Using mtDNA SNP Typing To Resolve Common HV1/HV2 Types in Highly Degraded Samples From a Missing Persons Case

Rebecca S. Just1, Christopher W. Los1, Christina M. Miller1, Mark Leney2, Suzanne M. Barritt-Ross1, and Thomas J. Parsons1

1) Armed Forces DNA Identification Laboratory (AFDIL), 1413 Research Blvd. Bldg 101, Rockville, MD 20850, USA. 2) Joint POW/MIA Accounting Command-Central Identification Laboratory (JPAC-CIL), 310 Worchester Ave. Bldg 45, Hickam AFB, HI 93853-5530, USA.

Mitochondrial DNA (mtDNA) can be used to identify degraded skeletal remains by comparing the sequence of an unknown sample to that of a known maternal reference. However, common mtDNA sequences can be problematic when trying to distinguish one sample from another. Single Nucleotide Polymorphisms (SNPs), found in the control and coding region of the mitochondrial genome, can be used to differentiate samples that share a common HV1/HV2 sequence and can be combined into multiplex panels (Coble et al, 2004). Multiplex panel A targets 11 SNP sites that are useful for distinguishing individuals matching the most common Caucasian HV1/HV2 type (263G, 315.1C) (Vallone et al, 2004). This poster illustrates the application of multiplex panel A to mtDNA extracted from degraded skeletal remains for resolving common HV1/HV2 types. Cases that used mtDNA SNP typing to exclude previously inconclusive results will be presented. One case involved three references that all shared a common HV1/HV2 type with an unknown sample. The references could not be excluded/included using traditional mtDNA sequencing so multiplex panel A was used to differentiate the family reference samples. Another case was more complicated involving multiple unknown samples sharing common HV1/HV2 types. This 11-plex assay is suitable for use with degraded skeletal remains because it targets short amplicon sizes and requires a small amount of sample, which is helpful in cases where DNA extract is limited. The assay is extremely sensitive, ranging between 1-2 pg, and has the ability to detect mixtures and heteroplasmy more consistently than with conventional mtDNA sequencing (Just et al, 2004).