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**Standard for Training in Statistical Calculations Used for
Forensic Short Tandem Repeat (STR) DNA Data**



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Standard for Training in Statistical Calculations Used for Forensic Short Tandem Repeat (STR) DNA Data

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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.

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

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Standard for Training in Statistical Calculations Used for Forensic Short Tandem Repeat (STR) DNA Data

1 Scope

This standard outlines the minimum requirements for a training program in the use of statistical calculations and values reported for forensic autosomal and Y short tandem repeat (STR) DNA 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

The likelihood ratio that evaluates the hypothesis that the tested individual is the biological uncle (or aunt or niece or nephew) of the profile donor versus the hypothesis that the tested individual is unrelated to the profile donor.

3.2

Combined Probability of Exclusion CPE

The probability that a randomly selected individual would be excluded 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 exclusion is $1 - \prod_j (1 - P_j)$.

3.3

Combined Probability of Inclusion CPI

The 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 $\prod_j (1 - P_j)$.

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.

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

37 **3.5**38 **haplotype**

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

43 **3.6**44 **Hardy-Weinberg equilibrium**

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

52 **3.7**53 **Identical by Descent**54 **IBD**

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

57 **3.8**58 **Identical by State**59 **IBS**

60 Identical alleles that may or may not be copies of the same ancestral allele.

61 **3.9**62 **inbreeding**

63 Mating of two persons who are more closely related than if they were chosen at random. It
64 increases the frequency of homozygous genotypes above the expected for a randomly mating
65 population in Hardy-Weinberg equilibrium.

66 **3.10**67 **kinship analysis**

68 Comparison of genetic profiles of two or more individuals to evaluate alternative degrees of
69 relatedness.

70 **3.11**71 **Likelihood Ratio**72 **LR**

73 A likelihood ratio is defined by a ratio of two conditional probabilities: the probability of the
74 evidence given each of two mutually exclusive and competing propositions. In forensic science
75 applications, the likelihood ratio is used as an expression for the meaning of scientific evidence and
76 as measure for its value.

77 *ASTM E1732-12[mod]²*

² Available from ASTM, at www.astm.org

78 3.12**79 linkage equilibrium**

80 Linkage equilibrium describes the situation in which the haplotype frequencies in a population
81 have the same value that they would have if the alleles at each locus were combined at random. If
82 both Hardy-Weinberg and linkage equilibrium hold, then random match probabilities may be
83 multiplied over loci.

84 3.13**85 mutation rate**

86 The relative frequency at which mutations have been observed at a specific genetic locus; generally
87 estimated as the number of mutations observed in parent-offspring pairs divided by the total
88 number of pairs examined.

89 3.14**90 Paternity or Maternity Index****91 PI/MI**

92 The likelihood ratio that evaluates the hypothesis that the tested individual is the biological mother
93 or biological father of the profile donor versus the hypothesis that the tested individual is unrelated
94 to the profile donor.

95 3.15**96 Random Match Probability****97 RMP**

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

100 3.16**101 reverse parentage**

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

106 3.17**107 sibship index**

108 Likelihood ratio in which the numerator is conditioned on the hypothesis that a sibling of the
109 source of the questioned profile in a specimen, and the denominator is conditioned on the
110 hypothesis that an unrelated individual is the source.

111 3.18**112 source attribution**

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

116 3.19**117 theta correction**

118 A value used to adjust statistical calculations that rely on population databases to correct for
119 substructures within populations.

120 4 Requirements

121 4.1 General

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

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

128 4.2 Knowledge-Based Training

129 **4.2.1** At a minimum, the knowledge-based portion of the training program shall require review of
130 the following:

- 131 a) the laboratory's protocols for statistical applications;
- 132 b) the laboratory's applicable validation studies;
- 133 c) literature used to support specific calculations and their use in appropriate circumstances;
- 134 d) applicable literature as assigned by the trainer;
- 135 e) literature on the effects of cognitive bias in decision-making processes associated with
136 statistical calculations used for forensic STR DNA data.

137 **4.2.2** The knowledge-based training component of the laboratory's training program shall
138 provide the trainee with a basic understanding of probability and statistics applied to autosomal
139 and Y-STR data to include, at minimum, the following topics.

- 140 a) *Population Genetics*
 - 141 1) laws of Mendelian genetics (law of segregation and the law of independent assortment);
 - 142 2) Hardy-Weinberg equilibrium;
 - 143 3) linkage equilibrium/disequilibrium;
 - 144 4) use of theta correction to adjust for inbreeding and population substructure;
- 145 b) *Statistical Foundations*
 - 146 1) frequency;
 - 147 2) probability;
 - 148 3) odds;
 - 149 4) the laws of probability (e.g., the addition rule and product rule);

- 150 5) Bayes' theorem.
- 151 6) Sources of uncertainty (e.g. modelling uncertainty and sampling variability)
- 152 c) *Population Allelic Frequency Databases*
- 153 1) population database size relative to the population size;
- 154 2) sample collection, to include:
- 155 i) number of samples,
- 156 ii) how population group was determined,
- 157 iii) how the database was created, maintained and reviewed,
- 158 iv) sampling uncertainty;
- 159 3) differences in allele frequencies observed between population databases;
- 160 4) mechanisms to account for alleles not observed in the database.
- 161 d) *Suitability of data for statistical application*
- 162 1) when to perform statistical analyses and which statistical calculation is validated for the
- 163 type of data obtained and comparison performed;
- 164 2) instruction on which loci to include in the statistical analyses when the following
- 165 circumstances are observed, at a minimum:
- 166 i) no allelic data,
- 167 ii) partial allelic data,
- 168 iii) tri-alleles, duplications/triplications, null alleles, and mutations,
- 169 iv) STR artifacts.
- 170 e) *Statistical analysis for autosomal STR data*
- 171 1) proper use of statistical calculations to include derivation and applicable equations;
- 172 2) statistical calculation method(s) in use by the laboratory, to address:
- 173 i) population substructure,
- 174 ii) mutation rates,
- 175 iii) known relatedness;
- 176 3) equation(s) in use by the laboratory, to include:

- 177 i) combined probability of inclusion,
 178 ii) combined probability of exclusion,
 179 iii) random match probability (or modification),
 180 iv) likelihood ratio including formulating propositions;
- 181 4) operation of the software program(s) in use by the laboratory, including the underlying
 182 equations and review of the output data;
- 183 5) source attribution statements, if applicable;
- 184 6) limitations and assumptions of statistical method(s) in use by the laboratory.
- 185 f) *Statistical analysis for Y-STR data*
- 186 1) detailed instruction on the calculation of haplotype frequencies using the counting method,
 187 to include:
- 188 i) consideration of the differences between the loci that the database samples are typed
 189 with and the loci in the amplification kit used by the laboratory,
- 190 ii) instruction on confidence intervals, Y-STR profile probabilities and Y-STR match
 191 probabilities,
- 192 iii) instruction on combining statistical values from autosomal and Y-STR data;
- 193 2) operation of the software program(s) in use by the laboratory, including the underlying
 194 equations and review of the output data.
- 195 g) *Kinship analysis*
- 196 1) statistical calculations for kinship associations including derivation and use, to include, as
 197 applicable:
- 198 i) the difference between alleles that are identical by state (IBS) or identical by descent
 199 (IBD),
- 200 ii) how to set-up competing propositions for kinship calculations;
- 201 iii) how to account for mutations in the kinship calculations;
- 202 2) determination of appropriate calculation for the case (identifying the unknown in the
 203 relationship scenario), to include:
- 204 i) maternity or paternity index,
 205 ii) reverse parentage,
 206 iii) sibship, avuncular or single grandparent index,

207 iv) complex family reconstruction.

208 **4.3 Practical Training**

209 **4.3.1** The practical component of the laboratory's training program shall provide the trainee with
210 sufficient practical instruction for the trainee to obtain the skills for calculating statistical values for
211 DNA data used by the laboratory, to include, at minimum the components in 4.3.2 through 4.3.4:

212 **4.3.2** At a minimum, the practical portion of the training program shall include the observation of
213 a trained analyst performing the processes at least once or until clearly understood with exercises
214 representative of the range, type, and complexity of DNA data from routine casework or database
215 samples processed by the laboratory.

216 **4.3.3** Practical exercises shall be representative of the range, type, and complexity of routine DNA
217 data from casework samples processed by the laboratory. Practical exercises shall include the
218 following:

- 219 a) the application of statistical analysis to the laboratory's own data;
- 220 b) hand calculations for the following, as appropriate: RMP (or modification), single source LR,
221 CPI/CPE, and kinship analysis likelihood ratios;
- 222 c) exercises to understand the derivation of the equations involved in the calculation for the
223 following, as appropriate: parentage and kinship analysis likelihood ratios.

224 **4.3.4** The practical exercises performed shall be sufficient to demonstrate the trainee's ability to
225 follow the laboratory's protocols and produce appropriate statistical values.

226 **4.4 Competency Component**

227 **4.4.1 General**

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

232 **4.4.2 Knowledge-Based Competency**

233 As applicable to the trainee's job responsibilities, the trainee shall successfully complete a
234 knowledge-based test covering the critical information obtained during the training on case record
235 management, forensic DNA report writing, and performing technical and administrative reviews.
236 The test(s) shall cover, at a minimum, the topics outlined in 4.2 and its subsections.

237 **4.4.3 Practical Competency**

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

242 **5 Conformance**

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

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247
248

Annex A (informative)

249

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251 in defining the breadth and scope of the materials to be reviewed by the trainee. This list is not
252 meant to be all inclusive.

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