

# Mitochondrial DNA Heteroplasmy

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**REFERENCE:** Melton T: Mitochondrial DNA heteroplasmy; *Forensic Sci Rev* 16:1–20; 2004.

**ABSTRACT:** Heteroplasmy, the presence of more than one type of mitochondrial DNA (mtDNA) in an individual, holds implications for forensic analysis of specimens such as blood, hair, and skeletal material. That is, what can we conclude about the likelihood that heteroplasmic specimens could or could not be from known individuals? Originally believed to be quite rare in healthy individuals, we now know that heteroplasmy exists at some level in all tissues on a predominantly homoplasmic background. A substantial body of general literature covers the biological origins of heteroplasmy, especially its transmission to new offspring and during life, the methodology for its detection, and its distribution in different tissues. In addition, the forensic community has contributed many observations on the characteristic appearance of heteroplasmy in relevant regions of the mtDNA control region and its appropriate treatment in forensic science. As a result of this growing understanding of a relatively simple biological phenomenon, we conclude that heteroplasmy can be expected to play a role in forensic interpretation on a regular basis, and that knowledge of its biological underpinnings contribute to just, conservative, and scientifically appropriate interpretational guidelines.

**KEY WORDS:** DNA sequencing, forensic DNA interpretation, heteroplasmy, mitochondrial DNA, mtDNA.

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