B90 An Evaluation of Mitochondrial DNA Variation: From Linear Arrays to Whole Genome Sequencing

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After attending this presentation, attendees will learn about the assessment of genetic variation in the control and coding regions of the entire mitochondrial DNA (mtDNA) genome. This will be discussed in relation to Linear Arrays and the identification of coding region polymor- phisms for increased forensic discrimination of common types.

This presentation will impact the forensic community and/or humanity by enhancing the knowledge of the forensic community in regards to mitochondrial DNA genetic variation and its impact on mtDNA Linear Array and coding region analyses.

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. Recent research has focused on two of the limitations for mtDNA testing: the cost of mtDNA testing and the low power of discrimination associated with common mtDNA types. To overcome the cost prohibition of mtDNA testing, Linear Arrays have been evaluated as a screening tool (1, 2). The identification of polymorphic mutations in the mtDNA coding region has also been proposed as a way to increase the forensic discrimination of common mtDNA types (3, 4).

An evaluation of mtDNA Linear Array data compared to the control region sequence information from 666 population samples were analyzed to determine the underlying source of null alleles (blanks). An examination of coding region variation in a global dataset of mtDNA genomes has also been studied to determine the feasibility of sequencing segments of the mtDNA genome for increased forensic discrimination.

Linear Array analysis for over 600 population samples from self- described Caucasian, African American, and Hispanic individuals were conducted (1). Control region sequences were generated at the Armed Forces DNA Identification Laboratory (Rockville, MD) using established, published protocols (5). An assessment of coding region variation for increased forensic discrimination utilized published data from the literature available at the mtDB website (http://www.genpat.uu.se/mtDB/). Haplogroup associated polymorphisms were determined from the literature.

Most of the null alleles observed in Linear Arrays were created by mutations associated with mtDNA haplogroups rather than private poly- morphisms. This suggests that null alleles can provide useful information in Linear Array analysis. In addition, most of the "highly polymorphic" mutations in the coding region, potential targets of increased discrimi- nation, are sites associated with mtDNA haplogroups. These sites would therefore be uninformative for forensic discrimination since much of this information is redundant with sequence information determined from the control region.

An appreciation of mtDNA haplogroups can be useful for mtDNA screening using linear arrays for HV1 and HV2, and for avoiding highly redundant, uninformative polymorphisms in the mtDNA coding region.

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2. Divne, A-M, Nilsson, M., Calloway, C., Reynolds, R., Erlich, H., Allen, M. (2005) Forensic casework analysis using the HVI/HVII mtDNA linear array assay. *J. Forensic Sci.,* 50: 548-554.

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5. Edson, S.M., Ross, J.R., Coble, M.D., Parsons, T.J. and Barritt, S.M. (2004) Naming the dead — confronting the realities of rapid identifi- cation of degraded skeletal remains. *Forensic Science Reviews* 16(1): 64-89.

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