DNA Report Glossary of Terms and Important Concepts

The following supplemental information is to provide our clients with a more comprehensive description of the methods used by our laboratory for the examination/analysis of items submitted for forensic testing.

Definitions

DNA Analyses and Results Interpretation

Allele. A form of a gene, or region of DNA that is located at a specific physical location or site (a locus, plural is loci) on a particular chromosome. Humans generally have either one or two alleles at each locus. When individuals inherit two copies of the same allele, they are **homozygous** at that locus. When they inherit two different alleles, they are **heterozygous** at that site.

Analytical Threshold. The peak height value below which a peak cannot be reliably distinguished from background machine noise.

Autosome. A chromosome that is not one of the sex chromosomes—chromosomes 1–22 in humans.

CODIS. Combined DNA Index System. A compilation of state and national DNA databases (all 50 states, FBI, USACIL, DC, Puerto Rico) containing STR profiles from convicted offenders, evidentiary samples, missing persons, and other categories. CODIS is maintained by the FBI.

Deconvolution. The virtual separation of DNA mixtures into the genotypes of the individual contributors based on quantitative peak height information and assumptions pertaining to the profile, such as the number of contributors and any known/assumed contributors.

Deoxyribonucleic acid (DNA) testing involves multiple steps, including DNA extraction, quantitation, amplification, electrophoresis and analysis of the resulting data.

DNA extraction. The recovery of purified DNA from biological samples.

DNA quantitation. A means of measuring the amount of DNA (total human DNA, male DNA, and degradation index) recover from biological samples by a specialized PCR method called quantitative polymerase chain reaction (qPCR) or real-time PCR.

DNA Profile. The DNA test results (single source or mixture) generated at 1 or more loci.

Differential DNA extraction. A method of separating semen-containing mixed body fluid samples into two fractions, a sperm-rich fraction (SF) and an epithelial-rich fraction (EF), based on differences between sperm cells and other cell types.

Electropherogram. A representation of the DNA profile (PCR products separated on the basis of their size by capillary electrophoresis) in the form of peaks on a graph. The height of the peaks is measured in relative fluorescent units (RFUs).

Fusion 6C PCR Amplification Kit (F6C). F6C is a 6 dye system manufactured by Promega that amplifies 27 loci (23 autosomal STRs, 3 Y-STRs, and the Amelogenin gender marker) in a single reaction. F6C contains all 20 loci now required for CODIS.

GlobalFiler PCR Amplification Kit (GF). GF is a 6 dye system manufactured by Thermo Fisher Scientific that amplifies 24 loci (21 autosomal STRs, 1 Y-STR, 1 insertion/deletion polymorphic marker on the Y chromosome, and the Amelogenin gender marker) in a single reaction. GF contains all 20 loci required for CODIS.

Known/Assumed contributor. The assumption that a person is the source of (or contributor to) a DNA profile. The assumption must be consistent with the DNA profile results and there must be objective support for the assumption i.e., there must be a reasonable scientific expectation that an individual's DNA is in the evidentiary profile. In general, this would apply to non-probative comparisons for intimate samples and other items that have been regularly handled or worn by the assumed contributor.

Loci. Specific locations in the DNA molecule (singular is locus).

Polymerase Chain Reaction (PCR). Also known as DNA amplification. PCR can be thought of as "molecular photocopying", where small segments of DNA (typically, short tandem repeats or "STRs" in forensics) are copied many times in a thermal cycler.

Probabilistic Genotyping (PG). A DNA profile analysis tool. Probabilistic genotyping is the use of biological modeling to infer genotypes for the DNA typing results and to calculate likelihood ratios. PG analysis provides a statistical weighting to the different genotype combinations that could explain the evidentiary profile.

STRmix. A probabilistic genotyping DNA analysis software system that models (analyzes) DNA profile results. STRmix is a software tool used to assist the DNA analyst in interpreting DNA profiles and in performing statistical calculations. STRmix can accurately deconvolute DNA mixtures—a virtual separation of mixture profiles into their individual components (genotypes) based on quantitative peak height information.

Stochastic threshold. The peak height value above which it is reasonable to assume that allelic dropout has not occurred at any given locus. Allelic dropout is when one or more alleles present in the sample are not detected above the analytical threshold.

Yfiler PCR Amplification kit. The Yfiler kit, manufactured by Applied Biosystems, amplifies 17 Y-STRs in a single reaction. Yfiler is a 5 dye system containing the SWGDAM recommended Y-STRs and other highly variable markers.

Yfiler Plus PCR Amplification kit. The Yfiler Plus kit, manufactured by Thermo Fisher Scientific, amplifies 27 Y-STRs in a single reaction. Yfiler Plus is a 6 dye system containing the SWGDAM recommended Y-STRs and other highly variable markers.

DNA methods for the analysis of biological material employ two basic types of DNA testing:

- **Autosomal STR testing** (loci not on the X or Y chromosomes) offers the greatest potential for individualization. Such tests detect both male and female DNA equally, but an excess of female DNA (e.g. >50:1) may render a male profile undetectable.

● **Y-STR testing** detects only male DNA. As a result, a male DNA profile can be detected even in the presence of a significant excess of female DNA (>100:1). Since all paternally-related males (and an unknown number of unrelated males in the general population) have identical Y-STR profiles, a Y-STR profile cannot be individualized to a single male.

General Categories of Testing Conclusions

Mixture. A DNA profile that contains results from more than 1 individual.

Major contributor. A DNA profile that is present in a higher quantity within a DNA mixture.

Minor contributor. A DNA profile(s) that is present in a lower quantity within a DNA mixture.

Inconclusive. Per DSS SOPs, no conclusion can be drawn from the comparison between the known sample and the evidentiary sample.

Insufficient profile results/Insufficient for comparison. When insufficient data is obtained for the questioned DNA Profile for comparison purposes, the following statement is reported: "Due to limited data detected from the item, the comparison to the known profile is inconclusive." This statement is made for low-level, partial DNA profiles when there is insufficient data from the evidentiary profile for comparison to a particular known.

Consistent with Source. The results are consistent with the tested individual being the source of a single-source DNA profile.

Inclusion. The results demonstrate that the tested individual is a potential contributor to a DNA mixture.

Cannot be eliminated. Analogous to a "partial match". An individual cannot be eliminated (CBE) as the source of (or contributor to) a DNA profile if there is a strong positive association (LR is at or above the DSS inconclusive threshold of 1,000 - 10,000, dependent on amplification kit) between the known profile and the evidentiary profile but some of the alleles present in the known sample are not detected in the evidentiary profile.

Eliminated. The DNA profile for a questioned sample is not consistent with originating from the known sample tested.

General Categories of Statistical Conclusions

The Likelihood Ratio (LR) assesses the probability of the evidentiary profile occurring given two alternate (mutually exclusive) hypotheses; (H_1 and H_2). H_1 is typically where the data is explained by an inclusion of the person of interest (POI). H_2 is typically where the data is explained by a person selected at random from the general population and not the POI (match is coincidental). An LR is typically calculated using STRmix™ Analysis software for forensic unknown DNA profiles when the known individual(s) is not eliminated as the source of or a contributor to the evidence profile by a manual comparison. The LR is the probability of the profile occurring if hypothesis 1 (H_1) is true compared to the probability of the profile occurring under the alternate hypothesis (H_2).

The Y-STR statistic (counting method) reflects the number of times that a given Y-STR profile is observed in a search of the National Y-STR Database. The counting method provides an estimate of the random match probability for a Y-STR profile.

Random Match Probability (RMP). RMP estimates the rarity of a DNA profile in the general population (unrelated individuals). It is the probability that a person selected at random from the general population (unrelated) would match the evidentiary profile.

Criminal Parentage Testing. The random man not excluded (RMNE) statistic is calculated when the tested individual is not eliminated. The statistic calculated is the expected frequency (parentage inclusion probability) of individuals who could contribute the paternally (or maternally) transmitted alleles.

Combined Kinship Index (CKI). CKI compares the relative support for two competing hypotheses (non-criminal cases): the two individuals are related, versus the hypothesis that they are unrelated individuals selected at random from the population.