



Informativity of ancestry-sensitive markers from autosomes, Y-chromosome and mitochondrial DNA in U.S. populations

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Outline

- Markers and selection
- NIST U.S. sample set
- SNP assay development
- Y SNPs and Mitochondrial data
- Results
- Conclusions

Ancestry-Sensitive Markers (ASM)

- Estimate genetic ancestry
- Biogeographical population structure
- Mitochondrial genome
- Y chromosome (STRs, SNPs)
- Autosomal SNPs (population specific)

Marker Selection

- 24 autosomal SNPs selected
 - Affymetrix SNP array
 - CEPH–Human Genome Diversity Project Cell Line Panel (CEPH-HGDP)
- Selection criteria (high Fst, low het, limit redundancy)
 - Intended for a broad scope...
- Bi-allelic, spread over 13 chromosomes

Extension of work by Lao et al. AJHG 2006 78: 680-690

NIST U.S. Sample Set

- 710 NIST samples typed
- African American = 259
- U.S. Caucasians = 262
- U.S. Hispanics = 140
- U.S. Asians = 49



NIST samples have been extensively typed on other forensically relevant markers
Autosomal STRs (CODIS and additional),
Y STRs, Y SNPs, Autosomal SNPs, mtDNA sequencing

Anonymous
Self Identified Ancestry

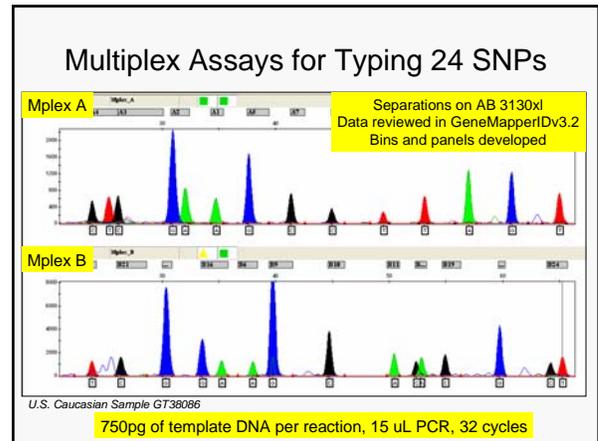
<http://www.cstl.nist.gov/biotech/strbase/NISTpop.htm>

Initial Questions

- How will the 24 autosomal ASM SNPs characterize the NIST U.S. sample set?
 - How does Self ID ancestry correlate with ASMs
 - Sample set provides a practical forensic test
- How will the different ASMs compare?
 - 24 autosomal SNPs
 - Mitochondrial haplogroup
 - Y haplogroup

SNP Typing Assays

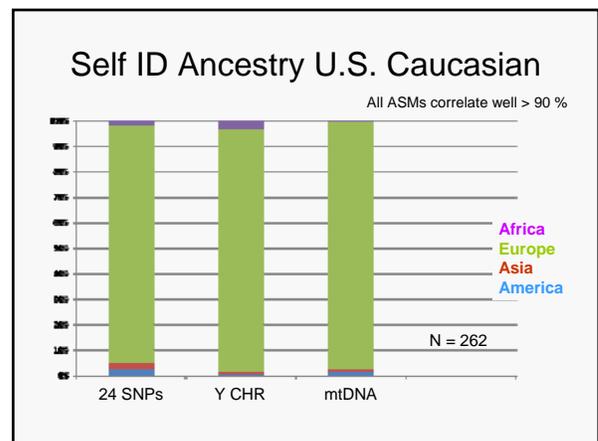
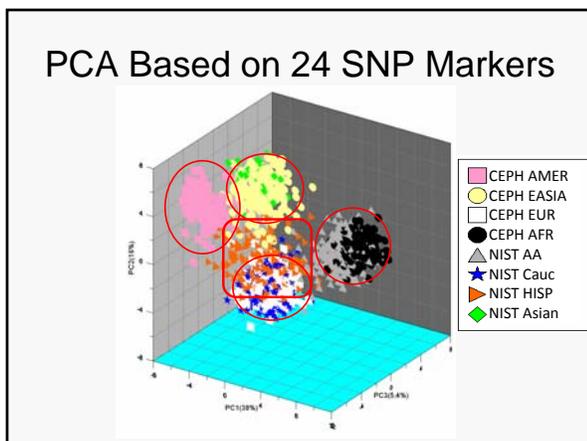
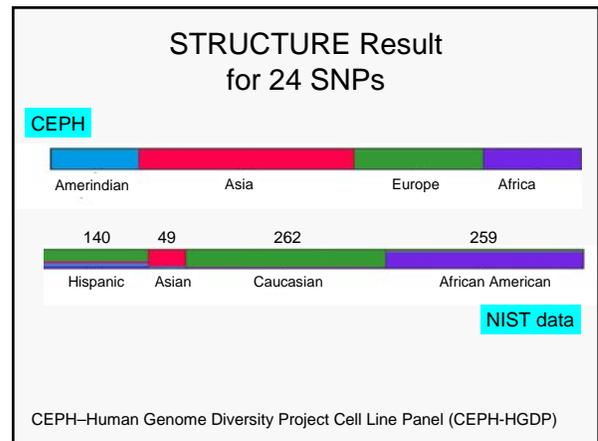
- SNPs were typed allele-specific primer extension (minisequencing /SNaPshot™)
- 2 x 12plex multiplex PCR and extension reactions
- Assays were optimized for typing the 710 NIST U.S. samples

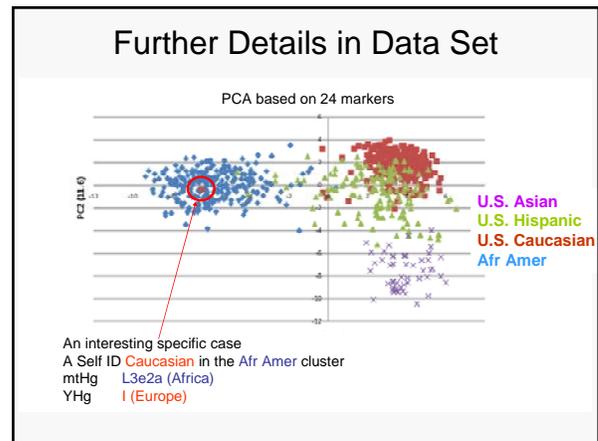
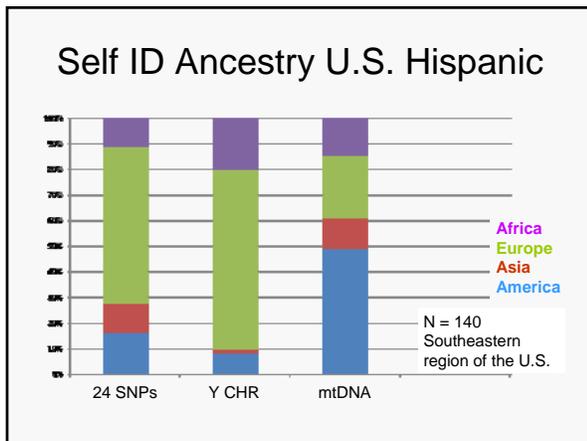
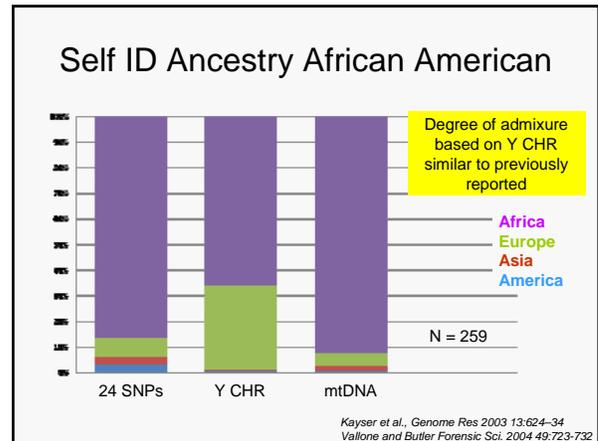
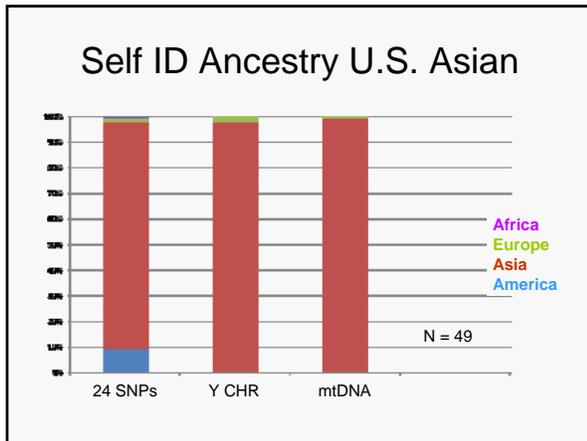


Y SNP and Mitochondrial Data

- Mitochondrial sequencing
 - AFDIL performed control region sequencing
 - Haplogroups were assigned (Dr. Mike Coble and Ms. Jodi Irwin)
- 24 Y SNP markers were typed in the laboratory of Peter DeKnijff (minisequencing)

Details P. de Knijff et al. in prep





Conclusions

- Minisequencing SNP assays developed for 24 autosomal ASM markers
- Further characterization of NIST U.S. sample set – ASMs (24 SNPs and uniparental lineage markers)
- Degree of correlation between Self Identified Ancestry and ASMs
 - U.S. Caucasian, U.S. Asians, African Americans
 - ASMs reflect admixture in Hispanic and AA samples

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