Important 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.
Allele – one of two or more alternative forms of a gene.
Allele frequency – the relative occurrence of a particular allele in a population.
ASCLD (ascld.org) – American Society of Crime Laboratory Directors; involved with accreditation of DNA testing labs.
Autoradiogram (autoradiograph; autorad) – a recording of the positions on an X-ray film of where radioactive probes have bound to alleles.
Band – a representation of a particular DNA fragment on an autoradiogram.
Base pair – two complementary nucleotides in DNA; base pairing occurs between A and T and between G and C.
Blind proficiency test – a proficiency test in which the laboratory personnel do not know that a test is being conducted.
Capillary electrophoresis – a method for separating utilizing a narrow polymer-filled tube to perform DNA size separation.
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 1998 and containing the STR DNA profiles of tens of thousands of convicted offenders.
COfiler – PCR Amplification Kit (AmpFLSTR® COfiler™) that provide human identification laboratories with the ability to generate information for the STR loci (CSF1P0, D16S539, TH01,TP0X, D3S1358, D7S820 and Amelogenin) as required by the Combined DNA Index System (CODIS).
Controls – tests performed in parallel with experimental samples and designed to demonstrate that a procedure worked correctly.
Degradation – the chemical or physical breaking down of DNA.
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.
Dye blobs – occur when fluorescent dyes come off of their respective primers and migrate independently through the capillary in STR testing.
Electrophoresis – a technique in which different molecules are separated by their rate of movement in an electric field.
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 (usually agarose or acrylamide) used in electrophoresis to separate molecules.
Gene – the basic unit of heredity; a sequence of DNA nucleotides on a chromosome.
Genetic drift – random fluctuation in allele frequencies due to small population sizes (sampling error).
Genome – the sum total of an organism's genetic material.
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.
Hardy-Weinberg equilibrium (HWE) – allele frequencies in populations of organisms that are in HWE do not change from one generation to another and there are no statistical correlations between any pairs of alleles within individuals in the population.
Heterozygous – a heterozygous organism has two different alleles at a particular locus; for forensic DNA typing, two alleles (not one) are reported.
Homozygous – a homozygous organism has two copies of the same allele at a particular locus; for forensic DNA typing, one allele (not two) is reported.
Identifiler – PCR Amplification Kit (AmpFLSTR® Identifiler™) that provides human identification laboratories with the ability to generate information on 15 polymorphic STR loci plus Amelogenin (for determination of a contributor's sex).
Kilobase (kb) – 1,000 bases (or nucleotides or base pairs).
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); equivalent to the inbreeding coefficient of a (perhaps hypothetical) offspring; designated by F.
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 frequency than would be predicted if they were in HWE.
Locus (pl. loci) – the physical location of a gene on a chromosome.
Matrix failure (pull up) –a result of the inability of the detection instrument to properly resolve the dye colors used to label STR amplicons. Often due to off-scale peaks.
Mitochondrial DNA (mtDNA) – DNA found in the mitochondria inside of the cell (not associated with the nuclear chromosomes), transmission is only from mother to child.
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.
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 aware; internal proficiency tests are conducted by the laboratory itself and external tests are conducted by an agency independent of the laboratory being tested.
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.
Pull up – in STR testing, the result of color bleeding from one spectral channel to another; see also matrix failure.
Quality assurance – a program conducted by a laboratory to ensure accuracy and reliability of test performed.
Quality control – internal activities or activities according to externally established standards used to monitor the quality of DNA typing to meet and satisfy specified criteria.
Random match probability – the chance of a random match; as used in DNA profiling, it is the probability that the DNA in a random sample from the population has a DNA profile that cannot be distinguished from that observed in an evidence sample.
Restriction endonuclease, restriction enzyme – an enzyme that cleaves DNA molecules at particular strings of nucleotides.
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 – the discipline concerned with the immunologic study of body fluids.
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.
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.
SWGDAM (formerly TWGDAM) - Scientific Working Group on DNA Analysis Methods
Taq polymerase – a DNA polymerase 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.

Loci amplified and corresponding dyes in each of three commonly used STR-typing kits - in the order in which they appear on electropherograms.

Profiler Plus®

BLUE	GREEN	YELLOW
D3	Amelogenin	D5
vWA	D8	D13
FGA	D21	D7
	D18	

COfiler®

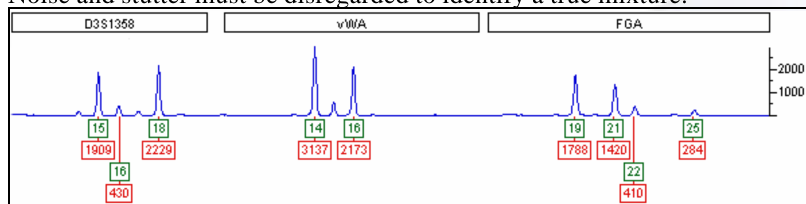
BLUE	GREEN	YELLOW
D3	Amelogenin	D7
D16	THO1	
	TPOX	
	CSF	

Identifiler®

BLUE	GREEN	YELLOW	RED
D8	D3	D19	Amelogenin
D21	THO1	vWA	D5
D7	D13	TPOX	FGA
CSF	D16	D18	
	D2		

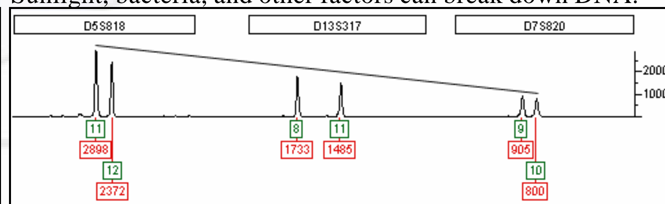
Mixture

A mixture is identified by observing more than two alleles in any locus. Noise and stutter must be disregarded to identify a true mixture.



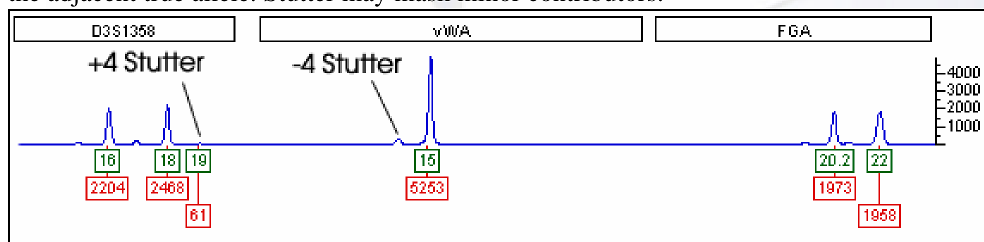
Degradation

Degradation is marked by consecutively falling peak heights. Sunlight, bacteria, and other factors can break down DNA.



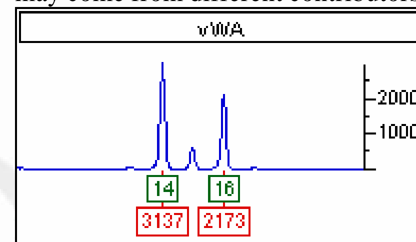
Stutter

Stutter peaks appear before or after true alleles and typically have heights less than 15% of the adjacent true allele. Stutter may mask minor contributors.



Peak height imbalance

Peaks differing by more than 30% may come from different contributors.



Twelve important questions always need to be asked about DNA evidence:

1. Has the prosecution documented the entire history of the key evidentiary samples from the time of collection to ultimate disposition, including records of all examinations and tests performed on those samples?

2. Is it possible to determine with certainty the nature of the biological material from which the DNA originated? (Particularly in sexual assault cases, it may be important to know whether a sample linked to a suspect originated from semen or some other biological material.)

3. Has the testing laboratory been audited or evaluated by an outside agency? If not, why not? If so, has the prosecution provided copies of the audit documents?

4. Is the testing laboratory accredited? If so, by what agency? If not, why not? (Did the laboratory seek accreditation and fail? If so, has the prosecution provided a copy of the report of the accreditation committee?)

5. Has the laboratory participated in a proficiency testing program? If not, why not? If so, has the prosecution provided documentation of the results?

6. Are there any inconsistencies between the DNA profiles that the lab declared to "match"? Are there any "missing" alleles or "extra" alleles that complicate the interpretation of the test results?

7. Did the laboratory run all necessary control samples? Did the control samples produce the expected results?

8. Did the laboratory employ "blind" procedures for interpreting the test results? (Failure to use blind procedures can result in "examiner bias" – i.e., the tendency for an analyst to interpret ambiguous data in a manner consistent with the expected or desired outcome and may therefore be an unreliable/incorrect scientific procedure.)

9. How much DNA did the evidentiary samples contain? (Knowing how much DNA was present may help you evaluate whether the results could be explained by contamination or inadvertent DNA transfer.)

10. Do any of the key actors in the case have close relatives who might have been involved? (Labs typically estimate the frequency of DNA profiles among unrelated individuals. The probability of a chance match between DNA profiles is always higher for relatives than for unrelated individuals.)

11. Have the statistical estimates been computed properly in accordance with generally accepted methods? Do they address the right issue? (There continues to be considerable controversy surrounding the proper way to generate statistical estimates for comparisons involving mixed samples and partial or incomplete profiles. Labs often choose methods that are unfairly slanted against the accused.)

12. Is there evidence of unreported additional contributors to any samples? (Labs sometimes overlook or fail to report weak results that may indicate the presence of an additional contributor to evidentiary samples.)

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