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INCREASING MITOCHONDRIAL DNA DISCRIMINATION AMONG CAUCASIAN AND HISPANIC POPULATION SAMPLES USING A PANEL OF 56 IMMOBILIZED SSO PROBES

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Polymorphic regions of the mitochondrial genome, most notably the hypervariable regions I and II (HVI/II), have proven valuable in the genetic analysis of forensic samples which are limited and/or highly degraded. Currently, a 31 probe panel that targets 18 sequence polymorphisms within 10 regions of HVI and II is available for commercial use (LINEAR ARRAY mtDNA HVI/HVII Region-Sequencing Typing Kit). However, there are inherent limitations to just targeting the HVI and HVII regions independent of the method of analysis (HVI/HVII sequencing or linear array assay). The power of discrimination is limited for all population groups as a result of a few relatively common HVI/HVII sequences. Seven percent of Caucasians share the same common HVI/II sequence and 13 additional sequences are shared among >0.5% of the population. Also, 11% of Caucasians share the same common HVI/II type, as determined by the linear array assay. Similarly, several common HVI/II types can be found amongst the Hispanic population. To further distinguish these common HVI/II types and increase the power of discrimination, an 84 probe panel consisting of 53 additional probes targeting the most discriminating polymorphic sites is in the process of being developed. We currently have a 56 probe panel which consists of the existing 31 probes for sites within HVI/HVII, as well as 25 additional probes for polymorphic sites located within the coding region, HVII and Variable Regions (VR). This assay has been tested on Caucasian and Hispanic population samples in order to estimate the improved power of discrimination of the 56 probe panel compared to HVI/HVII mtDNA typing and sequencing; results from this population study will be presented here, including frequency and genetic diversity calculations. A small population study consisting of 88 randomly selected Caucasian samples was conducted using the 56 probe panel assay. Results from the study indicate that the overall genetic diversity (h value) can be increased following the addition of the 25 new probes. When typed with only the HVI/HVII mtDNA linear array an h value of 0.973 is obtained for this population. However, the h value is increased to 0.992 using the 56 probe panel assay, which is nearly as informative as the h value obtained from HVI/HVII sequencing (0.994). A separate population study consisting of 91 Hispanic samples was also conducted using the 56 probe panel assay. Results from this study show that with the 56 probe panel, the two most common Hispanic HVI/HVII types can be further distinguished and genetic diversity (h value) can be increased. When the 56 probe panel assay is used, the most common Hispanic HVI/HVII type can be further subdivided into ten groups. Similarly, the second common Hispanic HVI/HVII type can be further subdivided into five smaller groups. Results also indicate that when these 91 samples are typed with only the LINEAR ARRAY HVI/HVII assay (31 probes) an h value of 0.950 is obtained. However, with the 56 probe panel assay, the h value is increased to 0.990. We are confident that the discrimination power of the fully developed 84 probe panel will be greater than that of HVI/HVII sequencing for all populations.