ASB Standard 081, First Edition

~~2022~~2024

# Standard for Training in ~~the Use of Statistics in Interpretation of~~ Statistical Calculations Used for Forensic Short Tandem Repeat (STR) DNA ~~Evidence~~Data

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ASB Approved Xxxxx ~~2022~~2024

ANSI Approved Xxxxxx ~~2022~~2024

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# Foreword

This standard defines the minimum requirements for a forensic DNA analyst training program in the application of statistics to autosomal and Y-STR DNA profiling results. The aim is to provide a framework for quality training that will result in consistency in the forensic DNA community.

This document was revised, prepared, and finalized as a standard by the DNA Consensus Body of the AAFS Standards Board. The draft of this standard was developed by the Human Forensic Biology Subcommittee of the Organization of Scientific Area Committees (OSAC) for Forensic Science.

The American Academy of Forensic Sciences established the Academy Standards Board (ASB) in 2015 with a vision of safeguarding Justice, Integrity and Fairness through Consensus Based American National Standards. To that end, the ASB develops consensus based forensic standards within a framework accredited by the American National Standards Institute (ANSI), and provides training to support those standards. ASB values integrity, scientific rigor, openness, due process, collaboration, excellence, diversity and inclusion. ASB is dedicated to developing and making freely accessible the highest quality documentary forensic science consensus Standards, Guidelines, Best Practices, and Technical Reports in a wide range of forensic science disciplines as a service to forensic practitioners and the legal system.

Questions, comments, and suggestions for the improvement of this document can be sent to AAFS-ASB Secretariat, asb@aafs.org or 401 N 21st Street, Colorado Springs, CO 80904.

All hyperlinks and web addresses shown in this document are current as of the publication date of this standard.

ASB procedures are publicly available, free of cost, at www.aafs.org/academy-standards-board.



**Keywords:** *random match probability, likelihood ratio, DNA interpretation, statistics*, *training, DNA standard*

# Table of Contents *(to be finalized prior to this document's publication)*

# Standard for Training in ~~the Use of Statistics in Interpretation of~~ Statistical Calculations Used for Forensic Short Tandem Repeat (STR) DNA ~~Evidence~~Data

## 1 Scope

This standard ~~defines~~outlines the minimum requirements for a training program in the use of statistical ~~methods approved within the laboratory~~calculations and values reported for ~~interpretation of~~ forensic autosomal and Y short tandem repeat (STR) DNA ~~evidence~~data.

## 2 Normative References

The following reference is indispensable for the application of the standard. For dated references, only the edition cited applies. For undated references, the latest edition of the referenced document (including any amendments) applies.

ANSI/ASB Standard 022, *Standard for Forensic DNA Analysis Training Programs*[1].

## 3 Terms and Definitions

For purposes of this document, the following definitions apply.

**3.1**
**avuncular index**
~~Likelihood~~The likelihood ratio ~~in which~~that evaluates the ~~probability of a questioned person's profile is evaluated under alternate propositions - they are an uncle/aunt/niece/nephew of a known~~hypothesis that the tested individual is the biological uncle (or aunt or niece or nephew) of the profile donor versus ~~they are~~the hypothesis that the tested individual is unrelated to the ~~known individual. This calculation also applies to the questions of possible half-siblings, grandparent and grandchild.~~ profile donor.

**3.2**
**Combined Probability of Exclusion**
**CPE**
The ~~product of the probabilities of exclusion calculated for each~~ probability that a randomly selected individual would be excluded as a contributor to the DNA ~~locus~~mixture. If the single-locus exclusion probabilities are independent, and if $P_j$ is the probability of exclusion at locus j, then the combined probability of exclusion is $1 - \prod_j (1 - P_j)$.

**3.3**
**Combined Probability of Inclusion**
**CPI**
**The** ~~product of the probabilities of inclusion calculated for each DNA locus.~~**probability that a randomly selected individual would not be excluded (i.e., is included) as a contributor to the DNA mixture.** If the single-locus exclusion probabilities are independent, and if $P_j$ is the probability of exclusion at locus j, then the combined probability of inclusion is $\Pi_j (1 - P_j)$.

[1] Available from: www.aafs.org/academy-standards-board.

**3.4**
**counting method**
A method for estimating genotype, sequence, or haplotype frequency by direct counting of the number of times a genotype, sequence, or haplotype is observed in a database and dividing by the number of samples in that database. This method is commonly used for estimating frequencies in populations for mitochondrial DNA and Y STR DNA haplotype results.

**3.5**
**haplotype**
A set of linked DNA variations, or polymorphisms, that tend to be inherited together (e.g.~~,~~ commonly used for human Y-chromosome or mitochondrial analysis). A haplotype can refer to a combination of alleles or to a set of single nucleotide polymorphisms (SNPs) found on the same chromosome.

**3.6**
**Hardy-Weinberg equilibrium**
A state in which allele and single locus genotype frequencies do not change (on average) from one generation to the next in a population. When alleles in a population are independent, allele and genotype frequencies are related through the Hardy-Weinberg principle: for a locus with 2 alleles P and Q at frequencies of p and q, homozygotes for P are found at frequency $p^2$, homozygotes for Q are found at a frequency of $q^2$, and heterozygotes are found at a frequency of 2pq. Use of the theta correction removes the need to assume Hardy-Weinberg equilibrium in the population for which a frequency database is constructed. See ~~**Theta Correction**~~**theta correction**.

**3.7**
**Identical by Descent**
**IBD**
Identical alleles that are copies of the same ancestral allele without mutation; this is a subset of identical by state (IBS).

**3.8**
**Identical by State**
**IBS**
Identical alleles that may or may not be copies of the same ancestral allele.

~~**3.7**~~**3.9**
**inbreeding**
Mating of two persons who are more closely related than if they were chosen at random. It increases the frequency of homozygous genotypes above the expected for a ~~random breeding~~randomly mating population in Hardy-Weinberg equilibrium.

~~**3.8**~~**3.10**
**kinship analysis**
Comparison of genetic profiles of two or more individuals to evaluate alternative degrees of relatedness.

~~3.9~~3.11
**Likelihood Ratio**
**LR**
~~The probability~~A likelihood ratio is defined by a ratio of ~~the evidence under one proposition (hypothesis), divided by~~ two conditional probabilities: the probability of the evidence ~~under an alternative,~~given each of two mutually exclusive ~~proposition (hypothesis). The magnitude of~~and competing propositions. In forensic science applications, the likelihood ratio is used as an expression for the meaning of scientific evidence and as measure for its value ~~expresses the weight of the evidence.~~.
*ASTM E1732-12[mod]*[2]

~~3.10~~3.12
**linkage equilibrium**
~~Two loci are in linkage~~Linkage equilibrium describes the situation in which the haplotype frequencies in a population have the same value that they would have if the ~~probability an individual jointly receives particular~~ alleles at ~~the loci is the product of the probabilities of receiving each of the alleles separately.~~each locus were combined at random. If both Hardy-Weinberg and linkage equilibrium hold, then random match probabilities may be multiplied over loci.

~~3.11~~3.13
**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.

~~1.1~~
**Paternity or Maternity Index**
**PI/MI**
The likelihood ratio ~~for observing~~that evaluates the ~~data in a parentage case. More specifically,~~ hypothesis that the ~~probability of observing this data if the alleged father is~~ tested individual is the biological ~~father of the child, divided by the probability of observing the data if a random, unrelated male in the population is the~~ mother or biological father~~. The identity of the mother may or may not be known, and her genotype may or may not be included in the evaluation. One could also calculate a maternity index if the identity of the mother is in question.~~

~~Note This is not to be confused with the abbreviation "PI" that refers to the probability of inclusion in autosomal STR analysis.~~

[2] Available from ASTM, at www.astm.org

~~3.12~~3.14

~~**Probability of Inclusion**~~

~~**PI**~~

~~The probability a randomly selected, unrelated individual is not excluded from being a source of DNA evidence. In human forensic DNA testing, this is often referred to as the probability a random man is not excluded (RMNE). The commonly used calculation is (SPI)2, the square of the sum of the relative frequencies (PI) of the observed alleles at a locus. If the randomly selected~~ of the profile donor versus the hypothesis that the tested individual is ~~assumed to be related to the person of interest, this formula is inappropriate.~~ unrelated to the profile donor.

~~Note: This is not to be confused with the abbreviation "PI" that refers to a "paternity index" commonly used in paternity testing.~~

~~3.13~~3.15

**Random Match Probability**

**RMP**

The probability of ~~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 the~~randomly selecting an unrelated individual from the population who could be a potential contributor to an evidentiary profile ~~probability.~~.

~~3.14~~3.16

**reverse parentage**

Likelihood ratio in which three individuals have been profiled ~~:~~ the child and two questioned biological parents. More specifically, the probability of observing the data if the child is the biological child of the alleged parents, divided by the probability of observing the data if two randomly selected people are the parents of the child.

~~3.15~~3.17

**sibship index**

Likelihood ratio in which the ~~probability of a~~numerator is conditioned on the hypothesis that a sibling of the source of the questioned ~~person's~~ profile ~~is evaluated under alternate propositions – he/she~~in a specimen, and the denominator is ~~a sibling of a known individual versus they are~~conditioned on the hypothesis that an unrelated ~~to the known~~ individual is the source.

~~3.16~~3.18

**source attribution**

A ~~decision made based on laboratory policy~~declaration which identifies an individual as the source of the DNA that produced an evidentiary single-source or ~~major~~deduced contributor profile. This statement is based on a statistical estimate that meets or exceeds a laboratory-defined threshold.

~~1.2~~

~~**theta 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. It avoids the need to assume Hardy-Weinberg equilibrium at the whole-population level.~~

**3.19**
**theta correction**
A value used to adjust statistical calculations that rely on population databases to correct for substructures within populations.

## 4 Requirements

### 4.1 General

~~Based upon the laboratory procedures, some of the requirements in this section may be omitted from the training program.~~

ANSI/ASB Standard 022, *Standard for Forensic DNA Analysis Training Programs* shall be used in conjunction with this document because ANSI/ASB Standard 022 provides the foundational training program requirements upon which additional specific requirements, such as this document, will be based.

The laboratory's training program shall include all requirements applicable to the work conducted by the laboratory and by the individual in training.

### 4.2 Knowledge-Based Training

**4.2.1** At a minimum, the knowledge-based portion of the training program shall require review of the following:

a) the laboratory's protocols for statistical applications;

b) the laboratory's applicable validation studies;

c) literature used to support specific calculations and their use in appropriate circumstances; ~~and~~

d) applicable literature as assigned by the trainer;

~~d)~~e) literature on the effects of cognitive bias in decision-making processes associated with statistical calculations used for forensic STR DNA data.

**4.2.2** The knowledge-based training component of the laboratory's training program shall provide the trainee with a basic understanding of probability and statistics applied to autosomal and Y-STR data to include, at minimum, the following topics.

a) *Population Genetics*

1) laws of Mendelian genetics (law of segregation and the law of independent assortment);

2) Hardy-Weinberg ~~Equilibrium~~equilibrium;

3) linkage equilibrium/disequilibrium;

4) use of theta correction to adjust for inbreeding and population substructure; ~~and~~

b) *Statistical Foundations*

1) frequency~~,~~;

2) probability~~,~~;

3) odds~~,~~;

4) the laws of probability (~~i.~~e.g., the addition rule and product rule~~) and~~ );

~~1)~~5) Bayes' theorem.

6) Sources of uncertainty (e.g. modelling uncertainty and sampling variability)

~~b)~~c) *Population Allelic Frequency Databases*

1) population database size relative to the population size;

2) sample collection, to include:

i) number of samples,

ii) how ~~racial origin~~population group was determined,

iii) how the database was created, maintained and reviewed~~;~~,

~~1) population group;~~

iv) sampling uncertainty;

3) differences in ~~alleles~~allele frequencies observed between population databases;

4) mechanisms to account for alleles not observed in the database.

~~c)~~d) *Suitability of data for statistical application*

1) when to perform statistical analyses~~; and~~ and which statistical calculation is validated for the type of data obtained and comparison performed;

2) instruction on which loci to include in the statistical analyses when the following circumstances are observed, ~~to include~~at a minimum:

i) no allelic data,

ii) partial allelic data,

iii) tri-alleles, duplications/triplications, null alleles, and mutations~~.~~,

iv) STR artifacts.

~~d)~~e) *Statistical ~~Analysis~~analysis for autosomal STR data*

1) ~~general principles~~ proper use of ~~autosomal STR~~ statistical ~~methods~~calculations to include derivation and applicable equations;

2) ~~underlying theory of~~ statistical calculation method(s) in use by the laboratory, to ~~include~~address:

i) population substructure,

ii) mutation rates,

~~ii)~~iii) known relatedness;

3) equation(s) in use by the laboratory, to include:

i) combined probability of inclusion,

ii) combined probability of exclusion,

iii) random match probability~~,~~ (or modification),

iv) likelihood ratio including formulating propositions;

4) operation of the software program(s) in use by the laboratory, including the underlying equations and review of the output data;

5) source attribution statements, if applicable; ~~and~~

6) limitations and assumptions of statistical method(s) in use by the laboratory.

~~e)~~f) *Statistical ~~Analysis~~analysis for Y-STR data*

1) detailed instruction on the calculation of haplotype frequencies using the counting method, to include:

i) consideration of the differences between the loci that the database samples are typed with and the loci in the amplification kit used by the laboratory,

ii) instruction on confidence intervals, Y-STR profile probabilities and Y-STR match probabilities,

iii) instruction on combining statistical values from autosomal and Y-STR data;

2) operation of the software program(s) in use by the laboratory~~.~~, including the underlying equations and review of the output data.

~~f)~~g) *Kinship ~~Analysis~~analysis*

1) statistical calculations for kinship associations including derivation and use, to include, as applicable:

i) the difference between alleles that are identical by state (IBS) or identical by descent (IBD),

ii) how to set-up competing propositions for kinship calculations~~, and~~ ;

iii) how to account for mutations in the kinship calculations;

2) determination of appropriate calculation for the case (identifying the unknown in the relationship scenario), to include:

i) maternity or paternity index,

ii) reverse parentage,

iii) sibship, avuncular or single grandparent index, ~~and~~

iv) complex family reconstruction.

### 4.3 Practical Training

**4.3.1** The practical component of the laboratory's training program shall provide the trainee with sufficient practical instruction for the trainee to obtain the skills ~~in~~for calculating ~~statistics~~statistical values for DNA data used by the laboratory ~~for the interpretation of forensic DNA evidence~~, to include, at minimum the components in 4.3.2 through 4.3.4:

~~**1.2.1** The protocol(s)~~At a minimum, the practical portion of the training program shall ~~be observed~~include the observation of a trained analyst performing the processes at least once~~.~~

**4.3.2** ~~NOTE This can be done by direct observation and supplemented with case file review~~ or until ~~the protocols are~~ clearly understood with exercises representative of the range, type, and complexity of DNA data from routine casework or database samples processed by the laboratory.

**4.3.3** Practical exercises shall be representative of the range, type, and complexity of routine DNA data from casework samples processed by the laboratory. Practical exercises shall include the following~~.~~:

a) ~~The~~the application of statistical analysis to the laboratory's own data~~.~~;

b) ~~Hand~~hand calculations for the following, as appropriate: RMP~~,~~ (or modification), single source LR, CPI/CPE, and kinship analysis likelihood ratios~~.~~;

c) ~~Exercises~~exercises to understand the derivation of the equations involved in the calculation for the following, as appropriate: parentage and kinship analysis likelihood ratios.

**4.3.4** The practical exercises performed shall be sufficient to demonstrate the trainee's ability to follow the laboratory's protocols and produce appropriate statistical values.

### 4.4 Competency Component

#### 4.4.1 General

The laboratory's training program shall include knowledge-based and practical competency in the laboratory's protocols for statistical applications. The format of the test(s) and the criteria for passing the competency test shall meet Section 4.3 of the ANSI/ASB ~~Std~~Standard 022, *Standard for Forensic Training DNA Analysis Training Programs*.

#### 4.4.2 Knowledge-Based Competency

~~The~~As applicable to the trainee's job responsibilities, the trainee shall successfully complete a knowledge-based test covering the critical information obtained during the training ~~in the application of statistics. The format of the test(s) shall be at the discretion of the~~on case record management, forensic DNA ~~Technical Leader.~~report writing, and performing technical and administrative reviews. The test(s) shall cover, at a minimum, the topics outlined ~~under~~in 4.~~1~~2 and its subsections.

#### 4.4.3 Practical Competency

The trainee shall successfully complete a practical competency test covering each of the statistical applications the trainee will be independently authorized to perform. DNA data from samples representative of the range, type, and complexity for which the trainee will be authorized to perform statistical calculations shall be included in the practical competency test.

## 5 Conformance

In order to demonstrate conformance with this standard, the laboratory shall meet the requirements outlined in section 5 of ANSI/ASB Std 022 Standard for Forensic DNA Analysis Training Programs and all the requirements set forth in this document.

# Annex A
(informative)

# Bibliography

The following information provides a list of the resources that may assist the DNA ~~Technical~~technical leader in defining the breadth and scope of the materials to be reviewed by the trainee. This list is not meant to be all inclusive.


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