A90 Method Development for Analyzing SNPs Associated With Stature

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The goal of this presentation is to demonstrate to attendees how SNPs (Single Nucleotide Polymorphism) can be analyzed to predict stature using biological evidence.

This presentation will impact the forensic science community by demonstrating that long bones are no longer needed to predict stature, instead more commonly seen biological evidence samples have the potential to be analyzed to predict stature.

The field of forensic molecular photofitting provides methods for the prediction of physical characteristics by the analysis of associated genetic markers. The prediction of physical characteristics presents scientists with the opportunity to provide investigators with reliable investigative leads and may prevent innocent people from being convicted. Several genetic markers for physical characteristics have already been thoroughly researched by the forensic community including ethnicity, red hair color, and eye color; however, researchers have been less successful with characteristics including stature, body weight, hair color (other than red), and age. Researchers have identified many potential genetic markers associated with stature; however, only limited populations studies have been performed to determine if a true correlation is noted between genotype and adult stature. To provide a predictive range from samples of unknown origin, the additive effect of multiple genetic markers related to stature needs to be evaluated. This study aimed to develop and evaluate a method that could be easily adapted by forensic DNA laboratories that is designed to specifically detect single nucleotide polymorphisms (SNPs) known to have a strong association with adult stature. The SNPs focused on in this study included two confirmed variants associated with stature, rs1042725 within HMGA2 and rs6060369 within GDF5, as well as two other variants found in regions or hotspots within the genome that have been associated with stature, PTCH1 (rs10512248) and BMP2 (rs967417). The SNaPshot Multiplex Kit, a primer based single-extension assay which analyzes up to ten SNPs in one reaction, was used to analyze HMGA2, GDF5, PTCH1 and BMP2. The method development using the SNaPshot Multiplex Kit involved designing and optimizing general primers and SNaPshot primers for HMGA2, GDF5, PTCH1, and BMP2. A sensitivity study was then performed to determine the optimal input of template DNA and to compare the full and reduced volume SNaPshot reaction. Following the method development, the PTCH1 and BMP2 height associated SNPs were successfully incorporated into a SNaPshot assay. The optimal input of template DNA for BMP2 was found to be 2.0ng for both the full and reduced volume SNaPshot reaction while PTCH1 had an optimal input of template DNA of 1.0-1.5ng for the full volume reaction and 2.0ng for the reduced volume reaction. All subsequent reactions were performed using the 2.0ng input of template DNA with the reduced volume SNaPshot reaction. Additionally, the data produced from a stature test pilot study of both males and females ranging from 196-149cm implicated the A allele as the height increasing allele for the PTCH1 SNP rs10512248, which was previously unknown. Lastly, the combined predictive power of only PTCH1 and BMP2 successfully predicted height from known samples with an error rate of 10.05cm. With the analysis of additional SNPs associated with stature, the combined predictive power of SNPs has the potential to produce an error rate similar to, if not better than, the current gold standard anthropological methods for height estimation. The availability of this type of assay for height prediction would eliminate the need for long bone and extend the application for height prediction to more commonly analyzed biological evidence samples including saliva, semen, and blood.

Molecular Photofitting, Stature, Single Nucleotide Polymorphism

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