OSAC RESEARCH NEEDS ASSESSMENT FORM

Title of research need: Software solutions for low template and high order DNA mixture interpretation in sequence and fragment-based methods

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.

Keyword(s): probabilistic genotyping, DNA mixture, number of contributors, artifacts, NGS, MPS

Submitting subcommittee(s): Human Biology **Date Approved:** 10/05/2021

(If SAC review identifies additional subcommittees, add them to the box above.)

Background Information:

1. Does this research need 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:

1) Alladio, E. et al. DNA mixtures interpretation – A proof-of-concept multi-software comparison highlighting different probabilistic methods' performances on challenging samples. Forensic Sci. Int. Genet. 37, 143–150 (2018).
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4) Bille, T. W., Weitz, S. M., Coble, M. D., Buckleton, J. & Bright, J. A. Comparison of the performance of different models for the interpretation of low level mixed DNA profiles. Electrophoresis 35, 3125–3133 (2014).
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6) Bleka, Ø., Storvik, G. & Gill, P. EuroForMix: An open source software based on a continuous model to evaluate STR DNA profiles from a mixture of contributors with artefacts. Forensic Sci. Int. Genet. 21, 35–44 (2016).
7) Marciano, M. A. & Adelman, J. D. PACE: Probabilistic Assessment for Contributor Estimation- A machine learning-based assessment of the number of contributors in DNA mixtures. Forensic Sci. Int. Genet. 27, 82–91 (2017).
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12) Adelman, J. D., Zhao, A., Eberst, D. S. & Marciano, M. A. Automated detection and removal of capillary electrophoresis artifacts due to spectral overlap. Electrophoresis elps.201900060 (2019). doi:10.1002/elps.201900060
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14) Coble, M. D. et al. DNA Commission of the International Society for Forensic Genetics: Recommendations on the validation of software programs performing biostatistical calculations for forensic genetics applications. Forensic Sci. Int. Genet. 25, 191–197 (2016).
15) Gill, P., Kirkham, A. & Curran, J. LoComatioN: A software tool for the analysis of low copy number DNA profiles. Forensic Sci. Int. 166, 128–138 (2007).
16) Gill, P. et al. DNA commission of the International Society of Forensic Genetics: Recommendations on the evaluation of STR typing results that may include drop-out and/or drop-in using probabilistic methods. Forensic Sci. Int. Genet. 6, 679–688 (2012).
17) Perlin, M. W. et al. Validating TrueAllele® DNA Mixture Interpretation*,†. J. Forensic Sci. 56, 1430–1447 (2011).
18) Bauer, D. W., Butt, N., Hornyak, J. M. & Perlin, M. W. Validating TrueAllele ® Interpretation of DNA Mixtures Containing up to Ten Unknown Contributors. J. Forensic Sci. 65, 380–398 (2020).
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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 need advocates for the continual development of software that offers quality improvements to the "toolbox" that forensic DNA analysts use to aid in judgement 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.