

OSAC RESEARCH NEEDS ASSESSMENT FORM



Title of research need:

Methods in Forensic Investigative Genetic Genealogy

Describe the need:

While close family relationships may be inferred from STR data using familial searching of STR databases, more distant relationships can be found using Forensic Investigative Genetic Genealogy (FIGG) (dependent on the country/state legislation). FIGG combines the use of dense genome-wide single nucleotide polymorphism (SNP) profiles with traditional record-based family tree genealogy. A familial genetic connection is determined by searching the SNP profile [generated from whole genome sequencing (WGS), microarray hybridization, target hybridization capture, and targeted sequencing methods] obtained from a crime scene sample or unidentified human remains against a publicly-available database [i.e., profiles from direct-to-consumer (DTC) kits] and then performing genealogy searches with birth, death, marriage and other records and social media, with the intention of generating actionable intelligence. Currently, database searching can only occur in privately-held databases (such as FamilyTree DNA and GEDmatch PRO) with proper user consent. There are only limited assessments of the database algorithms and the quality of the match lists of potential relatives, despite the active research and improvements in the technical aspects of generating SNP typing data. Potential areas of evaluation to be explored include:

1. Performance metrics appropriate to the SNP genotyping method (e.g., WGS, microarray, targeted sequencing) to assess the quality of the profile (e.g., number of SNPs, heterozygosity), and what the expected metric ranges are for high- and low-quality samples to establish thresholds.

2. Performance metrics for generated match lists from the database searches and how they relate to the genotype performance and to the downstream genealogy family trees. Potential metrics to consider include the length of the lists (i.e., possibly highlighting false positives), are matches reproduced with replicate processing, and what is the furthest distance relative that can be detected.

3. Evaluate performance metrics for SNP profiles and the generated match lists from database searches for deconvoluted profiles from mixture samples.

FIGG assessments often disconnect between the generation of the SNP profile and the genealogical results and subsequent investigation. To fully understand the performance of SNP genotyping methods for application to FIGG, researchers will need to embrace this technique's multidisciplinary nature.

Keyword(s):

SNP Genotyping, Massively Parallel Sequencing, Microarray Hybridization, Whole Genome Sequencing, FIGG, genealogy,

Submitting subcommittee(s):

Human Biology

Date Approved:

05/16/2025

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, in the use of Forensic Investigative Genetic Genealogy for casework.

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, but not enough in detail.

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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3. Galván-Femenía, I., Barceló-Vidal, C., Sumoy, L. *et al.* A likelihood ratio approach for identifying three-quarter siblings in genetic databases. *Heredity.* 2021;126, 537–547.
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5. Thomson J, Clayton T, Cleary J, Gleeson M, Kennett D, Leonard M, Rutherford D. An empirical investigation into the effectiveness of genetic genealogy to identify individuals in the UK. *Forensic Sci Int Genet.* 2020;46:102263.
6. Turner SD, Nagraj, VP, Scholz, M, Jessa, S, Acevedo, C, Ge, J, Woerner, AE, & Budowle, B. Evaluating the Impact of Dropout and Genotyping Error on SNP-Based Kinship Analysis With Forensic Samples. *Frontiers in Genetics.* 2022;13: 882268.
7. Wang, M, Chen, H, Luo, L, Huang, Y, Duan, S, Yuan, H, Tang, R, Liu, C, & He, G. Forensic investigative genetic genealogy: Expanding pedigree tracing and genetic inquiry in the genomic era. *Journal of Genetics and Genomics.* 2024;S1673852724001589.
8. Woerner, AE, Novroski, NM, Mandape, S, King, JL, Crysup, B, & Coble, MD. Identifying distant relatives using benchtop-scale sequencing. *Forensic Science International: Genetics.* 2024;69:103005.

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, “Development of genetic genealogy testing procedures for use by the crime labs”.

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

It is important to understand the performance of different genotyping methods with routine and challenging sample types. Further, it is necessary to understand what can be expected with the match lists in terms of the potential of what degrees of relationship could be found (if present in the database) and how many potential

false positives are present that could extend the genealogical research and investigation. This will allow laboratories to make informed processing decisions.

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

Detailed characterization of the performance of SNP genotyping methods and the generated match lists to generate investigative leads is critical for understanding this technology and to guide standards around its implementation.

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

It will provide characterization that can help lead to standards for current and future implementation of FIGG as a tool for generating forensic investigative leads.

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.