Glossary of Forensic DNA Terms

Definitions are from Butler, J.M. (2010) *Fundamentals of Forensic DNA Typing*. San Diego: Elsevier Academic Press and other sources.

3' ("three prime"): refers to the end of a DNA molecule bearing a free hydroxyl group on the 3' carbon of the sugar ring

5' ("five prime"): refers to the end of a DNA molecule bearing a free hydroxyl group on the 5' carbon of the sugar ring; a DNA sequence is typically read from the 5' to 3' end

9947A: a cell line derived from a human female; DNA from this cell line is used as a positive control in some commercial DNA test kits

ABI: Applied Biosystems Inc.; an instrument and reagent manufacturing company based in South San Francisco, California that is now called ThermoFisher Scientific

ABI Genetic Analyzer: a capillary electrophoresis instrument sold by Applied Biosystems and widely used throughout the forensic DNA community since its introduction in 1995; an ABI 310 uses a single capillary filled with a viscous polymer solution to separate DNA molecules based on their relative size; laboratories often use a multi-capillary instrument such as the ABI 3130 (4-capillaries), ABI 3130*xl* (16-capillaries), ABI 3500 (8-capillaries), or ABI 3500*xl* (24-capillaries) Genetic Analyzer

Accreditation: the formal process by which a laboratory is evaluated, with respect to established criteria, for its competence to perform a specified kind of measurement; the evaluation takes place through an audit by an outside, qualified agency and helps provides confidence that the laboratory meets minimum professional standards for general operations

Accuracy: the degree of agreement or conformity of a measured value with its actual (true) value

Adventitious DNA: contaminating DNA that unintentionally becomes amplified along with the intentional template DNA obtained from evidence or reference samples

Adventitious match: an association of an evidence DNA profile to the profile of a person who is not the true donor of that profile; this situation may arise when the DNA profile contains information from a limited number of loci (e.g., from a damaged DNA sample) that are insufficient to distinguish the profiles of two different individuals

Allele: one of two or more versions of a genetic sequence at a particular location (a locus) in the genome; typically, multiple alleles are possible with STR markers

Allele drop-in: allelic peak(s) in an electropherogram that are not reproducible across multiple independent amplification events; also a hypothesis/postulate for the observation of one or more allelic peaks in an electropherogram that are inconsistent with the assumed/known contributor(s) to a sample

Allele drop-out: failure of an otherwise amplifiable allele to produce a signal above the analytical threshold because the allele was not present or was not present in sufficient quantity in the aliquot that underwent PCR amplification

Allele frequency: the number of times that an allele appears in a data set is the absolute frequency; the proportion of a particular allele among the chromosomes carried by individuals in a population

Allelic ladder: in STR testing, a measurement calibration tool, consisting of the most commonly observed alleles, used for assigning an allele designation to a peak in an electropherogram at a particular genetic locus

Amelogenin: a commonly used sex-typing genetic marker because the gene, which codes for tooth enamel, occurs on both the X and Y chromosomes; the X-chromosome region amplified in commercial STR kits is 6 bases shorter than the Y-chromosome region such that female (X,X) samples result in a single peak while males (X.Y) have two peaks

Amplification: an increase in the number of copies of a specific DNA fragment; can be *in vivo* or *in vitro*; in forensic DNA testing laboratories, this refers to the use of the PCR technique to produce many more copies of DNA (typically alleles) at specific genetic loci

Amplicon: the product of polymerase chain reaction DNA amplification

Analyst: an individual trained and qualified to report DNA results

Analytical threshold: a RFU level determined to be appropriate for use in the PCR/STR DNA typing process; a minimum threshold for data comparison that is identified in each laboratory through validation studies

Artifact: any non-allelic product of the amplification process (e.g., stutter or non-template addition), anomaly of the detection process (e.g., pull-up or spike), or a by-product of primer synthesis (e.g., dye blob) that may be observed in an electropherogram; may complicate interpretation of a DNA profile when they cannot be distinguished from actual allele(s) data

Audit: evaluation of a laboratory based on a set of criteria established by an outside, qualified agency usually for establishing accreditation

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 (bp): two complementary nucleotides joined by hydrogen bonds; base pairing occurs between A and T and between G and C

Base sequence: the order of nucleotide bases in a DNA molecule; typically read from the 5'-end to the 3'-end

Bayesian approach: defines the probability of an event as the degree of belief in the truth of a proposition that asserts it will happen

Bin: a range of sizes established empirically that is used to establish allele designation for length-based differences in STR alleles

Buccal swab: a relatively non-invasive technique of scrapping the inside of a mouth with a cotton swab or similar collection device to collect cells from the inner cheek lining; a common method for collecting and preserving samples for DNA testing from known individuals

Capillary electrophoresis (CE): an electrophoretic technique for separating DNA molecules by their size based on migration through a narrow glass capillary tube filled with a liquid polymer

Cell: the basic building block of an organism; humans have approximately 100 trillion cells in their body, most containing DNA

Chelex extraction: a method of removing DNA from cells involving Chelex resin; since one step of the method involves boiling, the extracted DNA is single-stranded

Chromosome: the structure by which hereditary information is physically transmitted from one generation to the next

CODIS: Combined DNA Index System, which under the direction of the FBI Laboratory, is the software architecture that runs the U.S. national DNA database

CODIS loci: an established set of STR loci required for inclusion of a DNA profile at the national level in CODIS; originally the core set involved 13 STRs but after January 1, 2017, 20 core STRs will be required for entry at the national level

Cold hit: an association made between a crime scene DNA profile and a DNA profile found on a DNA database in the absence of any prior investigative leads; this association may be to another crime scene DNA profile or to a profile from a known individual

Combined Probability of Inclusion (CPI): the product of the probabilities of inclusion calculated for each locus; the probability of inclusion at each locus estimates the probability that a randomly selected, unrelated individual is not excluded from being a source of the DNA mixture profile and is calculated as the square of the sum of the relative frequencies of the observed alleles at the locus; sometimes referred to as Random Man Not Excluded (RMNE)

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

Complete profile: a full DNA result with values being obtained from all attempted loci

Complex mixture: DNA profile resulting from a sample containing three or more contributors often having only a small amount of DNA from at least one of the contributors

Concordance: obtaining the same value when testing multiple times

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Conservative: an assignment of the weight of evidence that is believed to favor the defense

Consensus profile: the resulting DNA profile made up from the list of alleles that are repeated or reproduced a specified number of times when a DNA extract is divided into several replicate samples that are amplified concurrently

Contamination: exogenous DNA or other biological material in a DNA sample, PCR reaction, or item of evidence; the exogenous DNA or biological material could be present before the sample is collected or introduced during collection or testing of the sample

Controls: samples of known types, run in parallel with experimental, reference, or evidence samples, that are used to demonstrate that a procedure is working correctly

Critical reagents: chemicals or other materials used in testing whose performance is vital to the success of the test as determined by empirical studies or routine practice

Deconvolution: separation of component DNA genotypes of contributors to a mixed DNA profile based on quantitative peak height information and any underlying assumptions (e.g., the number of contributors to the mixture, mixture ratios, or known contributors)

Deduced profile: inference of an unknown contributor's DNA profile after taking into consideration the contribution of a known/assumed contributor's DNA profile based on quantitative peak height information in a mixed DNA profile

Degradation: the fragmenting, or breakdown, of DNA by chemical, physical, or biological means; a common occurrence when biological samples containing DNA encounter warm moist environments or excessive UV light

Deoxyribonucleic acid (DNA): a genetic material of organisms, usually double-stranded; a biopolymer composed of nucleic acids, identified by the presence of deoxyribose, a sugar, and the four nucleobases; DNA is a fairly stable molecule and variations in DNA sequence between individuals permits DNA testing to distinguish individuals from one another

Detection limit: the smallest amount of some component of interest that can be measured by a single measurement with a stated level of confidence

Differential extraction: a DNA extraction procedure where the sperm cells are physically separated from the DNA of other cells before the sperm DNA is isolated; generally results in a sperm and non-sperm (epithelial) fraction

Diploid: having two copies of each autosome; the normal constitution of most mammalian somatic cells

Direct PCR: the process of amplifying the DNA present in biological samples without prior extraction or purification

DNA databank: a repository of stored bloodstain cards or DNA samples used to generate DNA profiles

DNA database: a computer repository of DNA profiles

DNA profile: a string of values (numbers or letters) compiled from the results of DNA testing at one or more genetic markers; can be single-source or a mixture from multiple contributors

DNA profiling or typing: any methodology for generating data from a biological sample at one or more DNA loci; DNA test results can be compared to results obtained from other samples

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 native form of DNA, which looks like a twisted ladder; two linear strands of DNA assume this shape when held together by complementary base pairing (ladder rungs)

Double-stranded DNA: form of DNA in which the individual strands are held together by complementary base pairing

Dye blobs: artifact peaks in capillary electropherograms arising from fluorescent dye molecules coming off their associated PCR primer during primer synthesis (and failing to be purified); generally, dye blobs are lower in signal intensity, and can be characteristically sized in each color channel

Electropherogram: graphic representation of the separation of molecules by electrophoresis in which data appear as "peaks" along a line; the format in which DNA typing results are presented with the X-axis displaying the observed alleles in order of increasing size and the Y-axis recording the relative amount of DNA detected based on the fluorescent signal collected during analysis

Electrophoresis: a technique in which molecules are separated by their velocity in an electric field

Elimination database: collection of DNA profiles held in a searchable format whose access/role/activities are deemed to be a potential DNA contamination risk; DNA profiles from an elimination database may be used to identify instances of inadvertent contamination

Elimination sample: a sample collected from an individual who had lawful access to the crime scene (e.g., the spouse of a rape victim, police investigators, laboratory staff); results from this sample may be helpful in deconvoluting a DNA mixture

Enhanced DNA detection methods: those steps or methods employed during or subsequent to the PCR amplification step that increases the sensitivity of the standard method and are typically employed with low-quantity and/or low-quality samples; may include increased number of PCR amplification cycles, increased CE injection time or voltage, or post-amplification desalting or concentration

Epithelial cells: skin cells, vaginal cells, or other cells that are normally found on an inner or outer body surface

Eukaryote: an organism with cells containing a nucleus

Evidence sample: biological sample collected from a crime scene or people or objects associated with a crime scene; sometimes referred to as the "Q" or question sample

Exclusion: a conclusion statement that a biological sample did not originate from a particular source or individual

Exon: segment of a gene that is represented in the final mRNA product

Expert system: a software program or set of software programs that interprets the data generated from a DNA analysis instrument platform in accordance with laboratory-defined quality assurance rules and accurately identifies the data that does and does not satisfy such rules

Fluorescence: the emission of light from a molecule following its excitation by light energy; in the context of STR DNA analysis, different fluorescent dyes permit simultaneous detection of similar size PCR products through fluorescence emission in different wavelengths (colors); the relative amount of light emitted is recorded on the vertical axis of an electropherogram in relative fluorescence units, RFUs

Forensic science: the application of scientific knowledge to questions of civil and criminal law, typically through presentation of results from evidence in court

Frequentist approach: defines the probability of an event as the frequency with which the event occurs in the long-run of a sequence of identical trials where the event may or may not occur

F_{ST}: the between-person inbreeding coefficient used in subpopulation corrections; commonly equated with the theta (θ) correction

Gene: the basic unit of heredity; a sequence of DNA nucleotides on a chromosome passed from parents to offspring that specifies traits

Genetics: branch of biology that deals with the heredity and variation of organisms

Genome: the entire DNA sequence found in a cell; the human genome consists of approximately 3.1 billion base pairs of DNA sequence

Genotype: the genetic makeup of an organism, as characterized by its physical appearance or phenotype; with STR DNA testing, a lcous genotype generally consists of two alleles, inherited from one's mother and father

GlobalFiler: a multiplex STR typing kit from Applied Biosystems that co-amplifies 21 autosomal STRs, DYS391, a Y indel, and the sex-typing marker amelogenin

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Guidelines: a set of general principles used to provide direction and parameters for decision making

Heredity: transmission of characteristics from one generation to the next

Heterozygosity: the presence of different alleles at one or more loci on homologous chromosomes; a measure of the diversity of a locus in a population

Heterozygote: an individual having different alleles at a particular genetic locus

Homologous chromosomes: a pair of chromosomes containing the same linear gene sequences, each derived from one parent

Homozygote: an individual having the same (or indistinguishable) alleles at a particular locus due to the inheritance of the same allele from each parent

Identifiler: a multiplex STR typing kit from Applied Biosystems that co-amplifies 15 STRs and the sex-typing marker amelogenin; the STR loci amplified are D8S1179, D21S11, D7S820, CSF1PO, D3S1358, TH01, D13S317, D16S539, D2S1338, D19S433, VWA, TPOX, D18S51, D5S818, and FGA

Inclusion: a conclusion statement that a biological sample may have originated from a particular source or individual

Inconclusive: data are inadequate to draw any meaningful conclusions; a statement provided as the conclusion when testing results are insufficient or lacking in quality and/or quantity, as defined by the laboratory, for comparison purposes

Indistinguishable mixture: a DNA mixture in which relative peak height ratios/areas as determined from the electropherogram are insufficient to attribute alleles to individual contributor(s); generally observed when a similar amount of DNA from each contributor is present in the original extracted sample such that no clear major or minor contributor can be discerned

Inheritance: the reception of genetic qualities by transmission from parent to offspring or all of the genetic characters or qualities transmitted from parent to offspring

Inhibitor: as related to PCR, any substance that interferes with or prevents the synthesis of DNA during the amplification process

Internal size standard (ISS): specific DNA fragments of known length that are used to size other DNA fragments in a sample being measured; in STR testing, an ISS is commonly labeled with a different fluorescent dye not found in the amplification process enabling mixing of the ISS and amplicon without interference in the analysis; sometimes referred to as an internal lane standard (ILS)

Intimate sample: a biological sample from an evidence item that is obtained directly from an individual's body such as a vaginal swab

Intron: non-coding DNA, which separates neighboring exons in a gene; intron sequences are removed during transcription (the creation of mRNA) so that only exons contribute to gene expression

In vitro: Outside a living organism; literally "in glass" meaning biochemical reactions conducted in a test tube or other laboratory apparatus

In vivo: within the cell or organism

ISFG: International Society for Forensic Genetics; an organization of scientists from more than 60 countries that meet bi-annually; conference proceedings have been entitled "Advances in Forensic Haemogenetics", "Progress in Forensic Genetics", and "Forensic Science International: Genetics Supplement Series".

Kinship analysis: comparison of genetic profiles of two or more individuals to evaluate alternative degrees of relatedness; performing DNA evaluations using biological relatives to predict expected genotypes in missing individuals in order to serve as an indirect form of human identification when no direct reference samples are available

Known sample: biological material for which the identity of the donor is established and used for comparison purposes; sometimes referred to as the "K" sample

Length polymorphism: variation observed in the length of alleles (e.g., STR loci) between individuals in a population

Likelihood ratio: the probability of the evidence under one proposition divided by the probability of the evidence under an alternative, mutually exclusive proposition; the magnitude of its value expresses the weight of the evidence

Linkage: the non-independent transmission of two genetic units, usually because of proximity on the same chromosome

Linkage disequilibrium: the non-random association, in a population, of alleles at different loci

Loci: plural of locus; pronounced /LOW-sigh/

Locus: a unique physical location of a gene (or a specific sequence of DNA) on a chromosome

Low-copy-number (LCN) DNA testing: the analysis of a small quantity of DNA often conducted by increasing the number of PCR amplification cycles

Low-level or low-template DNA: usually defined as less than approximately 100 picograms (pg) or about 15 human diploid cells

Major contributor: the source of the predominant portion of the DNA in a mixed sample that generates a mixture profile

Marker: a gene or specific DNA sequence of known location on a chromosome; used as a point of reference in the mapping of other loci

Massively parallel sequencing: one of a number of high-throughput DNA sequencing techniques; another name given to next-generation sequencing

Match: genetic profiles show the same types at all loci tested and no unexplainable differences exist

Match probability: uses conditional probabilities to address the question "given that a particular DNA profile has been seen in the crime scene evidence and in the suspect, what is the chance of it occurring again?"

Minor contributor: source of the lesser portion of the DNA in a mixed sample that generates a mixture profile

Mitochondrial DNA (mtDNA): a small, circular DNA molecule located in the mitochondria that contains approximately 16,500 nucleotides; the abundance of hundreds of copies of mtDNA in each cell make it useful with samples originating from limited or damaged biological material

Mixed DNA sample: any biological sample containing DNA from more than one individual

Mixture: DNA typing results originating from two or more individuals

Mixture ratio: relative ratio of the DNA contributions of multiple individuals to a mixed DNA typing result as determined by the use of quantitative peak height information; can be used to discern major and minor contributors to a DNA profile

Multiplex PCR: co-amplification of multiple regions of a genome with more than one set of primers; enables information from the different target sequences to be collected simultaneously

Mutation: any inheritable change in DNA sequence; an alteration or change of an allele at a genetic locus resulting in genetic inconsistency between a biological parent and offspring

Mutation rate: the relative frequency at which mutations have been observed at a specific genetic locus; generally estimated as the number of mutations observed in parent-offspring pairs divided by the total number of pairs examined

Nanogram (ng): 10^{-9} grams (a billionth of a gram); there are approximately 15 human cells in 1 ng of DNA

Negative control: a sample containing only PCR amplification reagents without the addition of template DNA; can be used to detect contamination introduced into the assay during the testing process via reagents, disposables, or handling errors

Next-generation sequencing: non-Sanger-based high-throughput DNA sequencing technologies where millions or billions of DNA strands can be sequenced in parallel; also called massively parallel sequencing

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Non-sperm cell fraction: the portion of a sample produced during differential extraction containing DNA from non-sperm cells; also referred to as the epithelial cell or female fraction

Non-template nucleotide addition: the addition of an extra nucleotide not specified by the template to the 3'-end of a PCR product resulting in a fragment that is one base longer than the actual target sequence

NRC I: the first National Research Council report issued in 1992 entitled "DNA Technology in Forensic Science"

NRC II: the second National Research Council report issued in 1996 entitled "The Evaluation of Forensic DNA Evidence"

Nucleotide: a unit of nucleic acid composed of phosphate, ribose or deoxyribose, and a purine or pyrimidine base

Nuclear DNA: DNA contained within a nucleus of eukaryotic organisms

Nucleus: the cellular organelle in eukaryotes that contains the genetic material

Null allele: an allele that exists in an individual but is not detected during testing usually due to a mutation in the DNA template that prevents a PCR primer from binding properly; can result in a true heterozygote being called a homozygote

Obligate allele: an allele that must be carried by an individual as determined by relationship analysis of an individual's ancestry; in a mixed DNA sample, an allele that is foreign to the assumed or known contributor(s)

Off-ladder (OL) allele: STR allele that is of a different length than found in the allelic ladder utilized in typing a particular locus

Off-scale (STR) data: data produced when the emitted fluorescence from the PCR products being measured saturates the detector; may result in flat-topped peaks in the electropherogram for STR alleles and pull-up peaks in one or more color channels corresponding to the off-scale peak; makes data interpretation more complicated

Organic extraction: a method of isolating DNA from cells involving phenol and other organic chemicals

Partial profile: a DNA profile for which complete typing results are not obtained at all tested loci; may be due to limited DNA template, DNA degradation, inhibition, preferential amplification, and/or stochastic effects

Peak height: the maximum y-axis value obtained for a data peak; for STR analysis using CE and fluorescence detection, it is measured in relative fluorescence units and reflects the quantity of the PCR product being measured

Peak height ratio (PHR): the relative ratio of two peaks at a given locus; may be calculated by dividing the peak height of the longer length allele by the peak height of the shorter length allele or by dividing the shorter peak height by the taller peak; also called heterozygote balance (Hb)

Picogram (pg): 10^{-12} grams (a trillionth of a gram); there are approximately 6 pg of DNA in a single human cell

Polymerase chain reaction (PCR): an *in vitro* process that yields millions of copies of desired DNA through repeated cycling of a reaction involving the DNA polymerase enzyme

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

Population substructure: the existence of subpopulations with different allele frequencies

Positive control: an analytical control sample that can be used to determine if a test works properly

Post-amplification purification: cleanup of PCR product to reduce or eliminate amplification reaction components from competing with CE electrokinetic injection, which results in higher peaks in the electropherogram

Power of discrimination: the potential power of a genetic marker or set of markers to differentiate between any two people chosen at random; equal to one minus the sum of the square of the genotype frequencies

PowerPlex 16: a multiplex STR typing kit from Promega Corporation that co-amplifies 15 STRs and the sex-typing marker amelogenin; the STR loci amplified are D3S1358, TH01, D21S11, D18S51, Penta E, D5S818, D13S317, D7S820, D16S539, CSF1PO, Penta D, VWA, D8S1179, TPOX, and FGA

PowerPlex Fusion: a multiplex STR typing kit from Promega Corporation that co-amplifies 22 STRs and the sex-typing marker amelogenin

Precision: a measure of the closeness of results when experiments are repeated

Presumptive test: an initial examination of evidence to indicate the possible source of the sample (e.g., blood, saliva, semen, etc.); usually followed up by a confirmatory assay or DNA analysis

Primer: a short preexisting polynucleotide chain, usually 18-30 bases long, which targets a specific region of the template DNA and allows a DNA polymerase to initiate synthesis of a complementary strand

Probabilistic genotyping: use of statistical modeling informed by biological data, statistical theory, computer algorithms and/or probability distributions to infer genotypes and/or calculate likelihood ratios

Probability vs. odds: assuming that all outcomes are equally probable, the probability of event A is the number of ways event A can happen divided by the total number of possible outcomes; the odds of an event occurring is the ratio of two competing probabilities—the probability that an event will occur and the probability that it will not occur; if the probability of the event is p, the probability that it will not occur is 1 – p; the odds are therefore p/(1-p)

Probability of exclusion (PE): the percentage of the population that can be excluded as potential contributors to a DNA mixture result

Product rule: the combination of genotype frequency estimates from multiple loci through multiplying the individual locus genotypes together

Proficiency test: a quality assurance measure used to monitor performance of an analyst and identify areas in which improvement may be needed; can be internal (produced by the agency undergoing the test) or external (produced by an outside test provider); external proficiency tests can be either open or blind

Profile probability: answers the questions "what is the rarity of a specific DNA profile given the alleles observed?" or "what is the chance that a particular profile exists in a population based on allele frequencies?"

Profiler Plus: a commercial STR typing kit from Applied Biosystems that utilizes multiplex PCR to amplify and provide genetic information for 9 STRs and the sex-typing marker amelogenin; the STR loci examined are D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, and D7S820

Promega Corporation: a company based in Madison, Wisconsin that supplies STR typing kits and other reagents to perform DNA analysis

Pull-up: an artifact that may occur during analysis of fluorescently labeled DNA fragments when signal from one dye color channel produces artificial peaks in another, usually adjacent color, at a similar position on the horizontal axis in an electropherogram; sometimes referred to as bleed-through or spectral calibration failure

Qiagen: a German-based company that provides DNA extraction kits and other molecular biology supplies

Q-K comparison: evaluation of question and known sample DNA profiles with one another to reach a decision of inclusion, exclusion, or inconclusive

Quality assurance (QA): a system of activities whose purpose is to provide to the producer or user of a product or service the assurance that it meets defined standards of quality

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Quality Assurance Standards (QAS): FBI Director-issued requirements established initially under the DNA Identification Act of 1994 by the DNA Advisory Board; now revised by SWGDAM

Quantitative PCR (qPCR): method used for determining the quantity of DNA template present in a sample through measuring the point at which samples cross a detection threshold relative to a set of standard samples; also known as real-time PCR

Random Match: a match in the DNA profiles of two sample, where one is drawn at random from the population

Random Match Probability (RMP): the probability that an unknown individual in a given population has a particular profile; more appropriately the random match probability is computed conditioned on a known individual observed to have the profile; the unconditional probability is also known as the profile probability

Random Man Not Excluded (RMNE): the probability that a random person would be excluded as a contributor to the observed DNA mixture (DNA profile); answers the question - "how often would a random person unrelated to a true contributor be excluded?"

Rapid DNA testing: fully automated process of generating a STR DNA profile ("swab in – profile out") without human intervention; current methods take about 90 minutes and cost about ten times more than conventional DNA testing

Reference sample: a sample (typically blood or buccal swab) taken from a known person that is used for comparison purposes to an evidentiary sample; sometimes referred to as the "K" or known sample

Reproducibility: the ability to obtain the same result when a test or experiment is repeated

RFU: relative fluorescence units

Sanger sequencing: a method of DNA sequencing named after its inventor Frederick Sanger that involves determining the order of bases in a DNA molecule following selective incorporation of chain-terminating dideoxynucleotides by a DNA polymerase during *in vitro* DNA replication

Sex chromosomes (X and Y chromosomes): chromosomes that are different in the two sexes and involved in sex determination

Sexual assault kit (SAK): a set of items used by medical personnel to collect and preserve physical sexual assault evidence for use in a criminal investigation

Short tandem repeats (STR): multiple copies of an identical (or similar) DNA sequence arranged in direct succession where the repeat sequence unit is 2 bp to 6 bp in length; because STRs generally occur in the "junk" DNA outside of the constraints of genes, the number of repeat units can vary between individuals in an accordion-like fashion

Single nucleotide polymorphism (SNP): any polymorphic variation at a single nucleotide; most SNPs are biallelic (e.g., either C or T) with the minor allele being observed at least 1% of the time

Source attribution: a decision which identifies an individual as the source of the DNA that produced an evidentiary single-source or major contributor profile; the statement if often based on profile frequency estimates that are rarer than some defined value, generally more than the Earth's population

Spatial calibration: performed when a new capillary array is installed on a CE instrument in order to map the location of fluorescence from each capillary onto the charged-coupled device (CCD) detector

Spectral calibration: an examination of the contribution of overlap in the emission spectrum of fluorescent dyes used for a specific DNA test on a CE instrument; permits the color deconvolution necessary for multi-color STR typing or DNA sequencing; a poor spectral calibration may cause artifact peaks or inaccurate peak height determinations

Sperm cell fraction: the portion of a sample produced during differential extraction containing DNA from sperm cells; generally, from the male perpetrator in the case of sexual assault evidence

Standards: criteria established for quality assurance purposes that place specific requirements on laboratories and analysts; also refers to well-characterized samples that can aid calibration of measurements

Stochastic effects: the observation of intra-locus peak imbalance and/or allele drop-out resulting from random, disproportionate amplification of alleles in low-quantity template samples

Stochastic threshold: the peak height value in a DNA profile above which it is reasonable to assume that, at a given locus, allelic dropout of a sister allele in a heterozygous pair has not occurred in a single-source DNA sample; often used to disqualify loci from consideration with CPI statistical calculations due to the possibility of allele dropout; due to the possibility of shared alleles in mixtures, the presence of allele peaks above the stochastic threshold is no guarantee that allele dropout did not occur in mixed DNA sample profiles

STRmix: a probabilistic genotyping software program sold by ESR (Auckland, New Zealand)

Stutter product: a minor peak primarily appearing one repeat unit smaller than the primary STR allele; results from strand slippage during the amplification process; usually <15% of the height of the true allele

SWGDAM: Scientific Working Group on DNA Analysis Methods; formerly known as TWGDAM, Technical Working Group on DNA Analysis Methods; an FBI sponsored group that develops quality assurance standards and guidelines for laboratories in the United States

Theta (θ) correction: a method for calculating match probabilities, first described by Balding and Nichols (1994), to allow for population structure in the population for which a frequency database is constructed; it allows match probabilities for subpopulations to be calculated from whole population allele frequencies and avoids the need to assume Hardy-Weinberg equilibrium at the whole population level

Touch DNA: DNA that is transferred to or from surfaces via contact

Tri-allelic pattern: an occasional detection of three alleles at a particular STR locus in single-source samples; these copy number variants (CNVs) result from extra chromosome fragments being present in unbalanced (Type 1) or balanced (Type 2) peak heights

TrueAllele: a probabilistic genotyping software program sold by Cybergenetics (Pittsburgh, PA)

Uninterpretable: the inability to interpret or use results for comparisons due to poor or limited data quality, or data that fail to meet quality assurance requirements

Validation: the process of performing and evaluating a set of experiments that establish the efficacy, reliability, and limitations of a method, procedure or modification thereof; establishing recorded documentation that provides a high degree of assurance that a specific process will consistently produce an outcome meeting its predetermined specifications and quality attributes before acceptance for routine use; may include developmental and/or internal validation

Variant allele: a non-standard form of an allele due to a point mutation, an insertion or a deletion relative to other commonly seen alleles

Verification: confirmation by examination that specified requirements have been met

X-chromosome: one of the sex chromosomes; normal females possess two copies and males one copy

X-STR: short tandem repeat markers found on the X-chromosome that may be useful in addressing some kinship analysis scenarios

Y-chromosome: one of the sex chromosomes; normal males possess one copy and females none

Yfiler: a multiplex STR typing kit from Applied Biosystems that co-amplifies 17 segments of the Y-chromosome; the Y-STR loci examined are DYS19, DYS385a/b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, and Y-GATA-H4.

Y-STR: short tandem repeat markers found on the Y-chromosome that enable male-specific DNA testing; can be useful in cases involving sexual assault and with genetic genealogy to trace male lineages

Zygote: cell formed when the nuclear DNA from a father's sperm cell combines with nuclear DNA in a mother's egg restoring the diploid chromosome count; a fertilized egg