

IDENTIFICATION OF SINGLE NUCLEOTIDE POLYMORPHISMS (SNPS) INVOLVED IN THE DETERMINATION OF PHYSICAL APPEARANCE

van Daal A¹, Hughes-Stamm S¹, Barash M¹ and Kumar K²

¹Faculty of Health, Science and Medicine, Bond University, Gold Coast, QLD 4229, Australia

²Faculty of Business, Bond University, Gold Coast, QLD 4229, Australia

Forensic DNA profiling is a rapidly evolving field and in the last few years the concept of Forensic Molecular Photofitting has emerged. This new area seeks to obtain additional information from a DNA sample regarding the physical appearance of a person, such as skin, eye and hair pigmentation and more recently, facial morphology.

The goal of this study is to identify a set of single nucleotide polymorphisms (SNPs) involved in pigmentation phenotype as well as normal craniofacial variation and subsequently develop a robust, phenotypically informative forensic assay.

In order to achieve the project goals, DNA samples along with pigmentation phenotype information and 3-D facial images are collected. The samples are (pigmentation) or will be (face) genotyped at a set of candidate SNPs and evaluated for statistically significant associations with pigmentation and anthropometric craniofacial measurements, including cephalic, facial and nasal indices.

To date more than 200 genes and 800 SNPs, potentially involved in craniofacial development have been selected, based on literature review and web resources. SNPs with high Fst values as well as non-synonymous SNPs, SNPs located in splicing sites and transcription binding sites of genes shown to be associated with craniofacial development in human or model organisms have been selected.

This research aims to develop a molecular identikit and enable 'a face to be put to a DNA sample'. This should assist in solving criminal cases, identifying mass disasters victims and investigating historical skeletal remains.