Important DNA testing terminology

ABI 310/3100 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.

Amelogenin – a locus on the human sex chromosomes (XX=female, XY=male) that allows sex determination.

Base pair – two complementary nucleotides in DNA; base pairing occurs between A and T and between G and C.

Capillary electrophoresis – a method that utilizes a narrow polymer-filled tube to separate DNA molecules by size.

Chromosome – a large piece of DNA. Humans have pairs of 23 different chromosomes in most of their cells.

CODIS – COmbined DNA Index System, established in 1998 and containing the STR DNA profiles of millions of convicted offenders and arrestees.

COfiler – PCR amplification kit (AmpFLSTR® COfiler®) commonly used to generate information for six STR loci and Amelogenin.

Combined probability of inclusion (CPI) – the chance of a random match with a mixed sample; it is the probability that a randomly chosen person has a DNA profile that cannot be excluded from contributing to a mixed evidence sample.

Controls – tests performed in parallel with experimental samples and designed to demonstrate that a test was reliable.

Degradation – the chemical or physical breaking down of DNA.

DNA (Deoxyribonucleic acid) – the genetic material.

DNA polymerase – an enzyme that catalyzes the synthesis of double stranded DNA.

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.

Genome – the sum total of an organism's genetic material.

Genophiler® – an automated, objective system for reviewing and presenting DNA profiling data.

GenoStat®– a free software package used to calculate forensic DNA match statistics and resolve DNA mixtures.

Genotype – the genetic makeup of an organism, as distinguished from its physical appearance or phenotype.

Hardy-Weinberg equilibrium (HWE) –populations of organisms that are in HWE have no significant correlations between any pairs of alleles within individuals in the population.

Heterozygote – a heterozygous organism has two different alleles at a particular locus.

Homozygote – a homozygous organism has two copies of the same allele at a particular locus.

Identifiler – PCR amplification kit (AmpFLSTR® Identifiler®) commonly used to generate information on 15 STR loci and Amelogenin.

Linkage – the association of alleles at two or more loci due either to their residing on a single chromosome or their prevalence in a particular ethnic group that causes them to appear together at a higher than expected frequency.

Locus (pl. loci) – the physical location of a gene on a chromosome.

Low copy number (LCN) / Low-template (LT-DNA) DNA – DNA test results at or below the stochastic threshold. Typically involves less than 200pg of starting material.

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.

Mini-STRs – STR testing that uses a different set of primers to create shorter amplicons. Designed to get more complete profiles from degraded samples.

Mitochondrial DNA (mtDNA) – DNA found in the mitochondria inside cells (not associated with the nuclear chromosomes); transmission is only from mother to child.

Nucleotide – chemical units that are strung together in long chains to make DNA molecules.

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.

Profiler Plus – PCR Amplification Kit (The AmpFLSTR® Profiler Plus®) commonly used to generate information for nine polymorphic STR loci and the Amelogenin locus.

Random match probability (RMP) – the chance of a random match; as used in DNA profiling, it is the probability that a randomly chosen person has a DNA profile that cannot be distinguished from that observed in an evidence sample.

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.

Sequential unmasking – the process of evaluating an item of evidence in a blind fashion with no knowledge of potential contributor profiles.

Stochastic effects – random fluctuations in the testing results that can adversely influence DNA profile interpretation (e.g., exaggerated peak height imbalance, exaggerated stutter, allelic drop-out, and allelic drop-in).

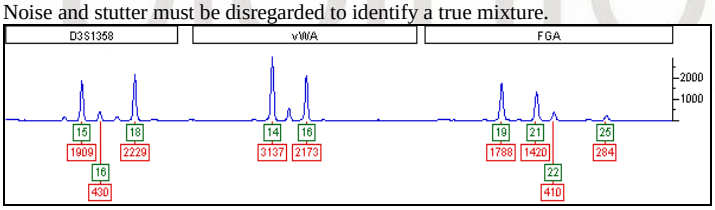
STR (short tandem repeats) – a locus where alleles differ in the number of times that a string of four nucleotides are tandemly repeated.

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.

Y-STR DNA – DNA found on a male's Y chromosome; transmission is only from father to son.

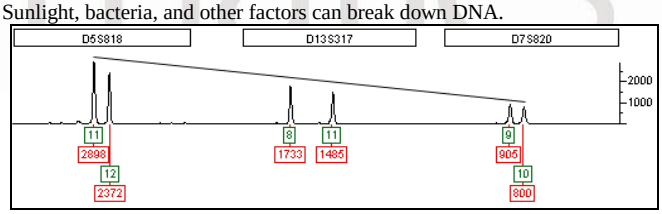
Mixture

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.



Loci amplified and corresponding dyes in STR-typing kits – in the order they appear on electropherograms.

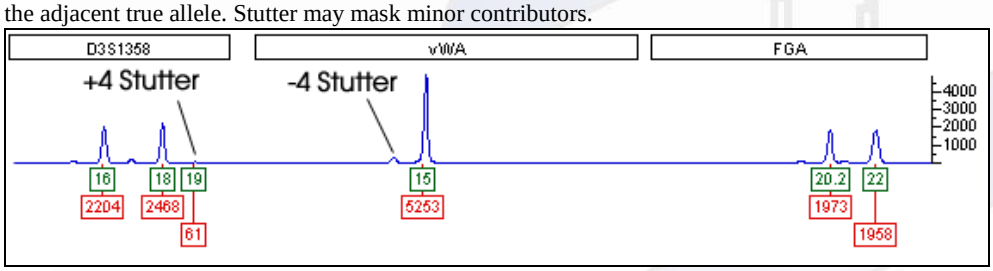
	Profiler Plus®					COfiler®					Identifiler®				
BLUE		D3	vWA	FGA			D3		D16		D8	D21	D7	CSF	
GREEN	AM	D8	D21	D18		AM	THO1	TPOX	CSF		D3	THO1	D13	D16	D2
YELLOW		D5	D13	D7					D7		D19	vWA	TPOX	D18	
RED											AM	D5	FGA		

	MiniFiler®				Yfiler®				
BLUE	D13	D7	vWA		DYS456	DYS389I	DYS390	DYS389II	
GREEN	AM	D2	D21			DYS458	DYS19	DYS385a/b	
YELLOW	D16	D18			DYS393	DYS391	DYS439	DYS635	DYS392
RED	CSF	FGA			H4	DYS437	DYS438	DYS448	

	PowerPlex16®							SMG+™			
BLUE	D3S1358	TH01	D21S11	D18S51	Penta E			D3S1358	vWA	D16S539	D2S1338
GREEN	D5S818	D13S317	D7S820	D16S539	CSF1PO	Penta D		AM	D8S1179	D21S11	D18S51
YELLOW	AM	vWA	D8S1179	TPOX	FGA			D19S433	TH01	FGA	

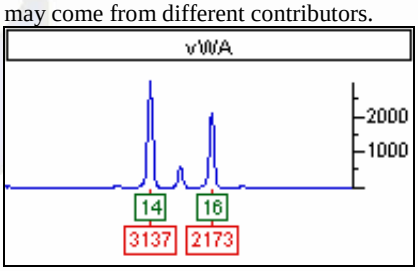
Stutter

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 by an outside agency? If not, why not? If so, have copies of the audit documents been provided?

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, have the results been provided?

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 do so can result in "examiner bias," a tendency to interpret ambiguous data in a manner consistent with the expected or desired outcome – 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. Does anyone in the case have a close relative that might be 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.)