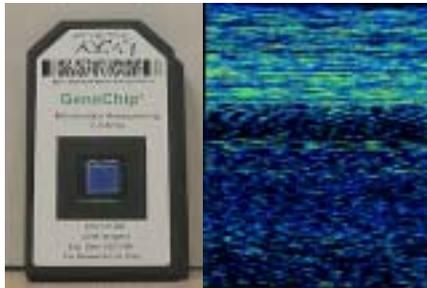


Human Identity Testing: Forensic Application of the Affymetrix Human Mitochondrial Resequencing Array

The goal of this research is to evaluate the potential of the Affymetrix GeneChip Mitochondrial Resequencing Array (ver 2.0) for forensic applications. The results will allow the forensic scientists to determine if the platform is robust enough for their specific purpose and allow the manufacturer to make improvements in future generations of the mitochondrial GeneChip.

P.M. Vallone, J.P. Jakupciak, M.D. Coble (831)

In the field of forensic DNA testing, sequencing regions of the mitochondrial genome is performed when insufficient genomic DNA is present for traditional autosomal short tandem repeat (STR) testing. Typically only subsets of the mitochondrial control region (HV1 and HV2) are sequenced for forensic analysis. Sequencing coding region polymorphisms in the mitochondrial genome can be useful for resolving individuals who have the identical HV1 and HV2 control region sequence. Various methods and strategies have been established to interrogate coding region polymorphisms. These range from single nucleotide polymorphism (SNP) assays probing sites most likely to differentiate individuals based on their HV1/HV2 sequence, to the use of mass spectrometry to pyrosequencing.



Picture of the Affymetrix GeneChip Mitochondrial Resequencing Array (left) A fluorescent scan of the array post DNA hybridization (right). Base calls are obtained from software aided analysis of the fluorescent cell intensities.

NIST is assessing an array based platform for its potential in forensic usage. Accuracy of the Affymetrix commercial resequencing array technology and its robustness (accurate and reproducible base calls) for routine usage within the forensic community is being determined.

The NIST team is investigating the reproducibility, base call accuracy, sensitivity, and general forensic utility of the array platform. This was done by resequencing 'challenging' samples (of non western European origin) and also resolving Caucasian samples that contain the identical HV1 and HV2 region. All results were compared to full genome sequencing using traditional fluorescent sequencing techniques and capillary electrophoresis as a detection platform. Initial results have been presented at the European Mitochondrial DNA Population Database Project

(EMPOP) meeting held in Innsbruck, Austria in the fall of 2006.

Impact: Forensic typing laboratories that perform mitochondrial DNA have expressed an interest in using the GeneChip array platform. The results will allow these researchers to determine if the platform is robust enough for their specific purpose. The information we have obtained will also allow the manufacturer to make improvements in future generations of the mitochondrial GeneChip.

Future Plans: Further testing of the GeneChip to determine the performance on degraded DNA materials. We are also interested in alternative PCR amplification of the mitochondrial genome that may work to increase the sensitivity of the system.

Publications:

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