

# MITOCHONDRIAL DNA TESTING: CASEWORK IN THE PRIVATE SECTOR, HETEROPLASMY AND GENETIC DIVERSITY WITHIN THE UNITED STATES

**Kimberlyn Nelson,<sup>1,3</sup> Mark Stoneking,<sup>2</sup> and Terry Melton<sup>1</sup>**

<sup>1</sup>*Mitotyping Technologies, LLC, State College, PA*

<sup>2</sup>*Max Planck Institute for Evolutionary Anthropology, Leipzig, Germany*

<sup>3</sup>*Department of Biology, Pennsylvania State University, University Park, PA*



Up until 1998, forensic mitochondrial DNA analysis in the United States has been performed primarily by government agencies with specific mandates such as criminal casework or identification of military remains. In the private sector, although retention by prosecution attorneys, defense attorneys, and law enforcement is common, there are opportunities to perform casework with a wide range of purposes such as missing person identification, post conviction exoneration, genealogical investigation of maternal relatedness, and the resolution of historical puzzles. We will describe the full range of case types performed by Mitotyping Technologies, one private sector lab performing mtDNA analyses, and the only lab providing mtDNA analyses exclusively.

Heteroplasmy has gained increased awareness in mitochondrial DNA casework. Heteroplasmy may be present as either single nucleotide site variation or variation in the length of an amplified region. The occurrence of both types of heteroplasmy in casework at Mitotyping Technologies will be presented. In addition, the results of a familial study of length heteroplasmy in the homopolymeric C stretches of HV1 and HV2 are summarized.

The value of any molecular marker for forensic analysis relies on an understanding of the population level polymorphism. Previously studied populations have shown nearly uniformly high estimates of genetic diversity that result from a few relatively common haplotypes and many rare haplotypes. Diversity estimates and haplotypes counts for mtDNA haplotypes derived from SSO typing of 2282 individuals from three ethnic groups within the United States are presented and support the utility of mtDNA typing for forensic work.

