



DNA for Genealogists

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- **DNA (DeoxyriboNucleic Acid)** is the genetic material of organisms. Nuclear DNA is present in the cell nucleus and is usually structured into double strands, hence the familiar double helix.
- A **GENE** is the basic unit of heredity; a sequence which is part of the long strands of DNA.
- A **CHROMOSOME** is a piece of DNA containing genes. It is the structure by which hereditary information is physically transmitted from one generation to the next.
- A **GENOME** is all the genetic material of a particular organism – may refer specifically to the DNA inside the cell's nucleus. In humans, the nuclear genome comprises 46 chromosomes (23 pairs), with a complete copy found in each cell of the body. The term genome may also be used to refer to mitochondrial DNA.
- An **AUTOSOME** is a chromosome not involved in sex determination. The human genome consists of 23 pairs of chromosomes, 22 pairs of autosomes (numbered 1-22), and one pair of sex chromosomes (the X and Y chromosomes).
- **SEX CHROMOSOMES** (X and Y-chromosomes) are the chromosomes involved in sex determination. Females have two X chromosomes; males have one X and one Y-chromosome.
- **DOMINANT / RECESSIVE:** In a matched pair, each chromosome has genes that control the same function (eg freckles/ no freckles). A dominant gene on one chromosome will override a recessive gene on the other.
- A **MUTATION** is any inheritable change in DNA sequence. Mutations usually occur as a result of miscopying by cell enzymes.
- A **MARKER** (or Locus - pl. loci) is a gene at a known location on a chromosome. The greater the number of markers tested between 2 individuals, the better defined is their relationship.
- A **SNP** (*pronounced 'snip', think 'typo'*) (**S**ingle **N**ucleotide **P**olymorphism) is where a single DNA 'letter' differs from person to person. SNPs account for 90% of the genetic variation in humans. Nearly all SNPs have mutated only once in human history so large proportions of the population have the same SNP.
- An **STR** (Short Tandem Repeat, *think 'stutter'*) **MARKER** is a stretch of DNA where a small sequence repeats itself several times. An STR mutation adds or subtracts one or more repeats. STRs of the Y-chromosome are useful for tracking exclusively male lines over the last few hundred years.
- An **ALLELE** is the number of repeats at an STR marker.
- The **HAPLOTYPE** is the set of numbers that are the alleles at specific STR markers. Two or more haplotypes may be compared as a check for a genetic relationship. Closely related individuals have similar haplotypes.
- **DYS** (**D**NA **Y**-chromosome **S**ingle copy sequence) numbers refer to particular STR marker locations on the Y-chromosome (eg DYS388, DYS390).
- **HAPLOGROUPS** define large groups of genetic populations and are often geographically orientated. For example, the Y-chromosome haplogroup most common to males living along the western coasts of Europe is Haplogroup R1b. The Y-chromosome **haplogroup** of an individual is **not** defined by their **haplotype**, because haplotypes are determined by STR markers. Instead, **haplogroups** are determined by SNPs.
- **MITOCHONDRIAL DNA (mtDNA)** is the circular DNA contained inside the mitochondria. Mitochondria are small organelles residing in animal cells that provide the power to the cell. The mtDNA is passed from mother to her offspring (both sons and daughters), but only her daughters pass it on. Deep but very broad genealogies (i.e. several thousands of years) can be deduced by considering mtDNA. In addition tests of the 'full sequence' of mitochondria indicate recent relatives with a common ancestor on the all-female line. A mtDNA haplogroup (or **clade**) can usually be assigned for any given sequence.
- **CAMBRIDGE REFERENCE SEQUENCE** is a standard sequence of mitochondrial DNA against which all other mtDNA is compared. The results of a mtDNA test cite only the markers that differ from this standard.
- **NON-PATERNITY EVENT** is a situation where a family member does not have the same genetic profile as his alleged father, perhaps due to adoption, illegitimacy, etc.

- **MOST RECENT COMMON ANCESTOR (MRCA)** between two people can be predicted by their DNA. For example, two 1st cousins share a grandparent as their MRCA. If the cousins were both boys, they would share their grandfather's Y-chromosome. The time when the common ancestor lived can be approximated by the number of markers tested, the mutation rate and the number of mismatches.
- **AUTOSOMAL DNA (atDNA)** tests between 2 people compare the linked blocks of DNA across the 22 pairs of autosomal chromosomes. As autosomes contain random mixtures of DNA from ancestors, closer relatives share larger fragments of DNA in common. Autosomal tests can be used to test for common ancestors across all lines (male and female) but are most useful if the common ancestor is within 6 generations.
- **GENETIC GENEALOGY** is a new field of family history research, comparing Y-chromosomes, mitochondrial or autosomal DNA to prove or disprove biological connections. The Y-chromosome links the strictly paternal line (father to son), whilst mtDNA links the all-maternal line. Autosomal DNA tests compare all family lines.

FOR FURTHER INFORMATION

Books

- *DNA for Genealogists*, Kerry Farmer, Unlock the Past, Modbury, S. Aust., 2011
- *DNA and Social Networking*, Debbie Kennett, The History Press, Stroud (GLS, UK), 2011
- *DNA & Genealogy*, Colleen Fitzpatrick & Andrew Yeiser, Rice Book Press, USA, 2005
- *DNA and Family History*, Chris Pomery, the National Archives, Kew (UK), 2004
- *Family History in the Genes*, Chris Pomery, the National Archives, Kew (UK), 2007
- *DNA & Tradition*, Rabbi Yaakov Kleiman, Devora Publishing Company, New York, 2011
- *How to Interpret Your DNA Test Results for Family History & Ancestry*, Anne Hart, Writers Club Press, NY, 2002

Major testing companies *(also see their information pages)*

- Family Tree DNA – <http://www.familytreedna.com>
- 23 and Me – <http://www.23andMe.com> *(health scan as well as genetic ancestry)*
- Ancestry.com – <http://dna.ancestry.com>
- GeneTree – <http://www.genetree.com> *(useful for those who had the Brigham Young test years ago)*
- Oxford Ancestors – <http://www.oxfordancestors.com>

Free DNA databases *(for entering and/or comparing DNA test results)*

- Mitosearch – <http://www.mitosearch.org/> *(search mtDNA)*
- Ysearch – <http://www.ysearch.org/> *(search by Y-DNA or by surname)*
- Sorenson Molecular Genealogy Foundation – <http://www.smgf.org/> *(search Y-DNA or mtDNA databases)*

Further reading

- Autosomal DNA – <http://www.familytreedna.com/faq/answers/default.aspx?faqid=17>
- Autosomal percentage estimates – <http://www.yourgeneticgenealogist.com/2010/09/known-relative-studies-with-23andme.html>
- DNA forums – <http://dna-forums.org/>
- DNA Genealogy mailing list – <http://lists.rootsweb.ancestry.com/index/other/DNA/GENEALOGY-DNA.html>
- DNA glossary – <http://www.dnaheritage.com/glossary.asp> *(common genetic terms)*
- DNA Testing – <http://www.buzzle.com/articles/dna-testing/> *(articles)*
- DNA Tutorials – <http://www.genetree.com/tutorials> *(short online videos)*
- Forensic Genealogy – <http://www.forensicgenealogy.info/> *(Most Recent Common Ancestor lookup tables)*
- GEDmatch – <http://www.gedmatch.com> *(tools for comparing Y-DNA, mtDNA, atDNA from FTDNA & 23andMe)*
- Genetics 101 – <https://www.23andme.com/gen101/>
- Human Genome Project – http://www.ornl.gov/sci/techresources/Human_Genome/project/info.shtml
- International Society of Genetic Genealogy – <http://www.isogg.org/> *(includes free newsletter)*
- Interpreting Genetic Genealogy results – http://www.familytreedna.com/pdf-docs/Interpreting-Genetic-Genealogy-Results_web_optimized.pdf *(eBook)*
- Mitomap – <http://www.mitomap.org/>
- Most Recent Common Ancestor – http://en.geneawiki.com/index.php/Most_recent_common_ancestor
- National Geographic Project – <https://genographic.nationalgeographic.com/genographic/>
- Online Journal of Genetics & Genealogy – <http://jgg-online.blogspot.com/>
- Short Tandem Repeat Internet DNA Database – <http://www.cstl.nist.gov/biotech/strbase/index.htm>
- The Genetic Genealogist Blog (Blaine Bettinger) – <http://www.thegeneticgenealogist.com/> *(eBook)*
- Time to Most Recent Common Ancestor calculator – <http://nitro.biosci.arizona.edu/ftdna/TMRCA.html>
- Tour of the DNA basics – <http://learn.genetics.utah.edu/content/begin/tour/>
- What can you learn from a DNA test – <http://www.nationalarchives.gov.uk/podcasts/dna-test.htm>