ISFG Short Term Fellowship 2019 – Post-Visit Report

Applicant:
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Research visit to the Institute for Legal Medicine, Innsbruck Medical University
October 17th 2022 until October 21st 2022

The main goal of this visit was to further the ongoing collaboration to develop high-quality mitochondrial DNA (mtDNA) haplotypes for submission to the EDNAP mtDNA Population Database (EMPOP), particularly whole mtDNA genomes (mitogenomes) using massive parallel sequencing (MPS) technology. Additionally, it was desired to obtain training in statistical tools and methods for the analysis of population mtDNA data. This visit was originally scheduled for March of 2020, but due to the COVID-19 pandemic, the visit was delayed until October 2022.

The Armed Forces DNA Identification Laboratory (AFDIL) has collaborated with the Institute for Legal Medicine (Institut für Gerichtliche Medizin, GMI) at the Innsbruck Medical University for over 20 years. The primary focus of this collaboration has been the development of high-quality mtDNA reference data for submission to EMPOP for the estimation of haplotype frequencies. Over this time period, AFDIL has submitted over 25,000 mtDNA control region (CR) haplotypes from over 200 global populations. More recently, AFDIL has received funding to sequence 8,000 mitogenomes using MPS and these haplotypes will ultimately be submitted to EMPOP. Prior to the release of these data, multiple quality control (QC) checks are performed at both AFDIL and GMI. This visit enabled in-person review and discussion of both CR and mitogenome data submitted to EMPOP by AFDIL over that last two decades.

During this visit, concordance of the mtDNA data in the internal AFDIL database and EMPOP was assessed. This included 24,183 CR samples from 238 populations and 1915 mitogenomes from 14 populations. Additionally, the final QC was performed of 9 populations, which will now allow the release of an additional 1502 mitogenomes in EMPOP. This includes the replacement of CR data with MPS-generated mitogenomes. Furthermore, 2019 mitogenomes from 32 populations were submitted for EMPOP QC. As the data were reviewed, the collaborator information and sample set ethical status was assessed for all populations. In total, mtDNA data from nearly 30,000 samples was reviewed and any discrepancies between the two databases were resolved. Lastly, training was obtained on the use of Arlequin in order to perform the population comparisons and statistic calculations for a large Swedish mitogenome dataset. Overall, this in-person visit was invaluable for the continued effort to develop the highest quality mtDNA reference data for use by the forensic community.