Chapter 8: Genetic Genealogy

Introduction

Some ancestors are difficult to trace. Adoptions illegitimacies, name changes and migrations can present brick walls in our research that seem impregnable. In other cases, documentation that would shed light on our family history is missing, destroyed or non-existent. These challenges can be discouraging, but they are also possible to overcome. Despite the obstacles that may arise through the historical record, each one of us also carries an additional biological record of our genealogy. Every cell of our body carries part of our family story and the story of the larger human family. Our own DNA assists us in understanding and interpreting that story and breaking down walls that conceal our hidden history.

In recent years, genetics has grown to be one of the most important and rapidly developing fields of science. The effects of this growth are evident in agriculture, medicine, criminal investigation, anthropology and countless other fields and disciplines. Paralleling this growth, genetics has also become an important tool in genealogy and family history. Difficult-to-trace ancestors who migrated, changed their names, avoided the written record, or who were adopted, orphaned or illegitimate can be identified through the genetic signatures of their living descendants. By linking living individuals who share common DNA, genetic testing facilitates collaboration among distant relatives as they seek to overcome obstacles in their traditional research. Though genetic genealogy is a very useful and insightful tool, it cannot tell us everything about our family history. DNA analysis can direct our efforts in traditional historical research which in turn provides the necessary context and interpretation of relationships proposed through genetic testing.

Genealogists should be interested in DNA for the same reason they are interested in wills and heirlooms: inheritance. Some of the most useful genealogical records demonstrate family relationships by the names, privileges, education and property that individuals pass on to their descendants. Probate records, land transfers, and most other genealogical resources show inheritance in some aspect. As a result they help to confirm connections between generations of family members. One of the most important aspects of turning our hearts to our fathers is recognizing what we have inherited as a legacy from them. James E. Faust declared, “In many ways each of us is the sum total of what our ancestors were. The virtues they had may be our virtues, their strengths our strengths, and in a way their challenges could be our challenges. Some of their traits may be our traits” (Faust October 2003). In a very real sense, genetic genealogy investigates the “sum total of what our ancestors were” and connects us to our distant family members based on what we have inherited from them. While genealogical records may show the distribution of land or physical possessions, genetic inheritance instead shows the distribution of DNA among descendants of a common ancestor. However, because of its universal, equal and independent nature, genetic inheritance is not bound by some of the limitations connected with legal inheritance.
**Universal Inheritance**

Genetic inheritance is universal. From conception, each individual receives a genetic legacy from his or her parents regardless of age, birth order or gender. In some societies, legal inheritance customs favored or gave exclusive right to a single heir. Often the heir may not have even been a direct descendant of the family, but might have been a nephew, uncle, or other relative. As a result, some descendants of these families were left a considerably smaller portion or no inheritance at all. This phenomenon is impossible in genetic inheritance. No matter what the social or economic situation of a child may be, they will always receive a genetic inheritance from their parents.

**Equal Inheritance**

Genetic inheritance is equal. Although some children clearly inherit different genetic traits than their siblings, they all will receive the same amount of genetic material. Situations that might exclude a child from legal inheritance do not apply to genetic inheritance. Children who were abandoned, adopted, disowned, disinherited, estranged, orphaned, absent at their parents’ deaths, or who died before their parents all received a genetic inheritance despite the disadvantages they and their descendants may have had in legal inheritance. Every person receives half of their DNA from their mother and half of their DNA from their father and every person carries the same amount of DNA.

**Independent Inheritance**

Genetic inheritance is independent. If one child inherits certain genes from a parent, this does not exclude other children from inheriting the same genes. Because legal inheritance deals with the distribution of material objects among family members, when one heir receives an item exclusively, no other heir can exclusively inherit that same item. For example, if one child inherits complete rights to a portion of the family land, no other child can inherit complete rights to that same plot. Although they might inherit other portions of the land, the fact that one child has already inherited that land excludes the other siblings from also inheriting that land. Because genetic inheritance actually passes copies of each parents’ genes to their children, parents retain the original DNA, and multiple children can inherit a copy of the same portion of their parents’ DNA. In fact, it is this shared inheritance that makes genetic inheritance so useful to the genealogist.

Because genetic inheritance is universal, equal and independent, DNA analysis can help to solve family mysteries that traditional genealogical resources cannot on their own. DNA analysis reveals the portions of genetic material that multiple individuals have inherited from common ancestors.

**DNA Basics**

DNA is the basic unit of inheritance in living organisms and is responsible for the transmission of

![Figure 1: Complimentary Base Pairs](image-url)

Figure 1: Complimentary Base Pairs
physical and behavioral traits to subsequent generations. It is composed of four different chemical bases: adenine, thymine, guanine and cytosine, which are commonly referred to by the one-letter abbreviations A, T, G and C. DNA bases are linked together on a sugar backbone and then paired against another strand that is complementary, thus forming a series of “base-pairs”. Adenine pairs with Thymine (A-T) and Guanine pairs with Cytosine (G-C) (See Figure 1). When the two strands of DNA interact, they form a double helix structure that helps to preserve and protect the integrity of the genetic code (See Figure 2).

Just as a binary computer code provides the basic level of instructions for a computer program, DNA provides instructions for the assembly of proteins, and heritable traits. Many of an individual’s physical and behavioral characteristics are dependent on the proteins their body produces. A protein’s role and function is dependent on its structure. Its structure is dependent on its amino acid sequence. Its amino acid sequence is dependent on the DNA sequence used to assemble it. In this sense, DNA is the “biological binary” which encodes for the characteristics and traits that combine to make each individual unique.

Each human has billions of cells, and each cell, with the exception of sex cells and red blood cells, has a complete copy of their DNA. Within the cell there are two places where DNA is stored: the nucleus and the mitochondria. (See Figure 3)

Among other functions, the mitochondria serve as the energy powerhouses for the cell. They have their own set of DNA which makes them unique from other cellular structures. Most cells have multiple mitochondria and each mitochondrion has multiple copies of the mitochondrial DNA.

The nucleus is the control center of the cell where DNA is copied, maintained and regulated. Most DNA in humans is stored in the nucleus. Every time a cell divides to form two daughter cells, a complete copy of the nuclear DNA is passed on to the new cell as well. Because DNA provides the instructions for how a cell should function,
this copy must be identical to the original. Even though each cell has a complete copy of an organism’s DNA, different types of cells read and express different portions of the genetic code resulting in different cell functions. This is what differentiates skin cells from eye cells or liver cells. Nuclear DNA is transmitted to daughter cells via structures called chromosomes. These structures are long strands of DNA wound tightly around proteins to allow for greater mobility. Each human has twenty-three pairs of nuclear chromosomes: twenty-three that they receive from their mother and twenty-three that they receive from their father (See Figure 7). Sex cells only have one set of twenty-three nuclear chromosomes.

Both mitochondrial and nuclear DNA are useful in establishing genealogical connections. However, they follow different patterns of inheritance and therefore answer different research questions. Mitochondrial DNA is inherited in a different pattern than nuclear DNA. Within the larger category of “nuclear DNA” there are three sub-categories of DNA that follow distinct inheritance patterns: the Y-chromosome, the X-chromosome and autosomal DNA (See Figure 4).
Types of DNA

Mitochondrial DNA (mtDNA)

Mitochondrial DNA (mtDNA) is unique from other DNA in the cell. It is circular rather than linear and contains about 16,500 base pairs making it much smaller than nuclear DNA. Both men and women inherit mtDNA, but only females pass it on to the next generation. mtDNA is passed intact from a mother to her children. This characteristic makes it particularly useful in tracing the direct maternal line of an individual. (See Figure 5) Every person receives mtDNA from their mother, who received it from her mother, who received it from her mother etc. Therefore, individuals who share the same mtDNA sequence share a common direct-line maternal ancestor.

Several recent historical investigations have utilized mitochondrial DNA analysis to confirm the identity of human remains and better understand the past. In 1994 investigators successfully identified the remains of the Romanov family, the last emperors of Russia (Gill, et al. 1994). By comparing mtDNA from the remains to mtDNA of documented maternal relatives, researchers identified each of nine family members buried in a mass grave in Yekaterinburg, Russia. In 2009 an additional two members of the family were identified from a nearby grave (Coble, et al. 2009). More recently, in 2013, mtDNA testing was used to identify the remains of King Richard III which were found underneath a parking lot in Leicester, England (Buckley, et al. 2013). Like the Romanov case, the mtDNA from the remains were compared to the mtDNA of maternal relatives. Even though these cases used DNA from deceased persons, mtDNA can also be

Figure 5: Inheritance Pattern of Mitochondrial DNA
helpful in genealogical research by comparing the samples of living descendants of a common ancestor.

**Nuclear DNA (nuDNA)**

*The Y-Chromosome. (Y-DNA)*

The Y-chromosome is a small portion of nuclear DNA which contributes to the biological gender of an individual. All males have one Y-chromosome that they receive from their father and one X chromosome that they receive from their mother. Like mtDNA, Y-DNA is passed on intact from generation to generation. Unlike mtDNA which descends through the maternal line, Y-DNA is passed from father to son along the direct paternal line (See Figure 6). Because surnames are often inherited in a similar fashion, Y-DNA is often studied in connection with individuals who share the same last name.

Y-chromosome analysis has also led to several high profile discoveries in recent years. One project demonstrated that Thomas Jefferson, or another close Jefferson family member, fathered at least one of the children of Sally Hemings, a slave living at Monticello. The Y-DNA of direct line paternal descendants of Jefferson and of Sally Hemings’ youngest son matched (Foster, et al. 1998). Another study done on the descendants of Joseph Smith revealed that the Smith family originally immigrated from Ireland rather than England (Perego 2010). Other Y-DNA lineages commonly found in Europe have been attributed to descendants of Genghis Khan (Zerjal, et al. 2003) and Niall of the Nine Hostages (Moore, et al. 2006). Because of the ties between surnames and the Y-chromosome, this type of DNA is particularly helpful for identifying ancestry in cases of abandonment, adoption, illegitimacy and changes of surname.

![Figure 6: Inheritance Pattern of the Y-Chromosome](image-url)
**The X-Chromosome (X-DNA)**

The X-chromosome is also a sex chromosome. However, unlike the Y-chromosome, both males and females inherit it. Males have one X-chromosome that they inherit from their mother and which they pass on to their daughters. Females have two X-chromosomes: one that they receive from their mother and one that they receive from their father. X-DNA differs from Y-DNA because it undergoes a process called “recombination” in which the genetic material from two paired X-chromosomes is shuffled before being passed on to the next generation.

Recombination or “crossing over” is a process that occurs during the creation of sex cells. As previously mentioned, most cells in the body contain twenty-three pairs of chromosomes making for a total of forty-six chromosomes. Each chromosome pair for an individual includes a chromosome inherited from their father and a chromosome inherited from their mother. Sex cells, meanwhile, only have one set of twenty-three unpaired chromosomes. Chromosomes located in sex cells have undergone recombination, meaning that the genetic material they carry is a mixture of genes from the associated pair of chromosomes in a normal cell. Each chromosome in an individual’s sex cell derives about half of its genetic material from the chromosome inherited from their father and approximately half from the chromosome inherited from their mother. Because of the random nature of recombination, a recombined chromosome could derive a little more or a little less from one of the paired chromosomes than from the other. Over the recombination of all twenty-three chromosomes in a sex cell, the ratio of genetic material derived from each parent is about 50/50.

*Figure 7: 23 Pairs of Human Chromosomes Including the X and Y Chromosomes*
Because males only have one X-chromosome, when they pass it on to their daughters, it is passed on intact with minimal recombination. Since females have two X-chromosomes, when they pass on an X-chromosome to their children, it is recombined and is actually a mixture of both of their own X-chromosomes. As a result, the X-chromosome that a female receives from her father and which represents 50% of her X-DNA is inherited intact from her paternal grand-mother, whereas the chromosome that she receives from her mother is recombined and therefore only carries approximately 25% from each of her maternal grandparents (See Figure 8).

The complicated inheritance pattern of X-DNA makes it difficult to apply to specific genealogical problems. More commonly, X-DNA is simply analyzed in connection with autosomal DNA. However, X-chromosome analysis is common in medical research for investigation of sex-linked diseases and conditions. Hemophilia (Camerino, et al. 1984) and red-green color blindness (Nathans, et al. 1986) are examples of sex-linked traits influenced by genes on the X-chromosome.

**Autosomal DNA (atDNA)**

Autosomal DNA refers to all other nuclear chromosomes in the cell that are not sex chromosomes. The term “autosomal” refers to the fact that these twenty-two pairs of chromosomes only pair with like chromosomes. Y-DNA pairs with X-DNA despite the fact that they are quite different. X-DNA can pair with like X-DNA or with Y-DNA and therefore neither Y-DNA nor X-DNA are considered to be autosomal. Meanwhile, each of the remaining twenty-two chromosomes inherited from the father will only pair with a like chromosome inherited from the mother. To maintain the integrity of the genetic code held by each cell, it is important that these chromosomes
pair correctly before cell division and replication. There are times when this pairing does not happen correctly, and genetic abnormalities can be the result.

Like the X-chromosome, autosomal DNA (atDNA) undergoes recombination. Unlike X-DNA however, autosomal DNA recombines every generation regardless of gender. Therefore, each individual receives exactly 50% of their atDNA from their father and exactly 50% of their atDNA from their mother. Because it recombines they inherit approximately 25% from each grandparent, approximately 12.5% from each great-grandparent, and approximately 6.25% from each great-great grandparent. (See Figure 9) Because of recombination and random chance, eventually there will be some ancestors in the more distant past from which an individual receives none of their atDNA.

The amount of DNA shared between descendants of a common ancestor quickly drops each generation. Although two people may have descended from a common ancestor, and even if they each have inherited portions of that ancestor’s DNA, they may have inherited different portions to the extent that they may not appear to be related genetically. Siblings share about 50% of their DNA, first cousins share about 12.5% of their DNA, second cousins share about 3% of their DNA and third cousins typically share less than 1% of their DNA (Family Tree DNA n.d.). Despite these low percentages, most autosomal DNA tests analyze more than 650,000 sites in a tester’s DNA, and even at low percentages can highlight genetic relationships between relatives within the range of 10-14 degrees of consanguinity (See Figure 10).

Whereas Y-DNA and mtDNA only reveal information about specific family lines, atDNA can reveal relationships across all lines of ancestry. As more family members participate in autosomal testing, more matches with other living relatives appear since some descendants may have received different portions of a common ancestor’s DNA than others. As with all genetic

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**Figure 9: Inheritance Pattern of Autosomal DNA**

[Diagram showing inheritance pattern of autosomal DNA with percentages for each generation and relative.]
genealogy tests, once genetic relationships have been established through testing, these finds should be confirmed with traditional genealogical research.

Autosomal DNA testing is particularly helpful for breaking past genealogical obstacles in recent generations and can help clarify many of the same situations addressed through Y-DNA research. However the conclusions that can be drawn through analysis of atDNA are typically limited to research in the first 6-8 generations of a genealogy.

Figure 10: Relationship Chart Showing Degrees of Consanguinity

**Genetic Mutation**

Since all humans descend from a relatively small pool of ancestors, then there should be few differences between their DNA. Because Y-DNA and mtDNA do not recombine, all individuals would have identical mitochondrial DNA and all males would have identical Y-DNA were it not for occasional genetic mutations.

In fact, all humans do share about 99.9% of their DNA in common (Lander 1999). The sequences they do not share universally result from genetic mutations which occurred at various times in genetic history. These differences are what make genetic genealogy so helpful in family history research. If it were not for the occasional mutations that have been passed down to descendants, there would be no purpose in analyzing genetics for the purpose of ancestral research.
Mutations occur in the genetic code when DNA is copied before the division of a cell. In order for the mutation to be passed on to subsequent generations, it must occur when the genetic code is copied for inclusion in a sex cell. The cellular machinery that copies DNA is regulated for accuracy since mutations can be dangerous and can cause harm to the cell and to the body. Even so, every time DNA is passed on to a subsequent generation there are approximately 70 new mutations in the inherited copy (Roach, et al. 2010). Some mutations are silent, meaning they have no effect on cell function. Other mutations are dangerous and can result in dysfunctional proteins and cells, cause cancer or result in other problems in the cell. Some portions of DNA are more prone to mutation than others and extensive research has sought to establish the mutation rates for different sections and sites of DNA. The two most commonly analyzed mutations in genetic genealogy are single nucleotide polymorphisms (SNPs) used in autosomal testing, and short tandem repeats (STRs) used in Y-DNA and Mt-DNA testing.

**Single Nucleotide Polymorphisms**

A single nucleotide polymorphism refers to a single base change at one site of the genetic code. One of the four bases (A, T, G or C) is changed for one of the other bases and is then passed on to other cells. SNPs have a fairly low mutation rate making them useful for deep ancestry analysis and estimated relationship to various ethnic groups (Nachman and Crowell 2000).

**Short Tandem Repeats**

Some sections of DNA are composed of a series of repeating DNA motifs typically 2-5 base pairs long. For example one segment of DNA on human chromosome 7 is a series of repeats of the DNA motif “GATA”. Depending on the individual this motif could be repeated anywhere from 6-15 times. As the cellular machinery reads and copies these sequences, it may make an error and include an extra repetition of the motif or exclude a repetition of the motif. STRs mutate at a faster rate than SNPs and can be indicative of shared ancestry in more recent generations (Chandler 2006).

**Haplotypes and Haplogroups**

**Haplotypes**

A haplotype is a “combination of DNA sequences at different places on a chromosome that are transmitted together” (Haplogroup 2013) Test results from Y-DNA and mtDNA testing constitute the haplotype of an individual because the markers analyzed were transmitted together on the same DNA segment through multiple generations. Test results for atDNA when compared with the test results of other relatives can reveal autosomal haplotypes or “sequences of DNA at different places on a chromosome that are transmitted together”. Autosomal testing requires comparison with other individual’s test results to identify unique haplotypes since recombination mixes the set of DNA sequences inherited from generation to generation. After comparison, if two relatives share a set of DNA sequences on the same chromosome, then they share haplotypes.

**Haplogroups**

A haplogroup is a group of individuals who share similar haplotypes. This definition is most useful for Y-DNA and mtDNA which is passed on intact from generation to generation with few mutations over time. Since autosomal DNA recombines every generation, autosomal haplotypes are rarely informative except for cases of extended population isolation in which case
they will likely reflect the same patterns as their associated Y-DNA and mtDNA haplogroups. Haplogroups are defined by SNPs, but can often be inferred by STR values. They are designated on the assumption that individuals who share similar SNP values are descended from the same ancestor. Haplogroups are useful for anthropological investigation and show an individual's deep ancestral roots (See Figure 11 and Figure 12).

Figure 11: Y-DNA Haplogroup Map (Family Tree DNA 2009)
Mutations and Relationship

Mutations can be used to estimate how closely related two individuals are. However, this principle applies differently to Y-DNA and mtDNA analysis than it does to atDNA analysis. In Y-DNA and mtDNA analysis, the fewer mutations that exist between two individuals, the more closely related they are. When there are more mutations between two individuals, they are either more distantly related, or not related within a genealogical time frame. Y-DNA and mtDNA analysis both depend on shared mutations between two descendants of a common ancestor. Each marker is associated with a different mutation rate and the number of marker differences between two individuals can contribute to an estimate of when their common ancestor lived, and how closely related they actually are. Analysis of atDNA depends on the comparison of thousands of SNP markers throughout an entire genome. The relationship between two individuals who participate in autosomal testing is estimated based on the number of markers they share in common as well as the length of DNA sequences that they share in common. When two individuals share a large amount of total DNA as well as several longer segments of DNA, they share a common family history and a common ancestor.
**DNA Testing Options**

Several companies offer genetic testing for genealogy purposes each with its own strengths and weaknesses. This chapter will review three of the most prominent and popular testing options: Family Tree DNA, 23andMe, and Ancestry.com

**Family Tree DNA**

Family Tree DNA is a personal genetics testing company based in Houston, Texas, and is a branch of the larger parent company Gene by Gene. Founded in 2000, it was the first company to offer genetic testing for genealogy purposes. It also has the largest database of Y-chromosome and mtDNA genetic results with which to compare. Family Tree DNA’s results are separated into three categories: Y-DNA, mtDNA, and atDNA. Although they have the largest collection of overall test results because they have been in operation the longest. However, they have the smallest database of autosomal test results of the three above mentioned companies. Both Y-DNA and mtDNA testing at Family Tree DNA can be performed at different levels of detail. They offer Y-DNA tests that compare across 12, 37, 67, or 111 markers. Their two types of mtDNA testing offer the option to analyze the entire mtDNA sequence or to only analyze the hyper-variable regions (the non-coding regions of the DNA where most mutations occur).

Family Tree DNA is the only company to offer a platform for organizing surname, haplogroup, and geographical projects. This allows for increased collaboration among participants and researchers. This platform coupled with the ability to upload personal GEDCOMs makes them one of the top choices for genealogists. They are also the only company that accepts genetic testing results from other companies. They provide analysis tools, contact information for genetic matches and files of the raw data for download. Family Tree DNA’s tests are available for shipment worldwide allowing for projects with foreign participants (Janzen 2013).

**23andMe**

23andMe is a personal genomics and biotechnology company based in Mountain View, California. Their mission is to be the world’s trusted source of personal genetic information. Their primary focus as a company is to offer personal genetic testing for health and medical information. Currently, they only offer a single test for autosomal DNA testing. However, they do also test portions of Y-DNA and mtDNA which provide haplogroup information. Their autosomal test is the most comprehensive of all the companies testing 967,000 autosomal markers in addition to another 30,000 markers from X-DNA, Y-DNA and mtDNA. They also have the largest database of autosomal test results with more than 350,000 participants. Because 23andMe mainly advertises genetic testing for medical purposes, it attracts a large audience not represented in the other databases which cater to genealogists.

Despite the fact that many participants test at 23andMe for medical information, the company still offers an advanced set of data analysis tools to investigate ancestry and ethnicity admixture. Due to complicated legal and ethical issues surrounding personal genetic health, 23andMe only ships to 56 countries worldwide, and their testing kits are illegal in some countries (Janzen 2013).
**Ancestry.com**

Ancestry.com offers an autosomal SNP test called AncestryDNA. They began offering this test in May 2012. Ancestry.com also offers two Y-DNA tests which analyze 33 and 46 markers as well as an mtDNA test which analyzes the hyper-variable region. However, their main marketing focus is currently the AncestryDNA autosomal test. Between the three major genetic testing companies, Ancestry.com currently offers the fewest number of autosomal markers in their testing. Despite this fact, they host one of the fastest growing databases of genetic samples.

Ancestry.com’s strength is its connection to billions of historical records and family trees, which creates an active conversation between genetics and documented history. Not only does Ancestry.com compare the genetics of each participant against all other participants, they also compare the family trees of genetic matches to estimate how they are related. Although they lack some of the analysis tools available at the other companies, their users are very responsive to collaboration. The AncestryDNA product is still quite young and is rapidly changing. They recently began allowing customers to download the raw data files of their test results. Currently, they only ship within the United States (Janzen 2013).

**Additional Resources**

Each of the major testing companies provides educational tools on its website. In addition, there are many wonderful resources to learn more about genetic genealogy and how to apply it to your own research.

- International Society of Genetic Genealogy (ISOGG) [isogg.org](http://isogg.org)
- Sorenson Molecular Genealogy Foundation (SMGF) [smgf.org](http://smgf.org)
- DNA eXplained – [dna-explained.com](http://dna-explained.com)
- The Genetic Genealogist – [www.thegeneticgenealogist.com](http://www.thegeneticgenealogist.com)
- The Legal Genealogist – [legalgenealogist.com/blog/](http://legalgenealogist.com/blog/)
- Haplogroup - [www.haplogroup.org](http://www.haplogroup.org>
- The Genetic Genealogy Consultant - [www.geneticgenealogyconsultant.com](http://www.geneticgenealogyconsultant.com)
- Beginners Lessons to Genetic Genealogy: [Beginners Guide to Genetic Genealogy](http://www.beginnersguide.com/)

**Conclusion**

One of the main goals of family history research is to shed light on our origins and where we came from. It seeks to show our place in the world and our relationship with the past. As a biological record of our own families, DNA is an invaluable resource for establishing family relationships and investigating the past, particularly when written records do not yield the information we seek. By linking us with living relatives, genetic genealogy helps us to better collaborate and encourage each other in the discovery of our families. As we embark on the quest to discover who we are, genetics can reveal our relationship to ourselves, to our families and to the world.


