What Your Chromosomes Can Tell You About Your Family

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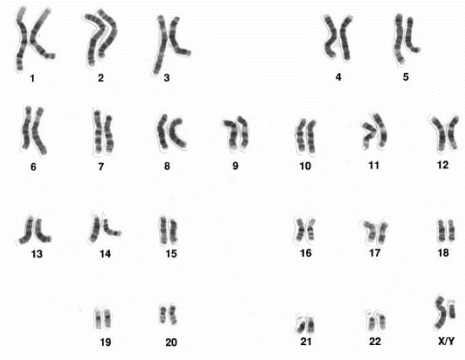
Of course we are all curious about where we come from or we would not be genealogists! It is no wonder then that many people are simply curious about their presumed ethnic breakdown. While the ethnic breakdown will vary dependent on testing company it is a good general guide to where your DNA originated from.

Whatever the reason, many people turn to DNA testing hoping it will answer all of their questions. Sometimes you do find the answers in the DNA. More often than not, you end up with more questions that you started with! However there are two truths to testing.

A word of caution. Don't test unless you want to know. DNA does not lie and it can uncover deeply buried family secrets. Some of these secrets can be humiliating, shameful or painful to older members of the family. Realize that not everyone may be as curious as you. You will need to respect the privacy concerns of other members of your family is you uncover something surprising.

Also, it is just a tool. Use it as you would another document to aid in your search for your family history. DNA can help, it can also confuse, just like a census record that makes no sense. When used in conjunction with paper genealogy it can open doors you did not know about.



Human Male Karyotype. By Courtesy: National Human Genome Research Institute [Public domain], via Wikimedia Commons

Autosomal DNA (atDNA) are the 22 pairs of non-sex chromosomes located in the nucleus of the cell in genetically healthy humans. The sex chromosomes (X and Y) are the 23rd pair of chromosomes and determine if you are male (XY) or female (XX) resulting in 46 total chromosomes for a healthy human. They are numbered 1 – 22 and arranged primarily by size. The combination of genes on the chromosomes, however, are not the same for each person and is what makes you and me a genetically unique organism.

X Inheritance

The inheritance pattern of the X-Chromosome is interesting and can be a gold mine of information when you receive a genetic match on it. Men (XY) inherit their X-Chromosome intact, without recombination, from their mother. This X is a combination of the 2 X-Chromosomes she inherited, one from her father and one from her mother. Since a man only has 1 X he passes it intact to his daughters giving them the ability to see large amounts of genetic information from her paternal great grandparents.

Women (XX) inherit one intact X from their father, passed thru him from her paternal grandmother, and one recombined X from her mother. They then pass on recombined X-Chromosomes unto their children, both boys and girls. The percent of DNA you have in common from parent to child is shown here in the following chart.

Relationship	% X DNA in Common
Father and Daughter	100%
Father and Son	0%
Mother to Daughter	50%
Mother to Son	50%

The further out you go in generations the less in common you have, just like in atDNA inheritance. However, you also begin to lose entire branches off of the male lines.

If you match someone on their X-Chromosome, and they are male, you now know you are looking for ancestry in their maternal line. That is 50% of their family tree wiped away that you do not need to look at. If they match you on the X-Chromosome and another autosome which you have determined to be from a specific line then you can drill down to an even more specific set of ancestors. Just like in atDNA inheritance you will lose DNA segments with each recombination. The further back in time you go the less in common you will share with your ancestors.

Not all of the testing companies analyze the xDNA. There is hope that as the field of study grows, and our understanding of genetic genealogy continues, more companies will offer these results. For now, the best company to test with for xDNA results is 23andMe. FTDNA offers 2 panels of STR testing, and you can see the results on your chromosome browser, but they do not calculate into the algorithm for determining kinship on the website.

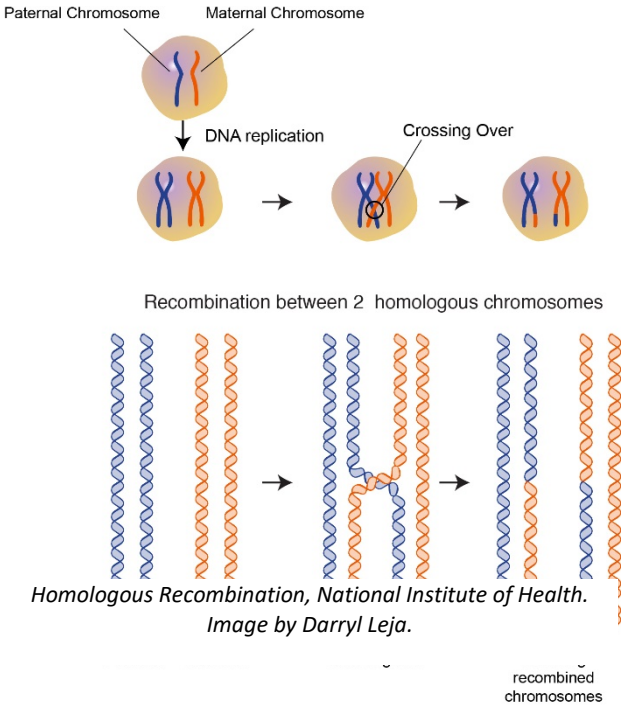
Why you are you

By now you know that 50% of your genetic make-up came from your father and 50% came from your mother. We have cells with a nucleus that contain genetic information along with other "stuff" within a cell wall. To create the cells which will eventually become sperm and eggs specific types of cells go through a process called meiosis. Through this process our cells can half themselves. If your father and your mother passed on a cell with 46 chromosomes each you would have 92 in your cells. That is not correct! Doing the math, our cells must somehow (through meiosis) divide down so they only pass on half of the chromosomes possible. There are three distinct steps to the process.

To begin the chromosomes in the cell pair up to replicate, or make a copy, so there is double the number of chromosomes in the cell. When the cell divides the first time there will then be the correct number of chromosomes (46) in the newly created daughter cells. At this time recombination occurs, which we as genetic genealogists are most interested in.

Recombination, also called crossing over, occurs when the chromosomes exchange genetic material with each other. To put it simply the "arms" of the chromosomes inherited from the person's mother and father touch at multiple points over the length of the chromosome.

Wherever they touch a connection is made and segments of the chromosome are exchanged. Through this process new combinations of chromosomal DNA are created, making each of us a unique individual.



Homologous Recombination, National Institute of Health. Image by Darryl Leja.

Finally in this process the cells divide again into cells that only contain one half of the information needed to create a new person. These cells are called gametes. One gamete from the father and one from the mother eventually combine to create a new chromosomally unique individual.

Not everyone in a family shares the same DNA sequences. Each child in a family, unless they are an identical twin, will have different combinations of DNA passed onto them from their parents. Now think about what happens in the next generation. The child's DNA mixes with a new parent and then even less is passed down from prior generations.

It is important to understand that you may be genealogically related to someone but NOT genetically. You can verify a paper connection for many, many people but you may not have inherited the same ancestral genes in large enough quantities (or at all) to show up in a DNA test.

Due to the odds you play when taking a DNA test you may hear from a variety of people about how important it is to test as many people as you can. Testing wide and deep can also aid in reconstructing ancestral DNA. Yes, you can build in your spreadsheets from segments you have, or discover, what an ancestor's DNA potentially contained.

Ethnicity and atDNA

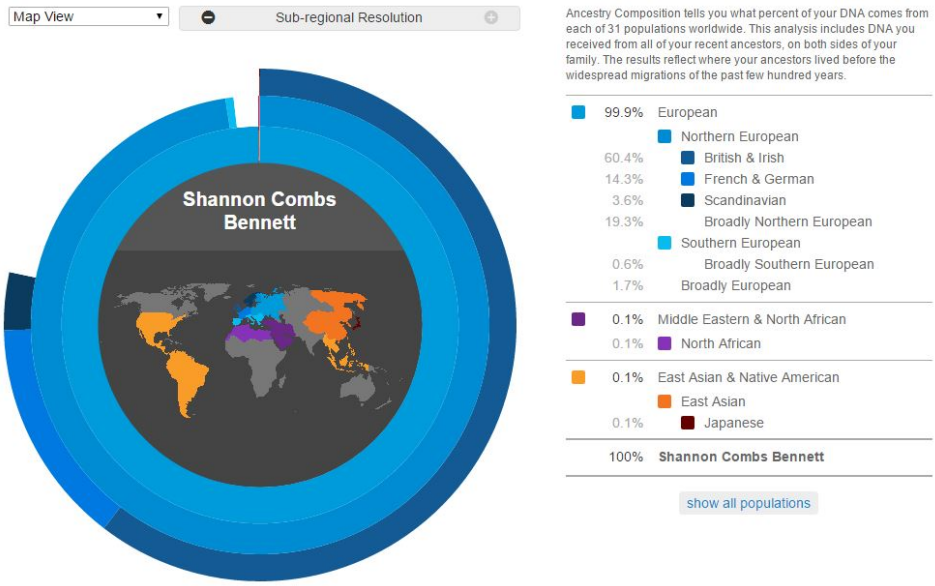
Using your atDNA to find out where in the world your DNA originated from is a fun and interesting side effect of finding your cousins. We all want to know if there is something hidden in our DNA and the surprises that come up when our ethnicity is shown through a testing company can be fascinating. We need to remember though that these results are only estimates, called biogeographical estimations or admixture. Ethnicity is determined through a multi-step process briefly outlined below.

Importance of Reference populations: A biographical estimation is created by a computerized comparison of personal DNA segments to DNA segments of populations from around the world (called a reference population). The more diverse a company's reference population the better it is for their consumers. If a company has a small reference population sample they will be severely limited with the results they can offer. For example, if they only have data for European populations the results will be meaningless for anyone who has African or Asian ancestry. Major global testing companies are continuously updating and adding reference populations as they become available so your results will be the best possible.

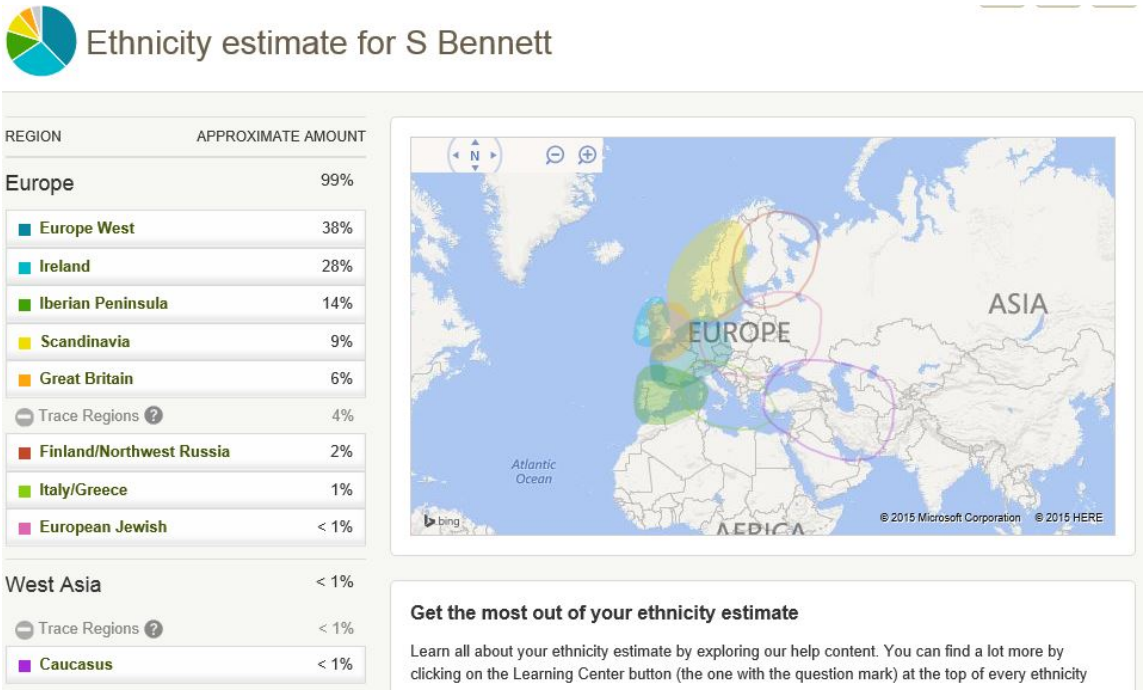
Genetic vs Genealogical Trees: The most important lesson you can take away is the understanding that you have 2 distinct, but overlapping, family trees. Your genealogical family tree is all of the "paper" research. The other family tree, your genetic tree, only contains only those who contributed to our DNA. You should be able to see that your genetic family tree will be a smaller subset of your genealogical family tree. There will be people on both, sure, but a majority will be only on one.

How companies compare: Each company creates their own reference populations and therefore may not be like another company at all. Sometimes it is also due to the process the company puts the segments of DNA through because they have different matching requirements, or a whole host of other variables. To show you how one person's results can vary the images below show my ethnicity results from AncestryDNA, 23andMe, and FTDNA.

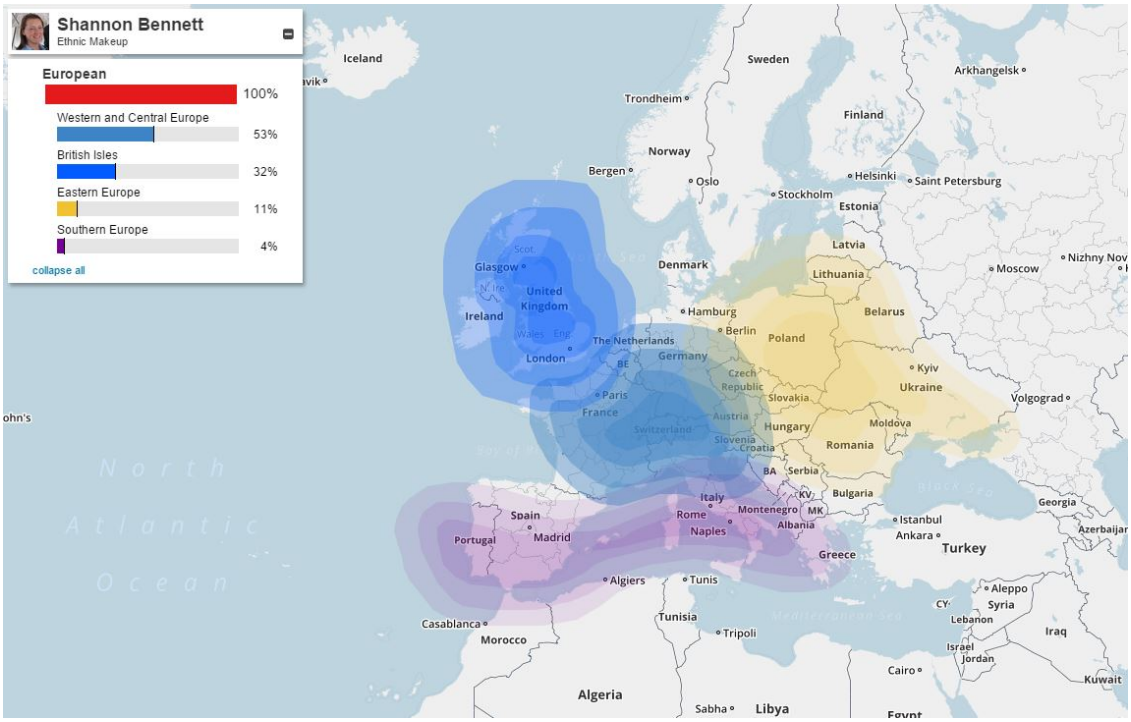
As you can see I am primarily of European descent, and I have the documents which show that. I have trace amounts of DNA from other countries depending on which company is doing the tested. Now, these could be calculation anomalies (especially the ones at less than 1%) or they could be actual trace amounts of ancestral DNA sifting through. Each company used different reference populations which can also alter the results slightly. Particularly 23andMe, which has a good track record of testing for Native American and African Ancestry. Could the small percent of Asian showing through on their result be that elusive Native American Ancestry my family lore says they have?



23andMe Ethnicity Results



AncesrtyDNA Ethnicity Results



Family tree DNA ethnicity Results

Third Party Tools for Analysis

There are a large number of resources and third party tools online to help you analyze your DNA better. Most of them are free, but a few do operate by donation or for a small subscription fee. If you find yourself doing deep analysis more and more you should look at investing in some of these applications.

- **David Pike's Utilities** (http://www.math.mun.ca/~dapike/FF23utils) (FREE) – A comprehensive suite of tools for analyzing raw data, searching for shared DNA in two files, and several advanced phasing tools.
- **GEDmatch** (www.gedmatch.com)
- **Genetic Genealogy Tools** (http://www.y-str.org) (FREE) – An impressive and ever-growing list of advanced tools for analyzing raw data, including an X-DNA Relationship Path Finder, Ancestral Cousin Marriages, Autosomal Segment Analyzer, a DNA Cleaner, a SNP Extractor, and etc. From Felix Jeyareuben Chandrakumar, an Australian software professional.
- **HIR Search** (http://hirs.snpology.com) (FREE) – Once your raw data is entered in the database, you can find HIRs (half-identical regions) that you share with others in the database.
- **Minor Allele Program** (http://www.ianlogan.co.uk/23andme/23andMe_index2.htm) (FREE) – A tool to identify rare SNPs in your 23andMe or Family Tree DNA raw data.
- **Segment Mapper** (http://kittymunson.com/dna/SegmentMapper.php) (FREE) – A tool to show specific DNA segments in a graphic chromosome-style chart.
- **SPA** (http://genetics.cs.ucla.edu/spa/index.html) (FREE) – Spatial Ancestry analysis (SPA) is a method for predicting ancestry or where an individual is from using the individual's DNA. 23andMe users can download the software and analyze their results with this admixture tool.
- **Tim Janzen's Phasing Program (Excel-based):**
  http://dl.dropbox.com/u/21841126/phasing%20program%20%28small%20version%29.xls
  Instructions: http://dl.dropbox.com/u/21841126/phasing%20program%20instructions.rtf
  Y-STR Tool (www.y-str.org/2013/07/phasing-utility.html)