B24 Detection of Sequence Variation in Caucasian and Hispanic Samples Across the Mitochondrial Genome Using an 83 Immobilized SSO Probe Panel

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The goal of this poster is to present results from a population study conducted to investigate sequence variation in Caucasian and Hispanic DNA samples using a panel of 83 SSO probes immobilized on a nylon membrane and to determine the power of discrimination for these populations.

This presentation will impact the forensic community and/or humanity by providing practitioners with additional data on the frequency of polymorphic sites outside of the commonly targeted HVI/HVII regions of the mitochondrial genome.

Currently, a 31 probe panel that targets polymorphic regions within hypervariable regions I and II (HVI/II) is commercially available. Although forensic laboratories have successfully used this assay to make exclusions in casework, there are limitations to targeting just the HVI and HVII regions independent of the method of analysis (HVI/HVII sequencing or linear array typing). Seven percent of Caucasians share the same HVI/II sequence and 11% share the same HVI/HVII type, as determined by the linear array assay. Similarly, there are two common HVI/II types shared amongst Hispanics. To further distinguish these common HVI/II types and increase the discrimination power, a probe panel targeting the most discriminating polymorphic sites located in the variable and coding regions was developed. Results from a population study will be presented here, including frequency and genetic diversity calculations.

An intermediate probe panel, consisting of mtDNA HVI/II probes and additional coding regions probes (56 probes total), has been developed and tested on Caucasian and Hispanic samples. A population study including 88 Caucasian samples was conducted using the 56 probe panel and preliminary results indicate that significant increases in the detection of overall genetic diversity can be obtained. When typed with only the HVI/HVII linear array an h value of 0.973 is obtained. However, the h value is increased to 0.992 using the 56 probe panel (HVI/HVII and 25 additional probes), which is nearly as informative as the h value obtained from HVI/HVII sequencing (0.994). A population study including 91 Hispanics was also conducted using the 56 probe panel. Preliminary results from this study show that with the addition of the 25 probes, the two common Hispanic HVI/HVII types can be further distinguished and an increased h value can be obtained. The most common Hispanic HVI/HVII type can be further subdivided into ten groups when using the 56 probe panel. Similarly, the second common Hispanic HVI/HVII type can be further subdivided into five smaller groups. Preliminary calculations indicate that when these 91 samples are typed with only the HVI/HVII linear array, an h value of 0.944 is obtained. However, with the addition of the 25 new probes the h value is increased to 0.989. Results from the combined 83 probe panel will also be presented here. The discrimination power of the combined 83 probe panel will be greater than that of HVI/HVII sequencing for all populations.

Mitochondrial DNA, Linear Array, Immobilized SSO Probes

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