B186 The Application of New Non-CODIS Mini-STRs to Highly Degraded Samples

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After attending this presentation, attendees will understand the limitations of mtDNA testing and the utility of new mini short tandem repeat markers (STRs) on degraded remains. Attendees will understand how mini-STRs can be employed to sort samples with matching mtDNA profiles, particularly in complex cases and/or cases with a large number of remains. Attendees will also learn how non-CODIS miniSTRs can be used to complement low copy number (LCN) nuclear typing results obtained with commercial kits and, thereby, add weight to the identification of remains.

This presentation will impact the forensic community and/or humanity by demonstrating the potential utility of non-CODIS miniSTRs in the analysis of highly degraded human remains.

Highly degraded human remains pose a challenge to many forensic laboratories due to the difficulty of producing short tandem repeat (STR) profiles using standard nuclear DNA typing protocols. Some laboratories perform mtDNA sequence analysis on such highly degraded samples. However, mtDNA is not a unique identifier, due to maternal inheritance and lack of recombination. Sample individuation using mtDNA sequence analysis is further confounded by the small region of the mtGenome typically examined (hypervariable regions I and II, approximately 610 base pairs, or the entire control region), and by the existence of common mtDNA types resulting from founder effects, population bottlenecks, and genetic drift in human history. These characteristics of mtDNA typing virtually ensure that with large numbers of samples there will be multiple non-related individuals whose mtDNA profiles are identical. This particular limitation of mtDNA typing becomes significantly problematic in situations where remains have been commingled. In such cases, with mtDNA data alone, it may be difficult or impossible to simply determine the total number of individuals represented in a particular case/incident.

The development of new non-CODIS miniSTR markers for use with highly degraded samples has the potential for significant impact in forensic casework.[1,2] Non-CODIS STRs will be particularly useful in sorting samples when multiple elements cannot be distinguished using mtDNA. Sorting can be accomplished by applying miniSTRs to those samples that share common mtDNA types. The results can then be used to reassociate remains and determine the number of individuals represented. The results of typing with 8 non-CODIS miniSTR markers (D2S441, D2S1776, D3S4529, D6S474, D9S2157, D10S1248, ATA63, and D22S1045) on samples from a large set of commingled remains repatriated from North Korea will be presented. The size range of these eight non-CODIS markers vary from ~70-170 base pairs. Also included within the 8-plex is a single CODIS marker (D16S539) having a size range from ~235-275 base pairs, and acts as an internal control to assess the level of DNA degradation for the sample. MtDNA control region sequencing was previously performed on many of these remains. However, a large number of the sequences matched mtDNA types common in the Caucasian population, preventing individuation or even an accurate assessment of the total number of individuals represented. The miniSTR data presented will demonstrate that the sorting of such samples can be accomplished through the use of these non-CODIS nuclear markers.

In addition to the use of non-CODIS miniSTRs for sample sorting, these new markers will be used to complement LCN nDNA results and add weight to the statistics generated with CODIS markers. In cases with highly degraded remains it can be challenging to obtain high likelihood ratios (LRs) from partial profiles developed using LCN nDNA typing. Furthermore, in many of these cases, ideal reference material from direct reference samples and/or immediate kin are unavailable. The addition of supplementary markers to the calculations can increase LRs significantly when only partial profiles are recovered with CODIS markers and/or only extended kin are available as references. Data will be presented that shows the benefit of adding 8 non-CODIS miniSTRs (D2S441, D2S1776, D3S4529, D6S474, D9S2157, D10S1248, ATA63, and D22S1045) to samples that have been typed using LCN protocols for PowerPlex16 (Promega) and Y-filer (Applied Biosystems), and have also been sequenced in the mtDNA control region and typed for mtDNA coding region SNPs.

References:

[1] Coble, M.D., Butler, J.M. (2005) Characterization of New MiniSTR Loci to Aid Analysis of Degraded DNA. *J. Forensic Sci.* 50(1): 43-53. [2] Hill, C.R., Coble, M.D., Butler, J.M. (2006) Development of additional new miniSTR loci for improved analysis of degraded DNA samples. *submitted*.

MiniSTR, Degraded DNA, Mitochondrial DNA

 ** Presenting Author*