Successful Illumina Array Genotyping on Serum Stored For Three Decades

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An integrated series of prospective cohort studies of drug users in medication assisted treatment across the US, begun in 1980’s, linked with a research biospecimen repository, has >11,000 visits. Serum was collected in SST tubes, centrifuged locally, and shipped insulated overnight to a central lab for aliquoting, with continuous storage at -80°C. The protocol, including genetic testing, was reviewed and approved by the Rutgers IRB. Coded vials were sent to RUCDR and processed as per standard NGC protocols, including DNA extraction from the entire supplied serum vial followed by amplification.

Of 5033 specimens (volume ranging from 1.0-2.0 ml), genotyping on Illumina Infinium OmniExpress-24 V1.3 BeadChip was successful in 5025 (99.84%). For ≥20% and ≥15% the overall miss rates were only 1.55% and 2.75%; miss rates monotonically declined with larger volume, from 4.97% & 8.15% for 1.0-1.3 ml, to 0.97% & 1.78% for 1.7-2.0 ml. Sample set includes 2% serial blood draws; call rates are being compared.

Parameters that might affect white cell count, and hence cellular material, such as HIV and HTLV infection status, are being explored.

In light of the high success rate with just 1.0ml, a set of 0.5ml vials is being processed.

Summary: Highly successful Illumina array genotyping is possible using limited serum volume even after three decades of storage.

The genotype and demographic & drug use phenotype data are being deposited by Dr. Weiss into dbGaP, will contribute to ongoing NGC analyses, and will significantly increase the number of samples available for joint analyses among Blacks and Hispanics.