B182 Development and Expansion of High Quality Control Region Databases to Improve Forensic mtDNA Evidence Interpretation

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After attending this presentation, attendees will understand the current limitations of mtDNA population databases for forensic mtDNA casework involving criminal evidence and missing persons specimens. This presentation will explain the current efforts underway at the Armed Forces DNA Identification Laboratory (AFDIL) to increase both the size and quality of mitochondrial DNA control region population databases for the forensic community.

This presentation will impact the forensic community and/or humanity by demonstrating that large, high quality mtDNA databases can and should be established to maximize the benefit to forensic mtDNA applications in criminal casework and missing persons investigations.

Forensic mtDNA analysis of highly degraded materials, or samples lacking sufficient quantity of nuclear DNA for STR testing (e.g., shed hairs) has found an important niche in DNA testing. Once a mtDNA sequence haplotype is generated for a case sample, it is necessary to assess the significance of the evidence using an appropriate population database. Such an assessment is similar to the evaluation of Y-STR evidence. However, mtDNA databases are significantly more costly and time consuming to generate than Y-STR databases, and the strength of the mtDNA evidence is often dependent upon the size of the population database used for comparison. Therefore, a sufficiently large population sampling is required for forensic mtDNA databases.

Recently, phylogenetic analyses of published mtDNA databases have been utilized to identify sequencing "phantom mutations" and other artifactual errors in forensic databases.[1] A series of high profile discussions has taken place in the scientific literature debating the seriousness of these errors,[2,3] culminating in a court case (US v. Ida Chase) that challenged the admissibility of the current forensic mtDNA database. Although the forensic mtDNA database was accepted in the resolution of this particular case, it is hoped that the current situation will be improved by developing high-quality mtDNA control region sequences for the forensic community.

AFDIL has developed a high-throughput automated system that utilizes robotic instrumentation for all laboratory steps from pre- extraction through sequence detection, and a rigorous 5-step, multi- laboratory data review process with entirely electronic data transfer. Since 2004 this laboratory has generated over 5000 control region sequences from both U.S. populations and underrepresented global populations (such as several from Central Asia).

The strength of this project is based on collaborations with colleagues, who provide the laboratory samples for sequencing and in return are provided co-authorship on any resulting publications. Furthermore, a strong relationship with the European DNA Profiling Group's was developed (EDNAP) Mitochondrial Population Database (EMPOP) team for additional quality control checking of the data and phylogenetic analyses. Additionally, EMPOP plans to provide these data to the forensic community.

Current progress on an effort being funded by the U.S. National Institute of Justice to generate over 3500 control region databases per year from U.S. populations will be presented. In addition to the high quality data generated from this effort, presented will be phylogenetic analyses of the data to determine the relative mutation rate of the control region. Some interesting nomenclature issues that have been encountered will be presented. Finally, some preliminary data that examines the substructure and heterogeneity of various regional or named population groups (such as "Hispanics") within the United States and component source populations will be given. In conclusion, large, high quality mtDNA databases can and should be established to maximize the benefit to forensic mtDNA applications in criminal casework and missing persons investigations.

References:

1 Bandelt, HJ, Lahermo, P, Richards, M, Macaulay, V (2001) Detecting errors in mtDNA data by phylogenetic analysis. *Int J Legal Med.* 115(2):64-69.

2 Bandelt, HJ, Salas, A, Bravi, C (2004) Problems in FBI mtDNA database. *Science.* 305(5689): 1402-1404.

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Mitochondrial DNA, Population Databases, Control Region

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