



A176 Craniometrics vs. Single Nucleotide Polymorphism (SNP)s: Is There a Correlation?

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Learning Overview: After attending this presentation, attendees will understand the relationship between craniometrics and specific DNA variations (SNPs) and its value for positive identification purposes.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by investigating specific regions of the human genome to determine how they influence the craniofacial skeletal structure in an effort to improve human identification and facial reconstruction efforts for positive identification of the deceased.

In a world where forensic countermeasures are sophisticated tools to deflect, detect, or solve a crime, it is important to consider the human aspect in forensic science. Human identification has long been a leading tool to hold perpetrators accountable, give families closure, and put a face on a skull. This project is a pilot study to merge three disciplines that fall under the human identification umbrella: forensic anthropology, forensic genetics, and forensic art. Current facial research in genetics focuses on data from living individuals where specific SNPs that influence specific regions of the face have been found.¹⁻³ This study determines if these same regions can be translated to craniometric dimensions (inter-landmark distances) of the underlying skull itself.

The goal of this project is to provide information regarding the correlation of craniometric measurements and SNPs, as well as how these relationships can decrease subjectivity in forensic facial reconstructions and encourage interdisciplinary work within the forensic sciences. This study examined a selection of candidate SNPs currently identified in the literature to determine if there were correlations between inter-landmark distances and those SNPs within the same individual. Craniometric landmark data were collected from 17 documented skulls from the Texas State Donated Skeletal Collection using a 3D MicroScribe® digitizer. Criteria for inclusion in this study included European ancestry, the presence of intact skulls, and the presence of associated donor blood cards collected at the time of body donation. Using these blood samples, DNA from each individual was extracted, amplified, and sequenced through next generation sequencing for the specific chosen SNPs. Afterward, bioinformatics tests were applied to observe the presence or absence of the major or minor alleles in the specific locations on the genome. After determining the presence or absence of an SNP, a set of statistical tests were performed, including: Spearman's correlation between the craniometric measurements and the individual's genetic data variables; two-way hierarchical clustering and Bootstrap Forest modeling between variables that demonstrated significant correlation; a principal component analysis was performed on the craniometric data (inter-landmark measurements) and genetic data (SNP presence/absence) in order to check the homogeneity of each data set; and finally, a pair-wise Procrustes analysis was completed on the correlation of the two data sets as different groups.

The results indicate a correlation in various degrees between the targeted craniofacial regions and the targeted SNPs. There were 11 SNPs that showed significant correlation ($p < 0.05$). However, the correlations were not as expected and showed some interesting results. By group level, there is no significant correlation; however, there is correlation at the individual level. While some SNPs affected the soft tissues only, others showed correlations with the skull (hard tissue), a finding that had not been previously known. These results show that combining craniometric and DNA analyses may provide new tools for forensic anthropological, genetic, and facial artistic approaches to human identification.

Reference(s):

1. K. Adhikari et al. A Genome-Wide Association Scan Implicates *Dchs2*, *Runx2*, *Gli3*, *Pax1* and *Edar* in Human Facial Variation. *Nat Commun* 7 (2016).
2. Peter Claes et al. Genome-Wide Mapping of Global-to-Local Genetic Effects on Human Facial Shape. *Nature Genetics* (2018).
3. J.R. Shaffer et al. Genome-Wide Association Study Reveals Multiple Loci Influencing Normal Human Facial Morphology. *PLoS Genet* 12, no. 8 (2016).

Forensic Genetics, Forensic Anthropology, Human Identification