

Report ISFG Short Term Fellowships 2019 (2023)

Applicant: Masinda Pascalina Rafael Nguidi, PhD student at LDD/UERJ

Coming from: DNA Diagnostic Laboratory (LDD), State University of Rio de Janeiro (Brazil)

Visiting at: Institute of Legal Medicine (GMI), Medical University of Innsbruck (Austria)

The initial purpose of the scientific exchange was to acquire knowledge and training in MPS technology and analyze mtDNA data from Nigerian populations with the obtained award in 2019. However, the fellowship was postponed due to the SARS-Cov2 pandemic and the visit was carried out in 2023 (from 22nd May to 16th June).

Therefore, apart from getting training in MPS technologies, a new topic was included as a fellowship purpose, namely, to perform a study on a recently developed MPS tool (the COMBO). The COMBO includes autosomal DNA markers, Y chromosomal SNPs and mitogenome, where a single sequencing reaction results in more than a thousand amplicons. The COMBO tool was developed in the Institute of Legal Medicine (GMI) and is a useful instrument for human individual identification, populational studies, analysis of remains of unidentified persons and so on. It can also be applied in criminal and forensic cases since it combines Forensic DNA Phenotyping markers that allow prediction of phenotype, with data on biogeographical ancestry and visible characteristics.

The fellowship allowed the processing samples that are part of my doctoral project/thesis, which aims to investigate the influences of African populations in South America. In that manner, Colombia is an interesting country to study since it has great contribution from Africa, with a high representation of Afrodescendant individuals, especially in the Caribbean and Pacific regions. Therefore, the goal was to investigate either if there is a contrast of the population from Coast, Central and Inland departments from the Caribbean region. Furthermore, South American countries usually present different admixture patterns and genetic investigations can help to elucidate their history and complex population dynamics.

The fellowship allowed a contact to Massive Parallel Sequencing Technologies, to acquire knowledge about processing genomic data, opportunity to learn about new technologies being developed in the Institute of Legal Medicine (the COMBO, for example) and introduction to analysis with new softwares. It also allowed to maintain the collaborative projects that were already established between the institutions. The fellowship granted me the experience of acquiring knowledge about different strategies in the laboratory, contributing to my personal and academic growth.

The final analysis of the generated data will be carried out at the LDD with support from the Innsbruck team. The results of this collaboration between UERJ and GMI will be included in a manuscript to be submitted for publication to a journal in the field of population and forensic genetics.