OSAC RESEARCH NEEDS ASSESSMENT FORM

Title of research need:	Software solutions for Y-STR mixture deconvolution
Describe the need:	Much progress has been made for the analysis of autosomal STR mixtures including the development of software solutions for mixture deconvolution that accommodate the probability of allelic drop out and drop in for low-level and degraded profiles. Very little research has progressed for best practices for the deconvolution of Y-STR mixtures and/or the comparison of known samples to Y-STR mixtures. This research should specifically include work on rapidly mutating Y-STRs and male relatives, where a low amount of differentiating is expected.
Keyword(s):	Y-STR mixtures, Continuous Probabilistic Genotyping, Likelihood Ratio

Submitting subcommittee(s):	Human Biology	**Date Approved:**	05/04/2021

(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)

Currently no standards address Y-STR mixtures. This is needed.

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)?

There are various Y-STR mixture tools, but most have no real support and there is no agreed upon model or calculations.

3. Key bibliographic references relating to this research need:

1) Scientific Working Group on DNA Analysis Methods Interpretation Guidelines for Y-Chromosome STR Typing (approved on January 9, 2014) - http://media.wix.com/ugd/4344b0_da25419ba2dd4363bc4e5e8fe7025882.pdf
2) Taylor D, Bright J-A, and Buckleton J (2016) Using probabilistic theory to develop interpretation guidelines for Y-STR profiles. *FSI-Genetics* 21:22-34.
3) Andersen MM, Eriksen PS, Mogensen HS, and Morling N (2015) Identifying the most likely contributors to a Y-STR mixture using the discrete Laplace method. *FSI-Genetics* 15:76-83.
4) Ballantyne KN, Keerl V, Wollenstein A et al (2012) A new future of Forensic Y-chromosome analysis: rapidly mutating Y-STRs for differentiating male relatives and paternal lineages. *FSI-Genetics* 6:208-218.

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*

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

Approximately 60% of the crime labs in the U.S. conduct Y-STR testing (Doug Hares, NDIS Custodian, personal communication). There is very little guidance to the forensic community on how to interpret indistinguishable mixed Y-STR profiles. The 2014 SWGDAM guidelines for Y-STR typing states, "SWGDAM has not yet reached consensus, however, on the appropriate statistical approach for estimating the occurrence of a combination of haplotypes in a population." Research to understand the underlying behavior of Y-STR mixtures is sorely needed and would allow laboratories to interpret and report these mixtures.

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

Many of the core Y-STR loci suffer from low resolution in a population much like some of the low discriminating autosomal loci (e.g. TPOX). Allele sharing can increase the uncertainty of being able to determine the number of contributors. There is no guidance on establishing mixture ratios for the deconvolution of two (or more) mixed males. For example, is the "major" contributor at a 5:1 ratio consistent across the entire profile? There is also not enough data on other mixture parameters like number of contributors, effect of degradation, and stutter. Presently, statistics for Y-STR mixtures use a counting method calculation of how many individuals in the database could not be excluded. This approach could be improved by pursuing probabilistic genotyping. Software solutions for deconvoluting mixtures and the interpretation of missing (drop out/in or the effect of null alleles) data is needed.

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

One advantage of Y-STR testing is the usefulness for resolving mixtures where there is a high female background and low male profile (in an autosomal STR mixture). Given the focus on processing untested sexual assault kits, having more research and software tools to help resolve multiple male contributor Y-STR mixtures would benefit the criminal justice system.

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

	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.