Genetic Genealogy DNA Vocabulary List-October 2019

Base pairs (bp): a pair of nucleotides on opposite strands of DNA. A always pairs with T and C always pairs with G

centiMorgans (cM): a term used in genetic genealogy to refer to the size of matched segments of DNA in autosomal DNA testing. The larger the number of shared segments, the more likely there is a shared ancestor or the closer the relationship. First cousins share an average of 850 cMs while third cousins share an average of 53 cMs.

Chromosome: a package of DNA. Humans typically have 23 pairs of chromosomes numbered 1 to 22 along with a pair of sex chromosomes.

- **Autosomes**: the chromosomes (numbered 1-22) except for the sex chromosomes. One autosome of each of the 22 pairs is inherited from each parent.

- **Sex chromosomes**: X and Y chromosomes which are involved in sex determination of an individual.

    - Females typically have two X chromosomes (one from each parent)

    - Males typically have one X chromosome (from the mother) and one Y chromosome (from the father).

    - Y chromosomes are inherited by a male from his father only as a mother does not have a Y chromosome.

DNA: **d**eoxyribo**n**ucleic **a**cid. This substance contains the genetic information on how to form and maintain the organism.

- In humans, DNA is typically present in both the nucleus of a cell and referred to as **nuclear DNA**) and in the mitochondria (energy-producing bodies in the cell outside the nucleus) referred to as **mitochondrial DNA**.

Genetic distance: expressed as the number of differences in the DNA between two individuals who have undergone the same testing such as mtDNA testing or Y chromosome testing. The closer the relation between two people, the fewer number of differences.

Genetic genealogy is based on DNA testing of different types. These include:

- **Autosomal DNA testing** analyzes the genetic material present in both males and females that is inherited from the mother and the father.

- **Mitochondrial DNA testing** analyzes material present in both males and females that is inherited only from the mother and through the maternal line (mothers to daughters).

- **Y-DNA testing** analyzes material present in males that is inherited only from the father and through the paternal line (fathers to sons)

Haplogroup: a group of individuals who share several genetic markers and presumably share a common ancestor. Used with mtDNA and Y-DNA testing to help determine ancestral origins. For example, if mtDNA testing showed the A2w haplogroup, the tested individual would likely have Native American ancestry through the maternal line.

Mitochondrial DNA (mtDNA): DNA found in the mitochondria consisting of a single strand in a circle of 16,569 nucleotides. mtDNA is inherited solely from the mother of an individual as it is present in the egg cell, but not in the sperm cell.

MRCA or most recent common ancestor: the most recent ancestor (such as a great-great grandparent) that two individuals share.

Mutation: any change in the sequence of the nucleotides on the strand of DNA. Some changes can affect an individual's health or appearance, but most changes are harmless and do not cause an observable change in the person. Changes in DNA are useful in determining the relatedness of two individuals.

Nuclear DNA: consists of two long strands in a double helix structure consisting of millions of nucleotides that occur in a specific order.

Nucleotide: a building block of DNA which comes in four types known as adenine, guanine, cytosine and thymine and referred to in shorthand as A, G, C and T.

SNP or single nucleotide polymorphism: a change in the DNA sequence involving a single nucleotide. SNPs are the most common type of genetic variation in humans, typically with no effect on health or development. SNPs are inherited from parents and can be used as markers to help determine how closely related one individual is to another.

STR or short tandem repeat: a marker consisting of a short string of nucleotides that is repeated a variable number of times. The number of repeats at several locations on the chromosome is used in Y DNA testing to help determine relationships between two individuals.