

Glossary

This is a selection of definitions from the ISOGG at <http://www.isogg.org/>

Allele: [*pron. a-lee-lee*] Broadly, one of the alternative forms of a gene or genetic marker. More narrowly, the term allele value refers to a count of the number of repeats in an STR (pronounced ess-tee-are). A list of marker labels and their associated allele values constitutes an individual's *haplotype*.

Autosomal DNA - The non-gender chromosomes that mix or recombine. Also known as admixture DNA.

Base: A small chemical molecule which is the information portion of the nucleotides in DNA. The chemical bases are: A (Adenine), T (Thymine) C (Cytosine) and G (Guanine).

Base Pair (bp): A (Adenine) pairs with T (Thymine) and C (Cytosine) pairs with G (Guanine). These base pairs form the ladder of the DNA molecule.

Clade: From the Greek word *klados*, meaning branch. A clade on the Y chromosome tree is also called a **haplogroup**.

CRS - Acronym for Cambridge Reference Sequence. The first mitochondrial DNA to be fully sequenced at Cambridge University in 1981. The anonymous donor of the CRS was haplogroup 'H'. Mitochondrial results are determined based upon where the results differ from the CRS.

DNA (deoxyribonucleic acid): The large molecule inside the nucleus of a cell that carries genetic instructions for making living organisms. See Y-DNA.

Haplogroup - A group of similar haplotypes that share a common ancestor with a SNP mutation. Because a haplogroup consists of similar haplotypes, this is what makes it possible to predict a haplogroup. A SNP test confirms a haplogroup. Haplogroups are assigned letters of the alphabet, and refinements consist of additional number and letter combinations, Example: R1b1. Y-chromosome and mitochondrial DNA haplogroups have different haplogroup designations. Haplogroups pertain to your deep ancestral origins dating back thousands of years.

Haplotype: Broadly, the complete set of results obtained from multiple markers located on a single chromosome. For the Y chromosome, the term is restricted by convention to allele values (number of repeats) obtained from microsatellite (STR) markers.

Junk DNA - Slang term usually used in referring to the non-coding region of DNA on the Y-chromosome. For more about: <http://www.psrast.org/junkdna.htm>

Marker: An identifiable physical location on a chromosome that is variable between individuals and whose inheritance can be monitored. A term commonly used along with allele values in describing an individual's haplotype. Marker labels, such as M173 or DYS388, have no intrinsic meaning.

Mitochondrial DNA - Energy releasing organelles located in the cytoplasm of cells, which contain their own DNA. Mitochondrial DNA is passed from mother to child, but only females continue to pass on their maternal mitochondria to their children.

MRCA - Acronym for Most Recent Common Ancestor.

mtDNA - Acronym for mitochondrial DNA.

Mutation - A change in the DNA that occurs at random. Mutation is a scientific term that often connotes a negative connotation as a result of 1950's 'B' movies, but in genetic genealogy, a mutation is usually beneficial. Mutations on the Y-chromosome are often used for distinguishing different ancestral line

Short Tandem Repeats (STR - pronounced ess-tee-are): Patterns in the DNA sequence which repeat over and over again in tandem, i.e., right after each other. Typically the repeat motif is less than six (6) base pairs long. By counting the repeats, one gets an allele value which is given in an individual's haplotype. They are also called microsatellites and Simple Sequence Repeats (SSRs).

Single Nucleotide Polymorphism (SNP which is pronounced 'snip'): Variation in the nucleotide allele at a certain nucleotide position in the human genome. When the change occurs it is called a polymorphism, and polymorphisms accumulate over time. A polymorphism can be very common (found in a significant fraction of global or localized populations) or very rare (found in a single

individual). Common variations are used to track the evolution of the human genome over time (population genetics) and can be graphically represented in a haplogroup or phylogenetic tree.

Unique Event Polymorphism (UEP): A mutation which is treated as if it occurred only once in all of human history, so that all persons sharing the mutation descend from a common ancestor. Most UEPs are **Single Nucleotide Polymorphisms (SNPs)**, while some are insertions or deletions (for examples, see LINE and YAP).

X chromosome: One of two types of sex determining chromosomes, the other being the Y chromosome. When two X chromosomes, one from each parents, are paired with each other in a fertilized egg cell, the resulting child will be female. If the fertilized egg cell contains both an X and a Y chromosome, the resulting child will be male. The X chromosomes become subject to cross-over effects during subsequent egg cell creation in the female offspring, and thus the homologous gene alleles and genetic marker alleles in both these X chromosomes can randomly swap positions in the next generations making it very difficult to track a particular X chromosome over more than a couple of generations. Determining a common ancestor for an X chromosome is very difficult beyond a couple of generations; therefore, the X chromosome is not a very useful tool for genetic genealogy purposes.

YAP: See Y Alu Polymorphism.

Y Alu Polymorphism (YAP): A Unique Event Polymorphism that is an insertion of a few hundred base pairs. There are about a million Alu inserts scattered throughout the human genome.

Y chromosome: The Y chromosome is the chromosome that makes a person a male and can be passed by a male only to his sons. It differs from all other chromosomes that the majority of the chromosome is unique and does not recombine during meiosis (see NRY or non-combining Y). This means the historical pattern of mutations can easily be studied.

Y-DNA: The DNA in the Y chromosome that can be passed by a male only to his sons. This DNA can be tested to determine both haplotype and haplogroup of the individual.

