ISFG Short-Term Fellowship – Short Report

Project title: X-chromosome markers: population data, mutation and segregation modelling

Name: Nádia Pinto, PhD, Institute of Molecular Pathology and Immunology of the University of Porto and Center of Mathematics of the University of Porto

Host laboratory: DNA Diagnostic Laboratory (LDD), State University of Rio de Janeiro (UERJ), Brazil.

Host: Prof. Dr. Leonor Gusmão

Dates: 07.01.2019 - 18.01.2019

The main goal of the visit was the analysis of the data collected under the framework of three GHEP-ISFG working commissions: 1. "Population data XSTRs", 2. "Segregation XSTRs", and 3. "Study of mutations in a set of 12 X-STRs". Data collected in working commissions 1. and 2. concerns markers of X-STR Decaplex (see https://ghep-isfg.org/pt-pt/working-commissions-pt/history-pt/sex-chromosomes-pt/), while in 3. markers of Investigator Argus X-12 QS Kit, Qiagen, were considered (see https://ghep-isfg.org/pt-pt/estudo-mutacoes-num-conjunto-12-x-strs/).

Due to practical reasons, we gave priority to the analysis of data obtained under the framework of working group 3. "Study of mutations in a set of 12 X-STRs". Data from 18 laboratories worldwide, comprising 1699 father/mother/daughter trios – 40,776 allelic transfers, was then analysed. Laboratories were contacted to provide clarifications on the genetic data whenever it was considered necessary. From the data, 165 genotypic inconsistencies compatible with the occurrence of mutation were detected, and locus specific mutation rates were computed for both maternal and paternal meiosis. Also correlations with the age of the parents in the time of the birth of the daughter were performed. Population analyses were also computed as we inferred from the data 2,756 unrelated haplotypes from 16 populations. A draft of one article was accomplished.

The framework how working groups 1 and 2 should be integrated and developed, in a near future, was also discussed.