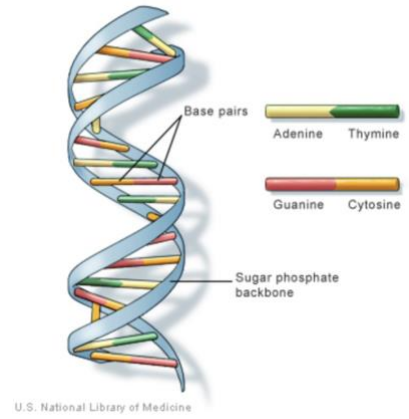
A Basic DNA Testing Primer
By Marty Brady, 15 Feb 2025

DNA, or deoxyribonucleic acid, is the hereditary material in humans and almost all other organisms. Nearly every cell in a person's body has the same DNA. Most DNA is located in the cell nucleus (where it is called nuclear DNA). It is the blueprint for our construction and our metabolic activity. The DNA is packaged in chromosomes. The autosomal (atDNA) chromosomes are numbered 1 through 22 by size with 1 being the largest and 22 being the 2nd smallest (21 is the smallest). The 23rd pair of chromosomes are the sex chromosomes X & Y. Females have two X chromosomes and males have an X and a Y chromosome. The nuclear DNA contains 2 copies of each chromosome (one copy from mom and one from dad) so there are 23 pairs of chromosomes. Mitochondrial DNA is arranged differently and will be described later.

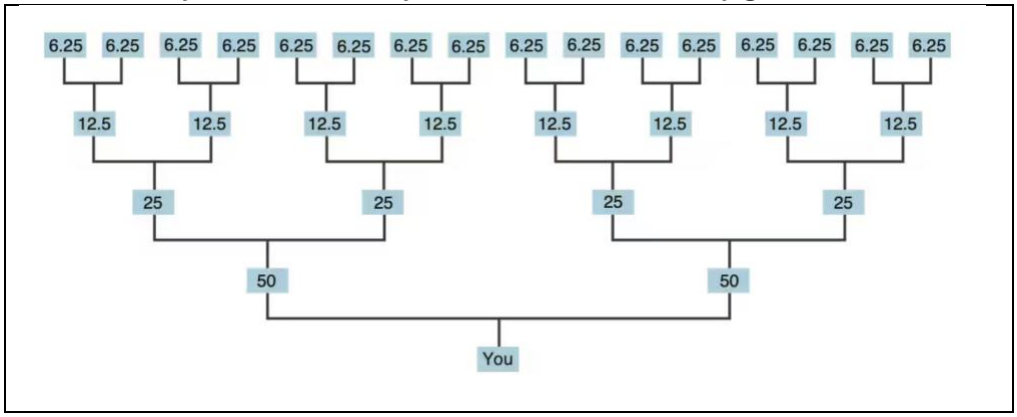
DNA is found as a double helix which is kind of a twisted ladder of 2 complementary strands of DNA. The steps of the ladder are base pairs (or nucleotide pairs). A (Adenine) always pairs with T (Thymine) and G (Guanine) always pairs with C (Cytosine). The DNA is described as a sequence of these bases (aka nucleotides), such as AGTCTAGC, etc.



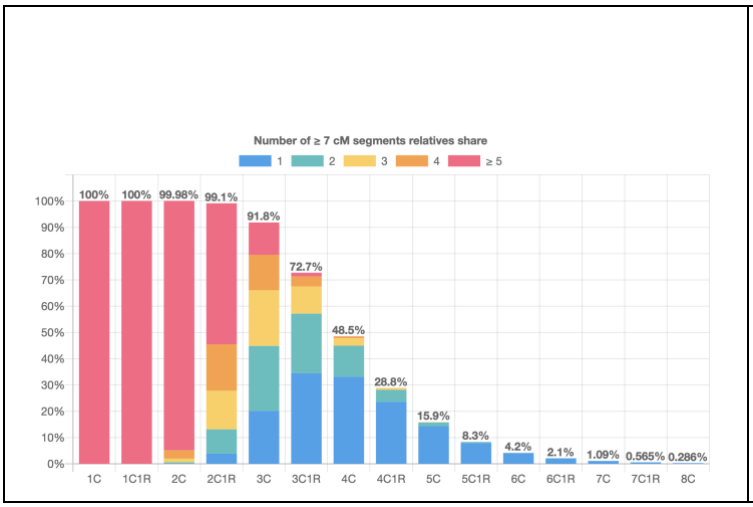
Credit: U.S. National Library of Medicine

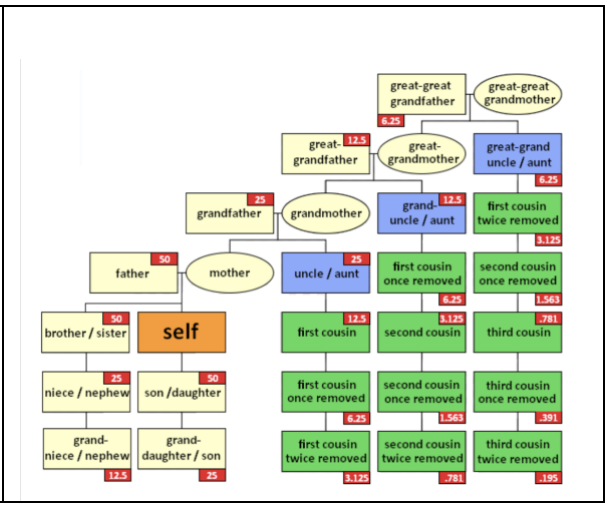
Phew..... enough of this science stuff.

We inherit 50% of our DNA from each of our parents. They in turn inherited 50% of their DNA from their parents and so forth back through all of their ancestors. So, we inherit roughly 25% (range of 18% to 32%) of our DNA from our 4 grandparents and roughly 12.5% of our DNA from our 8 great grandparents and so on. Because of this, at 7 generations back, less than 1% of your DNA is likely to have come from any given ancestor.



Also, only about 92% of your 3rd cousins will share atDNA with you and less than 50% of your 4th cousins will share atDNA with you.





The above graphics from the ISOGG website clearly demonstrate how we share DNA differently with different relatives. We can distinguish the amounts of DNA we share with relatives because of mutations. These are small changes to the DNA due to copying errors. The human body produces billions of cells a day and the DNA must be copied for each new cell. The average male produces over 500 billion sperm cells in his lifetime. When sperm cells are produced DNA is copied as well.

The two main types of mutations (or copying errors) in genealogy are STR's and SNP's.

- An STR is a **s**hort **t**andem **r**epeat. This is a place in your DNA code where a letter sequence is repeated. For example, AGTAAGTAAGTA is three repeats of the sequence AGTA. STRs have a fast mutation rate. Some STRs mutate faster than others. When they change, it is an increase or decrease in the number of repeats.

- A SNP is a **s**ingle **n**ucleotide **p**olymorphism. That means that it is a single small change in your DNA code. These changes are rare. Once they happen, they seldom change back (back mutate).

There are about 700,000 STRs and about 5,000,000 SNPs in the human genome. As more people around the globe test, we will probably find more. As of 10 Feb 2025, there are well over 700,000 known SNPs on the Y-chromosome alone. And the number is growing very quickly as more people test. Mom and dad always reshuffle the DNA they inherited from their parents before passing it on to their children. This shuffling is called recombination. The number of different ways the information on 23 pairs of chromosomes can be reshuffled is astronomical. Therefore, no two siblings will inherit the exact same segments of DNA from their 4 grandparents (except for identical twins).

There are five major DNA testing companies that are commonly used in genealogy. AncestryDNA, 23andMe, MyHeritage, FTDNA and LivingDNA. They all have their pros and cons. AncestryDNA has the largest database by far and many useful tools, but they don't offer a chromosome browser and they don't allow uploads of raw DNA info from other testing companies. Since you may have cousins that have tested in only one company, if you didn't test with that company you won't find that cousin. The general advice is to fish in as many ponds as possible. Get your DNA into as many databases as possible. You might want to test with Ancestry first and then upload your raw DNA to GEDmatch (free), then

upload to FTDNA, MyHeritage and LivingDNA (small fee). Later, you can test with 23andMe. Ancestry and 23andMe do not allow uploads of raw DNA from other companies.

There are many tools for working with cousin matches. Pro tools on Ancestry, chromosome browser information with several companies, DNAPainter tools, Leeds Method, Clustering, Visual Phasing and many others. I just learned on DNAeXplained about a new feature on FTDNA called matrix. To get the most out of your test, watch a lot of videos and play with those tools. And take advantage of the one-to-one online consultations offered by AGS. Remember though that a big limitation of atDNA is that you can usually only go back 5 to 7 generations.

Ethnicity

Some people only test with Ancestry (or other companies) to find out if they should wear lederhosen or a kilt. They are not interested in genealogy. You may come across these folks as you try reaching out to DNA matches. However, you can also use ethnicity estimates for family research and Paul Woodbury has some interesting information about this in his videos on Legacy Family Tree Webinars.

The goal of ethnicity estimates is to compare the DNA of a customer to the DNA of people with long family histories in a particular region or group (i.e., the reference panel). The testing companies are constantly improving their reference panels. Ancestry went from 60 regions to 71 then to 88 and now in October 2024 they have increased the number of regions to 107. Ancestry also has some excellent White Papers on the subject on their website. Essentially, they compare the haplotype (a grouping of SNPs) of small segments of your DNA to the same segment in the reference panel. Remember that these are only estimates.

Health traits

Some SNPs are associated with certain traits and health conditions. You can subscribe to testing companies' traits feature or you could download your raw DNA and look on SNPedia or Promethease to find what traits you may be predisposed to encounter. Remember, the expression of a trait is often a combination of environment and genetics. Not genetics alone. So having a particular SNP does not mean you will have that trait or health condition.

Y-DNA

Y-DNA is inherited from father to son relatively unchanged. So, instead of sharing 25% of his DNA with his paternal grandfather (as he does with atDNA), a male shares about 100% of his Y-DNA with his paternal grandfather. This makes it possible to do deep ancestry research with Y-DNA. You can find distant cousins with Y-DNA testing. In societies that associate surnames with the male line (as in European cultures), surname research can be done with Y-DNA information. Y-DNA can also be of use in questioned parentage cases. A caveat with Y-DNA testing is that you cannot tell which male in the patrilineal line contributed the Y-DNA (i.e., you can't distinguish brothers, uncles, etc.). Y-DNA can also be of use in Investigative Genetic Genealogy. Keep in mind that even females can do Y-DNA research. You just need to have a male in your family test (i.e., brother, father, uncle, etc.).

This also means that a male can explore more than his direct paternal line by enlisting the help of male distant cousins.

Mitochondrial DNA

A small amount of DNA is not in the nucleus of the cell and can be found in the mitochondria. Mitochondria are structures within cells that convert the energy from food into a form that cells can use. Mitochonrial DNA is passed relatively unchanged from mother to all her children. However, only her daughters will pass the mtDNA on to the next generation. We can then use mtDNA for deep ancestral searches on the maternal line. We can also use mtDNA to disprove or corroborate genealogical hypotheses. Because each mitochondrion may have 10 copies of its DNA and there may be 1000 mitochondria in a cell, there is lots of mtDNA available for testing.

The FTDNA website states the following: "The range of possible generations before you share a common ancestor with a match is wide. Your mtDNA (HVR1, HVR2 and Coding Region) exact matches may be recent, but they may also be hundreds or thousands of years in the past." So, we are probably not going to use mtDNA to find cousins. The best presentation on using mtDNA in genealogy is the Michael Lacopo presentation listed in the references. mtDNA can best be used to corroborate or disprove a genealogical hypothesis. The military also uses mtDNA for identifying MIA remains.

X-DNA

The X chromosome exhibits a unique inheritance pattern. A father has an X and a Y chromosome. The X and Y chromosomes are not homologous, so they do not recombine (i.e., no exchange DNA between them). The father transmits an X chromosome to his daughters, but only a Y-chromosome to his sons. Because the X and Y do not recombine, the X-chromosome that the father transmits to his daughters is essentially the paternal grandmother's X chromosome. The X chromosome experiences less segmentation than the 22 numbered autosomes. So, the bar is slightly higher for avoiding false matches. The generally accepted cutoff for atDNA is 7 cM, but for X-DNA the cutoff is 20 cM. Below that size of segment, there is an increased chance of false matching. Creating a personal X-DNA inheritance chart should help when working with X-DNA matches.

So, the major takeaways are:

1) Fish in as many ponds as possible for cousin matches. Use as many tools as possible to work with DNA matches.
2) Reach out to distant cousins for deep ancestry connections.
3) Use mtDNA to confirm or disprove your hypotheses that involve maternal heritage.
4) Create your own personal X-DNA Inheritance Chart to help you when working with X-DNA matches.

Resources

Books, papers and blogs

"The Family Tree Guide to DNA Testing and Genetic Genealogy" by Blaine Bettinger
"The Genealogist's Guide to Y-DNA Testing for Genetic Genealogy" 2nd Edition by David Vance
Ancestry's "Ancestral Regions 2024 White Paper"

FTDNA “Understanding Your mtDNA Full Sequence Results”
Sue Griffith’s website http://www.genealogyjunkie.net/downloads.html

Videos
“Foundations in DNA 1 of 5: Introduction to DNA and Genetic Genealogy” by Blaine Bettinger
“Covering Your Bases: An Introduction to Autosomal DNA Coverage” by Paul Woodbury
“Evaluating Shared DNA” by Paul Wodbury
“Broken Branches: How to Detect Cases of Misattributed Parentage in Your Family” by Paul Woodbury
“Using Mitochondrial DNA Testing for Genealogical Problem Solving” by Michael D. Lacopo, D.V.M.
“Understanding X-chromosome DNA Matching” by Michelle Leonard
“Anchors are the Way: Leveraging Multiple forms of DNA Evidence in Your Research” by Paul Woodbury