Fabio Oldoni - Short Report - ISFG Short-Term Fellowship Award 2017

Washington DC, 1/11/2017

Dear ISFG Fellowship Review Board and Treasurer,

My exchange visit to the Forensic Molecular Biology laboratory lead by Associate Prof. Podini at The George Washington University (GWU) in the US took place between mid-May and end-August 2017.

The main purpose of my visit was to acquire valuable hands-on experience on the use of the cutting-edge Ion Chef™ and Ion S5™ (Thermo Fisher Scientific) massively parallel sequencing (MPS) DNA technology. In particular, the main goal of the MPS project I worked on was to develop a novel microhaplotype (MH) forensic assay and preliminary evaluate its potential to provide valuable information on (1) biogeographic ancestry prediction and (2) mixture deconvolution.

Microhaplotype (MH) is a novel insightful marker that presents some advantageous features over conventional STRs encompassing same allele size, absence of stutter fragment, and lower mutation rate. A 74-MH panel was developed and run on the Ion Torrent™ MPS platform, which enables the clonal sequencing of each DNA strands, and thus parental haplotypes at a specific MH locus to be distinguished. The 74-plex MH assay was used to genotype an initial set of population samples of African-American, European-American, South West Hispanic origin, to predict the ancestry of some representative test samples and assess the MH's detection limit to target the minor contributor of few synthetic unbalanced two-person DNA mixtures.

The preliminary and promising results of this project presented by Prof. Podini at the ISFG congress 2017 held in Seoul and recently published on Forensic Science International Genetics: Supplement Series (https://doi.org/10.1016/j.fsigss.2017.09.209) demonstrate that MHs can be an effective tool for ancestry prediction and mixture analysis and also for enhancing and complementing further standard STR analysis for human identification purposes.

In conclusion, there is no denying that the ISFG Short-Term Fellowship Award 2017 was definitely a great financial support that allowed me to remarkably enrich my research experience and get acquainted with the cutting-edge MPS technology useful to address challenging forensic DNA-oriented questions.