



### **B59 Genomic Approaches to the Identification of Individuals Through Familial Database Searches**

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After attending this presentation, attendees will learn how to use familial searching to generate investigative leads that would otherwise go undetected by the current database searching methods. Additionally, attendees will learn how adding Y-STRs and mitochondrial DNA sequencing to the CODIS autosomal STRs improves the discrimination power of familial searching.

This presentation will impact the forensic community by providing an objective assessment of the utility and limitations of familial searching as an investigative tool to aid in the identification of perpetrators of no-suspect and cold cases and also to identify victims of mass disasters and human rights abuses.

Forensic DNA databases (e.g., CODIS) are routinely used to identify the perpetrator of a crime by comparing the unknown biological evidence profile to the known convicted offender profiles in the database. The most common type of database search is referred to as a high stringency match search, where the evidence and offender profiles must match at all loci to produce a hit. However, instances occur when two compared profiles share at least one allele at each locus; this type of hit results from a moderate stringency match, which requires at least one allele at all loci in common between the two profiles. Due to the highly similar yet non-identical profiles generated from these moderate stringency searches, the law enforcement and forensic science communities have recognized that these partial matches may indicate a potential close familial relationship, such as parent and offspring, and these matches could be valuable investigative leads to find the true perpetrator. Until recently, the FBI did not allow the release of the identity of an offender that partially matches a crime sample from another state's search. Furthermore, most states do not have policies regarding the reporting of partial profile matches resulting from database searches. Consequently, these moderate stringency matches—some of which may be true relatives—are not routinely evaluated in most states.

Although the CODIS search algorithm can identify individuals with shared alleles, CODIS was not purposely designed to identify relatives. An alternative approach that specifically aims to identify family members, called familial searching, has been used to identify victims of mass disasters, human rights violations, and missing persons cases. Before familial searching methods are employed in criminal cases, it is imperative to evaluate the discriminatory power of the CODIS STRs to identify potential relatives. It has been previously demonstrated the high proportion of false positive leads (i.e., unrelated individuals who appear related by chance) that result from familial searches of profiles with 13 STRs. A majority of these false positives will likely be dismissed upon law enforcement investigation. However, with the national database rapidly approaching 5 million profiles, investigation of the thousands of putative relatives generated from a familial search would be inefficient and ineffective.

In order to decrease the proportion of false positive matches and subsequent unnecessary investigation, Y-STR and mitochondrial DNA (mtDNA) analysis can be added to the CODIS STRs. Although these lineage markers are not fully informative for positive individual identification, Y-STRs, and mtDNA can be highly informative for identifying unique, family-specific haplotypes and sequences. We can exploit the lineage-specific nature of these markers and the high incidence of unique haplotypes to reduce the number of false positive matches that result from familial searches using only autosomal loci. The addition of Y-STR and mtDNA analysis to the CODIS loci enables more accurate identification of potential close relatives by excluding individuals who cannot be related to the true perpetrator either through a paternal or maternal lineage. Upon Y-STR and mtDNA analysis of individuals after a familial search of their autosomal STR profiles, we observe a dramatic decrease in the proportion of database individuals that require further investigation. The addition of lineage-specific markers to the CODIS autosomal STRs will allow greater discriminatory power than familial searching with CODIS markers alone. The acceptable level of discrimination has yet to be determined by the forensic and law enforcement communities.

#### **Familial Searching, Offender Database, DNA Identification**