

Peter M. Schneider ISFG Short Term Fellowship Final Report

Applicant:

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The purpose of this research collaboration was to gain knowledge related to bioinformatics, genotype calling methodologies, and genotype imputation for use in forensic investigative genetic genealogy (FIGG). This technology would be beneficial in fulfilling the Defense POW/MIA Accounting Agency's (DPAA) mission to provide the fullest possible accounting for missing personnel dating back to World War II. Many of these service members do not have a family reference specimen (FRS) on file or the reference(s) is beyond 4th degree, which is our current limitation when using the genotype likelihood approach with a custom ~95,000 SNP panel.

Prior to the visit, hybridization capture, with multiple bait panels, was performed at AFDIL on both low quality skeletal and reference samples from three proof of concept FIGG cases. These data were used during the ISFG fellowship to evaluate potential analysis approaches (i.e., genotype calling and imputation) and parameters (e.g., coverage thresholds, allele balance) necessary to generate a reliable profile that could qualify for GEDmatch upload for low quality sample data. Although this evaluation is still ongoing in collaboration with Dr. Andreas Tillmar, valuable theoretical and practical experience was gained in the aforementioned areas. I was introduced to the R software for the purpose of running various scripts for genotype calling and genotype imputation. I also gained hands on experience with the FamLink blind search feature using the low quality and reference sample data, which provided preliminary insight into the quality of the resulting genotypes from the different analysis approaches.

Beyond the main scope of the visit, I was able to learn more about the National Board of Forensic Medicine's (NBFM) implementation of the FORCE panel, ongoing research with the Twist 1.3 million SNP hybridization capture panel, as well as different statistical approaches for kinship prediction. Additionally, I was able to tour the National Forensic Center (NFC) of the Swedish Police Authority to learn more about their routine casework and current research projects. I also had the opportunity to give a brief presentation on the Armed Forces Medical Examiner System with a focus on AFDIL and the work currently being done by the Emerging Technologies Section.

I am very thankful for this opportunity provided by the Peter M. Schneider ISFG fellowship and the NBFM. This visit and the ongoing collaboration lay the foundation for implementing an in-house analysis pipeline for FIGG purposes to generate investigative leads as well as extending kinship analysis capabilities beyond 4th degree relatives.