



## Physical Anthropology Section – 2006

### H83 Applications of DNA Identification to Human Rights: Additional Informative Sites in the mtDNA Genome

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After attending this presentation, attendees will gain a broader understanding of the variability found in the mtDNA genome and how this knowledge can be applied to better discriminate individuals sharing common mtDNA sequence motifs.

This presentation will impact the forensic community and/or humanity by demonstrating the value of mtDNA analysis as a principal identification strategy in human rights and mass-disaster investigations.

Over the last decade, the use of the mtDNA genome in forensic identification has increased due both to its suitability for identification of degraded human remains and for its capacity to be used in a highthroughput, cost-efficient manner. High throughput and low cost are particularly important when developing strategies to identify those killed in extrajudicial executions, a situation where sheer numbers can overwhelm available resources. For over two decades, the authors have been working with human rights organizations and families of disappeared persons to identify human remains. The initial analysis compares HVI and HVII sequences retrieved from human remains with those from maternal families. Consistent with current forensic standards, remains are considered excluded from membership in a family if they differ at two or more sites in HVI and HVII. Those who cannot be excluded are analysed for nuclear markers to further resolve the probability of family membership.

There are, however, situations where human remains are found to differ from multiple families by only zero, or one sites in HVI and HVII and thus cannot be excluded from family membership. This study considers whether sufficient mtDNA variation exists outside HVI and HVII to be informative in these situations. To address this question, anthropologists considered a single test family and 28 unrelated individuals who all share the same HVI and HVII motif of 16223 16298 16325 16327 and 73 249D 263 290D and 291D respectively. Outside of this combined HVI and HVII motif, the 28 unrelated individuals each differ by only zero or one substitutions from the test family. Three regions outside of HVI and HVII were selected for sequencing. These included HVIII (nt 438 to 574) and portions of the MTATP8/MTATP6 (nt 8366-8900), and MTCYB (nt 14750-15887) genes. Sequence data produced from these regions revealed three common single nucleotide polymorphisms (SNPs); these were found in HVIII at nt 493, nt 8772 in MTATP6 and nt 15740 in MTCYTB. The HVIII nt 493 A/G substitution was particularly informative in this comparison. Of the 23 unrelated individuals with one difference from the test family in HVI and HVII, 18 were also found to differ from the test family at 493 and could thus be excluded from membership. Additional private polymorphisms were detected in MTATP at nt 8790 and MTCYTB at positions 15462, 15644 and 15700. When all of the SNPs detected in these three regions were included in the analysis, 24 of the 28 unrelated individuals (86%) were found to differ from the test family at two or more sites. These results suggest that there is value sequencing additional mtDNA loci outside of HVI and HVII in order to discriminate individuals with shared mtDNA motifs.

**mtDNA, Coding-Region SNPs, Human Rights**