Blind Testing and Evaluation of a Comprehensive DNA Phenotyping System

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Introduction

DNA phenotyping refers to the prediction of ancestry and/or physical appearance from DNA. In forensics, these predictions have the potential to generate new investigative leads in cases where DNA does not match a known suspect or a database, and to discover more information about unidentified remains. In this study, the Parabon® Snapshot™ DNA Phenotyping System, which predicts detailed biogeographic ancestry, pigmentation (eye color, hair color, skin color, and 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.

Methods

- 24 subjects recruited for phenotypic and ancestral diversity by the University of North Texas Health Science Center (UNTHSC)
- 25 anonymous DNA samples sent to Parabon, including one two-person mixture (not made known to Parabon, but Parabon readily detected the mixture and identified the contributors)
- Each sample genotyped on the Illumina CytoSNP-850K chip (851,274 SNPs) and run through the Snapshot algorithms
- Phenotype predictions compiled into a detailed report for each subject, including a predicted composite in which differences from the average face for the same sex and ancestry were emphasized
- Age and body mass index (BMI) values then delivered to Parabon, and subjects with large differences from default age (25) and BMI (22) age-progressed by a forensic artist
- Photographs and self-reported ancestry and phenotypes collected by UNTHSC, and predictions for each Level 1 phenotype (sex, pigmentation, ancestry) compared to actual phenotypes
- Next phase will incorporate 3D scanning and craniofacial measurements to assess accuracy of predicted face morphology

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Predictions Vs. Actual Appearance
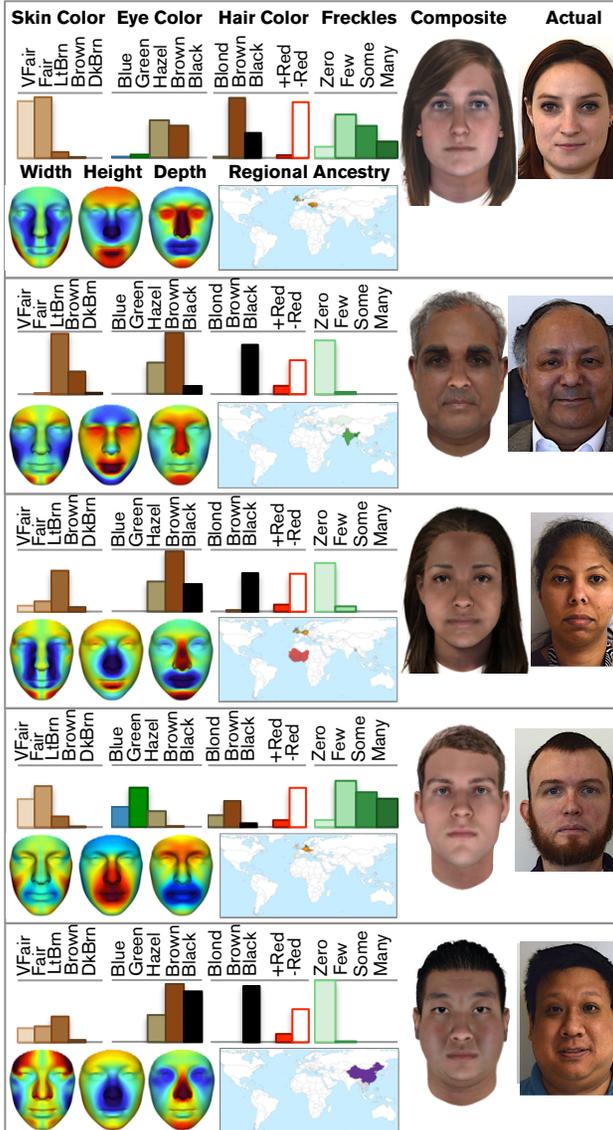
Prediction Results

Predicted Phenotype Consistencies vs. Actual Phenotype
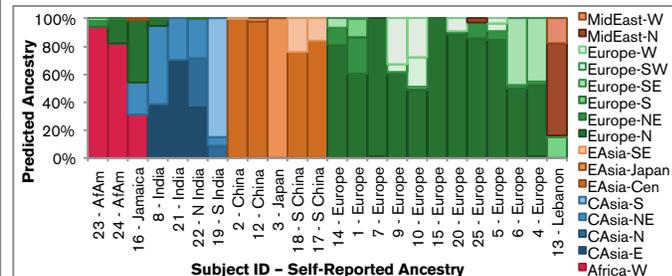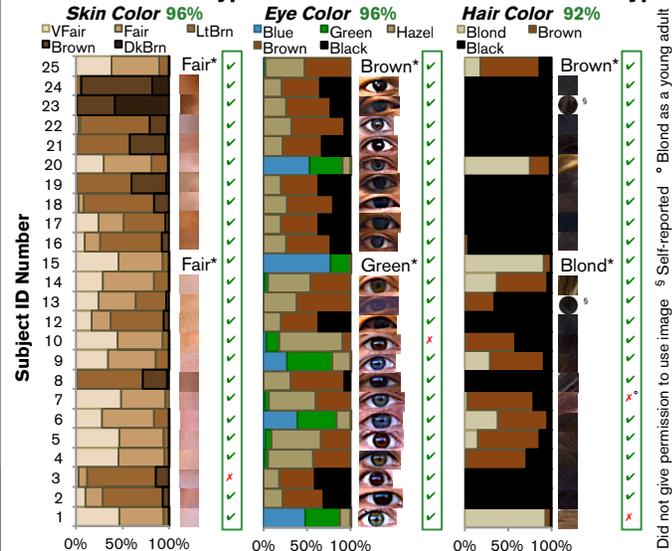
Conclusions

This study demonstrated the predictive performance of the Parabon Snapshot DNA Phenotyping system. Overall, the predicted features were consistent with the actual phenotypes: skin color, eye color, hair color, freckling, and ancestry. This phase of the study serves as a preliminary assessment of Level 1 detail so that strengths and limitations could be identified to set up a more in-depth analysis of face morphology in phase 2.