DNA: Glossary

Allele	Any one of the alternative variant forms at a particular genetic locus.
Allelic ladder	An artificial mixture of the common alleles present in the human population for a particular STR marker.
Amplification	Producing multiple copies of a chosen DNA region by PCR (Polymerase Chain Reaction).
Artifact	Any non-allelic product of the PCR amplification process (i.e. stutter, minus "A") or anomaly of the detection process (i.e. pull-up or spike).
Candidate match	A possible match between two or more DNA profiles discovered by CODIS software.
Capillary electrophoresis	The platform for CE uses narrow silica capillaries containing a polymer solution through which the negatively charged DNA molecule migrates under the influence of a high voltage electric field.
CODIS	The Combined DNA Index System administered by the FBI. CODIS links DNA evidence obtained from crime scenes, thereby identifying serial criminals. CODIS also compares crime scene evidence to DNA profiles from offenders, thereby providing investigators with the identity of the putative perpetrator. In addition, CODIS contains profiles from missing persons, unidentified human remains, and relatives of missing persons. There are three levels of CODIS: the *Local DNA Index System (LDIS*), the *State DNA Index System (SDIS*), and the *National DNA Index System (NDIS),* managed by the FBI as the nation's DNA database containing all DNA profiles uploaded by participating states.
Composite profile	A DNA profile generated by combining typing results from different loci obtained from multiple injections of the same amplified sample and/or multiple amplifications of the same DNA extract.

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DNA: Glossary, Continued

Contamination	Contamination is the unintentional introduction of DNA into a sample or PCR product.
Criminal paternity profile	A DNA profile comprised of the obligate paternal alleles deduced from the child and/or product of conception.
Critical reagents	Critical reagents are determined by empirical studies or routine practice and require testing on established samples before use on casework samples.
Deduced profile	The DNA profile determined for an unknown contributor(s) which includes at least one locus with multiple reasonable genotypes.
Degradation	The reduction of a chemical compound to one less complex. The fragmentation of an intact DNA molecule can occur through both enzymatic and chemical processes such as cellular nucleases and hydrolytic cleavage of the base-sugar bond. Environmental conditions such as heat, humidity, and UV exposure can also cause the DNA molecule to fragment.
Detection threshold	The detection threshold, determined by internal validation studies, defines the minimum peak height where a true allelic peak can confidently be distinguished from baseline noise or artifacts. Peaks that are above the threshold may be designated as an allele. Also known as the *Analytical threshold.*
Developmental validation	The acquisition of test data and determination of conditions and limitations of a new or novel DNA methodology for use on forensic samples.
Differential degradation	A DNA typing result in which contributors to a mixture are subject to different levels of degradation thereby impacting the mixture ratios across the entire profile.

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DNA: Glossary, Continued

Differential extraction	A procedure in which sperm cells are separated, or extracted, from all other cells in a sample.
DNA advisory board (DAB)	A panel formed as a result of the passage of the DNA Identification Act of 1994. The FBI incorporated their recommendations for forensic DNA testing into the *National DNA Quality Assurance Standards*. DAB was later replaced by SWGDAM.
DNA profile	The genetic constitution of an individual at defined locations (also known as loci) in the DNA. A DNA profile derived from nuclear DNA typically consists of one or two alleles at several loci (e.g. short tandem repeat loci).
Genetic analyzer	An instrument that analyzes PCR products from the amplification process using capillary electrophoresis. Alleles are separated according to size and detected using fluorescent dye labeling.
Heterozygous	Of or pertaining to an *individual* possessing two different *alleles* at a particular genetic locus.
Homozygous	Of or pertaining to an *individual* possessing the same *alleles* at a particular genetic locus.
Inhibitor	Substances which may co-extract with DNA in a sample and can interfere with or prevent the DNA amplification process from occurring properly. The presence of an inhibitor can lead to complete PCR amplification failure or a reduced sensitivity of detection usually at the larger PCR loci by: • interfering with the cell lysis necessary for DNA extraction • causing nucleic acid degradation • inhibiting polymerase activity preventing amplification of target DNA

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Internal validation	The accumulation of test data within the laboratory to demonstrate that established methods and procedures perform as expected in the laboratory. Prior to using a procedure for forensic applications, a laboratory shall conduct internal validations.
Material modification	An alteration of an analytical procedure that may have a consequential effect(s) on analytical results.
Methodology	The processes and procedures used to support a DNA-typing technology. For example, extraction methods (manual v. automated), quantitation methods (shot blot, fluorometry, real-time), typing test kit, and platform (capillary electrophoresis, real-time gel, and end-point gel instruments or systems).
Negative PCR control	This control consists of only PCR amplification reagents without the addition of template DNA and is used to detect contamination of amplification reagents.
Obligate allele	An allele at a locus that an individual must have in order to be included. In paternity testing, the obligate allele at a locus in a child must be present in the profile of the alleged parent in order to be included as a parent of the child. Obligate alleles can be identified by comparing the profile of the child to the profile of a known parent to deduce the contribution of the unknown parent. Due to the possibility of mutation, a maximum of two allele mismatches between the child and the alleged parent will be reported as a paternity inclusion.
Offender	Within CODIS, the term “offender” is intended to include arrestees, convicted offenders, detainees, and Legal Index specimen categories.
Off-scale data	When too much template DNA is added to the PCR reaction, the PCR products may exceed the linear dynamic range for detection by the instrument.

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DNA: Glossary, Continued

Organic extraction	The phenol-chloroform extraction method removes proteins and other cellular components from nucleic acids, resulting in relatively purified DNA preparations. This method results in double-stranded DNA that is suitable for amplification.
Platform	The type of analytical system utilized to generate DNA profiles, such as capillary electrophoresis, real-time gel, and end-point gel instruments or systems.
Polymerase chain reaction (PCR)	An enzymatic process by which a specific region of DNA is replicated during repetitive cycles which consist of the following: • denaturation of the template • annealing of primers to complimentary sequences at an empirically determined temperature • extension of the bound primers by a DNA polymerase
Positive PCR control	This control consists of the PCR amplification reagents and a known DNA sample and is used to determine if the PCR process performed properly.
Quality Control (QC) stain control	The QC stain consists of blood of a known type and acts as an internal positive control since the analyst performing the analysis does not know the DNA type. A QC stain control will be extracted at the same time as the reference samples. The QC stain control ensures that the extraction procedure worked properly.
Quantitation	A method used to determine the quantity of DNA in a sample, usually reported as ng/µl.
Reagent blank	The reagent blank sample contains no template DNA and is used to monitor contamination from DNA extraction to automated analysis. This blank is treated the same as, and parallel to, the forensic casework samples being analyzed.

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Reference sample	A material of a verifiable/documented source which, when compared with evidence of an unknown source, can show an association between an offender, crime scene, and/or victim.
SWGDAM	Scientific Working Group on DNA Analysis methods (SWGDAM) is a panel, replacing the DAB, which meets under the guidance of the Federal Bureau of Investigation (FBI). It is the body that proposes and recommends revisions to the *National Quality Assurance Standards (QAS)*. Formerly called TWGDAM (Technical Working Group on DNA Analysis and Methods).
Stochastic effects	Stochastic effects are a result of an unequal sampling of the two alleles present from a heterozygous individual when only a few DNA molecules are used to initiate PCR. Stochastic effects can be observed as peak height imbalances or allele dropout.
Stochastic threshold	The minimum allele peak height in RFUs, determined by internal validation studies, above which it is reasonable to assume that, at a given locus, allelic dropout of a sister allele has not occurred.
Short tandem repeat (STR) typing	DNA analysis method which targets regions on the chromosome which contain multiple copies of an identical DNA sequence occurring in succession.
Stutter	In addition to an allele's primary peak, artifactual minor "stutter" peaks can occur at four-base intervals. In tetranucleotide repeats, the most common stutter peaks are four bases smaller than the primary peak (n-4). It is also possible to see additional "n+4" peaks (four bases larger), especially when excessive amounts of DNA are amplified. These stutter peaks may be due to repeat slippage during amplification.
Substrate	Any background material upon which a biological sample has been deposited.

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DNA: Glossary, Continued

Target DNA profile	A DNA profile submitted by an NDIS participating laboratory for the purpose of searching DNA profiles maintained by NDIS which could match an indexed DNA profile.
Technical lead	An employee who is accountable for the technical operations of the DNA laboratory and who is authorized to stop or suspend laboratory operations.
Technology	A term used to describe the type of forensic DNA analysis performed in the laboratory, such as RFLP, STR, YSTR, or mitochondrial DNA.
Thermal cycler	An instrument used to amplify targeted segments of DNA via the Polymerase Chain Reaction (PCR) process.
Threshold value	A Relative Fluorescent Unit (RFU) value that must be exceeded to reliably make an interpretation. The threshold value is used to distinguish between background noise and true allelic peaks and is established within each laboratory through a validation process.
Y-STR	Short Tandem Repeat loci located on the Y chromosome.