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MITOCHONDRIAL DNA HETEROPLASMY AS A FUNCTION OF HAIR MORPHOLOGY

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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 the detection of heteroplasmy in hair tissue.

Bloodstains (127 individuals) and head hairs (128 individuals) representing four population groups were typed using the mitochondrial DNA LINEAR ARRAY ASSAY™. DNA sequencing analysis was also performed on a select number of hairs. 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. The study evaluates the frequency of heteroplasmy across racial population groups, and correlation with cosmetic treatment, age, gender, medulla morphology, region of the scalp, hair growth phase, hair pigmentation, or, when comparing living and deceased donors.