A General Introduction to DNA Testing for Family History

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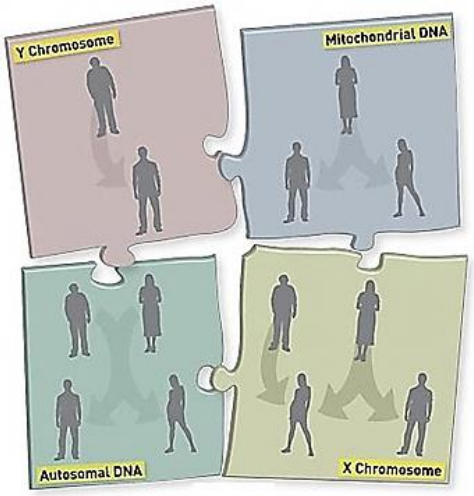
WHAT IS IN THE GENETIC GENEALOGIST TOOLBOX?



During the past 15 years, advances in technology and science contributed new and exciting ways to research our own family history using DNA. Therefore, genetic genealogy is the application of genetic data and utilities to learn more about a person's past and how we are connected to other people, even if the genealogical records might be scarce or missing to support these connections. The benefits of this approach are countless. Thanks to the field of genetic genealogy, individuals can now connect to distant genetic cousins, verify research that has already been done, link into other people family tree, discover the origin of specific family lines, understand the ancient migrations of their early ancestors, and so on. These explorations is mainly carried forward through four types of DNA testing:

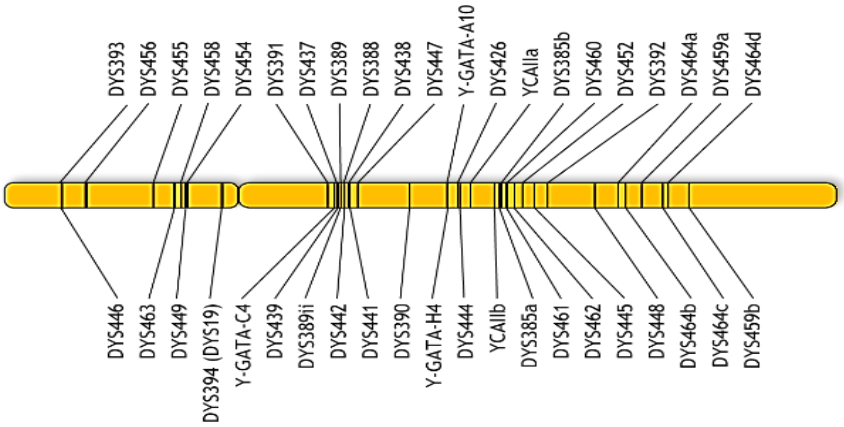
- Y chromosome;
- Mitochondrial DNA;
- Autosomal DNA;
- (X chromosome – not covered in this handout)

WHAT IS THE Y CHROMOSOME?

The Y chromosome (Ycs) is one of the 23 pairs of chromosomes. It determines the male gender in the offspring. It has a specific inheritance pattern that follows the unbroken paternal line, father to son, which in many western cultures would also be the surname line. Because the Ycs remains essentially intact generation after generation, comparison of two individuals sharing a common paternal ancestry would also result in a close or identical Ycs genetic profile. Data is reported in values called markers. Most of these markers have names that begin with the acronym DYS, which stands for DNA Y-chromosome Segment. The number following DYS indicates the position (singular LOCUS; plural LOCI) along the Ycs segment of a particular marker. Therefore, every man would have the same list of loci along their Ycs, but the actual value found at each location may vary from man to man. The combined set of values would be identical or

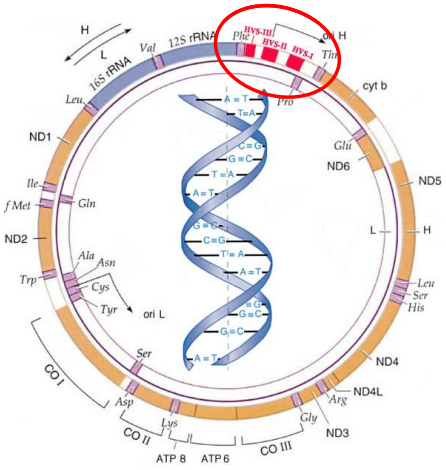
with very few variations between two men sharing a relatively close paternal ancestry, while two individuals that are not related would have sets with several differences.

Women interested in having Y chromosome testing done to study their paternal lines would need to identify a male relative sharing their maiden name that would volunteer to be tested in their stead.



WHAT IS MITOCHONDRIAL DNA?

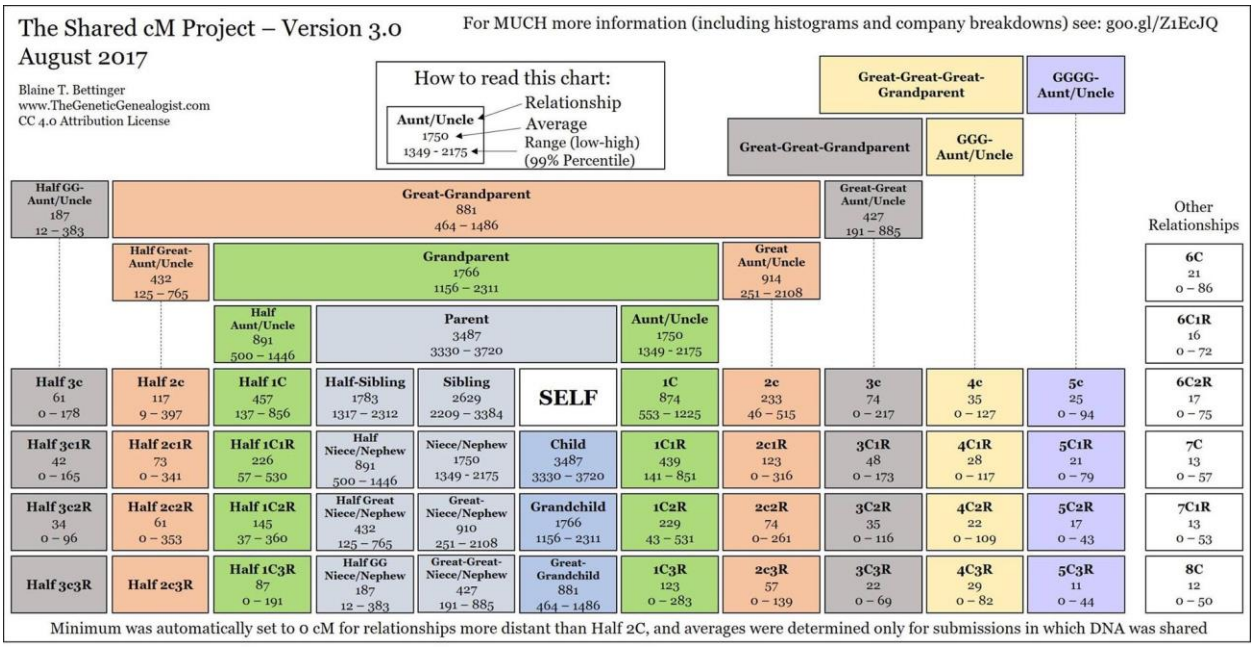
Mitochondrial DNA is a peculiar, ring-shaped piece of DNA found in both men and women that is inherited almost unchanged, generation after generation, along the unbroken maternal line. Copies of the mitochondrial DNA (mtDNA) genome are found in extra-nuclear organelle called mitochondria, which are used by the cell to produce the energy that is necessary to sustain life. This genome is "only" 16569 bases long, which is much shorter when compared to the nearly 3.2 billion bases found on the 23 pairs of chromosomes within the nucleus of each cell. However, even 16569 bases can be too many for a quick analysis. Therefore, after the sequencing of the first complete mtDNA genome in 1981 (which was revised and corrected in 1999), scientists decided to simply report ONLY the differences from the first mtDNA sequenced, which became the standard for comparison for ALL the mtDNA sequences produced since 1981. The standard reference sequence is often called CRS or rCRS. Additionally, only mutations from the reference sequence in the segment of mtDNA provided by the genetic lab are reported. For example, if a person purchases a test that would only provide the sequence of the mtDNA control region (approximately 1100 bases ranging from position 16000 to 16569 and from 1 to 570 and named HVR1, HVR2 and sometimes HVR3), only the mutations from the reference sequence within this range will be provided. If a person purchases the Full Genome Sequence (the full 16569) all the mutations from the reference sequence will be provided thus obtaining the HIGHEST level of molecular resolution for the mtDNA genome.



Lastly, specific mutations provide important details regarding the recent and ancient history of each mtDNA lineage (called haplogroup). These mutations are used to place every mtDNA profile on a specific branch of the world mtDNA tree. These branches (or haplogroups) are named following a straightforward pattern of letters and numbers. The most up to date mtDNA tree (called a phylogeny) is found at PhyloTree.org.

WHAT IS AUTOSOMAL DNA?

In humans, genetic information is contained in 23 pairs of chromosomes inside the cell's nucleus and in a small genome called mitochondrial DNA found in extra-nuclear organelles called mitochondria. The 23rd pair of chromosomes is referred to as sex chromosomes, as they are responsible for determining the gender of the subject. A mother would always contribute an X-chromosome, while the father will contribute either an X- or a Y-chromosome. The combination of two X's will result in female offspring, while a combination of Y and X will result in a male. Y chromosome DNA and mitochondrial DNA have been employed successfully for nearly 20 years in reconstructing genealogical information regarding the unbroken paternal and maternal lines respectively. In recent years, genetic genealogy testing has expanded into autosomal DNA testing, which includes a large survey of sites found across the remaining 22 pairs of chromosomes, known as autosomes.



Two characteristics of autosomal DNA help us understand how it is used in family history and also its limitations as compared to uniparental markers such as the Y chromosome and mitochondrial DNA. First, we receive it from both of our parents, their parents and so on back a few generations, which means that we can use it to trace all of our family lines independently from the gender of the ancestor. However, we also lose half of it at every generation so chances

are that after 5-6 generations things are getting quite blurry and many of our ancestors begin to be related to us only genealogically, but not genetically.

While the immediate benefit of autosomal DNA testing is the opportunity to recover genetic information useful to determine the ancestry of ALL our pedigree lines, regardless of gender, the abundance of data and the process of *recombination* (the shuffling and loss of DNA at each subsequent generation) of autosomal DNA has created new challenges in understanding and successfully applying genetic data to a genealogical context. The new field is quickly evolving and the companies offering these tests are eager to continue improving the way that results are delivered and explained. Genetic genealogy consulting is also growing due to the large number of individuals tested and the amount of data now available to reconstruct family histories and ancestry, which could be often confusing to the traditional genealogist (www.GeneticGenealogyConsultant.com).

What Autosomal Tests are Available?

A table indicating all the available autosomal DNA tests and their details is available at: http://www.isogg.org/wiki/Autosomal_DNA_testing_comparison_chart.

Further reading on genetic genealogy is available on the International Society of Genetic Genealogy website: www.isogg.org and www.GeneticGenealogyConsultant.com.