An Introduction to

DNA and Genetic Genealogy

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INTRODUCTION

Remember that hollow feeling in the pit of your stomach when you realized grandma was gone and she was the last person who could identify those unknown people in hundreds of photos? Don't let that happen again by putting off genealogical DNA testing until the last person in a line is gone. You might be fearful because you do not understand DNA. But you probably didn't understand what "third cousin, twice removed" was when you first started genealogical research either. Now is the time to

educate yourself, or find someone to help you decide what testing will contribute the most to your personal family history problem or brick wall.

While you add to your family history you can be part of a group that is advancing the science of DNA and genealogy—genetic genealogy. Astronomy and genetics are two of the hard sciences where amateurs can contribute as much to the knowledge base as scientists. Amateur astronomers using backyard telescopes discover comets and other astronomical bodies. Amateur geneticists, also known as genetic genealogists, are discovering new links between families, unraveling incorrect links, and even contributing to our understanding of the migration of humans across the world thousands of years ago.

DNA test results are used to support a lineage documented by a paper trail. DNA results do *not* replace a paper trail and cannot prove an exact relationship between two people. DNA results show a statistical probability of a link between two people to support conclusions based on your paper trail. Once you have DNA test results you play a matching game with others who have tested to find common ancestry.

This article gives an introduction to the types of DNA testing used for genealogy, what you can support statistically (not prove) with DNA information, how to get DNA testing (even free), where to get DNA testing, and resources for learning more.

DNA TESTS FOR GENEALOGY

There are currently four types of DNA tests commonly used in genealogical research.

1. Y-chromosome DNA (Y-DNA) direct paternal line testing that can be done only on DNA from a male. This testing is very popular because the Y-DNA follows the paternal line, the surname in most western cultures, and can support conclusions about links in the paternal line.

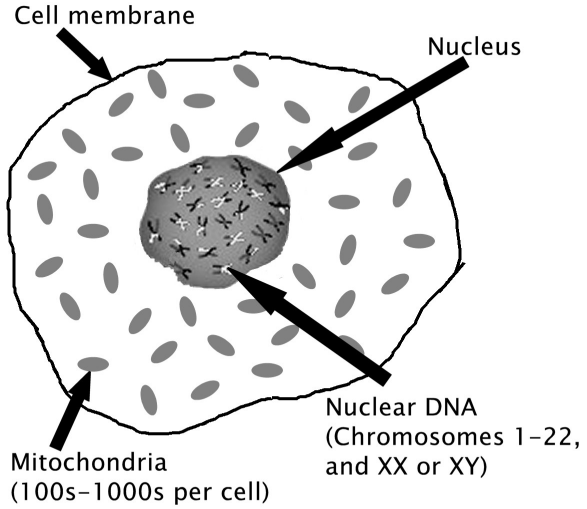
2. Mitochondrial DNA (mtDNA) direct maternal line testing can be done on DNA from male or female. This testing can support conclusions about links in the maternal line.

3. Autosomal or admixture DNA (atDNA or aDNA) testing analyzes the DNA inherited from the all ancestors including your direct paternal and maternal lines. Some testing companies use this type of testing to indicate ethnic makeup. It is also used in paternity, maternity, and siblingship tests.

4. SNP (pronounced snip) testing primarily is used for deep ancestry testing and confirms your branch of Y-DNA and mtDNA trees. Rare SNPs in Y-DNA and mtDNA can also indicate recent common ancestry.

DNA in Cells

This diagram illustrates a cell. The cell nucleus is where nuclear DNA is located. Nuclear DNA includes two copies each of twenty-three chromosomes; the autosomal DNA includes the X and Y chromosomes. Mitochondrial DNA is outside the cell nucleus and there are many copies in each cell.

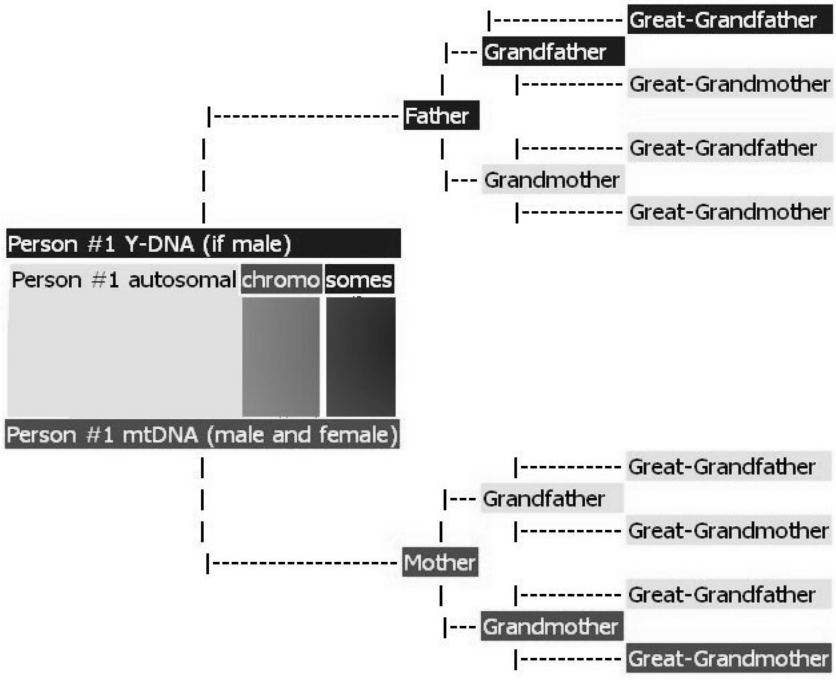


Cell diagram © 2009, Debbie Parker Wayne

DNA and Pedigree Chart

This pedigree chart indicates the line(s) along which each type of DNA is inherited. Y-DNA is inherited along the upper line of the pedigree chart—the direct male line through father, paternal grandfather, and so on. Mitochondrial DNA is inherited along the lower line of the pedigree chart—the direct female line through mother,

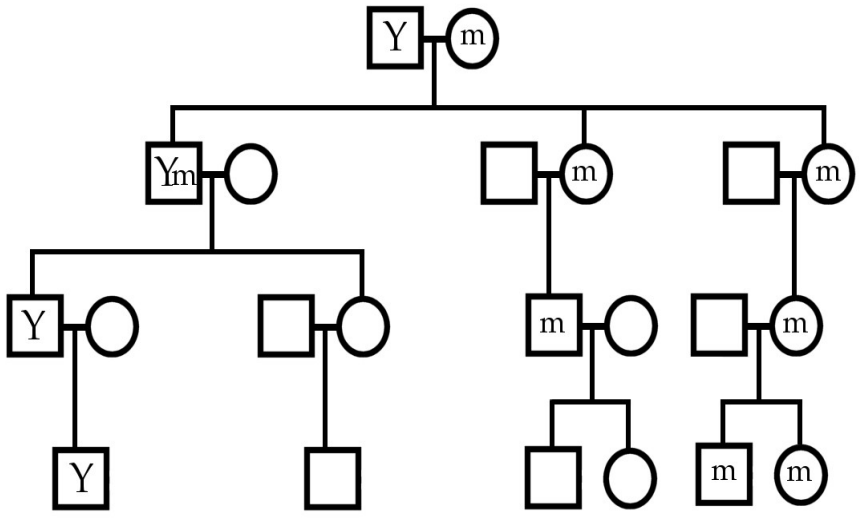
maternal grandmother, and so on. The nuclear or autosomal DNA is inherited from all lines on the pedigree chart. Due to DNA recombination it is impossible to know how much DNA any individual has inherited from any given line other than the Y-DNA and mtDNA lines. When a child is conceived the DNA of each parent mixes and combines creating a unique version of **that parent's** DNA. That unique combination is passed on to the child where it is paired with the recombined DNA inherited from the other parent. Statistically, each parent passes ½ of the DNA of two grandparents to a child. In actuality, the mix is not exactly 50–50 due to recombination.



DNA Inheritance Pedigree Chart
© 2009, Debbie Parker Wayne

Inheritance of Y-DNA and mtDNA

Understanding DNA inheritance is useful when determining which person to test to support or disprove any given hypothesis. The following chart illustrates the inheritance of Y-DNA and mtDNA from an ancestral couple. Males are indicated by a square, females by a circle in the chart. Male children inherit a Y chromosome from their father and pass it down to each son as indicated by the "Y" in the chart. All children inherit mtDNA from their mother. Only daughters pass it down to their male and female children as indicated by the "m" in the chart. Drawing a similar chart for your family can help locate a living person whose DNA test results can be used as evidence in your family history. Often when a parent or a brother is no longer living, a remote cousin can be found whose DNA will help prove or disprove a hypothesis.



Y-DNA and mtDNA inheritance © 2009, Debbie Parker Wayne

USING Y-DNA IN GENEALOGY

Y-DNA test results are most often used to determine if people with a common surname have common roots, to divide them into family groups, to determine deep roots of a lineage, and to test a theory about paternity. Rare surnames may obtain results by testing as few as twelve markers. For most surnames better results will be obtained when testing more markers. Many genetic genealogists now recommend a minimum of thirty-seven markers be tested. Some recommend testing as many markers as you can afford as this saves money in the long run. Your personal goals and finances should be considered when determining how many markers to test.

Y-DNA test results consist of what is called a haplogroup identifier, for example, R1a or R-M198, and a list of marker names and values. The haplogroup identifier is used to place a person in a general group. The marker values are compared with the values of other individuals in the group to form family groups. As we learn more about DNA we see that some markers change rapidly and some seem to rarely change. These changes, called mutations, can help link families as a difference on a rapidly changing marker might indicate a closer likelihood of a relationship than a difference on a slowly changing marker. It may be necessary to consult with a knowledgeable genetic genealogist to fully understand these subtleties.

USING mtDNA IN GENEALOGY

Mitochondrial DNA (mtDNA) can be used to determine a relationship along the maternal line—such as differentiating which of several wives is the mother of specific children and for other purposes. mtDNA is passed from a mother to all her children. The daughter's will pass that mtDNA to their children. A son's children will receive their mtDNA from their mother, not their paternal grandmother.

Mitochondrial DNA test results consist of a haplogroup identifier, such as U or U5, and a list of marker locations that differ from a reference sample. The haplogroup identifier, as with Y-DNA, is used to place a person in a general group. The markers values are compared with the values of other individuals in the group to form family groups. But since mtDNA changes at a slower rate than Y-DNA the groups go much further back in time. The slow change rate and the fact that women in western cultures change surnames with marriage make it more difficult to use mtDNA without a deeper understanding of exactly how it can and cannot be used.

USING atDNA IN GENEALOGY

Autosomal DNA, the twenty-three pairs of chromosomes in the cell nucleus that includes the X and Y chromosomes, is used to determine ethnic percentages in your lineage. Because nuclear DNA other than the Y chromosome gets shuffled, or recombined, each time a child is conceived the percentages of DNA from any particular grandparent or great-grandparent can never be known with accuracy. If an ancestor of a

particular ethnicity is more than a few generations removed from the person whose DNA is tested it is possible the ethnicity will show a low percentage or not even be indicated at all in the DNA.

Autosomal DNA testing can be useful to some and is encouraged to help contribute to the databases for future testers. But don't be shocked if it doesn't confirm your Native American or African American or other ethnic heritage. You may be one of the lucky ones who inherited much of your DNA from your Cherokee ancestor, but there is no guarantee. Of course, as more people are tested and we learn more about DNA the usefulness of this testing could improve in the future. Consider autosomal DNA testing as an investment in the future of genetic genealogy.

The CODIS markers used by North American law enforcement, other markers used by European law enforcement, and some called "miniSTR" markers are used not only for ethnicity testing, but also for paternity, maternity, and siblingship testing. These markers have been measured in the general population and are used to calculate the probability of a random person in the same ethnic group being one of the parents. Paternity, maternity, and siblingship tests are more accurate when more of the people involved are tested. It is difficult to prove siblingship when only testing two siblings without one or both parents also being tested despite what you see on the CSI shows on television.

Autosomal DNA test results consist of a marker name and pair of values. The value pair consists of one value inherited from the father and one from the mother. Relationship testing compares the values of multiple individuals and statistically computes the likelihood of a paternal, maternal, or sibling relationship between the two individuals. Ethnicity testing compares the values to those in a database and computes the statistical percentage of a specific ethnicity based on those marker values. It should be obvious that accuracy depends on having a very large number of accurately identified samples in a database for computations. Due to the inherent problems with ethnicity testing using the current state of technology, some testing companies will only test ethnicity on Y-DNA and mtDNA samples and will not do autosomal testing.

USING SNP DNA TESTS IN GENEALOGY

SNP (Single Nucleotide Polymorphism) tests are useful in genealogical research if a rare Y-DNA or mtDNA mutation exists in a line. The rare mutation indicates a close relationship between individuals. SNP testing is much more useful in showing migration of humans over thousands of years and is used by anthropologists to study the deep ancestry of humans.

BENEFITS OF DNA IN GENEALOGY

DNA testing will not provide you with everything you need to know about your family history. It is only useful to disprove a theory or to support conclusions based on

traditional research. DNA testing is most useful if you start with a hypothesis and use the results to disprove the hypothesis as in the Parker family story below. If the DNA results do not disprove a hypothesis they may be used to support a conclusion such as a link to a family. DNA results will not be able to tell you which of several brothers your line descends from, but it can show your line does descend from one of the brothers or a common ancestor of the brothers.

DNA testing can save time and money directing your future genealogical research. Anyone in Texas with the surname Parker has probably been told he or she is related to Quanah Parker, or the Parkers of the Fort Parker Massacre in 1836, or maybe Bonnie Parker, Clyde Barrow's companion in crime. My aunts told me all of these stories. But a Y-DNA test showed my Parkers do not have Native American Y-DNA and do not match the male Parkers descended from the Fort Parker folks. If we are related to these Parkers, it could be through a female relative who married into my Parker line. But I now know I do not need to spend a lot of time and money trying to find a Parker link to Quanah or his mother's male Parker relatives.

CAUTIONS WHEN USING DNA IN GENEALOGY

You should not take a DNA test unless you are willing to accept what the science shows. Remember our ancestors were people just like us—some good traits, some bad traits, maybe not always able to live up to the high standards of our better angels. DNA

tests can indicate an unexpected and unknown event causing a disagreement between the parentage indicated by a paper trail and that indicated by DNA.

A break in Y-DNA is often referred to as a non-paternity event (NPE). This is a poor descriptor for what might better be called "misattributed paternity" or a "surname discontinuity event." This was recently discussed on the Yahoo group list restricted to members of the International Society of Genetic Genealogists. Some people jump to the conclusion that mama must have “known” someone other than papa in the Biblical sense of the word. If infidelity was involved it could have happened anywhere in the lineage between two tested men. But it is also possible the family adopted a child or that step-children took the surname of a step-father, as well as several other possibilities that are not known to the living generations of the family.

Because of the way DNA recombines in each newly created child it is difficult to pinpoint ethnicity using DNA, unless the ethnicity comes down the straight male line (Y-DNA) or female line (mtDNA). If you saw the PBS program, “African-American Lives,” (http://www.pbs.org/wnet/aalives/) you will remember that some of the Black celebrities tested showed very low percentages of African American ancestry. Statistically, you should get ¼ of your DNA from each of your grandparents through your parents. But in reality you get more from some grandparents and less from others. And the farther you are from an ethnic ancestor the less chance you have of keeping a measurable amount

of their DNA to indicate that ethnicity. This should not discourage ethnic testing, but you must be aware of the limitations.

HOW AND WHERE TO GET DNA TESTING DONE

TSGS has partnered with Family Tree DNA to offer DNA testing to individuals at discounted group project rates. You can test through the TSGS project and receive the same discounted rate as other projects plus TSGS receives a small amount of revenue that can be used to promote other genealogical and preservation projects. Once your test results are received you can also join a surname, mitochondrial, and regional project. Family Tree DNA now allows each person tested to join an unlimited number of projects.[1] Use the TSGS Web page at http://www.rootsweb.ancestry.com/~txsgs/tsgsdnaone.html to order a DNA test through the TSGS partnership with Family Tree DNA.

Family Tree DNA (http://www.familytreedna.com/) and other cooperative ventures, including the National Geographic Society's Genographic Project and AfricanDNA.com, now comprise the largest non-medical DNA testing program in the world. Family Tree DNA was founded in 1999 by Bennett Greenspan, an entrepreneur and life-long genealogy enthusiast, turning a hobby into a full-time vocation. His effort and innovation created the burgeoning field now known as genetic genealogy.[2] With

[1] Steven Danko, "Updates on Family Tree DNA," Steve's Genealogy Blog (http://stephendanko.com/blog/2009/03/25/updates-on-family-tree-dna/ : accessed 14 April 2009).
[2] "Who's Who at Family Tree DNA," Family Tree DNA (http://www.familytreedna.com/about.aspx : accessed 14 April 2009).

over 200,000 records, Family Tree DNA has the largest database of its kind in the world.[3]

If you just cannot afford to pay for a DNA test, but want to contribute to the wonderful discoveries being made today, consider using Sorenson Molecular Genealogy Group (SMGF) (http://www.smgf.org/) for testing. SMGF will take your DNA sample and test it for research purposes at no charge. You must submit a four-generation pedigree chart with your sample. They do not send test results to you directly, but place them into a publicly available database on their Web site. It can be many months (or years) before results are posted to the public database. The pre-1900 family information from your pedigree chart is included in the database so you can search for your surnames to find your results. SMGF recently partnered with GeneTree to provide test results to those who contributed DNA samples, but a fee is charged for this service.

Considerations When Choosing a Testing Company

Probably the most important consideration when choosing a testing company is how big their database is for comparisons. Remember, you learn the most about your ancestry when you compare DNA results with others. It is also important to find a company that offers the particular test you are interested in at a cost you can afford.

[3] "Family Tree DNA," Family Tree DNA (http://www.familytreedna.com/ : accessed 14 April 2009).

Other things to consider are:

- Do you want your DNA sample stored for more tests in the future? This is illegal in the UK, but in the U.S. it is legal. Family Tree DNA keeps your DNA sample for twenty-five years allowing additional tests to be performed without sending a new sample.
- Does the company provide a secure database to protect your privacy?
- Does the company have a reputation for prompt and accurate customer service?

The International Society of Genetic Genealogists (ISOGG) provides pages comparing the testing done by different companies for Y-DNA (http://www.isogg.org/ydnachart.htm) and mtDNA (http://www.isogg.org/mtdnachart.htm). This information can help you choose a testing company if you do not wish to join the TSGS project.

GETTING HELP INTERPETING DNA TEST RESULTS

Unless you plan to educate yourself on using DNA test results to further your family history, you will need help interpreting and using the results. Project administrators can be very helpful in interpreting DNA test results. Some family groups have researchers who can offer assistance. For example, in the Parker DNA Surname Project, the project administrator helps everyone in the project and posts information on all project participants at http://web.utk.edu/~corn/parkerdna/. Two of us work together to help those whose test results place them in Parker family group #1. We reorganize and

publish the information at http://debbiewayne.com/parker/dnaproject_migrations.htm in ways that will help those in family group #1 find links between the members.

If you wish to learn more than the project administrator may be able to help with there are professional / genetic genealogists who can help. The Association for Professional Genealogists (APG) at http://apgen.org/ allows members to list genetics as a research specialty. APG does not certify or validate specialties so you should check each member's Web site or biography for their specific experience and affiliations before selecting a specialist. The International Society of Genetic Genealogists (ISOGG) lists DNA Consultants/Services on a link from their home page at http://www.isogg.org/.

PRIVACY CONCERNS

DNA projects you join, such as the TSGS project, surname projects, or regional projects, should protect your privacy. The generally accepted policy is to identify people in a project by a participant number that cannot be traced back to an individual. If you provide a pedigree chart to be publicly displayed it is good practice to list only the deceased individuals and state something similar to "this DNA sample was provided by a descendant of John Doe (born 1 January 1883, died 31 December 1946)." This should give a possible cousin enough information to make a link. You can decide whether or not to share more information once a cousin contacts you.

You should not worry about your DNA sample being used to deny insurance coverage, but there are very few medical conditions that are indicated by the DNA testing done for genealogical purposes. A Y-DNA test might detect infertility. The full mtDNA sequencing test, called mtFULLsequence, might detect mitochondrial disease. The mtDNA and mtDNAplus tests do not reveal any medical conditions. If this is a concern, you should take steps to protect your privacy.

PUBLIC DATABASES

No matter which company you test with you can also place your DNA results into a public database. The public databases allow you to compare results with those who tested with other companies.

Some testing companies make it easy for you to upload your results to a public database by providing you with a button that automates the upload process. With others you have to manually type in your DNA results. As the exact nomenclature and testing values vary between the companies it is very important to be sure you understand whether and how to convert your DNA marker values before entering them manually into any database. Some of the public databases are:

- www.ysearch.org/ - Family Tree DNA Y-DNA Public Database
- www.ybase.org/ - DNA Heritage Y-DNA Public Database
- www.yhrd.org/ - YHRD-Y Chromosome Haplotype Reference Database

- www.smgf.org/ - Sorenson Molecular Genealogy Foundation Public Database (Y-DNA and mtDNA)
- www.mitosearch.org/ - Family Tree DNA mtDNA Public Database

Several public sites list DNA of famous people and ancient remains. You can have fun with your DNA test results by comparing to these lists. Check out ISOGG at (http://www.isogg.org/) and click on "Famous DNA" in the navigation bar.

RESOURCES FOR LEARNING MORE

A good place to start your education on DNA is with any current high school biology textbook. The March 2009 issue of the Association of Professional Genealogists *Quarterly* has a good article on DNA basics by Stephen P. Morse.[4] Morse's article covers the basics in a very clear and understandable manner.

Several beginner-level books are available on genetic genealogy. *Trace Your Roots with DNA* by Megan Smolenyak Smolenyak and Ann Turner[5] and *Unlocking Your Genetic History* by Dr. Thomas Shawker[6] are good introductory books for using DNA for genealogical research. If you plan to administer a DNA project or just want to understand

[4] Stephen P. Morse, "From DNA to Genetic Genealogy: Everything You Wanted to Know But Were Afraid to Ask," Association of Professional Genealogists *Quarterly* Vol. XXIV, No. 1 March 2009, 27-37.
[5] Megan Smolenyak Smolenyak and Ann Turner, *Trace Your Roots with DNA: Using Genetic Tests to Explore Your Family Tree* (Emmaus, Penn.: Rodale Press, 2004).
[6] Thomas Shawker, *Unlocking Your Genetic History: A Step-by-Step Guide to Discovering Your Family's Medical and Genetic Heritage* (Nashville, Tenn.: Rutledge Hill Press, 2004).

more about the fascinating details of DNA testing and how to analyze the details using what are called cladograms study *Forensic Genealogy* by Colleen Fitzpatrick, PhD,[7] and *DNA & Genealogy* by Colleen Fitzpatrick with Andrew Yeisier.[8]

If you are interested in deep ancestry, human history going back thousands of years, check out the books *The Seven Daughters of Eve* by Bryan Sykes,[9] *Adam's Curse* by Bryan Sykes,[10] and *The Journey of Man* by Spencer Wells.[11]

Web sites and lists where you can learn more about genetic genealogy include:

- International Society of Genetic Genealogy – http://www.isogg.org/
- Journal of Genetic Genealogy – http://www.jogg.info/
- Charles Kerchner's site – http://www.kerchner.com/kerchdna.html
- The DNA-Newbie Yahoo Forum – subscribe and ask questions – http://groups.yahoo.com/group/DNA-NEWBIE/
- RootsWeb Genealogy DNA list – subscribe and ask questions – http://lists.rootsweb.ancestry.com/index/other/DNA/GENEALOGY-DNA.html
- RootsWeb also offers surname related DNA discussion lists – see http://lists.rootsweb.ancestry.com/index/other/DNA/

[7] Colleen Fitzpatrick, *Forensic Genealogy* (Fountain Valley, California: Rice Book Press, 2005).
[8] Colleen Fitzpatrick and Andrew Yesier, *DNA & Genealogy* (Fountain Valley, California: Rice Book Press, 2005).
[9] Bryan Sykes, *The Seven Daughters of Eve* (New York: W. W. Norton & Co., 2001).
[10] Bryan Sykes, *Adam's Curse* (New York: W. W. Norton & Co., 2004).

[11] Spencer Wells, *The Journey of Man* (New York: Random House, 2002).

CONCLUSION

This is only a brief introduction to how genealogists can use DNA testing and how to learn more. Like any tool, you must learn how to use DNA to achieve your research goals. As more testing is done and the DNA databases grow in size we will know more about our genetic heritage and our connections and make better use of DNA analysis. Don't you want to be part of this exciting new world and have no regrets that you waited too long? Use the TSGS Web page at http://www.rootsweb.ancestry.com/~txsgs/tsgsdnaone.html to order a DNA test through the TSGS partnership with Family Tree DNA. Then plan to join a surname, regional, and mitochondrial project and use the public databases; let DNA help you find new cousins and break down your brick walls.

