

ISFG report Short Term Fellowship 2020

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Research visit to University of Copenhagen, Faculty of Health and Medical Sciences, Department of Forensic Medicine, from February 17th 2020 until March 13th 2020

The ISFG Short Term Fellowship granted me the opportunity to travel to the University of Copenhagen, Faculty of Health and Medical Sciences, Department of Forensic Medicine. The main goal of this exchange was to assess the resolution for Southern-American specific haplogroups of a Y-SNP multiplex (Ralf et al., 2019), generating and analyzing Y-SNP MPS data, and to obtain training in massive parallel sequencing (MPS) technology.

The heterogeneity of South American populations varies among countries because of different admixture processes through years of colonization, involving South American inhabitants, European colonizers and African slaves.

The data generation and analysis involved DNA extraction of Southern American unrelated male samples, the amplification of the target DNA, library building, library quantification, sequencing and data analysis. DNA was extracted from FTA cards with the EZ1[®] DNA Investigator Kit. The samples were quantified using Qubit[®] and amplified in a PCR reaction following the Ion AmpliSeq[™] Library Preparation for Human Identification Applications user guide. DNA libraries were constructed using the Ion AmpliSeq[™] Library Kit. The purified libraries were quantified with qPCR. Template preparation and chip loading were executed by Ion Chef[™]. Samples sequencing was performed in the Ion S5[™] instrument.

In conclusion, the financial support from the ISFG Short Term Fellowship Award 2019 was crucial to remarkably enrich my research experience and get acquainted with the cutting-edge MPS technology useful to address challenging forensic DNA-oriented questions.

Reference:

Ralf, A., van Oven, M., González, D. M., de Knijff, P., van der Beek, K., Wootton, S., ... & Kayser, M. (2019). Forensic Y-SNP analysis beyond SNaPshot: high-resolution Y-chromosomal haplogrouping from low quality and quantity DNA using Ion AmpliSeq and targeted massively parallel sequencing. *Forensic Science International: Genetics*, 41, 93-106.