Mitochondrial DNA Typing in the FBI Laboratory: Casework Experience and Future Directions

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The FBI Laboratory implemented mtDNA sequencing on casework samples in June 1996. The technique is particularly useful for hair evidence. Initially, the FBI Laboratory only tested hairs which were associated through comparative microscopy. However, it soon became clear that the technique had a potentially wider application, specifically, in addition to other kinds of human biological evidence, it could be applied to hairs which are not suitable for significant microscopic comparison purposes.

Four overlapping segments of the human mtDNA control region are amplified by PCR and sequenced with an automated DNA sequencer. The quality and quantity of the PCR products are assessed through laser-induced fluorescence of DNA separated by capillary electrophoresis.

Two major areas of concern in mtDNA testing are the potential for contamination and heteroplasmy. Potential contamination is monitored through judicious use of reagent blank and negative control amplifications. Heteroplasmy is a biological aspect of mtDNA genetics which is becoming better understood by both the general scientific and forensic communities. In heteroplasmic individuals, point or length mutations are observed in differing ratios from tissue to tissue, and from hair to hair. The presence of heteroplasmy in an individual or maternal lineage can complicate sequence comparisons. However, careful assessment of known samples, as well as an understanding of the segregational mechanics of heteroplamy and mtDNA population genetics, assists the examiner in interpreting sequence information from such individuals. Properly understood and interpreted, the presence of heteroplasmy at a particular position greatly enhances the exclusionary power of a sequence match.

Approximately 70 cases utilizing mtDNA technology have been worked by the FBI Laboratory since June 1996. Expert testimony has been provided in seven states. The technique continues to be improved and refined, and has proven to be a robust and sensitive DNA typing technique.