Forensic Mitochondrial DNA Analysis: Current Practice and Future Potential

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ABSTRACT: Current practices for performing forensic mitochondrial DNA (mtDNA) sequence analysis, as employed in public and private laboratories across the United States, have changed remarkably little over the past 20 years. Alternative approaches have been developed and proposed, and new technologies have emerged, but the core methods have remained relatively unchanged. Once DNA has been recovered from biological material (for example, from older skeletal remains and hair shafts), segments of the mtDNA control region are amplified using a variety of approaches, dictated by the quality of the sample being tested. The amplified mtDNA products are subjected to Sanger-based sequencing and data interpretation is performed using one of many available software packages. These relatively simple methods, at least in retrospect, have remained robust, and have stood the test of time. However, alternative methods for mtDNA analysis remain viable options (for example, linear array assays and dHPLC), and should be revisited as the desire to streamline the testing process, interpret heteroplasmy, and deconvolute mixed mtDNA profiles intensifies. Therefore, it is important to periodically reassess the alternative methods available to the mtDNA practitioner, and to evaluate newer technologies being put forth by the scientific community, for example, next-generation sequencing. Although the basic mitochondrial DNA protocols and practices of public and private laboratories are similar, an overview of the current practices of forensic mtDNA analysis is provided, helping to frame the path forward.

KEY WORDS: Deep sequencing, DGGE, dHPLC, DNA damage, mass spectrometry, mtDNA mixtures, screening.