THE HIRISPLEX-S SYSTEM: COMBINED PREDICTION OF EYE, HAIR AND SKIN COLOR FROM DNA

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Forensic DNA Phenotyping (FDP) has become a fast growing topic in forensic research and practice. FDP is expected to aid police investigations by providing physical appearance information on unknown individuals when conventional DNA profiling, or other means of investigation are non-informative. Previously we developed the IrisPlex [1] and HIrisPlex [2] systems for eye and hair color prediction from DNA. Here, we add skin color by introducing the HIrisPlex-S system for combined eye, hair, and skin color DNA prediction. Eye and hair color prediction is obtained via the previously introduced single HIrisPlex SNaPshot multiplex assay targeting 24 SNPs and the eye and hair color prediction models, while a second SNaPshot multiplex is added that targets 18 skin color specific SNPs. For the new skin color prediction model, a total of 36 SNPs from both multiplexes provide population specific AUC prediction accuracy values ranging from 0.76 to 1 for five categories of skin color; very light, light, medium, medium to dark, and dark skin color in a validation set of approx. 240 individuals from nine populations while approx. 960 individuals were used for model building. With this study, we present the HIrisPlex-S system consisting of two multiplex assays and three statistical prediction models enabling combined predictions of all three human pigmentation traits in one DNA test system. Furthermore, we assess the full performance of the system on an additional set of individuals from a US population to substantiate its future application in forensic, missing person, as well as archaeological and anthropological cases.

1. Walsh et al. Forensic Sci Int Genet. 2011,5:170-180.
2. Walsh et al. Forensic Sci Int Genet. 2013,7:98-115.