

OSAC RESEARCH NEEDS ASSESSMENT FORM



Title of research need:

Improved software tools in support of forensic DNA interpretation

Describe the need:

DNA mixture interpretation remains one of the most significant challenges in forensic DNA analysis. Probabilistic genotyping, probabilistic number of contributors, and artifact identification systems have been developed. However, it is incumbent upon the forensic DNA community to continue the pursuit of high quality and reliable software as chemistries, methods, targets, and platforms may change. This need addresses both the evolution of current techniques and the innovation of new techniques. The focus of this research and development need includes the development of new, and comparison of current computational and statistical approaches used in the analysis of standard nuclear STR systems for both fragment and sequence analysis, Y-STRs, phenotyping, ancestry and mitochondrial DNA, SNPs, and microhaplotypes. Third party agnostic software to assist with the validation of existing software is currently lacking. Such tools would be useful for internal validations, both for planning validation experiments, and for documentation that can be easily reviewed and shared. It is expected that Artificial Intelligence/Machine Learning tools will be forthcoming, but it is important that these tools do not replace individual human expertise or be used as "black box" software programs that provide interpretations with no human interaction or ownership instead of assisting human experts with interpretation.

Keyword(s):

probabilistic genotyping, DNA mixture, number of contributors, artifacts, NGS, MPS

Submitting subcommittee(s):

Human Biology

Date Approved:

05/16/2025

Background Information:

1. Does this research need to address a gap(s) in a current or planned standard? (ex.: Field identification system for on scene opioid detection and confirmation)

Yes.

2. Are you aware of any ongoing research that may address this research need that has not yet been published (e.g., research presented in conference proceedings, studies that you or a colleague have participated in but have yet to be published)?

Yes.

3. Key bibliographic references relating to this research need: (ex.: Toll, L., Standifer, K. M., Massotte, D., eds. (2019). Current Topics in Opioid Research. Lausanne: Frontiers Media SA. doi: 10.3389/978-2-88963-180-3)

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4. Review the annual operational/research needs published by the National Institute of Justice (NIJ) at <https://nij.ojp.gov/topics/articles/forensic-science-research-and-development-technology-working-group-operational#latest>? Is your research need identified by NIJ?

Yes, “Improved methods for identifying the number of contributors and mixture interpretation algorithms for all markers (STRs, sequence-based STRs, Y-STRs, mitochondrial, microhaplotypes, SNPs) to include statistical considerations for combining marker types” and “Probabilistic haplotyping tool for mixture interpretation of lineage markers (Y-STRs, mitochondrial) and/or methods by which to statistically evaluate mixture profiles (Y-STRs, mitochondrial)”.

5. In what ways would the research results improve current laboratory capabilities?

The development of new software tools will meet the needs of an evolving technological climate in forensics, where new chemistries, instrumentation and targets are being evaluated and implemented. New methods may be developed that improve upon existing software solutions, thus enabling higher confidence in interpretation and conclusions. Finally, assessments of currently used software solutions will enable laboratories to make more informed decisions regarding implementation.

6. In what ways would the research results improve understanding of the scientific basis for the subcommittee(s)?

The development and assessment of software methods for forensic DNA interpretation lead to a better understanding of the underlying biology, the process related components such as sample collection, DNA isolation and purification, amplification and detection. This will also prepare the subcommittee for the development of new standards that will address the use of new chemistries, instruments or targets.

7. In what ways would the research results improve services to the criminal justice system?

The chemistries, methods, targets, and platforms used in forensic DNA analysis may change leading to a need to adapt or develop new methods for probabilistic methods to assess the resulting profiles. Ultimately this research needs advocates for the continuous development of software that offers quality improvements to the “toolbox” that forensic DNA analysts use to aid in judgment and decision making.

8. Status assessment (I, II, III, or IV):

IV

	Major gap in current knowledge	Minor gap in current knowledge
No or limited current research is being conducted	I	III
Existing current research is being conducted	II	IV

This research need has been identified by one or more subcommittees of OSAC and is being provided as an informational resource to the community.