

A Day Out With Your DNA

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www.yourDNAguide.com

Your testing company has evaluated around 800,000 locations on your DNA to help them determine your origins and your genetic cousins. While these reports detailing your percentage of this and that and your humongous list of newfound family is what you paid for, don't neglect your raw data. Your raw data is this list of 800,000 locations that were tested. These numbers are actually half of what you paid for. The other half is the company's interpretation of the numbers.

What is Raw Data?

Now, I am not prone to car analogies, but I think we could use one here to serve our purposes. Think of your raw data like all the parts of the car that actually make it work, and the testing companies' report of cousins and origins are like the body of the car, the part that we see and interact with. So while one company will show you a Jeep Grand Cherokee, another will show you a Mercedes-Benz M-Class, but underneath all that metal they are basically the same (or at least, that's what my husband told me!). Because you have this raw data file you can take it in between companies (i.e. Jeep and Mercedes-Benz) and get all sorts of new and interesting looks on the same raw data. The auto industry calls this platform sharing. In genetic genealogy, we call this transferring our data.

Downloading Raw Data

But before we get into all of that, we need to obtain that raw data file. This means you will want to download your raw data from wherever you were tested. This raw data comes in the form of a .csv file that you can open on Excel if you want (but really, if you have nothing better to do than scroll through 800,000 lines of data, please come talk to me - I have so many more productive things that need to be done!). For instructions on how to download your raw data from your testing company, head over to my website at ww.yourDNAguide.com/transfer.

Transferring Data

Transferring your data just means you are taking that data file from the company who generated it for you into a company who has some tools that you want to explore. These destination places generally fall into one of two categories: testing companies or third-party tools. As the genetic genealogy industry grows and changes we will see more and more of both kinds of companies cropping up to capitalize on the opportunities that are inherent in large amounts of new data being generated.

Caution!

Please please please remember that your raw data does contain your own personal information that does identify you uniquely from anyone else on the planet. While you shouldn't be afraid to try new tools and explore your personal genomics, it is very important to read the privacy information of each company carefully to be sure you know what you are consenting to when you are uploading your data. Most companies are fastidious about privacy, but many are also involved in research endeavors,

Big Ol' List of Tools: http://isogg.org/wiki/Autosomal_DNA_tools

Family Tree DNA: www.ftdna.com

AncestryDNA: www.ancestryDNA.com

My Heritage: www.myheritage.com

Living DNA: www.livingDNA.com

Gedmatch: www.gedmatch.com

DNA Land: www.DNA.land

Promethease: www.promethease.com

DNAGedcom: www.DNAGedcom.com

Livewello: www.livewello.com

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Ethnicity testing:

- Y-DNA or mtDNA haplogroup information provides information about only one ancestral line.
- Autosomal testing can provide information about the “admixture” of different ethnic groups from all of your ancestral lines.

Autosomal DNA testing:

- The 22 pairs of nuclear chromosomes that are not the sex-determining (X and Y) chromosomes are the autosomes.
- Each person inherits a unique mix of autosomal DNA from their parents. 50% is inherited from each parent. But the 50% provided by each parent is a random mix of the grandparents' DNA. This means (only on average) 25% from each grandparent, 12.5% from each great-grandparent, and so forth.
- Siblings will typically have the most DNA in common. First cousins will have less in common, second cousins will have even less, and so forth.
- Autosomal DNA can be tested to look for relatedness on all ancestral lines but is only reliable back to about 5th cousins.

Autosomal DNA

Each individual will inherit some DNA from each of their fourth great grandparents. Prior to this, they may have some genealogical ancestors who did not contribute genetic material to their genome. Collateral descendants of a common ancestor will inherit different portions of their ancestors' DNA; though they may share DNA with the common ancestor they may not share DNA with each other. Relatives closer than the level of second cousins should share detectable amounts of DNA with each other. Third cousin and more distant relationships are sometimes undetectable through autosomal DNA testing. The DNA inherited from a specific ancestor passes through all ancestors in between, so test collateral vs. hierarchical descendants. Tests for sibling sets, first cousins and second cousins can be helpful in reconstructing the genomes of

deceased ancestors since the segments of DNA they share in common and the segments of DNA they do *not* share in common can all be used to draw conclusions. The likelihood of identifying additional relatives by testing known relatives varies based on their relationship and can be assessed in the AncestryDNA help menus.¹ Since the amount of autosomal DNA shared with an ancestor decreases by approximately 50% each generation, prioritization of autosomal DNA testing should be given to the closest generational descendant and not necessarily the oldest living descendant. Search for closest generational descendants among the youngest children of the youngest children of the ancestor. These individuals will have the longest generation times.

X Chromosome.

Though there are no specific DNA tests for the X chromosome, it does have a unique inheritance pattern that can justify alterations in a research plan. When passed through males, the X chromosome does not recombine at significant levels; when passed through females it may recombine. Therefore it is equally likely that a female will inherit some X-DNA from her father's mother's father's mother's father's mother (4th great grandparent) as it is that they will inherit some X-DNA from their mother's mother's mother (great grandparent). Searching for descendants who follow the father-mother descent pattern can increase the chances of matching a relative on the X-chromosome. When performing research where several possible relationships could explain shared autosomal DNA, but shared X-DNA could eliminate some of those possible relationships, X-chromosome inheritance may affect the choices made in a testing plan

Mitochondrial DNA testing:

- mtDNA is passed from a mother to her children (so both men and women have it and can be tested for it).
- Not normally as useful as Y-DNA testing because there are no surname groups to serve as a comparison group.
- Primarily used in unique situations where a common maternal lineage needs to be supported or refuted.
- Tests can be for the non-coding Hyper Variable Regions (HVR1 and HVR2) and for the Coding Region (but the Coding Region may identify medical issues).

Mitochondrial DNA

As with Y-DNA, one individual's mtDNA test can represent a large number of relatives. Mitochondrial DNA mutates at a much slower rate. Though recent mutations are certainly possible, they are rarer than recent Y-DNA mutations, therefore it is not usually necessary to test the oldest representative, but still a good idea.

Y-DNA

If you are concerned about the possibility of recent mutations in your Y-chromosome line, consider testing older relatives. Once you have tested one relative, their Y-chromosome will be representative of their known male relatives who carry the same surname. Even if you do not have the Y-DNA that is pertinent to the research question, other family members and descendants might. Search for testing candidates who carry the same DNA as the research subject. Search Y-DNA surname projects at Family Tree DNA to determine if one of the direct line descendants of your ancestor may have already tested. Y-DNA signatures may vary from the expected if there was a case of misattributed paternity like an illegitimacy or undocumented adoption. If you suspect a situation like this in your own family, test another direct paternal descendant from a unique line to confirm or refute this hypothesis. If you are attempting to determine the paternity of an ancestor and there is at least one paternal candidate, test a direct paternal descendant or relative of that candidate. When performing Y-DNA testing, start out at lower levels of testing and then upgrade to higher levels after evaluating the test results. If DNA testing.

Determining Relatedness

If you add up the total of all cM values for the segments someone shares with you, you can get a rough calculation of how closely you are related to them. There is a total of around 6800cM in all 44 autosomal chromosomes. The following are expected cM matching values for various relationships:

- Identical twin - 6800cM (all chromosomes are identical)
- Parents - 3400cM (50% of the chromosomes are a match)
- Full siblings - 2500cM (37.5% match)
- Grandparents and aunts/uncles - 1700cM (25% match)
- Great-grandparents and first cousins - 850cM (12.5% match)
- Second cousins and first cousins twice removed - 212.5cM (3.125% match)

The cM match amount or overlap decreases as your relationship gets more distant. You might share only 13cM (.195%) with your fourth cousin (someone with whom you share a 3rd great-grandparent). Of course with the variability of many generations of recombination (or non-recombination) of chromosomes, you could share much more than that, or you could share 0cM and not be identified as a cousin match at all.

IMPORTANT!

There is much variability in DNA tests. Each company tests slightly different things in different ways. DNA inheritance is highly variable. For all of these reasons, keep in mind that the cM match values and predicted relationships are **VERY ROUGH ESTIMATES ONLY!** This is especially true for more distant cousins. Additionally, if you are related to someone on multiple lines - or if you or your match are related to your common ancestor on multiple lines (e.g., your grandparents were cousins) - then the total cM will suggest a closer relationship than is actually the case.

GEDmatch Tools (GEDmatch.com)

One-to-many Matches

Provides a list of people you share chromosome segments with in the GEDmatch data base. To view the report, click the 'One-to-many' matches link on the home page and select your kit # (found on the homepage) on the next page. We'll be comparing Autosomal chromosomes, not X, so make sure Autosomal is selected. Keep threshold at 7 cM and select Display Results

One-to-one Compare

A direct comparison of two test-taker's **autosomal** raw data. Remember, our chromosomes come in pairs. However, when the DNA testing tools do chromosome comparisons, they can't distinguish between the two chromosomes in a pair - they instead treat them essentially as one combined chromosome - as if the chromosomes have been laid on top of each other. This means that when you match someone on a chromosome segment, you can't be sure which of your chromosomes they match. It could be the chromosome you got from your father or the one you got from your mother.

X 'One-to-one'

A direct comparison of the X-DNA of two test takers.

Admixture

Many different ethnicity calculators which output results in graphical or percentage format.

People Who Match One or Both of 2 kits.

Phasing

Generates phased maternal and paternal data files, but MUST have a child and at least one parent tested.

Are Your Parents Related?

Determines whether a file has “Runs of Homozygosity” (ROH) which indicates that your parents were related.

Matching Segment Search

Other kits with segments that match yours.

Relationship Tree projection (highly experimental)

Calculates probable relationship paths based on autosomal and X-DNA sharing & genetic distancers.

Lazarus

Creates surrogate kits to represent close ancestors. (Consider joining the Lazarus Utility Group on Facebook..... [facebook.com/groups/818555341528048](https://www.facebook.com/groups/818555341528048))

Triangulation

Identify and confirm triangulation groups (TG) from your matches.

Genetic Triangulation, the key to our research [isogg.org/wiki/Triangulation](https://www.isogg.org/wiki/Triangulation)

Triangulation for autosomal DNA is kind of a chicken and egg thing. The goal is to associate and identify specific DNA segments to specific ancestors. The easiest way to do this, or to begin the process, is with known relatives. This gets you started identifying “family segments.” From that point, you can use the known family segments, along with some common sense tools, to identify other people that are related through those common ancestors. Through those matches with other people, you can continue to break down your DNA into more and more granular family lines.

The basics of triangulation for Y-DNA testing

Genetic genealogical triangulation is rather simple. Think of a triangle. /_\
Person A & B match genetically and that forms the base of the triangle. _

Person A has a paper trail (genealogy) that goes back in time. /

Person B has a paper trail that goes back in time. \
The top of the triangle is the MRCA or most recent common ancestor.

Person A is who you are testing. Some living biological male 2nd, 3rd or better cousin

Person B. The most common shared ancestor is the MRCA.

If the genetics of Person A & Person B match and the paper trail goes to the MRCA, then this helps prove they are related both genealogically and genetically. This is the goal of genetic genealogy. The genetics help confirm the paper trails (genealogy) back to the MRCA. When this is repeated several times back to a common ancestor, we then can recreate the DNA markers or genetic fingerprint of that ancestor. All without digging them up!

If there is a break in any point of the triangle, it should be noted appropriately. If Person A & B match genetically but either paper trail (genealogy) does not go back to the MRCA, then they match genetically but not genealogically. If Person A & B do not match genetically, but match with the paper trails, then they match genealogically, but not genetically. In this case the genealogy may be wrong or there is a formal or informal adoption of DNA into the genealogical line. The later is called a non-paternal event. When comparing any DNA test using triangulations, one should always cite the common test. For example, when comparing say a 37 marker Y-DNA test with 111 marker Y-DNA test, you should always cite the lower value. Using the example given, a proper statement of genetic triangulation would indicate that Person A & Person B matched genetically and genealogically at 37 Y-DNA markers.

Triangulation with autosomal DNA testing

In autosomal DNA testing triangulation is the term used to describe the process of reviewing the pedigree charts of people who match on the same IBD autosomal DNA segment to see if a common ancestor can be found. The technique is best used in conjunction with chromosome mapping.

Triangulation can be used going back many generations. However, well documented pedigrees are necessary for all the matching parties in order to rule out the possibility that the match is not on a more distant line which has not yet been researched. Caution still needs to be exercised when reviewing matches with smaller segments **under 15 cMs in size, and especially segments under 10 cMs in size, as many of these are false positive matches.**

The process of triangulation is greatly facilitated by the use of third-party tools such as those available from GedMatch.com and DNAGEDCOM (eg, Don Worth's Autosomal DNA Segment Analyser)...<https://isogg.org/wiki/Triangulation>

New Research Projects

The Adoptee Survey – a survey of 2,000+ adoptees to examine their experience with DNA testing

The Recombination Project – a project to examine recombination by comparing the DNA of grandchildren to their grandparents

This information is a compilation of research through the materials collected at the annual Southern California Genealogy Jambouree. I especially want to thank Blaine Bettinger, Ph.D., J.D. for the 3 hours spent introducing this to me. (I hope all of you will find a part of it useful to your ongoing genealogy projects. I will post this on our website: <http://www.blandheritage.org>

Jo Shannon2017 National Reunion....October 16, 2017

Some Definitions

cM

[Centimorgan](#) (abbreviated cM) is a measure of genetic linkage. Think of it as a measure of DNA information within a chromosome. Each chromosome contains different amounts of information. Chromosome 1 contains 281.5cM of information. Chromosome 2 has 263.7cM. Chromosome 21 has only 70.2cM.

SNP

SNPs, or [single-nucleotide polymorphisms](#), are tiny pieces of a chromosome that contain distinct blocks of information. There are thousands of them per chromosome. SNPs are compared between two people to see if they match. The amount of information in matching SNPs is measured in cM.

The cM values for SNP matches are sometimes referred to as "chromosome length" or "match length". However, information is more densely packed in certain areas or SNPs within chromosomes, so there's not a direct correlation between number of SNPs and cM amount. When you view GEDmatch's graphical depiction of chromosome matches, a bigger matching block does not always mean a higher cM value.

Segment

A "segment" refers to a section or block of contiguous SNPs. A "matching segment" is a section that is the same between two people.

Start and End Location

Individual markers (called base pairs - the things that SNPs are made of) within a chromosome are numbered. There are millions of these markers per chromosome. A segment of a chromosome can be identified by these location numbers.

IBS and IBD

Sometimes SNPs marker values match between two people simply by chance. This is called IBS or Identical By State. And sometimes they match because they were passed down from a common ancestor. This is called IBD or Identical By Descent.

MRCA

This is Most Recent Common Ancestor - the ancestor from which you and a DNA match received your common DNA segments.

DNAGedcom (www.dnagedcom.com)

An online suite of autosomal DNA tools created by genetic genealogists who were assisting the adoptee community.

Steps to Convincing People to Test Emily D. Aulicino, aulicino@hevanet.com

Nothing is fool-proof, and there are no guarantees of success, but doing nothing does guarantee nothing. Every relative you can convince to test will help you with unknown matches. Convincing a stranger to test whom you feel can help your brick wall is being pro-active

Before You Call ...

- Understand the basics of DNA testing
- Know which companies offer what, including the type of kit (spit or buccal)

- Be prepared to explain the different tests
- Do not misrepresent DNA testing
- Be sincere; be yourself and not over enthusiastic or pushy
- Know a few generations of the potential tester's lineage (three or more)
- Expect to spend much time on the phone and to call more than once
- Be interested in what the person is saying as they may wish to share family knowledge

Calling Etiquette

- Speak clearly and not quickly.
- Ask if this is a good time to call; do not interrupt a ball game, TV show, a meal or time with the children.
- Be interested in the person's occupation or avocation
- Be aware the person you called may be from another ethnic group, but still could be related and willing to test
- Be courteous to a person who is ill or in the middle of a project; ask when to call back
- Do not bore your potential tester with genealogy stories of your family

Alleviate Fears

- Some may be fearful of scams
- Worries about privacy in testing
- Justice system (CODIS)
- Medical and employment concerns (GINA)
- Results of company being shared
- Financial concerns

Basic Issues to Cover

- Introduce yourself as a genealogist and mention the relevant surname
- Ask the person if he or she is related to the ancestors you believe to be their grandparents or great-grandparents.
- Do not mention the parents
- Suggest there could be a relationship between their lineage and yours, but the paper proof is not there.
- Ask if the person knows the connection
- Ask if there is a family genealogist
- Offer to send a copy of their lineage, if interested.
- Use the US mail so you have their address unless they insist otherwise
- Obtain leads on their family and on contacting their relatives as another person may be more willing to test
- Thank the person for his/her time and interest in helping
- Ask if you can call again when you need to follow up and when you find more information on the family

Discussing DNA

- Refrain from mentioning DNA initially and find the paper trail that you will need later.
- Speak to the family genealogist before mentioning DNA to your potential tester. You may have to explain DNA and convince them how testing can show you are related.
- Be prepared to have several conversations
- Let the potential tester know that since no one can find the paper trail connecting the families, there is one other way...DNA}

Offer to pay for the test. Sometimes you or their family members may split the cost.

Remember that your goal is to educate a potential tester and to alleviate his or her concerns. When it comes to convincing people to test, "practice makes perfect". Understand that no one will ever be 100 percent successful so plan to contact more than one person for the test.