MITOSEARCH: SOFTWARE FOR MANAGING AND SEARCHING MITOCHONDRIAL DNA PROFILES

Keith L. Monson, Kevin W.P. Miller, and Bruce Budowle
Federal Bureau of Investigation, Forensic Science Research Unit, Quantico, VA

MitoSearch is an integrated software package designed for the management and analysis of mitochondrial DNA (mtDNA) nucleotide sequence data from various populations worldwide, and for the assessment of the relative frequency of forensic mtDNA profiles. The software is comprised of two main programs, presented on two screens, Search and Pairwise. The Search program allows the user to search an evidentiary mtDNA nucleotide sequence against nucleotide sequence data in the population database. The Pairwise module compares every profile to every other profile in the data sets selected. Customization of the database search or comparison features is accomplished via an Options screen in both programs.

Many options are available to define the search conditions and program output, including: defining the data set and which of the defined population groups within it are to be searched, limiting the profiles to be used for searching by their date of entry into the database, defining search ranges, specifying whether profiles that only partially overlap the sequence range are to be included, stipulating the treatment of sequence ambiguities and insertions, and denoting the degree of detail appearing in the output files. Three output file formats are produced; HTML (web browser), text (word processor) and comma-delimited (spreadsheet).

MitoSearch provides access to two population databases, forensic and published. The forensic (SWGDAM) database comprises anonymous population profiles of chiefly forensic interest. Quality of mtDNA nucleotide sequence data in the forensic database is closely controlled, and contributions from laboratories conducting forensic mtDNA analyses is encouraged. Before accepting database profiles from a laboratory that wishes to submit its results for inclusion in the forensic database, we ask that a series of control samples be typed, and that the results be returned to us. Exchange of these control samples assures compatibility of typing methods and uniformity of base calling conventions for the profiles in the forensic database. Upon successful completion, the FBI then provides the laboratory with a current version of MitoSearch and the current database of nucleotide sequence data.

Two additional databases that will be accessible with MitoSearch are in development. A “public” database will consist of published mtDNA sequence data that, on an ongoing basis, will be collected, cataloged, annotated and organized by the FBI. Another will provide the capability to store and search databases of mtDNA profiles of missing persons, unidentified human remains and living persons seeking a missing family member.

The FBI has used MitoSearch in casework for the past three years. In addition, MitoSearch and its associated database of population profiles have been distributed to approximately twenty forensic laboratories worldwide. The FBI currently updates and distributes MitoSearch and the database twice yearly. Currently, MitoSearch is a standalone program, but a CODIS version is under development. The CODIS version will allow searches of combined mitochondrial and nuclear DNA profiles, and searching of open case files across the CODIS network.