BLIND TESTING AND EVALUATION OF A COMPREHENSIVE DNA PHENOTYPING SYSTEM

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DNA phenotyping, a relatively new area of forensic genetics, predicts a person's physical appearance (phenotype) and ancestry from a DNA sample, by typing an array of single nucleotide polymorphisms (SNPs). 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. In this study, the Parabon Snapshot DNA Phenotyping System, which predicts detailed biogeographic ancestry, 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 a comprehensive 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 for phenotypic and ancestral diversity, and a buccal swab was collected from each individual. Twenty-five anonymous DNA samples were sent to Parabon for analysis with Snapshot. One sample was intentionally a mixture of two 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 emphasized. Age and body mass index (BMI) values were then delivered to Parabon, and composites for subjects with large differences from the default prediction age of 25 and BMI of 22 were age-progressed by a forensic artist. UNTHSC then compared the predictions of each phenotype with self-reported values and with the photographs. Detailed results will be presented along with a discussion of how phenotyping can be used in investigations. This poster will serve as a companion to the talk being given by the UNT PI, allowing for more in-depth discussion.