



PRE-CONGRESS WORKSHOP

MPS mtDNA: Analysis and interpretation

Monday, September 9, 2024 – 09:00 – 13:00 h

Walther Parson, Charla Marshall

In the past decade, the forensic field has witnessed major developments in mitochondrial DNA (mtDNA) analysis, particularly in association with Massively Parallel Next Generation Sequencing (MPS) techniques. While Sanger-based mtDNA control region sequencing is still suitable for many samples and widely performed in our field, many cases benefit from the increased sensitivity and discrimination power of full or partial mitogenome sequencing using MPS.

This workshop showcases the highlights of recent technical developments and discusses the benefits and limitations of applying MPS to human identification and criminal casework. This includes an overview of current methods from PCR-based to capture-based techniques, analytical details such as the interpretation of sequence data with particular focus on point and length heteroplasmy, NUMTs, damage patterns, artefacts and mixtures, as well as the generation of a consensus mitotype and its interpretation in forensic settings. We discuss features available on EMPOP, such as frequency searches, alignment and haplogrouping.

The workshop provides a brief introduction to the biology of mtDNA and is suitable for beginners and experts, practitioners, students and academics. The learning outcomes include

- i) understanding analysis and interpretation of mtDNA sequences,
- ii) the phylogenetic concept of mtDNA for frequency estimates, sequence alignment and haplogrouping and
- iii) learning how to report mtDNA evidence.

In order to demonstrate the implementation of the learned material in practice, we show the results of an ongoing joint research project on human identification in a complex historical case in a parallel track.

Additionally, participants are invited to send sequences or cases that they came across to be included in the workshop. Please contact the teachers prior to the workshop!