H13 The Application of Morphometrics to a Candidate Gene Approach for Identifying the Genetic Basis of Facial Morphology

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After attending this presentation, attendees will understand how morphometrics may be used to quantify aspects of facial morphology from 2D images. Attendees will also understand how the data produced may be used in conjunction with genotype information to identify genes affecting facial morphology.

This presentation will impact the forensic science community by introducing a low-cost method for the identification of genes that may prove useful in developing a method for the prediction of facial morphology from DNA. Such a method would provide a novel way to gain information from DNA samples from crime scenes or unknown remains that do not obtain a hit from database searches. Additionally, several of the techniques described herein may be generalized to other genotype-to-phenotype studies.

It has been shown that many aspects of the American criminal justice system result in bias that leads to false convictions. This bias often surrounds the identification of a suspect and may have a continued or compounded effect during the remainder of the criminal proceedings. The discovery of new, objective methods that use DNA to identify suspects could eliminate bias stemming from eyewitness testimony and malpractice in suspect identification. This could lower the number of misidentified suspects arrested and taken to trial, leading to a subsequent reduction in false convictions. It could also aid in the identification of unknown remains by providing an idea of the appearance of the deceased individual. This would help to solve cases that may have gone cold and bring closure to families.

In this study, candidate genes for facial morphology were identified from those implicated in craniofacial disorders and selected Single Nucleotide Polymorphisms (SNPs) found within them were genotyped. Facial morphology was captured from a set of landmarks mapped onto 2D photographs. Variation in the distances between pairs of landmarks was analyzed as a function of genotype for each SNP. Results revealed that specific inter-landmark distances varied significantly (alpha = .05) between genotype groups for two of the SNPs included in the study. This indicated that the genes may influence those aspects of facial morphology. The first of these SNPs is located in the POLR1C gene, which has been implicated in a form of Treacher Collins syndrome. This craniofacial disorder includes hypoplasia of bones in the mid-and lower face in its description.[1,2] The second is located within the LMNA gene, which has been implicated in Restrictive Dermopathy. This disorder includes abnormalities of the midface as part of its phenotype.[3,4] The SNP in the POLR1C gene indicated an effect on several measurements of the mid-and lower face. These measurements were a mixture of overlapping and discrete measurements and covered all three dimensions of the face. The other SNP, located on the LMNA gene, indicated an effect on measurements of the midface around the nose. LMNA's effect was only seen for measurements reflecting height and width of facial features. It was concluded that the POLR1C gene may play a role in the development of the bones of the mid-and lower face and that the LMNA gene may have some effect on the morphology of the midface, specifically the nasal region. The study is currently being expanded to a larger population. This will also involve the consideration of more polymorphisms.

Reference(s):

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Human Identification, Facial Morphology, Morphometric Polymorphism