Mitochondrial DNA Sequence Analysis — Validation and Use for Forensic Casework a,b


ABSTRACT: With the discovery of the polymerase chain reaction (PCR) in the mid-1980’s, the last in a series of critical molecular biology techniques (to include the isolation of DNA from human and non-human biological material, and primary sequence analysis of DNA) had been developed to rapidly analyze minute quantities of mitochondrial DNA (mtDNA). This was especially true for mtDNA isolated from challenged sources, such as ancient or aged skeletal material and hair shafts. One of the beneficiaries of this work has been the forensic community. Over the last decade, a significant amount of research has been conducted to develop PCR-based sequencing assays for the mtDNA control region (CR), which have subsequently been used to further characterize the CR. As a result, the reliability of these assays has been investigated, the limitations of the procedures have been determined, and critical aspects of the analysis process have been identified, so that careful control and monitoring will provide the basis for reliable testing. With the application of these assays to forensic identification casework, mtDNA sequence analysis has been properly validated, and is a reliable procedure for the examination of biological evidence encountered in forensic criminalistic cases.

KEY WORDS: DNA sequencing, forensic science, mtDNA, PCR.