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Att: ISFG Fellowship Review Board
Subject: Final Report on Short Term Fellowship of the International Society for Forensic Genetics.

After more than 2 years where the Covid-19 pandemic has postponed my planned Research Stay at the DNA Diagnostic Laboratory, State University of Rio de Janeiro in Brazil, I was finally able to travel to Rio de Janeiro from 21st January to 18th February this year. Since the original 2019 application, the initial plan of working on Ancestry Informative Markers had to be readjusted. It was agreed that the purpose of my visit would be to solidify and formalize the collaboration between the two institutions, while focusing on producing a manuscript on a topic relevant to both parties – X-chromosomal markers and their application in forensic casework.

X-chromosomal markers are useful in the analysis of biological kinship, especially in complex situations of identification through relatives, where autosomal markers are not able to produce conclusive results. The usefulness of X-chromosomal markers applied to complex kinship analyses prompted the development of genotyping methodologies for X-STRs, and the screening of allele/haplotype frequency distributions in many populations worldwide.
For any genetic marker to be used in forensic casework, it is crucial to know the genetic profile of the relevant populations. The evaluation of forensic genetic evidence is based on likelihood ratio principles, considering the probability of the observed profile(s) under two alternative and mutually exclusive hypotheses. The probability is usually calculated based on population allele frequencies. However, in the presence of linkage disequilibrium when marker independence can no longer be assumed, haplotype frequencies need to be estimated in the population rather than using the information of single locus allele frequencies. This usually means that very large population samples are needed for a proper estimation of the haplotype frequencies in a population.
Another important parameter to consider in the application of X-chromosomal markers in kinship analyses is the mutation rate. The studies carried out so far on X-STRs show a behavior similar to that of autosomal STRs. X-chromosomal marker mutation rates present great variation between and within STRs, that depends on the number and structure of the repetitive motif, and the gender and age of the parents.
During this month I had the opportunity of working on a study that aimed to generate and compile data on X-STRs in different populations worldwide, as well as data on segregation analyses in father/mother/son trios. These data comprised 19 X-STRs widely used in forensic genetics, which have been described in two PCR multiplexes with three overlapping loci: one with 10 X-STRs (X-Decaplex) and the other with 12 X-STRs (Argus X-12).
A joint analysis of the data obtained with those available in the literature allowed for the comparison of the different populations, and the establishment of diversity and substructure levels. Additionally, more thorough mutation rates were estimated based on segregation analyses on father/mother/daughter trios.
The ISFG fellowship for this short-term visit to the DNA Diagnostic Laboratory has given me an important opportunity to continue a collaborative work, and in particular, for the establishment of this project and to generate results and work on the manuscript resulting from this work.