Forensic DNA Testing Terminology

ABI 310 Genetic Analyzer – a capillary electrophoresis instrument used by forensic DNA laboratories to separate short tandem repeat (STR) loci on the basis of their size.
Adenine – a purine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter A.
Allele – one of two or more alternative forms of a gene.
Allele Frequency – the proportion of a particular allele among the chromosomes carried by individuals in a population.
ASCLD (ascld.org) – American Society of Crime Laboratory Directors; involved with accreditation of DNA testing labs.
Amino acid – Any of a class of 20 molecules that are combined to form proteins in living things. The sequence of amino acids in a protein and hence protein function are determined by the genetic code.
Amplification – An increase in the number of copies of a specific DNA fragment; can be in vivo or in vitro.
Autosome – A chromosome not involved in sex determination. The diploid human genome consists of 46 chromosomes, 22 pairs of autosomes, and one pair of sex chromosomes (the X and Y chromosomes).
Base pair – two complementary nucleotides in DNA; base pairing occurs between A and T and between G and C.
Base sequence – the order of nucleotide bases in a DNA molecule.
Base sequence analysis – a method, sometimes automated, for determining the base sequence.
Biotechnology – a set of biological techniques developed through basic research and now applied to research and product development.
Blind proficiency test – a proficiency test in which the laboratory personnel do not know that a test is being conducted.
Capillary electrophoresis – a method that utilizes a narrow polymer-filled tube to separate DNA molecules by size.
Ceiling principle – a conservative approach for estimating a DNA profile's frequency of occurrence in a population containing multiple ethnic groups.
Chromosome – a large piece of DNA. Humans have 23 different chromosomes in almost every type of cell.
CODIS – Combined DNA Index System, established in 1998 and containing the STR DNA profiles of many thousands of convicted offenders.
COfiler – PCR Amplification Kit (AmpFLSTR® COfiler™) that provide human identification laboratories with the ability to generate information for six STR loci and Amelogenin.

Complementary sequences – nucleic acid base sequences that form a double-stranded structure by matching base pairs; the complementary sequence to G-T-A-C is C-A-T-G.
Controls – tests performed in parallel with experimental samples and designed to demonstrate that a test was reliable.
Cytosine – a pyrimidine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter C.
Degradation – the chemical or physical breaking down of DNA.
Denaturation – the process of splitting the complementary double strands of DNA to form single strands.
DNA (Deoxyribonucleic acid) – the genetic material.
Diploid – having two sets of chromosomes, one from each parent (compare haploid).
DNA databank (database) – a collection of DNA typing profiles of selected or randomly chosen individuals.
DNA polymerase – an enzyme that catalyzes the synthesis of double stranded DNA.
DNA sequence – the relative order of base pairs, whether in a fragment of DNA, a gene, a chromosome, or an entire genome.
Double Helix – the shape that two linear strands of DNA assume when bonded together.
Dye blobs – a technical artifact associated with STR testing.
Electrophoresis – a technique in which different molecules are separated by their rate of movement in an electric field.
Enzyme – a protein that can speed up a specific chemical reaction without being changed or consumed in the process.
Gametic (phase) equilibrium – the state of loci on different chromosomes when the allele at one locus in the gamete varies independently of that at the other loci.
Gel – matrix (often agarose or acrylamide) used in electrophoresis to separate molecules.
Gene – the basic unit of heredity; a sequence of DNA nucleotides on a chromosome.
Gene frequency – the relative occurrence of a particular allele in a population.
Gene mapping – determination of the relative positions of genes on a DNA molecule (chromosome or plasmid) and of the distance, in linkage units or physical units, between them.
Genetics – the study of the patterns of inheritance of specific traits.
Genetic drift – random fluctuation in allele frequencies due to small population sizes (sampling error).
Genome – the sum total of an organism's genetic material.

Genome projects – Research and technology development efforts aimed at mapping and sequencing some or all of the genome of an organism.
Genophiler™ – an automated, objective system for reviewing and presenting DNA profiling data.
Genotype – the genetic makeup of an organism, as distinguished from its physical appearance or phenotype.
Guanine – a purine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter G.
Hardy-Weinberg equilibrium (HWE) –populations of organisms that are in HWE have no statistical correlations between any pairs of alleles within individuals in the population.
Heredity – the transmission of characteristics from one generation to the next.
Heterozygous – a heterozygous organism has two different alleles at a particular locus.
Homozygous – a homozygous organism has two copies of the same allele at a particular locus.
Identifiler – PCR Amplification Kit (AmpFLSTR® Identifiler™) that provides human identification laboratories with the ability to generate information on 15 STR loci and Amelogenin.
In vitro – outside a living organism.
Kilobase (kb) – unit of length for DNA fragments equal to 1000 nucleotides.
Kinship coefficient – the probability that two randomly chosen genes, one from each of two individuals in a population, are identical (i.e. both descended from the same ancestral gene, or one from the other).
Linkage – the association of alleles at two or more loci due either to their residing on a single chromosome or their abundance in a particular ethnic group that causes them to appear together at a higher than expected frequency.
Localize – determination of the original position (locus) of a gene or other marker on a chromosome.
Locus (pl. loci) – the physical location of a gene on a chromosome.
Marker – a gene of known location on a chromosome and phenotype that is used as a point of reference in the mapping of other loci.
Matrix failure (pull up) – a result of the inability of the detection instrument to properly resolve the dye colors used to label PCR amplification products. Often due to off-scale peaks.
Megabase (Mb) – unit of length for DNA fragments equal to one million nucleotides.
Mitochondrial DNA (mtDNA) – DNA found in the mitochondria inside cells (not associated with the nuclear chromosomes); transmission is only from mother to child.

Mitosis – the process of nuclear division in cells that produces daughter cells that are genetically identical to each other and to the parent.
Multiplexing – a sequencing approach that uses several pooled samples simultaneously, greatly increasing sequencing speed.
Mutation – any inheritable change in DNA sequence.
Nucleic acid – a nucleotide polymer that DNA and RNA are major types.
Nucleotide – chemical units that are strung together in long chains to make DNA molecules.
Nucleus – the cellular organelle in eukaryotes that contains the genetic material.
Oncogene – a gene, one or more forms of which is associated with cancer. Many oncogenes are involved, directly or indirectly, in controlling the rate of cell growth.
Physical map – a map of the locations of identifiable landmarks on DNA. Distance is measured in base pairs.
PCR (polymerase chain reaction) – an amplification process that yields millions of copies of desired DNA through repeated cycling of a reaction involving the enzyme DNA polymerase.
Peak height imbalance – a significant difference (usually 30% or more) in the amount of signal obtained for two alleles from a single STR locus that might be suggestive of more than one contributor to a sample.
Polymorphic – a locus is polymorphic if a population contains two or more detectable alleles.
Polymorphism – difference in DNA sequence among individuals. Genetic variations occurring in more than 1% of a population would be considered useful polymorphisms for linkage analysis.
Population – a group of individuals residing in a given area at a given time.
Primer – short preexisting polynucleotide chain to which new deoxyribonucleotides can be added by DNA polymerase.
Probe – single-stranded DNA or RNA of a specific base sequence, labeled either radioactively or immunology, that are used to detect the complementary base sequence by hybridization.
Proficiency tests – tests to evaluate the performance of technicians and laboratories; in open tests, the technicians are aware that they are being tested, but in blind tests, they are not.
Profiler Plus – PCR Amplification Kit (The AmpFLSTR® Profiler Plus™) that provides human identification laboratories with the ability to generate information for nine polymorphic STR loci and the Amelogenin locus.
Protein – a large molecule composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nuceotides in the gene coding for the protein. Proteins are required for the structure, function, and regulation of the body cells, tissues, organs, and each protein has unique functions.

Random match probability – the chance of a random match; as used in DNA profiling, it is the probability that the DNA of a randomly chosen person has a DNA profile that cannot be distinguished from that observed in an evidence sample.
Recombinant DNA technologies – procedures used to join together DNA sequences in a cell-free system. Under appropriate conditions, a recombinant DNA molecule can enter a cell and replicate there, either autonomously or after it has become integrated into a cellular chromosome.
Resolution – degree of molecular detail on a physical map of DNA.
Restriction enzyme – a protein that recognizes specific, short nucleotide sequences and cuts DNA at those sites. Bacteria contain over 400 such enzymes that recognize and cut over 100 DNA sequences.
Restriction fragment length polymorphism (RFLP) – variation between individuals in DNA fragment sizes cut by specific restriction enzymes; polymorphic sequences that result in RFLPs that are used as markers on both physical maps and genetic linkage maps. RFLPs are usually caused by mutation at a cutting site.
RFU (relative fluorescent units) – units of measure for the light intensity detected by a fluorescence detector, correlated with the amount of DNA associated with a particular STR allele.
Serology – a discipline that uses immunology to study body fluids.
Sequencing – determination of the order of nucleotides (base sequences) in a DNA or RNA molecule or the order of amino acids in a protein.
Sex chromosomes (X and Y chromosomes) – chromosomes that are involved in sex determination. In humans, XX corresponds to female and XY to males. In STR testing, typed at the amelogenin locus.
STR (short tandem repeats) – in DNA testing, a subset of polymorphic VNTR loci where alleles differ primarily in the number of times that a string of four nucleotides are tandemly repeated.
Southern blotting – transfer by absorption of DNA fragments separated in electrophoretic gels to membrane filters for detection of specific base sequences by raiolabeled complementary probes.
Stutter – PCR amplification products that are one or more repeat units less (or more) in size than a sample's true allele and arise during PCR because of strand slippage. Typically 15% or less of the height of the true allele.
Tandem repeat sequences – multiple copies of the same base sequence on a chromosome; used as a marker in physical mapping.
Thymine – a pyrimidine base; one of the four molecules containing nitrogen present in the nucleic acids DNA and RNA; designated by letter T.

Taq polymerase – a DNA polymerase (an enzyme) used to amplify a specific DNA template in the PCR technique.
VNTR (variable number of tandem repeats) – in DNA testing, a polymorphic locus where alleles differ primarily in the number of times that a string of nucleotides are tandemly repeated. Widely used throughout the 1990's but largely replaced by PCR-based STR testing today.