



DNA for Genealogists

by Kerry Farmer



- **ADMIXTURE** occurs when two or more previously separated populations inter-breed and may be loosely referred to as **ethnicity**.
- An **ALLELE** is the number of repeats at an STR marker.
- **AUTOSOMAL DNA (atDNA)** tests compare the linked blocks of DNA across the 22 pairs of autosomal chromosomes with that of others. As autosomes contain random mixtures of DNA from ancestors, closer relatives share larger fragments of DNA in common. Autosomal tests can test for shared ancestors across all lines (male and female) but are most useful if the ancestor in common lived within about 5 generations.
- An **AUTOSOME** is a chromosome not involved in sex determination. Humans have 23 pairs of chromosomes, 22 pairs of autosomes (numbered 1-22), and one pair of sex chromosomes (the X and Y chromosomes).
- **CAMBRIDGE REFERENCE SEQUENCE (CRS)** 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. mtDNA may also be compared to the **revised Cambridge Reference Sequence (rCRS)** or else to the **RSRS (Reconstructed Sapiens Reference Sequence)** which is the calculated mtDNA of the deepest common maternal ancestor of all people alive today.
- 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.
- **DNA (DeoxyriboNucleic Acid)** is the genetic material of organisms. Nuclear DNA is present in the cell nucleus and is usually structured into double strands, wound together as the familiar double helix.
- **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.
- **DYS (DNA Y-chromosome Single copy sequence)** numbers refer to particular STR marker locations on the Y-chromosome (eg DYS388, DYS390).
- A **GENE** is the basic unit of heredity; a sequence which is part of the long strands of DNA.
- **GENETIC GENEALOGY** is a field of family history research that compares 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 (recent) family lines.
- A **GENOME** is all the genetic material (although commonly refers to DNA on the chromosomes). In humans, 46 chromosomes (23 pairs) make up the genome, with a complete copy found in each cell of the body. (There is also a mitochondrial genome).
- **HAPLOGROUPS** define large groups of genetic populations (descended originally from one ancestor's mutation) 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 most common European mitochondrial haplogroup is haplogroup H. The **haplogroup** of an individual is determined by SNPs.
- A **MARKER** is a gene at a particular physical location (**locus**) on a chromosome. The greater the number of markers tested between 2 individuals, the better defined is their relationship.
- **MITOCHONDRIAL DNA (mtDNA)** is the circular DNA contained inside the mitochondria (but outside the cell's nucleus). Mitochondria are small organelles 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. Broad genealogies (across several thousands of years) can be deduced by considering mtDNA. However comparing the 'full sequence' of mitochondria can indicate recent relatives with a shared ancestor on the all-female line.
- **MOST RECENT COMMON ANCESTOR (MRCA)** between two people can be predicted by their DNA. For example, two first cousins share a grandparent as their MRCA.
- A **MUTATION** is any inheritable change in DNA sequence. Mutations usually occur as a result of miscopying by cell enzymes.
- **NON-PATERNITY EVENT** is a situation where the DNA of a family member indicates a different father, perhaps due to adoption, illegitimacy, etc.

- **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. Y-chromosome tests usually check STR markers. X-chromosome DNA is usually tested (and reported) with autosomal testing.
- A **SNP** (*pronounced 'snip'*) (**S**ingle **N**ucleotide **P**olymorphism) is where a single DNA 'letter' differs from person to person, produced by a copying mutation ('*typo*'). SNPs account for 90% of the genetic variation in humans. Nearly all SNPs have mutated only once in human history so large numbers of people have the same SNP.
- An **STR** (Short Tandem Repeat, *think 'stutter'*) 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.

FOR FURTHER INFORMATION

Books

- Bettinger, Blaine *The Family Tree Guide to DNA Testing & Genetic Genealogy*, Family Tree Books, Cincinnati USA, 2016.
- Bettinger, Blaine & Parker Wayne, Debbie *Genetic Genealogy in Practice*, National Genealogical Society, Arlington VA USA, 2016.
- Collins, Francis *The Language of Life: DNA and the Revolution in Personalised Medicine*, Profile Books, London, 2010 (Amazon)
- Farmer, Kerry *DNA for Genealogists*, 4th edition, Unlock the Past, Modbury, S. Aust., 2017 (Gould)
- Kennett, Debbie, *DNA and Social Networking*, The History Press, Stroud UK, 2011 (Amazon)

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

- 23 and Me – www.23andme.com (*atDNA tests*)
- Ancestry.com – www.ancestry.com (*atDNA tests*)
- Family Tree DNA – www.familytreedna.com (*Y-DNA, mtDNA & atDNA tests + projects*)
- Genographic Project Geno 2.0 – <https://genographic.nationalgeographic.com> (*deep ancestry*)
- Living DNA – www.livingdna.com (*detailed sub-regional ethnicity, currently not cousin-matching*)
- My Heritage – www.myheritage.com/dna/ (*atDNA tests, can upload from other companies*)

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

- Gedmatch – www.gedmatch.com (*tools comparing Y-DNA, mtDNA, atDNA from FTDNA & 23andMe*)
- Mitosearch – www.mitosearch.org (*search mtDNA*)
- Ysearch – www.ysearch.org (*search by Y-DNA or by surname*)

Further reading

- Autosomal percentage estimates – http://bit.ly/YGG_DNApercentages (*CeCe Moore*)
- Basic genetics – <http://learn.genetics.utah.edu/content/basics/> (*University of Utah*)
- Cruwys news – <http://cruwys.blogspot.co.uk> (*Debbie Kennett's blog*)
- DNA Lectures (Who do you Think You Are) – <http://bit.ly/DNA-WDYTYA> (*online lectures*)
- DNA Testing – www.buzzle.com/articles/dna-testing/ (*articles*)
- DNAeXplained – <http://dna-explained.com> (*Roberta Estes' blog*)
- DNAGedcom – www.dnagedcom.com (*extra tools for adoptees*)
- Family Tree DNA webinars – www.familytreedna.com/learn/ftdna/webinars/ (*recorded online lectures*)
- From DNA to Genetic Genealogy – <http://stevemorse.org/genetealogy/dna.htm>
- (The) Genetic Genealogist Blog (Blaine Bettinger) – www.thegeneticgenealogist.com (*blog & resources*)
- Genetics 101 – www.23andme.com/en-int/gen101/
- Genie1 – www.genie1.com.au/blog (*Louise Coakley's blog, especially for Australians*)
- Genome Mate Pro – www.getgmp.com (*tool for managing multiple DNA kits*)
- International Society of Genetic Genealogy (ISOGG) – www.isogg.org (*many resources*)
- ISOGG Wiki – www.isogg.org/wiki/ (*many resources*)
- Autosomal DNA statistics – https://isogg.org/wiki/Autosomal_DNA_statistics (*includes shared cM*)
- Genetics Glossary – https://isogg.org/wiki/Genetics_Glossary (*common genetic terms*)
- Interpreting Genetic Genealogy results – <http://bit.ly/FTDNAowwhat> (*eBook*)
- Jewish Story & a Reassessment of DNA evidence – www.jogg.info/11/coffman.htm
- Kitty's chromosome mapper graphic – <http://kittymunson.com/dna/ChromosomeMapper.php> (*tool*)
- Kitty's Overlapping Segment Mapper – <http://bit.ly/KC-SegmentMapper> (*tool*)
- (The) Legal Genealogist – www.legalgenealogist.com/blog/ (*legal implications, Judy Russell's blog*)
- Most Recent Common Ancestor calculator – <http://nitro.biosci.arizona.edu/ftdna/TMRCA.html>
- Promethease – www.promethease.com (*purchase genetic health predisposition report*)
- Your Genetic Genealogist – www.yourgeneticgenealogist.com (*CeCe Moore's blog*)