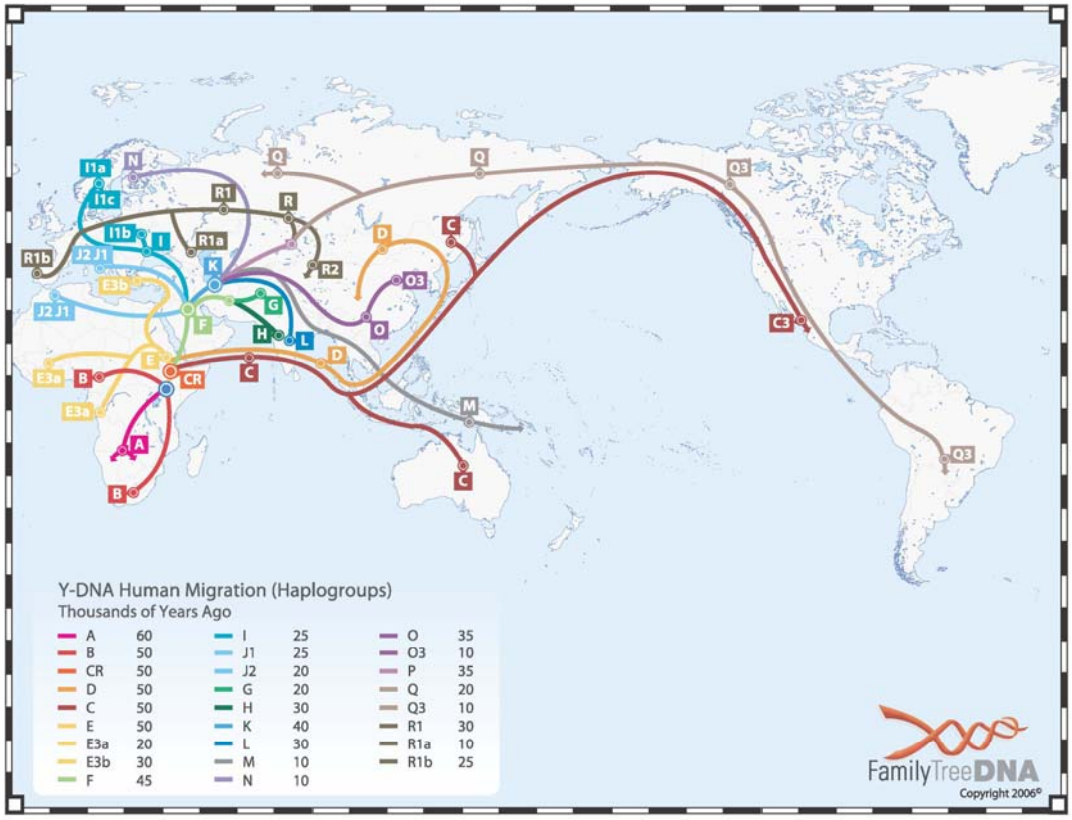
However, now with the results of our DNA, we can peer through that window. Based on the results of our tests, and the relative rarity of the combined numbers, humans are grouped together in clans. We know who was a member of which clan by both the tests shown above and a different kind of test, called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

Population geneticists are now using this information to determine how groups of people migrated, and when. We are often able to tell if a clan is Celtic, or Viking, or related to Genghis Khan.

Based on the clan type, we may be able to tell where the group resided during the last ice age, and then trace their path from there to England or America over hundreds or thousands of years or backwards in time to Africa, the native homeland of all humans living today.

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 individuals who have this very rare genetic combination are found in greater numbers than anyplace else on earth.

In the migration map below, we see how the various haplogroups migrated out of Africa and into the areas where they are found today. Remember when viewing this map that a new haplogroup is born when a mutation occurs that begins a new branch on the tree of all humanity.



How Does Yline Testing Help the Melungeon Project?

In order to increase the probability of success in the project, we decided to limit the initial focus of the study to the group of families who can be positively identified as Melungeon in Hawkins and Hancock County Tennessee. This is called the Core Melungeon group. Other people are also welcome to participate who feel they may be related, carry the same surname or otherwise feel they are connected, but they are in not in the core group at this point. This is not discriminatory, but administrative to facilitate the study of the core group, the results of which will extend to others as well.

The results of yline testing have been very interesting, if somewhat unexpected. Based on the oral history and legends of the core Melungeon families involved, it was expected that we would find a mixture of Native American and Indo-European, with possibly some African. By far the most prevalent oral history within the Melungeon population is that of being Portuguese, with admixture of Native American and some African.

While “Portuguese” is assuredly difficult to identify genetically, as Portugal and Spain are both mixing bowls given their political, social and geographic history, Native American and African are quite evident, although African DNA can be found in both Spain and Portugal.

The First Surprise

The first surprise was that some families of the same surname and clearly from the same social family do not have matching DNA. While we might be tempted to attribute this to something "Melungeon" in nature, we must first view this in the context of the typical "undocumented adoption", called in DNA circles an NPE, or Nonparental Event.

In a nutshell, this means that the yline DNA one would expect is not found in the participanting descendant. This leads to family questions that are all derivatives of "who's your Daddy?" These finds tend to be extremely uncomfortable and often cause disbelief and denial in those who are personally invested in the outcome, emotionally, financially or genealogically. At best, they are confusing.

The next step in the mystery generated by discovering an undocumented adoption is to find other people who are descended from the earliest documented ancestor, preferably through another son, and see who they match. In some cases, we have built an entire "genetic family" by testing descendants of all known sons of an individual.

While DNA can clearly solve some of these questions, it can't answer the question of why.

Most undocumented adoptions really have nothing to do with granny having a good time with the neighbor. The most common reason is a prior marriage and the children's usage of the stepfather's surname, or an illegitimate birth before marriage, or children "taken in" by a family member, or neighbor, and raised after their parents' death. At the time, it wasn't a secret, and it was so well known that it didn't even deserve comment, but over the succeeding generations, the knowledge has been forgotten and now via DNA we have "discovered" a family secret.

However, if we were to discover, for example, that we had a founding group of Melungeon women who all had children by several fathers but that the children shared a common surname, per mother, we might then begin to suspect that we have something peculiar to the Melungeon community and not the typical occurances of undocumented adoptions we see in every family.

Ongoing DNA testing and genealogical testing is in process to build these family trees. Success will depend on strong participation.

The Second Surprise

The second surprise is that we have found no Native American yline DNA, but we have found both Indo-European, which we expected, and quite a bit of

African, which we did not expect in the quantities and variety we have encountered.

Aside from the several European haplogroups, we find haplogroup A and multiple groups of haplogroup E, specifically both E1b1a and E1b1b1. All of these haplogroups are African in origin.

Haplogroup A is the haplogroup of the original Yline Adam found in mostly Southern Africa, not in the primary areas of the slave trade. However, ironically, this haplogroup is also found in a small pocket in England. A BBC article reporting this discovery can be found here:
http://news.bbc.co.uk/2/hi/uk_news/6293333.stm

How did this and other African haplogroups get to Great Britain? The two most likely avenues are through the slave trade and/or the Roman army garrisons thought to number more than 10,000 at their height who were stationed at and guarding Hadrian's Wall beginning in AD122.

Haplogroup E1b1a, formerly known as E3a, is nearly exclusively found in sub-Saharan Africa. Outside of that area, it is found only in areas associated with the slave trade or the Bantu expansion within African. A chart of the distribution of this haplogroup is found at Pubmed at this link:
http://www.pubmedcentral.nih.gov/articlerender.fcgi?artid=1976131&rendertype=figure&id=F3

Haplogroup E1b1b1 (formerly E3b1a) is also African, but has a very different distribution, focused in North African with its highest concentration and found routinely in the Near East and throughout Southern Europe and north into France. A distribution map of this haplogroup in Europe can be found here:

http://mbe.oxfordjournals.org/content/vol22/issue10/images/large/molbiolevolmsi185f04_ht.jpeg

While haplogroup E1b1a is likely of slave-trade origin, haplogroup E1b1b1 could very well exist in Europe, and via Europe in the America via population expansion combined with the expansion of the Roman Empire which regularly conscripted slaves from wherever the Army had been. Recent excavations in England near Hadrian's wall where the Roman legions camped, then settled, produced skulls of clearly African origin.

In the Melungeon project, we have 8 separate grouping of E1b1a and 2 groupings of E1b1b1 using only the core project names. We have more if include others. Furthermore, the surnames do not fall cleanly within either haplogroup nor within any subgroup. They are scattered among the groups.

Further testing is obviously in order within Melungeon family groups and within surname groups representative of the core Melungeons. These names are specifically, but not limited to:

Goins
Mullins
Collins
Moore
Miner
Bolin/Bolton

Mitochondrial DNA and the Melungeon Project

Unfortunately, mitochondrial dna testing has been the neglected step-child of DNA testing. Surname projects are sexy and offer immediate gratification. You can look at others of the same surname, who are easy to identify, and see immediately if you are related.

Mitochondrial DNA is not as easy to work with since the last name changes with every generation. Every genealogist is familiar with Jane or Elizabeth LNU on their charts, last name unknown.

Mitochondrial DNA testing is particularly important to the Melungeon project. It may tie the early female Melungeon family members together, and it is often the only way of determining a family affiliation when last names and records are completely unavailable. The Melungeon families are an extremely good candidate case for this type of familial association, because the families are known to have migrated and moved together and because they are also known to have married within their own group repeatedly.

As we research early Native American tribes of the Eastern seaboard and the Piedmont areas including both Virginia and North Carolina, we repeatedly hear of the male populations being reduced via warfare between tribes and with the English, as a result of disease and European introduced epidemics, alcohol, and for those who survived all of the above, via the slave trade. Indian males were considered unruly and often escaped, so they were sent to the West Indies where escape required a boat and hundreds of miles in the open sea.

Native females were often enslaved on plantations with males of African origin. Some escaped slavery altogether and others reclaimed their freedom through the court system. Many assimilated into either slave populations or fringe society as they were not readily accepted by white society. Many of these areas were on the ever moving westward frontiers where adventurous, typically white, males pushed the frontier lines forward and often took Indian wives either temporarily or permanently. Women of European origin probably weren't as comfortable in the wilderness as the native women were, and not nearly as useful either.

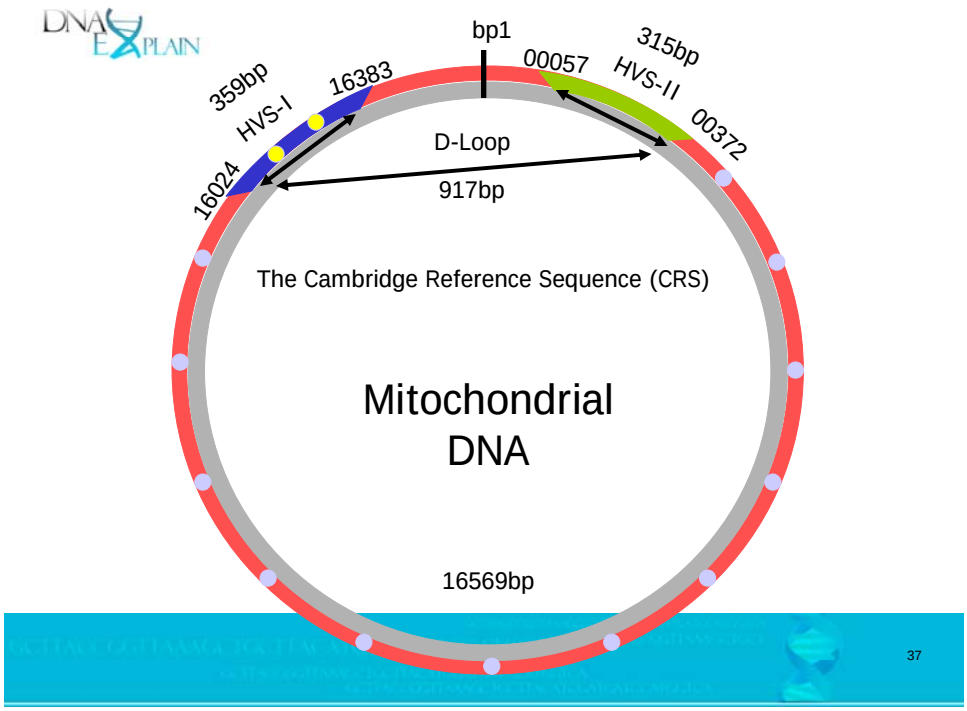
Given this scenario, if we are to find Native American admixture in the Melungeon families, it will most likely occur in the female lines. So let's take a look at how this works, as mitochondrial DNA testing and matching works a little differently than yline.

Mitochondrial DNA Analysis

When analyzing mitochondrial DNA, we compare individual results to the results of an individual whose DNA was sequenced in 1981 at Cambridge University. This set of results which has become the standard is called the Cambridge Reference Sequence, or CRS. Everyone else's DNA is compared against theirs, and the differences (mutations) duly noted.

Mitochondrial DNA is physically arranged in a circle resembling a clock. There are 16,569 base pairs that comprise mtdna. Think of each of those 16,569 locations as subsecond clicks on the clock face. At 12 noon on the clock face begins location 1 and at 11:59 we find location 16,569. Looking at the clock, these areas tested roughly cover the time from about 11:55 to about 12:05. This entire testing segment is called the D-Loop, or Diagnostic Loop, and the rest of the clock is called the Coding Region.

The purpose of mitochondrial DNA is the production and absorption of energy within cells, not genealogy as genealogists tend to believe. The Coding Region is where the instructions for energy production resides. Mutations do exist in the coding region, but they are rarer than in the D-Loop region, as cell energy production is essential to life itself. Mutations in the coding region are more likely to cause conditions that interfere with life itself, causing those organisms to not survive to reproduce, and therefore the mutation to not survive either.



Currently, there are three kinds of mtDNA tests you can order publicly. The first test, called the mtdna HVR1 test, tests an area known at the HyperVariable Region 1, beginning at 11:55 above (location 16024). From this test, we can sometimes discover which clan a participant descends from.

Further clarity can be determined from having analysis performed in HVR2, or HyperVariable Region 2, shown after noon above. This may break your clan into subgroups, so if HVR1 indicates that you are Haplogroup H (for example), then HVR2 analysis might tell us that you are in a subgroup of H called H1. Each additional area of your mitochondria that is analyzed gives us more information and narrows the list of your “cousins” significantly, eliminating false matches.

The third mtDNA test is the analysis of your full mitochondrial sequence (the entire circle above), called the FGS test. This testing provides the highest level of resolution, and as more people purchase this test, you will narrow your list of cousins to people who you are likely related to in the past few hundreds years.

However, you should know that while HVR1 and HVR2 tests do not reveal medical conditions (that we know of), full mitochondrial analysis MAY reveal some genetic medical information. If you do not want to be aware of this information, then you probably should not purchase the full mtDNA sequencing test.

For the Melungeon project, we are requesting only the HVR1 and 2 tests at this time.

The results returned to participants are the differences between their sample and the CRS. For example, the mutations below are from an individual who is a member of haplogroup U, and within that haplogroup subgroup U5a1a

16270T
16256T
16399G

Every clan has identifying mutations that all members of that clan have. Those mutations mean you descend from a female who had those mutations as well, back to the first female to develop that specific mutation, who began that particular clan, or subclan.

Mutation 16270T tells us that you are a member of haplogroup U.
Mutation 16256T tells us that you are a member of subgroup U5a1.
Mutation 16399G tells us that you are a member of subgroup U5a1a.

Each of these mutations identifies the participant's line more granularly from the super-haplogroup U.

Based on this comparison, Family Tree DNA lists individuals that you match on your personal page, along with their e-mail addresses. Particularly with mitochondrial DNA, don't depend on recognizing the surname of the applicant, because you likely don't know your 7th or 8th cousins. You will need to exchange and compare your genealogical data and also compare the general geographicsl regions in which your families were known to have settled. For this reason, geographic projects are particularly important to mitochondrial DNA research. In addition to the Melungeon project, the Cumberland Gap yline and mtdna projects encompass the geographic areas of interest to the Melungeon project.

The Cumberland Gap projects can be viewed at
www.familytreedna.com/public/Cumberlandgap-mtdna and
www.familytreedna.com/public/CumberlandGap-ydna.

Mitochondrial DNA Melungeon Results

Few individuals of Melungeon heritage are testing their mitochondrial DNA. At this time, there is only one Melungeon DNA project and it encompasses both yline and mtdna. Unfortunately, this means that the mtdna of individuals who test their Melungeon Yline also shows in the mtdna segment results, even though their mtdna may not be from the same family group or area.

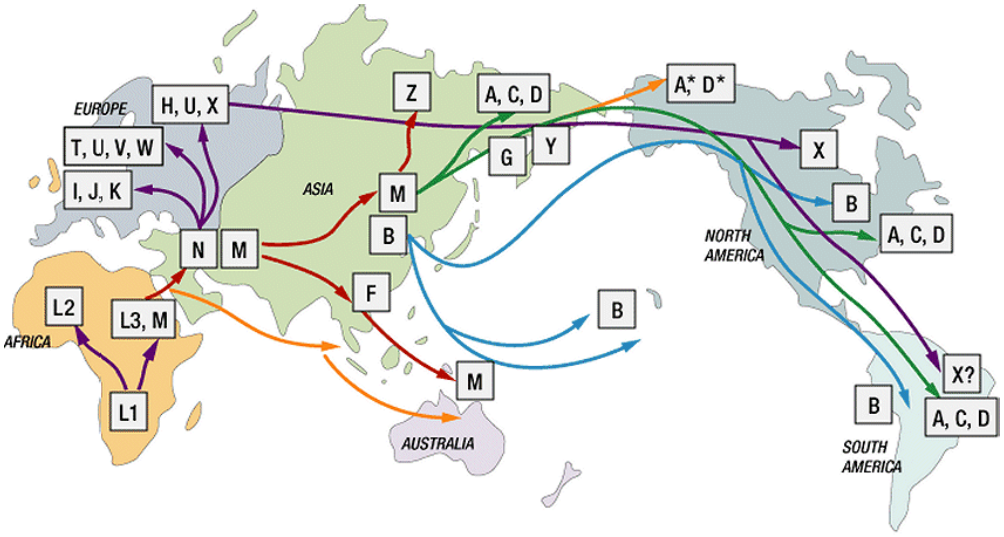
Discussions of a separate Melungeon mtdna project are underway which would isolate only Melungeon descended DNA in the Melungeon mtdna project. The message here is that the mtdna shown on the public page is not restricted to only Melungeon descendants, so should be not be analyzed with that in mind.

Having said that, we do find two individuals who have haplogroup B and two who have X, both Native American haplogroups. In addition, we have three showing African ancestry, haplogroup L.

We also have individuals who now have proven matches to a common ancestor, and in one case, a dna mismatch pointed out faulty genealogy.

We need more individuals to test and this along with recording the associated genealogies needs to become a focus for the Melungeon Historical Society.

The mitochondrial dna migration maps that show the migration out of Africa and the associated daughter haplogroups is shown below.



EXPANSION TIMES *(years ago)*	
Africa	120,000 - 150,000
Out of Africa	55,000 - 75,000
Asia	40,000 - 70,000
Australia/PNG	40,000 - 60,000
Europe	35,000 - 50,000
Americas	15,000 - 35,000
Na-Dene/Esk/Aleuts	8,000 - 10,000

FamilyTreeDNA
mtDNA Migrations Map

What About Tests that Tell Me What I Am?

The yline test and the mtdna test can tell us only about 2 of our genealogical lines. As exciting as this it, it leaves the rest of the lines on our pedigree chart blank, requiring us to find surrogate testers with those surnames or descended from those maternal ancestors. Sometimes, given surname projects, this is easy. Sometimes, particularly with mtdna, it is difficult, especially the further back in time one goes on the family tree. A little later, we'll talk about how to construct your own DNA pedigree chart to address this issue.

Biogeographical tests, or BGA tests, began being sold some years ago in the genealogical market space. These tests are targeted for the most part at the many of us Americans who either know or suspect we are of mixed Ethnic ancestry, meaning Indo-European, African and/or Native American.

It seems that every family in Appalachia carries the oral history of the Indian Princess and we all want to know if it‘s true.

While yline and mtdna will give us a definitive answer, we may need to go back in the family tree many generations, and our genealogy may give out before our native ancestor is discovered, or we may be unable to find a willing participant for dna testing.

Therefore, if there is a way to test the rest of our dna, our autosomal dna, that we receive admixed from both parents, for some hint or clue to our Native or African heritage, it would further our personal genealogical quest, not to mention the Melungeon project as a whole.

Autosomal DNA Testing

Unlike yline and mtdna testing where the DNA of the father or mother is passed to the offspring unmixed with that of the other parent, autosomal testing tests all portions of the DNA of an individual. As the field of genetic genealogy has moved forward, research has begun to indicate that certain markers are found in higher or lower amounts in different ethnic populations.

For example, if someone has the Duffy Null allele, or genetic marker, we know they positively have African admixture. We don’t know how much African admixture, or from which line, or when that individual with African admixture entered their family tree, but we know for sure they existed.

Attempting to determine the population frequency of varying markers and what that means relative to other populations is the key to this analysis. Few markers are simply present or absent in populations, but are found in varying frequencies. Some populations are widely studied in the research literature, and others are virtually untouched. The process of compiling this information in a meaningful manner so that it can be analyzed is a formidable task, as the information is often found in nearly inaccessible academic and forensic research publications. It’s difficult to determine sometimes if the DNA analysis of 29 individuals in a small village in northern Italy is, for example, representative of that village at all, of only that village, of a small regional area, of northern Italy in general, or more broadly for all of Italy as a whole (or maybe none of the above). Is it representative of Italy today or Italy historically? These and other similar questions have to be answered fully before the data from autosomal testing can be useful and reliable.

If the DNA tests being performed aren't mtdna or yline, then they are autosomal tests, meaning they are performed on the balance of the DNA contributed by both parents to an individual.

Before we discuss the varying kinds of autosomal tests and what they mean, let's take a look at the inheritance process and how it really works.

Inheritance

Everyone knows that you inherit half of your DNA from your mother and half from your father. However, this isn't the whole story. While each child does receive half from each parent, the actual inheritance pattern varies much more than that because of the random pairing of the grandparents DNA within the parent as their body prepares to combine both of the chromosomes from the grandparents into one for the parent to give to each child. That explains why each sibling may receive far more than 25% of their markers from either grandparent and why siblings genetic traits can vary so widely.

We don't understand today how inheritance traits are selected to be passed to children. Some "groups" of genetic material are inherited together, and you may wind up with more or less genetic material from one of your grandparents. In time, certain genetic "traits" will be lost in some descendants, while not in others.

Therefore, you can't figure actual inheritance percentages by using the 50% rule. This means that if your father was 50% Native American, you are not necessarily 25%, genetically speaking. You may receive 40% Native genes and your sibling may receive 10%.

Let's use the Duffy Null allele we mentioned earlier as an example. This marker could have entered your DNA pedigree chart with a grandmother who carried the allele but had no obvious visible African ancestral traits, or from your father who might have been visibly African in ethnicity. The Duffy Null allele, which is just one marker, could have been passed in the inheritance of DNA for many generations, far after any visible African traits had disappeared, or it could be one of many African traits passed from parent to child. Conversely, if that allele wasn't passed, it could be completely lost in just one generation, even if the individual clearly looks African.

The relevance of the Duffy Null allele is determined by the number of other "African" markers that appear in high quantity. If there are few other African markers, then your African ancestry was likely further back in time. If there are many, then your African ancestry was likely more recent. These statistical calculations are how the importance of autosomal markers are determined and how percentages or estimates of ethnicity are calculated.

Any one allele or marker can be lost permanently in any generation. Each child receives one gene from each parent. In the example below, let's say that the mother carried genetic markers A and B, and the father C and D, and D is the Duffy Null allele.

	Mother		Father	
	|	|	|	|
Markers	A	B	C	D

Child 1 – A and C
Child 2 – A and D
Child 3 – B and C
Child 4 – B and D

You can see that half the children received the D marker, but each inheritance event was a random recombination of the markers. It is also possible that none of the children would receive the D marker, or all of them would receive it. Statistically speaking, half will receive the marker, but statistics and individual inheritance are two different things. Random recombination is the reason why siblings who take autosomal tests sometimes show significantly different results.

You can also see how a marker that is very old ancestrally, meaning introduced many many generations ago, could be absent in one entire descendant line and present in another line.

From the above examples, we see that we have three variables that we need to deal with when attempting to use autosomal DNA for genealogy.

First, we need to take into consideration inheritance patterns which we can't determine retrospectively without testing several descendant lines. So, in essence, we can only deal with, and test, what we personally carry today as our genetic inheritance.

The second variable is determining population frequency for a particular marker and third, understanding its significance to us through comparative population genetics.

This is why autosomal testing can give us important hints, but in general is often considered "unreliable". The results are highly subjective today, but increase in accuracy as more research is completed, compiled, published and analyzed.

Types of Autosomal Tests

There are now four types of autosomal tests used today for genetic genealogy, although the newest market entrys are not specifically for genetic genealogy and include medical information as well.

One type of test uses the Codis forensic markers. The second type, biogeographical tests, use a much broader spectrum of marker results. The third type of test is an individual autosomal marker test and the fourth are much broader spectrum tests. Let's look at these various types of testing and the information they provide separately.

Codis markers are a standardized set of autosomal markers used for paternity testing. Additionally, they are used by police departments and forensics labs. The markers employed in these tests are selected specifically to differentiate between people in order to identify them individually, not to find common markers to place them in ethnic groups.

The results from these tests are only numbers, and the recipient is often left to their own devices as to how to interpret the results. These tests are available from numerous sources. Family Tree DNA offers these as Autosomal panels 1 and 2. When providing DNA analysis services for clients, I prefer to interpret these results in conjunction with Yline and mitochondrial DNA test results for as much of the genetic pedigree chart as can be provided in order to obtain a more complete genetic picture.

Below is an example of what Codis test results look like. They are very similar from any lab.

Location	Mother	Child
CSF1PO	10, 12	10, 12
D2S1338	17, 25	17
D3S1358	17, 18	17, 18
D5S818	11, 12	11, 12
D7S820	8	8, 9
D8S1179	12, 14	12, 13
D13S317	12, 13	13
D16S539	11, 12	11, 12
D18S51	12, 13	12, 20
D19S433	12, 14	14, 15
D21S11	30, 31.2	31, 31.2
FGA	20, 24	20, 24
TH01	6, 9.3	6, 9.3
TPOX	11	8, 11
vWA	17	17, 19

Analysis of Codis Markers

Unless you're using the Codis marker results to determine siblingship or some other personal reason, these numbers are fairly useless genealogically. It's the analysis of these markers that matters.

There are different avenues to analyze Codis results. None are "right" or "wrong", although even when using the same standardized analysis tools

sometimes different individuals produce strikingly different results which introduces the question of quality in the analysis process.

I use a combination of resources, both public and private, including Omnipop, which is publicly available, and other European and Canadian autosomal forensic data bases.

Tribes (www.dnatribes.com) has been compiling population data on the Codis markers for some years now and will compare your autosomal results with their data base. Take a look at their "sample" tab.

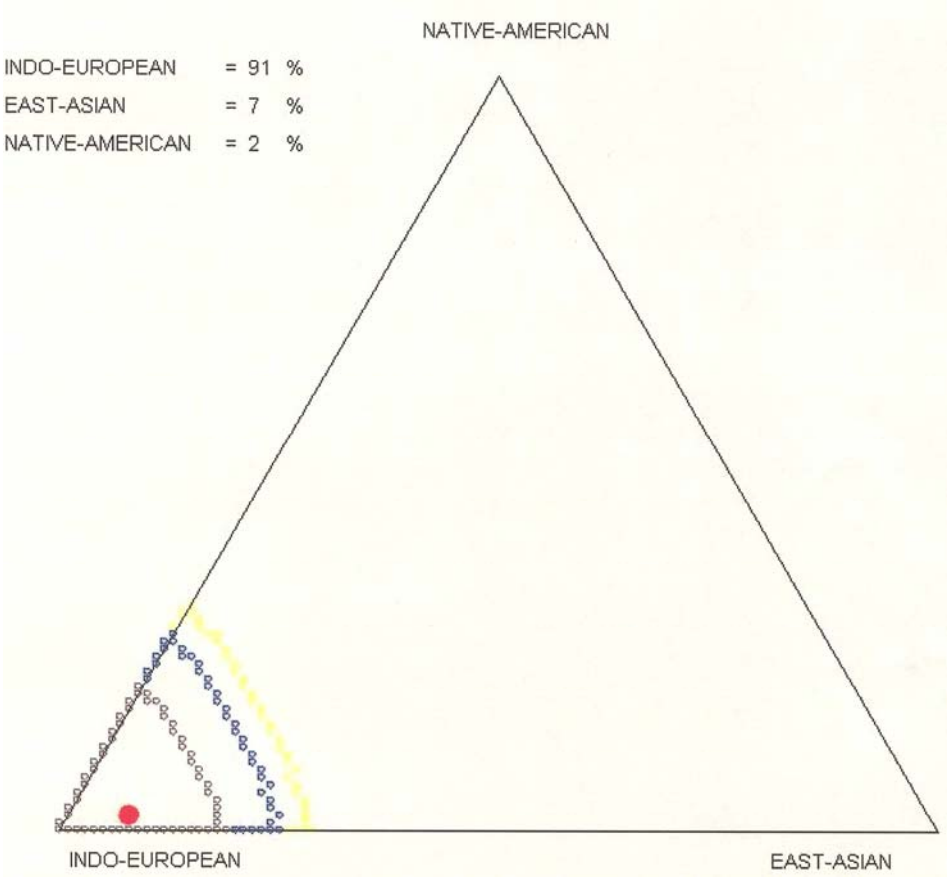
Ironically, the results may vary significantly between these resources. There is no "right" or "wrong" or "one" answer at this point. I encourage everyone to simply view these results as "data", hints to puzzle pieces. As the data bases improve and we better understand population migration and movement, the clarity of the results will improve too.

Tribes early population tables did not include data from the British Isles, so their results were highly skewed towards other world populations. Omnipop today relies on self-reported ethnicity of felons or their arresting officers and does not include normalized data (or a normalizing factor) for varying populations.

Because Tribes is a private company, we don't know much about their population data, whether it's widely representative of the world population distribution and whether it has been normalized or not.

Biogeographical ancestry testing, available from DNA Print Genomics (www.dnaprint.com), is the second type of autosomal testing. They test all of your genetic contributions for specific, proprietary markers that indicate geographical heritage, not just the Yline or mtdna. They do not use the Codis markers, but use, depending on your test selected, between 500 and 1349 markers they've discovered to be relevant to ethnicity.

This test is currently available from only one source, although the test is resold by several testing companies. Results from this test are returned as percentages of ethnic heritage as shown below.



Your results are reported within confidence bands, which indicate a range of percentages that might actually be accurate. This is shown above by the bands surrounding the red dot which shows the "most likely" result. The margin of error is often as high as 15%. Typically, there is no dispute over the majority ancestral type. However, minority types are apparently much more difficult to discern.

Various versions of their software have produced widely differing results, although hopefully that is because more current versions are increasing in accuracy.

Recently, this company has undergone a management change and possibly a change in ownership that has caused one of their laboratories to close. Their test results have been delayed for several months while their equipment is being relocated. Be vigilant before purchasing these tests.

Unfortunately, early adopters of the test are not offered an upgrade path from pre-2.5 versions, which has discouraged many individuals from continuing to pursue this testing option.

Wide Spectrum Testing – Personal Genotyping

Early in 2008, two firms introduced what is being referenced as wide-spectrum testing. This approach tests more than half a million dna locations and provides you with a wide range of information. Exactly what varies at these two companies, but the categories of information are similar.

The company's products are called 23andMe and deCodeMe and are offered by 23andMe and DeCode Genetics. www.decodeme.com and www.23andme.com

I tested initially at DeCode Genetics because they provided more information about the various ethnic groups. Both companys have made advances and just recently 23andMe announced a 50% reduction in the cost of their test which was previously $995.

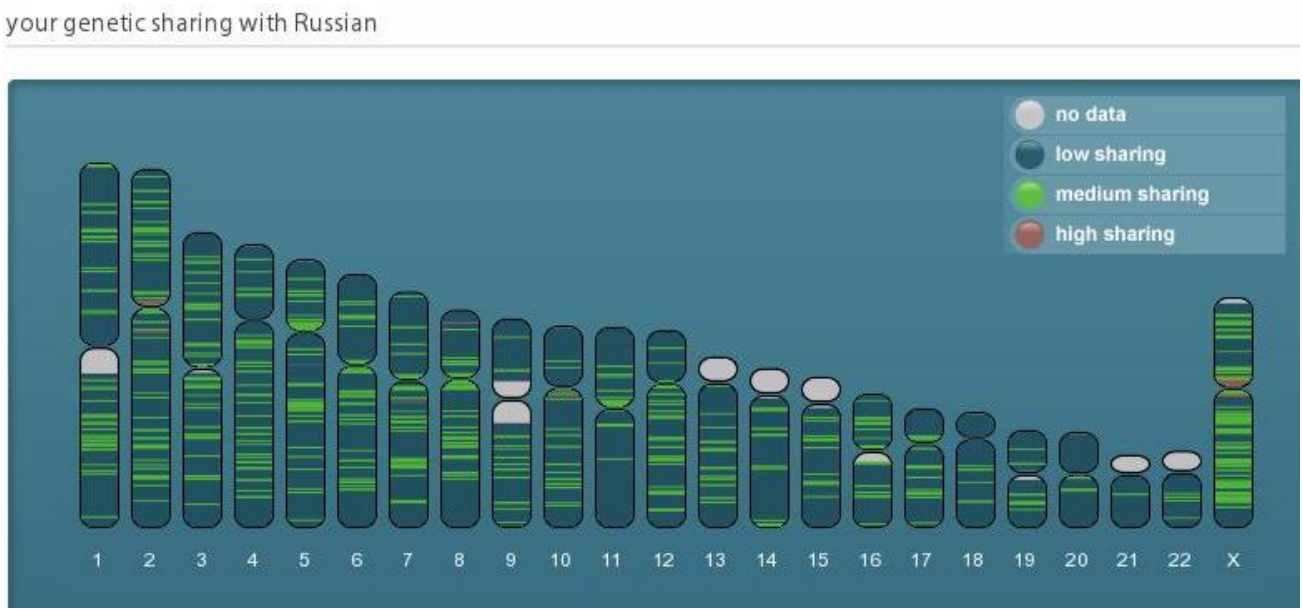
DeCodeMe

DeCodeme offers results that provide a probability for you of genetic predisposition to 29 various diseases. I was quite relieved to learn that I'm less than half as likely as the population as a whole to be predisposed to Alzheimers disease, for example.

The part of this test most applicable to the question of Ethnicity is the Ancestry portion of this product. My results, unsurprisingly were strongly European, as my heritage is more than half European. However, a close second of 6 possible global areas was SouthWest Asian, followed by Southeastern Asia which, without a category for Native American, would be as close as we can get.

Better yet, for each world area, matches are shown in order for various regions. For example my highest match is Orcadian (Northern British Isles) and my second highest is French (which seems to include the area of Germany) where my mother's family descends from, followed surprisingly by Russian.

Clicking on the Compare Me function shows your actual chromosomal comparison to their sample population, shown below:

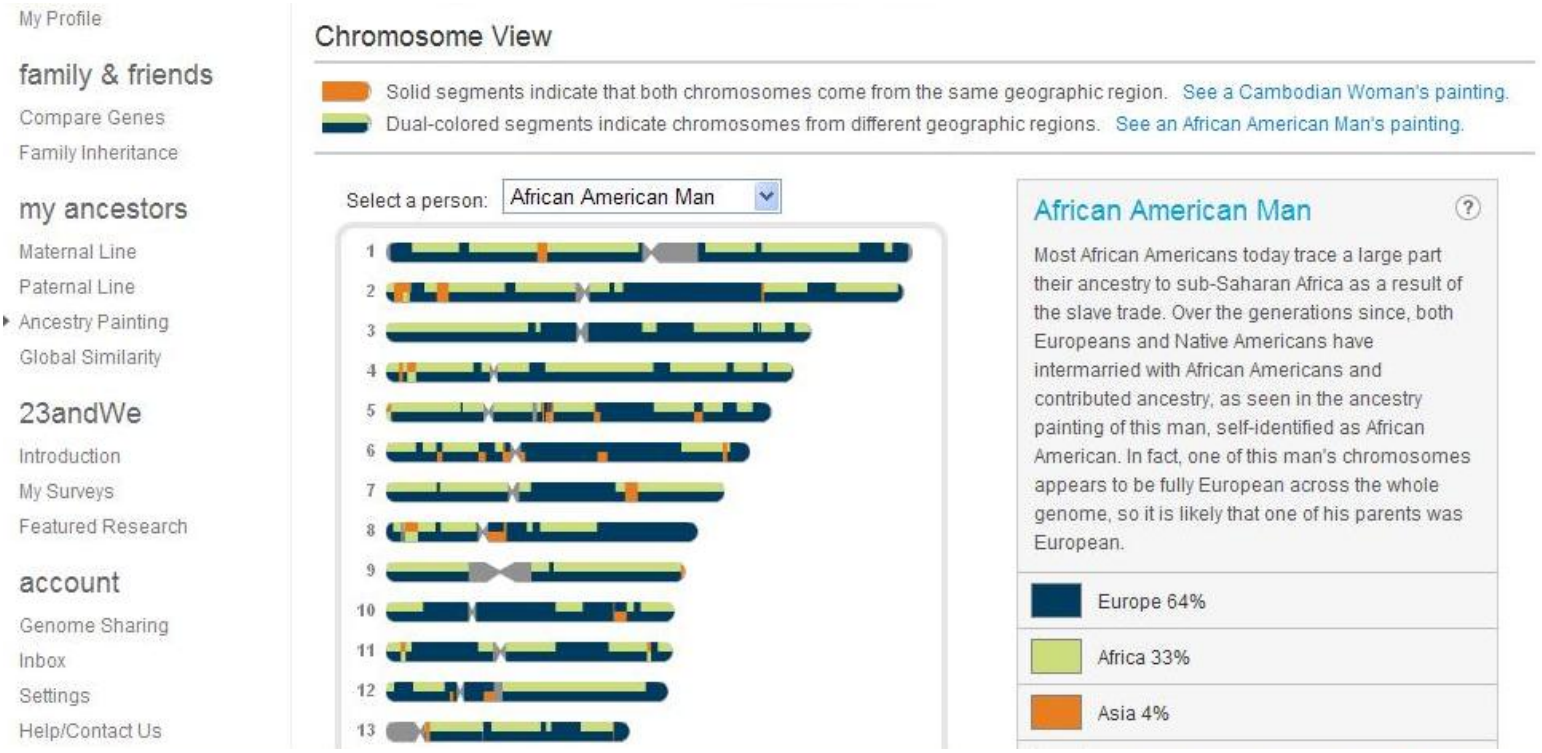


Working my way through my matches, I come to Africa, where I match best to the Mandenka people followed by the Yoruba. Both tribes are western African coastal tribes which would have been very susceptible to the slave trade.

Given that my ethnicity is primarily of European origin, followed closely by my Native heritage, I never thought that I would be able to have a tribal affiliation for my African heritage identified. I was absolutely thrilled with their ability to identify my closest tribal matches, and by delivering to the public the ability to see the locations of the matches, and to compare to others, it allows us to look for a higher than would be expected population match with those living in geographic groups, like Melungeons. It further will allow us to look for very specific allelic value matches on specific chromosomes that may by unique to a particular ethnic group. This holds the potential to be an extremely valuable tool moving forward in combination with the traditional yline and mtdna testing.

23andMe

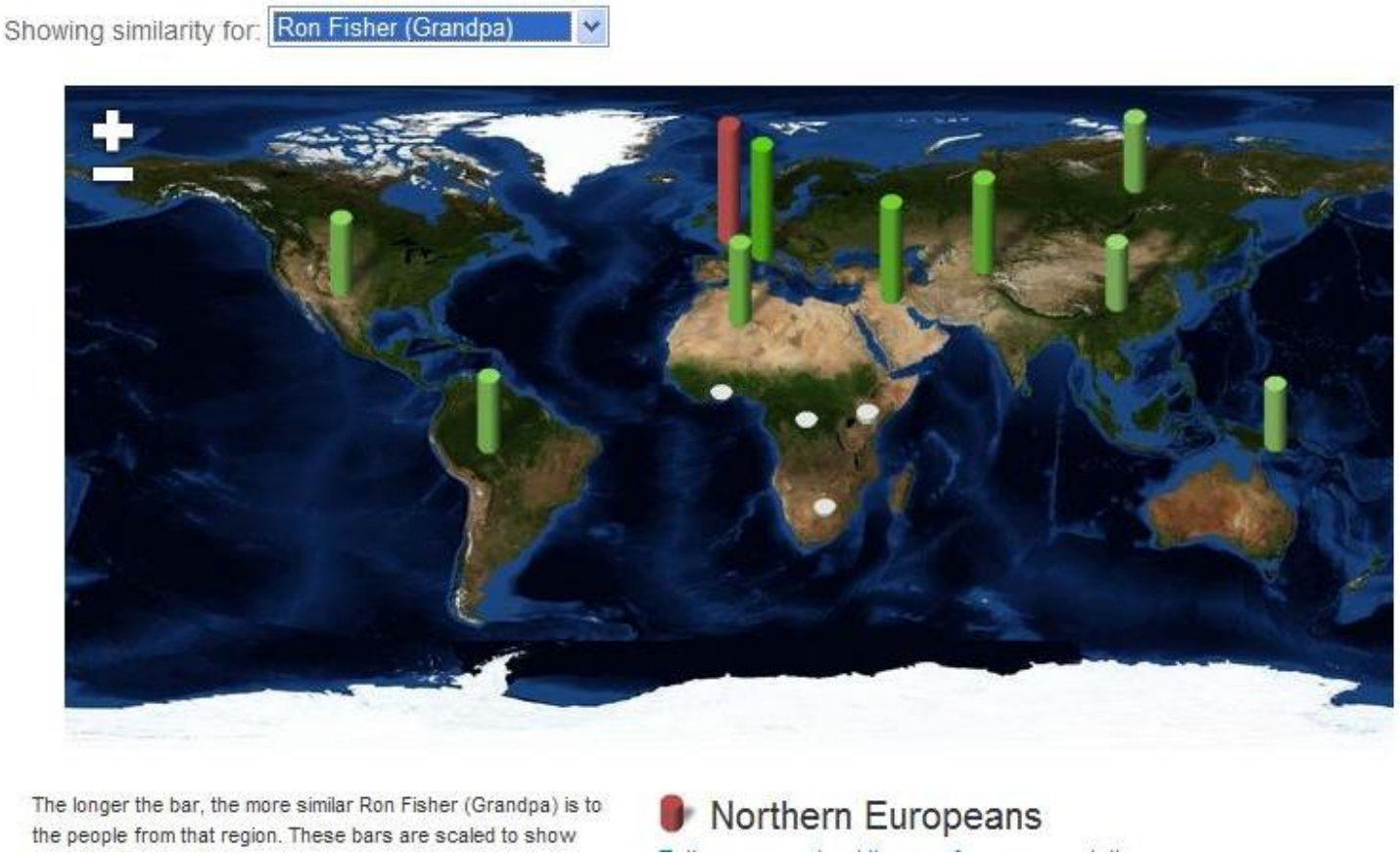
I only recently tested with 23andMe and don't have my results yet. However, they provide a demo ability and you can sign up as if you were a user and look at



sample data.

The data above is the sample data for an African American male. Each chromosomal segment is color coded, so you can see that he had many European and African segments, and since that data segment is split between tan and blue, he likely received half from each parent that is representative of

different ancestral homelands. However, he does have some that are completely blue and none that are completely tan, so one of his parents was mixed African and European and one had less or no African. This individual may well have Native American admixture as well, as there are several orange locations which could represent Native American admixture, and some are solid orange which would indicate that both parents carried this native allele combination. These locations would be target locations to look for others of a similar ethnic heritage for common inheritance patterns.



Using their example data, above we compare Grandpa to world reference populations. This display isn't nearly as informative as the deCodeMe results, but it is interesting. 23andMe's chromosomal view however, is superior and genealogically informative.

As you can see, great strides have been made in a very short time in this still infant field.

Individual Alleles

The fourth type of biogeographical testing is testing individual alleles. Ironically, it can be very informative and it is the least expensive of the various options. At

this point, only one individual allele identifying Native heritage is available for testing. At Family Tree DNA it is sold on their Autosomal Panel 3 and it is the D9S919 marker. This single marker only costs $15 to test.

The answer can be definitive. A value of 9 is a Native heritage indicator. A value of 15-18 means basicly European heritage. A value of 19 is quite rare and may also mean a Circumpolar Native heritiage, although that is not yet proven. Interestingly enough, one of our early Melungeon participants has a value of 19.

Only 37% of known native populations have the value of 9, but the value of 9 is not known in any other population. Therefore a value of 9 proves native heritage, but a value of anything else does NOT disprove the heritage. It simply means that this individual did not inherit this particular allele.

Since this test can positively confirm Native heritage and is inexpensive, I would hope that all of our Melungeon participants would test for this marker so that we can establish a base data set to work with.

Autosomal Summary

While the various autosomal BGA tests can indicate to you if there is ethnic heritage other than European, there are only two tests that can provide you with solid, indisputable, evidence of the source of your Native American or other ethnic ancestry. Those are yline and mitochondrial dna tests. It's important to try to fill in the blanks in your family tree pedigree chart by testing relatives who carry the yline and/or mtdna of the lines of your tree that you cannot personally be tested for.

Many people become frustrated with DNA testing when their goal is to determine Native, African or Asian minority ancestry and the best and only definitive tools they have at their disposal are the yline and mtdna tests.

In these cases, the best solution is to obtain the yline and mtdna of enough of the individuals in your pedigree chart to enable you to determine if they indeed had minority ancestry.

To assist my clients in this endeavor, I have constructed a color coded pedigree chart that I encourage them to use in order to find appropriate individuals to test, or who have tested already in surname projects perhaps, that will allow them to complete the boxes on their pedigree chart with names and haplogroups. As we know, haplogroups are the best indicator of the 4 basic admixtures (Indo-European, Native American (or other indigenous), African or Asian).

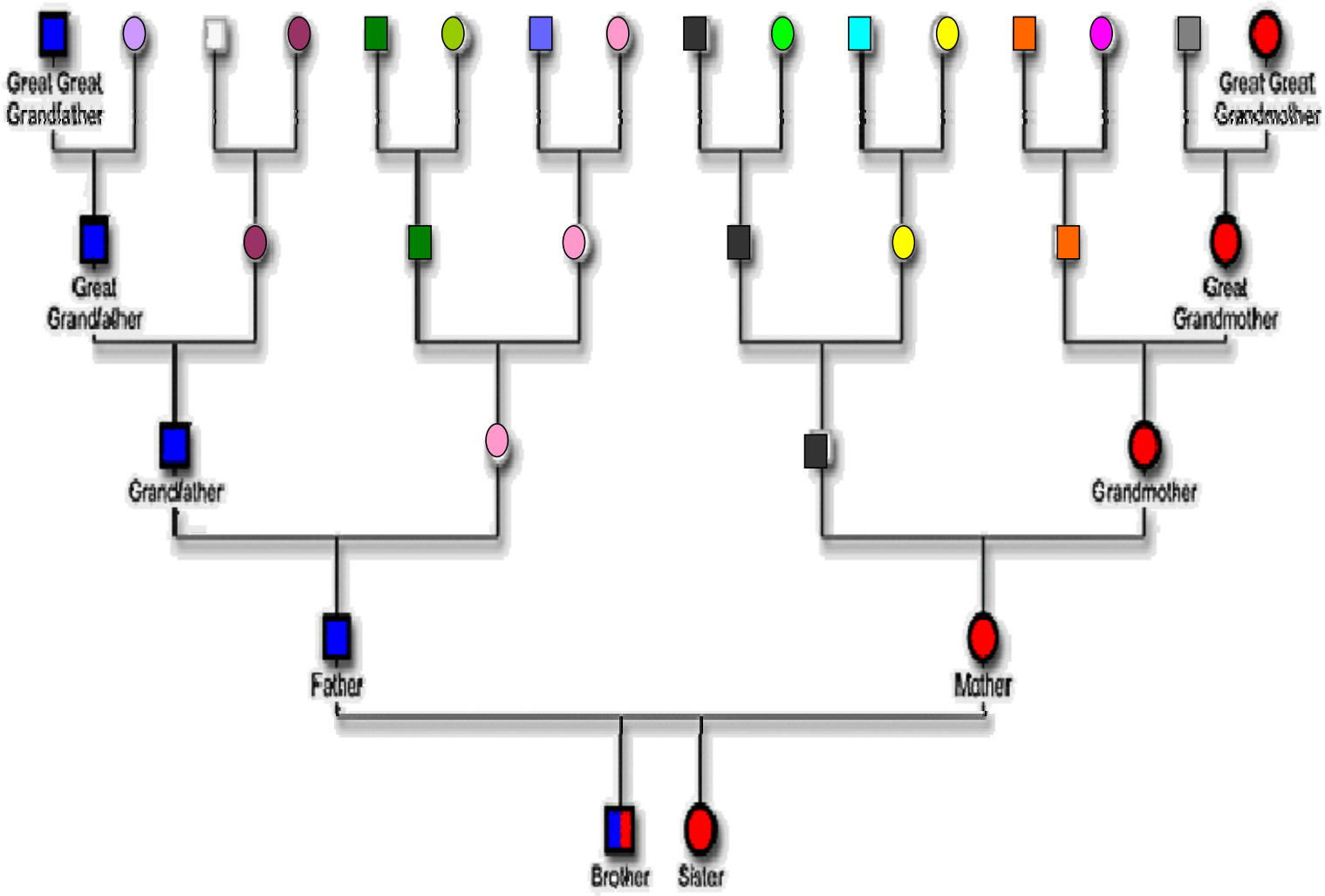
This information, combined with biogeographical tests, autosomal tests and even the newer tests at deCodeMe and 23andMe provide us with several tools to assess the presence or absence of minority ancestry. Only the mtdna and yline

tests can identify or eliminate the particular lines as Native heritage carriers on the maternal or paternal lines directly. What is does not tell us, of course, in these cases, is if the other (untested) parent is the contributor of the ancestry in question, so our goal is to go back as far as we can in our lines obtaining the appropriate tests to identify the haplogroups of our ancestors.

The methodology for doing this is much easier for yline than mtdna. With yline, the surname is tracked back in time, then forward, to identify living individuals carrying that surname to test. This can often be achieved using Rootsweb, Genforum, other mailing lists and resources such as the IGI and Ancestry.com.

Finding appropriate individuals to test for various mtdna maternal lines, is less obvious, but the same methodology is used. Going back in time, plain old fashioned genealogy is needed to identify the females who married and had female children. Track those female descendants to the current day and in the current generation, males or females who descend directly from a female carrying that DNA can be tested. This is more difficult, but it can and has been done quite successfully.

To assemble your dna pedigree chart, write the names of the appropriate ancestors on this chart in the appropriately designated areas. Then, by querying



the surname projects, ysearch, SMGF and other resources, see which of your yline surnames, represented by the squares, have surname projects and in those projects, if your lines may have already tested.

You can do the same thing with your female lines, although it may be more difficult to discern if descendants have already tested. Begin by recording their names, birth and death dates and locations near the appropriate boxes above. If you run out of space and have more known ancestors in a particular line, continue the line on another page by indicating that this is the continuation of the Blue line, or the pink line, or whichever line you are continuing.

It may help you with this exercise to obtain some colored highlighters or markers and print your pedigree chart using your genealogy software, coloring the various lines on your pedigree chart with differently colored markers. For analysis purposes, I try to work with a single page when possible to be able to visually see relationships and DNA admixture.

In the above color coded pedigree chart, males are square boxes and females are ovals.

The red/blue box on the bottom row represents a currently living male and his sister is shown as a red oval.

Males carry Yline dna inherited from their father which is represented by blue boxes. Males pass the Y chromosome to their sons, which is what makes them male. Males carry mitochondrial DNA inherited from their mother, but they do not pass it to their children.

Females carry only mitochondrial DNA, shows by the ovals. Females pass it to their children of both sexes, but only the females pass it on. Mtdna is not admixed with the DNA of the father.

The coloration tracks the DNA ancestral pattern of a particular individual or line.

In the first example, the male child carries the blue yline dna of his father, who carries the blue yline dna of his father, etc, until you climb the family tree until you run out of blue ancestors. Matches could come from descendants of any of the males in that family line with blue ancestry, or their brother, or their nephews, etc.

In the second example, the male and female children BOTH carry the mtdna of the red maternal line, but it is only tracked backwards up the tree in the red maternal line. This means that currently living males can test for both their maternal and paternal lines, but the maternal line is only passed to future generations via females. Men don’t pass it on.

Therefore the blue father carries the blue yline dna of his father, but he also carries the pink mtdna of his mother. If he is still living, he can test for both. If he has died, then another individual descended only through females from any of the individuals designated by pink ovals can be tested for the same mtdna that the father carried. These are known as proxy or surrogate tests, where another individual tests "in place of" the person whose DNA you would like to test, but can't.

All genetic genealogy results need to be accompanied by genealogical research to unravel the historical context for the lives and trials of our ancestors. DNA testing may well answer the question what and who, but the why is typically revealed only by studying the history of the times in which they lived.

What Do Melungeon Descendants Need to Do?

Melungeon descendants need to participants in both yline and mitochondrial DNA testing.

In addition, I would encourage them to purchase the D9S919 allele marker test for $15.

Each person needs to research their lines and provide the project administrators with their associated genealogy that is relevant to each line tested.

Participants need to construct a personal DNA pedigree chart for themselves and attempt to obtain the DNA signature for each person represented on the chart.

This is the only way a complete picture can be obtained for each genealogy line relative to Native, African and European admixture sources. It's not unlikely that several lines will have already tested, especially since the Melungeons tended to intermarry.

Join the Melungeon DNA project.

Continue to document your personal genealogy and historical research. DNA without genealogy and history is only half of the story.

Prepare to make new discoveries and meet cousins you never knew you had!!!

Links and Resources

MHS Blog – http://melungeon-historical-society.blogspot.com/ - subscribe for regular updates!

Melungeon DNA project – http://www.familytreedna.com/public/coremelungeon/

Melungeon DNA Project Administrators:
Penny Ferguson – pennyferguson@alltel.net
Jack Goins – jgoins@usit.net
Janet Crain – jcrain2@pgrb.com

DNA Testing – www.familytreedna.com

DNA Analysis – www.dnaexplain.com or contact Roberta at restes@comcast.net

Melungeon Web Page - http://www.jgoins.com/