B126 Inter-Laboratory Study on Bone Extraction for Mitochondrial DNA Analysis

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The goal of this presentation is to present the planned round-robin study to the forensic mitochondrial DNA community and encourage the participation of all commercial and government forensic mitochondrial DNA laboratories in the United States and abroad.

This presentation will impact the forensic community and/or humanity by increasing awareness of this study and maximize participation among government, commercial mtDNA laboratories in the U.S. and abroad

With the implementation of the National Missing Persons DNA Database, the forensic DNA analysis of remains consisting of bone evidence continues to increase. Since the DNA found in bone evidence is frequently limiting and/or degraded, mitochondrial DNA (mtDNA) analysis is often the analysis method of choice. In addition to the government and commercial laboratories currently conducting mtDNA analysis, several additional forensic mtDNA laboratories, including the FBI's Regional mitochondrial DNA laboratories, New York City Office of the Chief Medical Examiner, and California Department of Justice, are completing internal validation studies and are preparing to begin casework. It is anticipated that a substantial portion of this casework will deal with remains from missing persons. However, the availability of human bones for training purposes is limited. Furthermore, to date, no proficiency test is available using bones as the evidentiary material.

The Mitochondrial DNA Subcommittee of the Scientific Working Group on DNA Analysis Methods (SWGDAM) is assembling an inter-laboratory study comparing the extraction methodologies and sequencing results obtained from a single source of bone sample. The study is designed so that similar bones (ie. toe bones) or sections of a long bone (ie. femur) will be obtained from a single donor and distributed to the participating laboratories. Extraction, amplification, and sequencing of the resulting bone DNA will proceed according to the laboratories' standard protocols. Although full HV1 and HV2 sequencing results is desirable and recommended, partial mtDNA sequencing results from mini-primer sets may be acceptable. In addition to mtDNA HV1 and HV2 sequencing data, autosomal STR, Y STR, as well as mtDNA coding region and non-HV1/HV2 control region SNP data will be evaluated if obtainable from the bone sample.

In addition to submitting results of extraction yield and mtDNA sequencing data, participating laboratories will complete a questionnaire

regarding details of their extraction methodologies as well as amplification and sequencing strategies. Comparison of the results and extraction methodologies may highlight differences in methodologies that can be improved to give greater yield of high quality extracted DNA and/or amplified product. In addition, it is expected that this exercise will lend assurance to the field that subtle differences in amplification and sequencing protocols do not lead to differing mtDNA profiles.

In order to provide the most benefit to the forensic mtDNA community, all government and commercial forensic laboratories in the
U.S. and abroad currently conducting mtDNA testing are encouraged to participate in this study. In addition, laboratories that anticipate conducting mtDNA testing in the near future are also welcome to participate.

Contact information as well as a time-frame for participant response, sample distribution, testing, results submission, and study analysis will be discussed.

mtDNA, Bone, Inter-Laboratory

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