## B131 A Human Mitochondrial DNA Database Derived From Casework at Mitotyping Technologies

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After attending this presentation, attendees will know about the diversity and distribution of mitochondrial DNA control region sequences observed in a caseworking forensic mitochondrial DNA laboratory.

This presentation will impact the forensic community and/or humanity by demonstrating Forensic mitochondrial DNA analysis is being applied more broadly and frequently. However, forensic mtDNA practitioners rarely present DNA sequence data that they are gathering, and studies on the mtDNA variation present in North America are few. The data presented here will build the scientific understanding of all practitioners, and enhance the application of this type of DNA testing.

Between January 1999 and June 2004, Mitotyping Technologies collected 854 human mitochondrial DNA sequences for an internal database. Sequences were complete hypervariable region 1 and hypervariable region 2 nucleotide sequences encompassing positions 1599816400 and 30-407, respectively. A total of at least 783 nucleotides were available for each sequence. Identical sequences within a case (indicating that a match had been obtained between two or more samples within a case) were included only one time in the database. Therefore the samples are assumed to be randomly collected convenience samples derived from both questioned and known samples (blood, bone, hair, saliva, and other tissues) from most of the 50 United States, Canada, and the Caribbean. In most cases, an ethnic/biogeographical identity for the sequence (African/African American, Asian, Caucasian/European, Hispanic, and Native American) could be assigned based on either haplogroup-specific polymorphisms or information submitted with the samples.

The sequences were analyzed for sequence diversity, average number of nucleotide differences, haplogroup identity, and length heteroplasmy. These variables were examined at an overall level, within classically defined mtDNA haplogroups, and by state/geographic region. The results were compared to previously published studies on mitochondrial DNA diversity in the continental U.S. and internationally.

Overall, 72% (N=614) of the sequences could be classified as historically or biogeographically European while 17% (N=146) could be classified as historically or biogeographically African. Asian-, Hispanic-, Native American-type or unclassifiable sequences comprised the remainder of the sample. The electropherograms of unclassifiable sequences were reexamined for ambiguities or errors and were determined to be correct, indicating that the inability to classify these sequences was likely due to back mutations that have erased haplogroup-specific polymorphisms.

Diversity among the Europe-derived sequences was 0.995, with an average number of nucleotide differences of 8.2. Haplogroups H, I, J, K, T, U, V, W, and X were represented in this group, with diversity values ranging from 0.916 (V) to 1.0 (W, X). The average number of nucleotide differences within these European haplogroups ranged from 2.7 (V) to 7.9 (U). Haplogroup H members, the most frequently observed group in all published studies of European populations to date, comprised 38.4% of the European samples, and had a diversity value of 0.973 and an average number of nucleotide differences of 3.5. The “common” H1 haplotype (263A?G, 315.1insC) was observed at frequencies of 3.7% in the database overall and 5.7% within all European-derived sequences.

Among sequences determined to be historically or biogeographically African, diversity was close to 1.0 (0.999), indicating that nearly every sequence was unique in the database. The average number of nucleotide differences among these sequences was 15.1. Haplogroup L1 comprised 28% of the African-origin sequences, haplogroup L2 comprised 20% of these sequences, and haplogroup L3 comprised 36% of these sequences. Individuals from subgroups L1a, L1b, L1c, L2a, L2b, L2c, L2d, L3b, L3d, L3e, and L3f were represented from these three haplogroups. Diversity within the subgroups was uniformly high (above 0.977) with the exception of subgroup L1a (0.917).

**Mitochondrial DNA, mtDNA Sequence Database, Criminalistics**

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