

POSTER ABSTRACTS

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11:45 am – 2:00 pm
Tuesday, April 28th • Lobby

PS2 – 08

Development and Implementation of a Genetic Fingerprinting Assay for the Personalized Medicine Research Project

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Background: For any biorepository, it is necessary to develop measures that determine sample quality and ensure each sample can be correctly identified. One method of devising this type of quality control measure is to rely on DNA polymorphism panels developed for forensic applications. Although valid for identification, these panels are not useful for other purposes, such as medical research.

Aims: We developed an identification panel for the Personalized Medicine Research Project (PMRP) that uses medically relevant polymorphisms. This panel not only uniquely identifies samples and tests for sample quality but can also be used by investigators for candidate gene studies.

Methods: Polymorphism candidates were taken from investigator requests, PharmGKB (www.pharmgkb.org) and the disease association database (<http://geneticassociationdb.nih.gov/>). To be considered, a polymorphism had to be associated with disease in two independent populations and it had to have a reported minor allele frequency in a Caucasian population of at least 0.20. The final assay was developed on the Sequenom platform and all individuals within the PMRP cohort were analyzed. Probability of identity for the panel was calculated and each polymorphism was tested for Hardy Weinberg equilibrium. The allele frequencies found in our population of 20,000 were compared to reported allele frequencies. The allele frequencies in the PMRP population were compared between males and females and between different age categories.

Results: From a list of 116 potential polymorphisms we developed a single assay of 36 medically relevant somatic polymorphisms with at least one polymorphism on each chromosome and a single sex marker. The probability of identity for the assay was 6.132×10^{-15} and the sibling probability of identity was 3.077×10^{-8} . Three polymorphisms differed from previously reported allele frequencies by more than 10%. In our population, 4 polymorphisms were different in males and females and 4 polymorphisms varied in allele frequencies between age groups. Furthermore, the chosen polymorphisms have been used in two studies and as preliminary data in several other studies.

Conclusions: We were able to create a DNA panel of 36 medically relevant polymorphisms that successfully tested DNA quality, increased the knowledge of population wide allele frequencies, and is useful to investigators for candidate gene studies.