BLIND TESTING AND EVALUATION OF A COMPREHENSIVE DNA PHENOTYPING SYSTEM

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STR profiles are only informative if a suspect is available for comparison either through police investigation or a DNA database search. When this approach fails to yield an association, forensic DNA phenotyping provides an alternative approach for providing an investigative lead. DNA phenotyping, a relatively new area of forensic genetics, predicts a person's physical appearance (i.e., facial phenotype) and ancestry from a DNA sample, by typing an array of single nucleotide polymorphisms (SNPs) [1-6]. Several systems have been developed, ranging from simple multiplex SNP assays to predict eye color, hair color, and global ancestry to more comprehensive approaches for a complex set of traits [3-11]. In this study, the Parabon Snapshot DNA Phenotyping System, which predicts detailed biogeographic ancestry, sex, eye color, hair color, skin pigmentation, freckling, and face morphology, was evaluated in a blind experiment. This study represents the first public blind evaluation of the Snapshot DNA Phenotyping System, including side-by-side comparisons of the composite images and the actual photographs of each subject.

The University of North Texas Health Science Center (UNTHSC) recruited 24 subjects from a range of phenotypic and ancestral backgrounds. All sample collection was preformed in accordance with the approved protocols of the Institutional Review Board (IRB) at UNTHSC. A buccal swab was collected from each individual for phenotypic testing. Twenty-five anonymous DNA samples were sent to Parabon for analysis with Snapshot. One sample was prepared intentionally as a mixture of two female subjects, but this was not made known to Parabon. Self-reported ancestry and phenotypes were collected along with photographs of each subject for subsequent comparison with the phenotype predictions. Each DNA sample was genotyped at 851,274 SNPs and run through the Snapshot prediction algorithms. The phenotype predictions were compiled into a detailed report for each subject, including a predicted composite, in which the differences from the average face for the same sex and ancestry were displayed.

This presentation described the performance of Snapshot and its phenotype predictions for "Level 1" features, which include skin pigmentation, eye color, hair color, and freckling. Sex and bioancestry also were evaluated. The prediction of "Level 2" features (i.e., facial morphology) is currently being evaluated in an ongoing study. UNTHSC compared the self-reported values and subject photographs with each of the Level 1 phenotype and bioancestry predictions made by Snapshot. All predictions for sex and bioancestry were consistent with the participant's self-reported classifications. Skin pigmentation predictions were relatively consistent with 100% for Europeans (n=11) and approximately 92% for non-Europeans (n=13). Pigmentation predictions showed clustering of values (i.e., melanin index) within population groups for our sample set. Eye and hair color predictions for Europeans were 91% and 82% concordant, respectively,

while non-Europeans were 100% consistent. For Europeans, these phenotypes are known to be complex traits, unlike individuals of Asian or African-American descent, where these traits are considered fixed phenotypes (brown or black). Freckling predictions were 96% consistent with self-reported classifications.

All composites were generated at Snapshot's default values: 25 years of age and an average body mass index (BMI) of 22. Age and BMI values were delivered to Parabon following UNTHSC's initial evaluation, and composites for two subjects with large differences from the default prediction age of 25 and BMI of 22 were age-progressed by a forensic artist. Although a small sample size, the results support that the majority of Level 1 external feature and bioancestry predictions made using Snapshot are consistent with self-declaration of the donors and the UNTHSC Evaluators. Analysis of Level 2 features, i.e., facial morphology details, is underway.

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