VALIDATION OF MITOCHONDRIAL DNA ANALYSIS FOR FORENSIC CASEWORK UTILIZING CAPILLARY ELECTROPHORESIS DETECTION

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The use of mitochondrial (mt) DNA analysis in cases of body identification and hair comparison has become a matter of routine testing for the specialized laboratories that perform this analysis. The basic testing strategy involves PCR amplification of the HV1 (Hypervariable Region 1) and HV2 (Hypervariable Region 2) from the Control Region of the human mtDNA molecule. The amplified product is then subjected to cycle sequencing and detection via fluorescent dye labels.

The majority of validation studies thus far presented for this test have involved the use of sequencing gels and analysis with the Applied Biosystems 377 DNA Sequencer[™]. The purpose of this poster is to report the results of a validation study on mtDNA analysis involving the use of capillary electrophoresis. The detection device utilized for this study was the Applied Biosystems 310 Genetic Analyzer[™] in conjunction with the Applied Biosystems Big Dye Terminator Kit[™]. Internal validation studies included mixture analysis, sensitivity studies, precision studies, maternal inheritance study, tissue reproductability study, and non-probative casework (including ASCLD Lab approved proficiency tests). A final study involved an inter-lab comparison with two different mtDNA testing laboratories that have obtained results on the Applied Biosystems 377 DNA Sequencer[™]. The results of these validation studies revealed no deviation from reported results obtained from similar studies using the Applied Biosystems 377 DNA[™]. The conclusion is that both systems appear compatible and that either device can be used to perform mtDNA analysis with reliability.

Keywords: Mitochondrial DNA, Capillary Electrophoresis, Forensic Validation Study