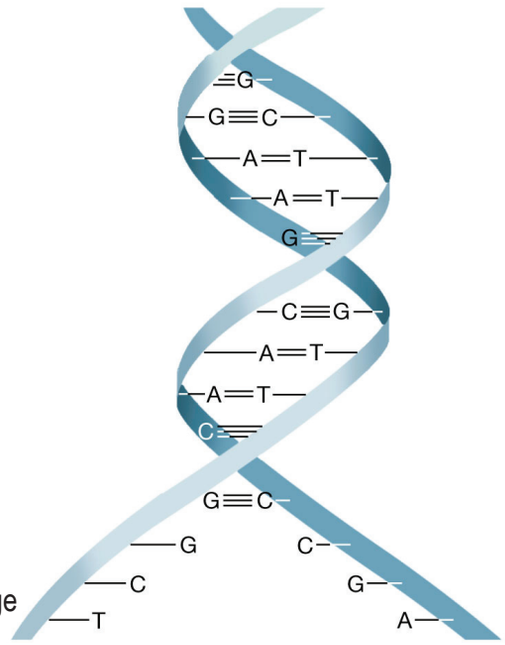
DNA and Genealogy

Brief summary

Tree Building - Genealogy Basics



- Build your tree through documents like BDMs
- Make sure you have evidence to support your tree
- DNA tests are a tool to help confirm or extend your tree

DNA Basics (see glossary)

- We have 2 very different types of DNA, Nuclear and Mitochondrial
- Nuclear DNA is coiled up into 23 pairs of chromosomes
- One set came from father, the other from mother
- 22 pairs - autosomal, the other pair are sex cells (X and Y)
- Males have YX sex cells, females XX

Inheritance

- Mitochondrial DNA passes from mother to all children with little change
- Y DNA passes from father to boys only with little change
- X DNA has a specific inheritance pathway
- Autosomal DNA is 'shuffled' during the making of sperm and egg cells
- This 'shuffling' (recombination) is random, but allows large segments of DNA to pass on unchanged

DNA testing

- 3 key tests (Y-DNA, mt-DNA, at-DNA)
- 3 suppliers of autosomal tests
- Each supplier has large database of potential matches
- Consider 'fishing in all three ponds' for matches

The tests in summary…

TEST	USED TO –	STRENGTHS	LIMITS
Y - DNA	Find male relatives that share common ancestor in **paternal** line	Close matches share paternal ancestor. If close male relatives don't match, proves there's been a mistake in the tree (a NPE?)	Only males can be tested Only checks paternal line Common ancestors may have lived 100s of years ago. Doesn't 'prove' paternal relationship
MT-DNA	Find relatives that share common ancestor in **maternal** line	Close matches share maternal ancestor. If close female relatives don't match, proves there's been a mistake in the tree	Only checks maternal line Common ancestors may have lived many 100s of years ago Doesn't 'prove' maternal relationship
AUTOSOMAL	Find relatives that share common ancestor **in any branch** of your tree	Most likely will find many cousins X-matches can be useful Tools available to do significant analysis	Some relationships (3rd cousins and beyond) may not show up Hard to tell which branch Multiple companies; your relatives may not be in their databases

Where to buy the tests…

TEST	AVAILABLE FROM	COMMENTS	COSTS (EXCHANGE RATE 1 JAN16)
Y - DNA	FamilyTreeDNA	Number of markers is important; the more markers the more $$$	Y37 = $US169 (**$A242**) Y67 = $US268 (**$A385**) Y111 = $US359 (**$A515**)
MT-DNA	FamilyTreeDNA	Early tests not very useful; current full sequence test can be very helpful	HVR1&2 = $US69 (**$A99**) Full = $US199 (**$A286**)
AUTOSOMAL	FamilyTreeDNA	"Family finder" test popular with UK and Australian customers	**$A152**
AUTOSOMAL	Ancestry	Only recently available to Australian customers. Largest database	**$A179**
AUTOSOMAL	23andme	Many customers use it for medical information, rather than genealogy	**$A238**

Test results

- Each site will display 'matches' with an estimate of closeness
- Each site has tools to help understand and analyse results
- There are other sites with analytical tools which can help

Useful websites

- International Society of Genetic Genealogy - good for back-ground, great Wiki — http://isogg.org
- Louise Coakley (Genie1) - Australian, also has a great Blog — http://genie1.com.au/
- FamilyTreeDNA - testing company: only source of Y-DNA and mt-DNA tests — https://www.familytreedna.com
- Ancestry - testing company, source of genealogical info and published trees — http://dna.ancestry.com.au
- 23andme - testing company, mostly focused on health rather than genealogy — https://www.23andme.com/en-int/
- Gedmatch - upload your raw data: free analytical tools (need to pay for Tier One tools) — http://v2.gedmatch.com/login1.php
- DNA Land - upload your raw data: free analytical tools — https://dna.land
- DNAGedcom - upload Gedmatch processed data: free ADSA report — https://dnagedcom.com

Glossary of useful terms

(adapted from John Chandler's glossary, Edmund Rice (1638) Association, http://www.edmund-rice.org/dnagloss.htm)

allele	variant of a gene or genetic marker. For STR markers, each allele is designated by the number of repeats of the short base sequence.
autosome	one of the non-sex-determining chromosomes. Autosomes occur in nearly identical pairs. See also X and Y.
base	the building block of DNA, one of four molecules that link up to make a DNA chain. The four are named adenine, cytosine, guanine, and thymine (A, C, G, T for short). These are also called nucleotides.
centiMorgan	a unit of recombinant frequency which is used to measure genetic distance. It is often used to imply distance along a chromosome, and takes into account how often recombination occurs in a region. A region with few cMs undergoes relatively less recombination.
chromosome	one of the DNA macromolecules found in the cell nucleus. Humans have 46 chromosomes. See also X and Y.
CRS	the Cambridge Reference Sequence for mtDNA, used for convenience to compare with all human mtDNA sequences, such that any sequence can be expressed concisely as a relatively short list of difference from the reference
DNA	deoxyribonucleic acid. The chemical constituent of genes and chromosomes. DNA has four different base units, designated A, C, G, and T, which are connected in long double chains, and the sequence of these bases encodes the genetic information.
DYS	DNA Y-chromosome Segment. A label for loci or genetic markers on the Y chromosome. Each marker is designated by a number, according to international conventions. Virtually all DYS designations discussed in the context of genetic genealogy are those of STR markers (which are useful for genealogy because of their relatively high mutation rate).
gene	a meaningful sub-unit of DNA, encoding a protein.
gene conversion	a process replacing one allele of a pair with a copy of the other. This term is used by extension for all kinds of loci, not just genes. See also recLOH.
genome	the entire inventory of nuclear DNA in an organism.
genotype	a set of allele values of one or more markers for one individual. Since most of the DNA consists of nearly identical pairs of chromosomes, a genotype typically has two alleles for each marker, one from each parent. See also haplotype.
haplogroup	a classification comprising many different haplotypes thought to be related. Haplogroups are defined in terms of markers that mutate so slowly they are treated as if they have occurred only once in all time. The major haplogroups originated thousands or tens of thousands of years ago.
haplotype	a set of allele values of one or more markers for one individual and identified as coming just from one parent, particularly markers on the Y chromosome or in mitochondrial DNA. See also genotype.
heterozygous	having two different alleles in the two copies of the same marker or locus existing on a pair of chromosomes. By extension, this term is sometimes applied to the two copies of a locus found on opposite arms of a palindrome on the Y chromosome. See also homozygous.
homozygous	having the same allele in the two copies of the same marker or locus existing on a pair of chromosomes. By extension, this term is sometimes applied to the two copies of a locus found on opposite arms of a palindrome on the Y chromosome. See also heterozygous.
HVR1	Hypervariable Region 1. A portion of the mtDNA molecule noted for its especially high mutation rate, consisting roughly of locations 16024-16569.

HVR2	Hypervariable Region 2. (See HVR1.) The limits of HVR2 are even more vague than for HVR1. HVR2 is generally said to start at location 1 and to extend for a few hundred bases, but part of this region is often called HVR3.
locus	(plural: loci) specific site on a DNA chain.
marker	a distinctive sub-unit of DNA, often not part of a gene. Often used interchangeably with locus, but referring to the **contents** of the site, rather than the **site** itself.
microsatellite	See STR.
mitochondria	organelles within the cell responsible for converting food into usable energy. Each mitochondrion has its own DNA. The mitochondria in a child come entirely from the mother, and so mitochondrial DNA ("mtDNA" for short) is a tracer of female-line ancestry, just as Y DNA is a tracer for male-line ancestry.
mtDNA	See mitochondria.
mutation	An event in which the DNA chain alters. In the case of STR markers, a mutation is almost invariably the gain or loss of one repeat of the basic short sequence (or, rarely, two repeats). Other types of mutations include the substitution of one base for another (known as a SNP) and the insertion or deletion of a whole segment (known as an indel. See also recombination.
nucleotide	a unit of DNA. See also base.
organelle	any small, compact, and cohesive entity within a cell. An organelle is to a cell what an organ is to a multi-celled creature.
PCR	Polymerase Chain Reaction. A chemical process that replicates a given sample of DNA many times, in imitation of natural replication. The process cycles between two stages: splitting the two strands of DNA apart and then forming new double strands by adding a mixture of the enzyme polymerase and the four DNA bases. By adding primers as well, the process can be used to replicate just the one or more DNA segments of interest.
polymorphism	The occurrence of more than one form of DNA in different individuals, or even in different cells within one individual. Such diversity arises through the occurrence of mutations.
recLOH	recombinant loss of heterozygosity. See also gene conversion.
recombination	a process of "mixing and matching" of paired chromosomes that takes place at cell division. One or more segments may be swapped between the two chromosomes, or occasionally a segment may replace the corresponding segment on the other chromosome. This process can also occur on palindromic segments of the Y chromosome and may affect paired loci, such as DYS385a and DYS385b.
SNP (pronounced "snip")	Single-Nucleotide Polymorphism. A type of mutation characterized by the substitution of one base for another, or the outright loss of a base.
STR	Short Tandem Repeat. Also known as microsatellite. This is a genetic marker consisting of multiple copies of a short motif, (a sequence of DNA bases). Occasionally, a microsatellite will mutate by the gain or loss of one repeat. So-called "simple" STRs have just one contiguous set of repeats; "complex" STRs may have multiple sets of repeats separated by short patches of non-repeating DNA and may even have repeats of more than one motif.
X	one of the two sex-determining chromosomes, See also Y.
Y	one of the two sex-determining chromosomes. A person with the combination XX is female, while a person with XY is male. Most of the Y chromosome, unlike the others, does not trade DNA with a "partner chromosome," and it therefore passes essentially intact from father to son. This property leads to a minimum of ambiguity in interpreting the results of Y DNA analysis.