OSAC 2021-S-0029 Standard for Familial DNA Searching

Forensic Human Biology Subcommittee
Biology SAC
Organization of Scientific Area Committees (OSAC) for Forensic Science

Draft OSAC Proposed Standard

OSAC 2021-S-0029
Standard for Familial DNA Searching

Prepared by
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This OSAC Proposed Standard was written by the Human Forensic Biology Subcommittee of the Organization of Scientific Area Committees (OSAC) for Forensic Science following a process that includes an open comment period. This Proposed Standard will be submitted to a standards developing organization and is subject to change.

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To be placed on the OSAC Registry, certain types of standards first must be reviewed by a Scientific and Technical Review Panel (STRP). The STRP process is vital to OSAC's mission of generating and recognizing scientifically sound standards for producing and interpreting forensic science results. The STRP shall provide critical and knowledgeable reviews of draft standards or of proposed revisions of standards previously published by standards developing organizations (SDOs) to ensure that the published methods that practitioners employ are scientifically valid, and the resulting claims are trustworthy.

The STRP panel will consist of an independent and diverse panel, including subject matter experts, human factors scientists, quality assurance personnel, and legal experts, which will be tasked with evaluating the proposed standard based on a comprehensive list of science-based criteria.

For more information about this important process, please visit our website at: https://www.nist.gov/topics/organization-scientific-area-committees-forensic-science/scientific-technical-review-panels.

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Standard for Familial DNA Searching

1. Scope

This standard requires laboratories that perform familial DNA searches to have a written policy with criteria for:

a) accepting a familial DNA search request, administrative structure, and responsibilities;
b) the search process, data verification, and validation;
c) reporting results; and
d) safeguarding individual privacy and confidentiality of the results.

2. Normative References

The document contains no normative references. See Annex A, Bibliography for other references.

3. Terms and Definitions

For the purposes of this document the following definitions apply:

3.1
familial DNA search
familial DNA searching
A deliberate search of a DNA database using software to detect and statistically rank a list of potential candidates in the database who may be close biological relatives (e.g. parent, child, sibling) to 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 a 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. For example, a laboratory may decide to investigate only those candidates above a certain likelihood ratio.

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. For example, a laboratory may decide

to investigate only a set number of the best ranked candidates regardless of the likelihood ratio.

3.6

sensitivity study

An assessment of the proportion of familial searches that detect true relatives of the contributor of the evidence profile when one or more relatives is present in the database.

3.7

specificity study

An assessment of the proportion of familial searches that exclude non-relatives of the contributor of the evidence profile. Practical limitations may restrict this study to the initial statistical comparisons of simulated forensic unknowns to DNA database profiles.

4. Requirements

4.1 Policy and Procedure: The laboratory shall have documented policies and procedures for conducting familial DNA searching that shall contain the information detailed in 4.1.1 through 4.1. To aid the public and law enforcement in understanding the laboratory's familial search results, the familial search policy should be publicly available.

4.1.1 The policy shall define the process for case submission and acceptance for conducting DNA familial searching. The following may 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 laboratory notes provided as available;
d) investigative stage to initiate a familial search, such as, when viable reasonable; investigative strategies have been exhausted; and
e) the availability of additional DNA evidence.

4.1.2 The policy shall define the roles of the individuals involved in the DNA familial search process and oversight. The familial DNA search process may involve individuals in the following roles:

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

b) an administrative representative from the source testing laboratory;

c) an administrative representative from the searching laboratory;

d) a technical representative from the source testing laboratory;

e) a technical representative from the searching laboratory;

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

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

h) a representative from the requesting law enforcement agency; and

i) a representative from the prosecuting agency.

4.1.3 The policy shall define the interactions between the individuals defined in 4.1.2 so as to safeguard privacy. The policy may include the use of a documented memorandum of understanding or equivalent.

4.1.4 The policy 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 those family members who could not be the perpetrator.

4.1.5 The policy shall ensure that all the individuals defined in 4.1.2 are informed of the process, likely outcomes, limitations, and the need for additional investigation.

4.1.6 The policy shall address the collection and testing of the following comparison samples:

4.1.6.1 surreptitiously collected samples associated with the person of interest

4.1.6.2 samples known not to be the perpetrator but may provide additional confirmatory information (e.g. possible relatives of the perpetrator), whether collected surreptitiously or voluntarily

4.1.6.3 confirmatory samples

4.1.7 The policy shall define the acceptance requirements of the evidence profile to be searched to include the information in 4.1.7.1 through 4.1.7.5.

4.1.7.1 That the DNA sample was recovered directly from the crime scene and is attributed to the putative perpetrator.

4.1.7.2 The minimum threshold for the number of searchable loci and/or profile rarity

4.1.7.3 The acceptable level of potential allelic dropout in the profile submitted for the familial DNA search

4.1.7.4 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

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

4.1.8 The policy shall define which database categories will be searched (e.g. convicted offenders, arrestees, suspects).

4.1.9 The policy shall define which relationships will be considered.

4.1.10 When possible, the laboratory shall 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. The laboratory shall document situations where, and justify conditions when conducting additional testing is not required.

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

4.1.12 The policy 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).

4.1.12.1 The use of metadata for the evaluation of a potential relative

4.1.12.2 Assessing kinship using non-invasive investigation

4.2 Technical

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

4.2.2 Internal validation shall include sensitivity and specificity studies.

It is impractical to perform both sensitivity and specificity studies on the entire process with a meaningfully sized dataset when the process includes extensive additional testing and investigative work.

A reasonable sensitivity test would seed a database of unrelated individuals with the profiles of true relatives 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 rank. Alternatively, the ranked list of unrelated individuals' LRs could be seeded with the LRs for each of the true family members, thus avoiding the insertion of experimental profiles into the database.

A reasonable specificity test would 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 would meet the requirements to be tested with additional lab work, this will give an estimate of how likely it would be to see a false positive. Knowing whether the statistical process the lab has put in place will lead to tens of candidates, hundreds of candidates, or thousands of candidates will give a sense of whether or not the subsequent lineage testing (or additional autosomal markers) might adventitiously include a non-relative.

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

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

4.2.3 The laboratory shall determine if the process will incorporate the following:

4.2.3.1 mutation model

4.2.3.2 silent (null) allele model

4.2.3.3 correction for linkage

4.2.3.4 probabilistic genotyping

4.2.4 The laboratory shall have a documented training program specific to familial DNA searching including 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:
    i. the flow of DNA through pedigrees; and
    ii. lineage vs. autosomal patterns of inheritance
b). The calculations performed in familial searching, including:
    i. likelihood ratios; and
    ii. 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.
h). The benefits and limitations of lineage vs. autosomal loci.

Annex A

(informative)

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