

## ISFG Short Term Fellowship Report:

### Purpose and outcomes of the visit to Institute of Legal Medicine, Medical University, Innsbruck, Austria

The research visit to the Institute of Legal Medicine, Medical University, Innsbruck, enabled by the ISFG Short Term Fellowship award, was realised in the period from 17<sup>th</sup> to 29<sup>th</sup> September 2018. Visit was supervised by Professor Walther Parson.

The main purpose of this collaboration was to perform more detailed bioinformatics analysis on sequences of whole mitochondrial genomes obtained by massively parallel sequencing (MPS) from Croatian population samples, with particular attention to forensically relevant aspects of analysis, as well as to interpretation of results. During my time in Innsbruck, through discussions and practical work in data analysis (performed on Croatian population dataset itself, comprising 300 samples), the following goals were achieved:

- Population samples were quality checked;
- All samples with quality warnings were reviewed;
- Review of samples included alignment check, nomenclature corrections, confirmation of newfound mutations (transitions, transversions, insertions, deletions, ambiguous bases) in separate software tool;
- Comparison with Sanger sequencing data of the same Croatian samples was also employed;
- Mitochondrial haplogroups of samples were checked, and differences to the original calls were discussed;
- Quasi-median networks software was used to detect errors in the dataset;
- Discussions with host-laboratory experts covered review of raw MPS data, thresholds for analysis, different software solutions used for analysis, interpretation of mtDNA-specific phenomena (point heteroplasmy, length heteroplasmy, appearance of NUMTs, anomalies in homopolymeric stretches) as well as recognition of sample degradation, phylogenetic alignment of sequences, phylogenetic analysis of whole dataset, recognition of errors in dataset using quasi-median networks and how to interpret them, different protocols for preparation of samples for sequencing, manual check of haplogroups determined by software tool, consideration of candidates for novel branches of global mtDNA phylogenetic tree, databases for forensic purposes (both in-house-developed and one available online).

Collaboration with experienced forensic scientists in Innsbruck resulted in more finely defined guidelines for analysis and interpretation of data obtained from MPS of whole mtDNA. We ascertained that Croatian population sample produced good quality data, which, after a more complete analysis with newly acquired knowledge, will be used for implementation of MPS method into routine forensic casework in Croatia and foundation of national forensic mtDNA database. It is planned for this collaboration to result in a joint publication of Croatian population study on whole mtDNA using MPS between Institute of Legal Medicine (Innsbruck) and Forensic Science Centre (Zagreb).

In conclusion, the ISFG Short Term Fellowship provided great opportunity to deepen the knowledge and skills in analysis and interpretation of MPS data, through work and discussions with forensic experts in the field.

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