Report ISFG Short Term Fellowships 2017

Applicant: Filipa Isabel Figueira Simão, PhD student at State University of Rio de Janeiro, *Laboratório de Diagnóstico por* DNA (LDD).

Research visit to Institute for Legal Medicine, Innsbruck Medical University, November 2nd 2017 until January 17th 2018.

The main goal of this exchange period was to give continuity to an ongoing collaboration concerning the study the mtDNA profile of South American populations, and to acquiring training in massive parallel sequencing (MPS) technology in the analysis of mitogenomes, for a future implementation in LDD.

South American populations are the most diverse in the world. Their heterogeneity varies from country to country because of different admixture processes along the years of colonization between native Americans, European colonizers and African slaves. For this reason, the inhabitants of South America have been targeted in different studies based on linguistic and anthropological evidences, in an attempt to understand the ancestry of these populations.

Genetic studies demonstrate high diversity within and among South American populations, which is still unrepresented in forensic mtDNA databases.

During my visit we were able to add information on 100 full mitogenomes from Paraguay to the EMPOP database, for forensic purposes, since no mtDNA data has been so far published for the general population of Paraguay. The samples were previously extracted in LDD, Rio de Janeiro, Brazil. The genotyping was performed during the exchange period in the Institute of Legal Medicine in Innsbruck. Library preparation was performed with Precision ID mtDNA panel (Thermo Fisher Scientific), chips were load with Ion ChefTM and samples sequenced using Ion S5TM (Thermo Fisher Scientific).

The obtained results are now being analyzed in collaboration between *Laboratório de Diagnóstico por* DNA and the Institute of Legal Medicine of Innsbruck, hoping for future publication, contributing to a better understanding of the maternal genetic background of Paraguay.

This short-Term Fellowships allowed not only my training in MPS technology in the analysis of mitogenomes, but also to strengthen the cooperation between the two Institutes.