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Standard for Familial DNA Searching

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DRAFT

Foreword

This document provides requirements for a laboratory conducting (or planning to conduct) familial DNA searching to identify a potential biological relative of an unknown individual whose DNA has been found at a crime scene or in a forensic investigation. Familial searching may be used in criminal investigations, missing persons cases, and cold cases after other investigative leads have been exhausted. It involves the use of STR-based likelihood ratio calculations to develop a candidate list of potential familial relationships between the unknown contributor of the evidence DNA sample and individuals in a DNA database. This document is designed to outline the procedures and requirements for familial DNA searching, without focusing on the source DNA testing or any additional confirmatory steps that may be necessary. Requirements for generating the initial searchable DNA profile and any confirmatory additional testing can be found in other published ANSI/ASB standards and the FBI Quality Assurance Standards for Forensic DNA Laboratories.

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.

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.

Questions, comments, and suggestions for the improvement of this document can be sent to AAFS-ASB Secretariat, asb@aafs.org or 410 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.

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Keywords: *familial searching, DNA, database*

Table of Contents *(to be finalized prior to publication)*

Standard for Familial DNA Searching

1 Scope

This standard provides the requirements for laboratories that perform familial DNA searches (searching laboratories) to have a policy specifying criteria for:

a) accepting a familial DNA search request;

b) administrative structure and responsibilities;

c) the search process;

d) reporting results; and

e) safeguarding individual privacy and confidentiality of the familial DNA search results.

This standard also defines validation requirements for familial DNA searches.

This standard does not apply to Forensic Investigative Genetic Genealogy or to the investigation of partial matches that may occur during the normal course of forensic database searches.

2 Normative References

The document contains no normative references. Annex A, Bibliography, contains informative references.

3 Terms and Definitions

For purposes of this document, the following definitions apply.

3.1
familial DNA search
familial DNA searching
A deliberate search of an offender DNA database [e.g. Combined DNA Index System (CODIS)] using software to detect and statistically rank a list of potential candidates whose DNA profile is in the database who could be close biological relatives (e.g., parent, child, sibling) of the unknown individual contributing the evidence DNA profile.

3.2
likelihood ratio
LR
The probability of the evidence under one proposition (hypothesis), divided by the probability of the evidence under an alternative, mutually exclusive proposition (hypothesis). The magnitude of its value expresses the weight of the evidence.

3.3
likelihood ratio threshold
The likelihood ratio below which a database profile specific to the relationship(s) under consideration would not be further investigated.

3.4

lineage testing

Genetic testing, such as YSTR, XSTR, or mitochondrial DNA analysis, used to trace maternal or paternal inheritance.

3.5

ranking threshold

The rank below which a database profile specific to the relationship(s) under consideration would not be further investigated.

NOTE For example, a laboratory may decide to investigate only a set number of the best ranked candidates regardless of the likelihood ratio.

3.6

searching laboratory

A laboratory that performs familial DNA searches.

3.7

sensitivity study

A validation assessment used to determine the proportion of familial searches within the test that detect true relatives of the evidence profile's known contributor at each ranking threshold.

3.8

source testing laboratory

An accredited laboratory that performs DNA testing on forensic evidence samples that are submitted for familial DNA searching and has ownership of the DNA data.

3.9

specificity study

A validation assessment used to determine the proportion of familial searches within the test that exclude true non-relatives of the evidence profile's known contributor.

NOTE Practical limitations can restrict this study to the initial statistical comparisons of simulated forensic unknowns to DNA database profiles.

4 Requirements

4.1 Policy and Procedure

4.1.1 The searching laboratory shall have documented policies and/or procedures ("the document") for conducting familial DNA searching that contains the information detailed in 4.1.2 through 4.1.11, as applicable to the laboratory's involvement in the elements described in each substandard. To aid the public and law enforcement in understanding the laboratory's familial search results, the familial search documents should be publicly available.

4.1.2 The document shall define the process for case submission and acceptance for conducting DNA familial searching. The following should be considered in the case acceptance policy:

a) seriousness or seriality of the crime;

b) commitment to proceed with investigation and prosecution;

c) case metadata and source testing laboratory notes provided as available;

d) investigative stage to initiate a familial search, such as, when other apparently viable investigative strategies have been exhausted;

e) the availability of additional DNA evidence; and

f) out of state search requests.

4.1.3 The document shall define the roles of the individuals involved in the DNA familial search process and oversight. An individual may fill more than one role. The familial DNA search process may involve individuals in the following roles:

a) a review committee (to evaluate requests, prioritize testing, verify results, and determine the propriety of moving forward with an investigation or disclosure of a name);

b) an individual or committee who has the ultimate control for the search and directs the release of investigative lead(s) and any follow-up;

c) an administrative representative from the source testing laboratory;

d) an administrative representative from the searching laboratory;

e) a technical representative from the source testing laboratory;

f) a technical representative from the searching laboratory;

g) a representative with access to investigative databases (metadata) typically restricted to law enforcement personnel;

h) a representative from the requesting law enforcement agency;

i) a representative from the prosecuting agency; and

j) a representative of the defendant when the search is being performed at the request of the defense.

4.1.4 The document shall define how information obtained through the familial search process is shared and released, and should include:

a) safeguards for the disclosure of sensitive information to the proper individuals; and

b) provisions protecting the privacy of possible family members in the search database who could not be the perpetrator.

4.1.5 The document shall ensure that all the individuals with roles established in 4.1.3 are informed of the process, likely outcomes, limitations, and the need for additional investigation, including source testing laboratory policies and procedures for acceptance, testing, and comparison of possible confirmatory samples. This could be achieved through the use of a documented memorandum of understanding or equivalent.

4.1.6 The document shall define the acceptance requirements of the evidence profile to be searched to include the following:

a) the DNA sample was recovered directly from the crime scene (or associated with the crime scene) and is attributed to the putative perpetrator;

b) the minimum threshold for the number of searchable loci and/or profile rarity;

c) the acceptable level of potential allelic dropout in the profile submitted for the familial DNA search;

d) the number of possible contributors, e.g., single source, deduced single source from a mixture, or mixtures up to a defined maximum number of contributors; and

e) a requirement that a direct comparison search of the profile has occurred in relevant databases (e.g., CODIS) prior to the familial DNA search being conducted.

4.1.7 The document shall define which database categories will be searched (e.g., convicted offenders index).

4.1.8 The document shall define which relationships (e.g., 1st degree relatives only) will be considered.

4.1.9 The searching laboratory shall document situations where, and justify conditions when, conducting additional testing is not required.

NOTE Where needed, the source testing laboratory may conduct expanded autosomal STR (beyond the original 13 CODIS core loci) and/or lineage testing (e.g., YSTR, XSTRs and/or mitochondrial DNA) of potential relatives.

4.1.10 The document shall define the thresholds (e.g., likelihood ratio and/or ranking) for proceeding with the release of information regarding potential relatives.

4.1.11 The document shall define the criteria for the release of the name(s) of the potential relative(s). The following criteria may be considered prior to the release of the name(s) of the potential relative(s):

a) the use of metadata for the evaluation of a potential relative, and

b) assessing kinship using non-invasive investigation, i.e., one that does not require direct contact with a person of interest.

4.2 Technical

4.2.1 The searching laboratory shall use internally validated analytical procedures/systems and software.

4.2.2 Internal validation shall include sensitivity and specificity studies to include the range of search profiles defined in 4.1.6 b), 4.1.6 c), and 4.1.6 d).

4.2.2.1 A reasonable sensitivity test will seed a database of unrelated individuals with the profiles of true relatives, at a minimum, those defined in section 4.1.8, of the experimental search profile. Ranking the LRs for all comparisons from highest to lowest, sensitivity is defined as the proportion of true relatives, real or in silico, included at each ranking threshold. Alternatively, the ranked list of unrelated individuals' LRs can be seeded with the LRs for each of the true family members, thus avoiding the insertion of experimental profiles into the database.

4.2.2.2 A reasonable specificity test will examine how many individuals remain as candidates after the statistical process (e.g., the initial LR rankings based solely upon autosomal STR loci). Coupled with how many of those individuals meet the requirements to be tested with additional laboratory work, will give an estimate of how likely it will be to see a false positive. Knowing whether the statistical process the searching laboratory has put in place will lead to tens of candidates, hundreds of candidates, or thousands of candidates will give a sense of whether the subsequent lineage testing, or additional autosomal markers, might adventitiously include a non-relative.

4.2.3 Search criteria developed from sensitivity and specificity studies should be established to err on the side of minimizing false positives. This approach enhances the probability of releasing reliable leads.

NOTE Search criteria developed from sensitivity and specificity studies can be impacted by the capacity and resources of the searching laboratory in addition to the size of the database being searched.

4.2.4 The searching laboratory shall determine if the sensitivity and specificity process will incorporate the following:

a) population substructure (Fst or theta);

b) mutation model;

c) silent (null) allele model;

d) correction for linkage;

e) probabilistic genotyping; and

f) prior odds/probability.

4.2.5 The searching laboratory shall have a documented training program specific to familial DNA searching that includes training in kinship analysis.

Training of an individual evaluating familial search results should include the following concepts:

a) the expected levels of DNA sharing for various relationships, including the flow of DNA through pedigrees and lineage vs. autosomal patterns of inheritance;

b) the calculations performed in familial searching, including likelihood ratios and conditional probabilities given an assumed level of relatedness;

c) likelihood ratio distributions for various relationship categories;

d) evaluating pedigrees larger than two-person comparisons;

e) addressing the issues of linked loci and meiotic mutations;

f) the effect of population database selection on the likelihood ratio;

g) the effect of database size on the ranking of true relatives; and

h) the benefits and limitations of lineage vs. autosomal loci.

Annex A
(informative)

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