Unusual and/or Problematic Sequence Variants in the mtDNA HyperVariable Regions 2 and 3: A Catalog, and Calling Guidelines

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The Armed Forces DNA Identification Laboratory, USA (AFDIL) performs mitochondrial DNA analysis in support of the identification of remains repatriated from Vietnam, Korean, and World War II era sites. Mitochondrial sequences recovered from these remains are compared to current forensic databases to determine the relative uniqueness of those sequences. AFDIL's Research Section is sequencing the mtDNA control region (bp16024-576) of populations under-represented in the forensic databases to support the mitochondrial DNA Global Database Initiative. As of June 2004, over 3000 control regions have been sequenced as part of this effort. Analysis of these sequences has identified a large number of sequence variations in the three polycystine stretches located in the control region. This poster will address the nineteen (see below) more interesting sequence variations observed within the HyperVariable Region 2 (HV2) polycystine stretch (bp 303-315) and the HyperVariable Region 3 (HV3) polycystine stretch (bp 568-573). Electropherogram layouts will be provided for each sequence variation, as well as alternate calling schemes where applicable. A copy of the Sequencher Calling Guide produced by AFDIL's Research Section will also be displayed. This book contains every insertion, deletion, and variation of specified regions observed during this database effort, together with guidelines for calling nomenclature. Consistent calling guidelines are necessary for databases to be searched, and shared by the forensic community. Our guidelines are consistent with, and extend, recommendations by others (Wilson et al. May 2002, Wilson et al. Oct 2002) to establish standardized mtDNA nomenclature. The nineteen sequence variations to be displayed will include the following: HV2 · 315.1 T · 315.1 C, 315.2 C · 309 D, 315.1 C · 308 D, 309 D, 315.1 C · 309 C-T, 315.1 C · 310.1 T, 315.1 C · 309.1 C, 310.1 T, 315.1 C · 309.1 C, 309.2 C(N), 310.1 C, 310.2 T, 315.1 C · Heteroplasmy between 309.1 C, 309.2 N

and 307 D, 308 D, 309 D HV3 · 573 D · 573.1 C · 573.1 C, 573.2 C · 573.1 C. 573.2 C, … 573.6 N (varying number of inserted C's) · 571 C-T · 573 C-T · 574 D · 575 C-T · 574 A-C, 575.1 C, 575.2 N · 574 A-C, 575.1 C, 575.2 C, 575.3 N