

DNA GLOSSARY

Term	Description
Allele	One of two or more possible forms of the DNA sequence at a particular locus. Each individual has only two alleles per autosomal STR locus, one inherited from their father and one from their mother, but more than two alleles may exist within the broader population. Males have only one allele per Y-STR locus, inherited from their father, but more than one allele may exist within the broader population.
Amelogenin	A locus on each of the X and Y sex chromosomes, used in forensic DNA analysis to determine gender.
Amplification	The copying and tagging, by PCR, of alleles at specific loci such that they may be detected and discerned in subsequent stages of DNA analysis.
Autosomal	Term used to describe 22 of the 23 pairs of human chromosomes, on which are located many of the test sites employed in forensic DNA analysis. The PowerPlex® 16 HS typing system, for example, produces STR test results from 15 autosomal loci (as well as one on the sex chromosomes which indicates gender).
Cannot Be Excluded	The term used to describe an individual who, upon comparison of his/her DNA profile from a reference sample to a DNA profile from an unknown sample, cannot be ruled out as the contributor of the unknown sample.
Capillary Electrophoresis	A method whereby copies of tagged DNA sequences generated during PCR are sorted and discerned in order to ascertain the DNA profile associated with a sample.
Chromosome	An organized structure of DNA and proteins found in the cell nucleus. Humans have 23 pairs of chromosomes (22 autosomal and 1 gender determining) – one of each pair is contributed by one's father while the other is contributed by one's mother.
Conventional Extraction	A procedure that utilizes chemicals to remove cells from their substrate (e.g. fabric) and to remove and purify DNA from cells so that it may be further processed.
Differential Extraction	An extraction procedure designed to separately collect DNA from epithelial cells and sperm cells, if present. A differential extraction may be used, as a precaution, on samples where sperm have not otherwise been confirmed. The fact that such a procedure has been employed is not necessarily indicative of the presence of sperm cells in a sample.
DNA	Deoxyribonucleic acid. Genetic instructions for living organisms.
DNA Profile	The combination of DNA test results at each of the loci tested for any given sample. A DNA profile may be from a single source or from a mixture of sources. Single source profiles are also referred to as genotypes, in the case of autosomal analyses, or as haplotypes, in the case of Y-STR analyses.

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Epithelial Cells	Generally, cells other than sperm. In a differential extraction, much (though not necessarily all) of the DNA from these cells is expected to partition into the so-called 'epithelial' fraction. Some may also partition into the so-called 'sperm' fraction.
Excluded	The term used to describe an individual who, upon comparison of his/her DNA profile from a reference sample to a DNA profile from an unknown sample, is ruled out as the contributor of the unknown sample.
Genotype	See DNA profile.
Haplotype	See DNA profile.
Inconclusive	The term used to describe the outcome, or potential outcome, of a comparison between an unknown profile and the profile of a person of interest, where an inference as to whether the person of interest is or is not excluded cannot be drawn due to uncertainty with respect to the reliability and validity of the unknown profile.
Likelihood Ratio	A ratio of two probabilities. The probability of having observed test results under one hypothesis divided by the probability of having observed the same results under a different hypothesis.
Locus	A physical location on a chromosome, or test site (plural loci).
Match	DNA test results from two profiles that, upon comparison, are indistinguishable.
Maternity Test	A test to determine whether a particular female can be excluded as the biological mother of a child. DNA analysis may be used to assess other potential relationships as well.
Major Profile	A DNA profile, in a sample containing a mixture of DNA, which is present in a greater quantity than the other profile(s).
Minor Profile	A DNA profile, in a sample containing a mixture of DNA, which is present in a lesser quantity than one or more other profiles.
Mixture	A sample containing the DNA of two or more people.
Paternity Test	A test to determine whether a particular male can be excluded as the biological father of a child. DNA analysis may be used to assess other potential relationships as well.
PCR	Polymerase Chain Reaction. An <i>in vitro</i> process that produces copies of DNA sequences at specific loci and tags them to allow for their ready detection in subsequent DNA analysis processes.
Phenotype	The physical features of a person, as determined by their DNA, for example, eye colour or hair colour.

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Plexor® HY System	A system used to estimate both the total quantity of human DNA in a sample as well as the quantity of male DNA in the same sample.
Population Database	Collection of DNA profiles from individuals belonging to a specific population group. For the PowerPlex® 16 HS system, individual allele frequencies within databases are used in the calculation of random match probabilities and likelihood ratios. For the PowerPlex® Y23 system, Y-STR profile frequencies within databases are used in these calculations.
PowerPlex® 16 HS System	 A DNA profiling system which allows for the amplification, by PCR, of 15 autosomal STR loci in a single test reaction: D3S1358, THO1, D21S11, D18S51, Penta E D5S818, D13S317, D7S820, D16S539, CSF1PO, Penta D Amelogenin, vWA, D8S1179, TPOX, FGA
PowerPlex® 23Y System	A DNA profiling system which allows for the amplification, by PCR, of 23 STR loci from the male-specific Y chromosome in a single test reaction: DYS576, DYS389I, DYS448, DYS389II, DYS19 DYS391, DYS481, DYS549, DYS533, DYS438, DYS437 DYS570, DYS635, DYS390, DYS439, DYS392, DYS643 DYS393, DYS458, DYS385 (a and b), DYS456, YGATAH4
PrepFiler™ Forensic DNA Extraction Kit	A commercial kit containing the necessary components and chemicals for the extraction of forensic DNA samples.
Probability of Paternity/Maternity	A term used, often in addition to a likelihood ratio in relationship testing cases, to express the probability of paternity/maternity. This probability (expressed as a percentage) assumes equal prior odds (all of the information outside of the DNA testing results) of paternity/maternity. Similar probabilities may also be included when relationships other than paternity/maternity are assessed.
QPCR	Quantitative PCR. The Plexor® HY system is an example of a QPCR system. An estimate of the quantity of DNA present in a sample is made by monitoring PCR products in real-time.
SNP	Single nucleotide polymorphism. A DNA sequence variation in which a single nucleotide (A, T, G or C) in the genome differs. SNPs are the most common genetic variations among humans.
Sperm	The male reproductive cells found in semen. In a differential extraction, much (though not necessarily all) of the DNA from these cells is expected to partition into the so-called 'sperm' fraction. Some may also partition into the so-called 'epithelial' fraction.
STR	Short Tandem Repeat. Repeating sequences of DNA arranged in succession at a given locus.
Y-STR	STRs on the male-specific Y-chromosome.

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