DETECTION OF HETEROPLASMY IN HUMAN HEAD HAIR AND BLOODSTAINS FROM 132 INDIVIDUALS USING A PROTOTYPE MITOCHONDRIAL DNA LINEAR ARRAY ASSAY

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This study examines mitochondrial DNA polymorphisms in human head hair and bloodstains with respect to their potential for forensic application. Mitochondrial DNA was isolated and polymorphisms were detected by applying sequence-specific oligonucleotide probe analysis, a technique that provides objective, timely and cost-effective results. The particular focus was to characterize the morphological features of human head hair in order to further the understanding of the factors that influence amplification success rate and detection of heteroplasmy in hair tissue.

131 bloodstains and 2554 head hairs from 132 individuals representing four population groups were amplified. The hair samples were characterized according to their hair growth phase: 1303 were identified microscopically as telogen in origin and 1251 were classified as anagen. Amplification success was assessed as a function of several independent variables: morphological characteristics; hair growth phase; donor age and gender; scalp origin; use of cosmetic hair treatments; and race of donor.

Samples that successfully amplified were typed using a prototype mitochondrial DNA linear array assay. The genetic diversity value for each population group was analyzed and the frequency of each mtDNA haplotype was determined. For the purpose of this study a sample was scored as heteroplasmic if two probe signals were visible within a single probe region (either with equal or uneven intensity). The results of this study demonstrate differences in heteroplasmic expression between hair and blood tissue. Further, differences in expression were also observed within each respective tissue. Finally, the study evaluates the frequency of heteroplasmy across racial population groups and assesses whether the heteroplasmic condition differs significantly with age, gender, medulla morphology, region of the scalp, hair growth or, when comparing living and deceased donors.