

B40 The Development of a Biogeographic Ancestry and Phenotype Single Nucleotide Polymorphism (SNP) Panel Using Ion Torrent™ Chef System

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After attending this presentation, attendees will have a better understanding of the performance of the Ion Torrent™ Chef Next Generation Sequencing (NGS) system and development process of an NGS panel for SNP.

This presentation will impact the forensic science community by providing results from a novel development of an NGS panel for SNP in order to predict biogeographic ancestry and phenotype of an individual. This presentation will also increase understanding of the practical utility and benefits of the NGS technology.

It is essential to perform fast and accurate identification of all types of traces that belong to mass disaster victims or unknown biological evidence collected from crime scenes. Only gender identification can be achieved by using standard STR kits. Additional physical characteristics could provide important information regarding suspects/individuals. Recently, SNP markers have been used for identification, determination of phenotype, and prediction of ancestry. “Molecular eyewitness” is defined as the determination of the suspect’s or individual’s population origin by using Ancestry Informative SNPs (AISNPs) and the determination of the suspect’s or individual’s physical characters by using Phenotype Informative SNPs (PISNPs). Since there are a few studies related to geographic region that includes the interest region of Southwest Asia, it is essential to study novel and informative markers for accurate ancestry assignment for this region. Hence, the objective of this study was to develop an SNP panel on Next Generation Sequencing (NGS) devices for use in predicting biogeographic ancestry and phenotype (i.e., eye, hair, and skin color) of an individual.

In this developed panel, this study sought to determine a set of AISNP markers that could be used to differentiate Southwest Asia and Mediterranean regions from Europe and other continental regions; therefore, SNPs were selected from previous research.¹⁻³ The set of PISNP markers was integrated into the set of AISNP markers in order to develop one reaction system on the NGS instrument. PISNPs were used to determine physical characteristics such as eye, hair, and skin colors and were chosen from previously published papers.⁴⁻⁶ As a result of this study, a biogeographic ancestry and phenotype panel, including 160 SNP markers, was developed by using the Ion Torrent™ Chef instrument. The optimization and validation of the system was performed by using commercially available reference samples (e.g., 9947a, 007). Sensitivity and reproducibility parameters were determined for the optimization and the validation of the Ion Torrent™ Chef panel.

In conclusion, the developed NGS SNP panel, which can be used for predicting the biogeographic ancestry and physical characteristics of an unknown, will contribute to the direction of the investigation and to clarify the incident quickly for unknown contributors, cold cases, or the identification of missing persons and disaster victims. In this presentation, results from the NGS panel will be presented in addition to additional evaluations of individual prediction results using statistical regression and Bayesian methods for the forensic use of this panel.

Reference(s):

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