Phylogenetics and Mitochondrial DNA

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ABSTRACT: Phylogenetic analysis can be conducted using a variety of methods, generally classified as distancebased or character-based approaches. Patterns found through phylogenetic analysis of human mitochondrial DNA (mtDNA) sequences have revealed a wealth of information in such disparate fields as the human evolution; the movement of human lineages throughout history (phylogeography); and the susceptibility of certain groups to devastating diseases. Forensic mtDNA analysis has also benefited from the use of these methods. Phylogenetic assessment of forensic mtDNA databases has revealed a consistency with published data at a depth of analysis that is not attainable with basic population genetic methods. The detailed characteristics of specific sites within a mtDNA sequence are best assessed using phylogenetic methods. These studies have identified the most informative sites for individual differentiation, while also providing quality assurance metrics to apply to individual mtDNA profiles or entire databases. Such a level of evaluation and understanding enhances the interpretation of forensic casework.

KEY WORDS: Haplogroup analysis, human genetic diversity, mitochondrial DNA, mtDNA, phylogenetics.