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THE APPLICATION OF MTDNA SNPS TO FORENSIC CASEWORK

Rebecca S. Just[1], Michael D. Coble[1], Mark D. Leney[2,3,4], Suzanne M. Barritt[1] and Thomas J. Parsons[1,5]

[1]Armed Forces DNA Identification Laboratory, Armed Forces Institute of Pathology, AFIP Annex, 1413 Research Blvd., Rockville, MD 20850, USA

[2]Joint POW/MIA Accounting Command – Central Identification Laboratory, 310 Worchester Ave. Bldg. 45, Hickam AFB, HI 96853, USA

[3]Massachusetts Biologic Laboratories, University of Massachusetts Medical School, Jamaica Plain, MA 02310, USA

[4]Scientific Advisor, The National League of Families of American Prisoners and Missing in Southeast Asia,1005 Glebe Road, Suite 170 Arlington, VA 22201

[5]International Commission on Missing Persons, Alipasina 45 A, 71000, Sarajevo, Bosnia

The Armed Forces DNA Identification Laboratory (AFDIL) routinely uses mitochondrial DNA (mtDNA) sequence data for sample sorting and individuation in Vietnam, Korea, and WWII-era cases where the DNA is expected to be highly degraded. mtDNA, as a circular genome and present in approximately 1000 copies per cell, has a much higher rate of recovery from degraded human remains than nuclear DNA. Sequencing of the two hypervariable regions (HV1 and HV2) and additional portions of the control region (CR) are well-established techniques for the identification of remains in missing persons cases, mass disasters, and other situations where circumstances dictate the use of mtDNA. However, a significant limitation to the use of mtDNA is the low power of discrimination when common mtDNA hypervariable region types (HV types) are encountered. To address this limitation we performed entire mitochondrial genome (mtGenome) sequencing of samples matching common HV types and identified single nucleotide polymorphisms (SNPs) in the CR outside of HV1 and HV2 and in the coding region of the mtGenome. SNPs which resolve common W. European Caucasian HV types were organized into multiplex panels targeting specific HV types, and allele-specific primer extension assays using SNaPshot chemistry (Applied Biosystems) were optimized for each panel (Coble *et al.* 2004, Vallone *et al.* 2004). One of these multiplex panels, panel "A", targets the most common W. European Caucasian HV type, "H:1" (263G, 315.1C), which occurs in ~8% of that population. We will report on the use of this SNP multiplex at the AFDIL to assist in the resolution of four cases in which samples matched or closely matched HV type H:1 and the identifications could not be made on the basis of HV1/HV2 sequencing alone. In two of the cases, the SNP typing successfully excluded one of two reference families that could not be excluded on the basis of HV1/HV2 sequencing, and resulted in the final resolution of both decades-old cases. In a third case, SNP typing confirmed the sorting and re-association of multiple commingled skeletal elements. In a fourth case, though the mtDNA evidence alone was not enough to support identification, the addition of the SNP data more than doubled the discriminatory power of the mtDNA evidence. The application of a specific mtDNA SNP assay in these cases demonstrates its utility in sample individuation when the most common Caucasian HV type is encountered in forensic casework.

References

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