

ISFG Pre-congress workshop

Bioinformatics MPS: Basic and/or advanced

Tuesday September 10th 2024

This workshop consists of a basic (morning session) and an advanced part (afternoon session). Participants can subscribe to one or both workshops. It is not mandatory to follow the basic course to sign up for the advanced course.

The advanced workshop will include hands-on exercises, for which it is required that participants bring their own laptops on which the software is installed. Further instructions will be provided prior to the workshop.

Bioinformatics MPS: Basic

This workshop will focus on the availability of sequencing technologies and commercial kits, the laboratory procedures utilized to generate sequence data, first- and third-party software workflows for data analysis, best practices of reporting sequence data in forensic genomics, and basic analysis of Massively Parallel Sequencing (MPS) data with the STRait Razor Suite and FDSTools software will be demonstrated. Attendees will be instructed on the usage of primary (STRait Razor v3 and FDSTools2.0) and secondary analysis (STRait Razor Online) tools to analyze data from commercially available kits. Finally, recommendations will be provided regarding MPS Short Tandem Repeat (STR) allele nomenclature and some useful online resources will be introduced, including STRSeq, STRidER, and the Forensic Sequence STRucture Guide.

Attendees should have general understanding of massively parallel sequencing of DNA markers including, but not limited to, concepts such as DNA libraries, STR repeats, and single-nucleotide polymorphisms (SNPs).

Intended audience

- Having an understanding of general constructs of DNA markers (e.g., STR repeats and SNPs)
- Anyone with general interest in the use of MPS in the forensic field
- No prior experience is required

Learning outcome

- Get an overview of sequencing platforms and (commercial) forensic kits available
- Get a broad overview of the lab protocols and software workflows
- Learn about sequence nomenclature, reported range, compatibility with capillary electrophoresis
- Learn about online resources such as STRidER and STRSeq

Preliminary program

Time schedule (CEST)		Subject
5 min	9:00 – 9:05	Hello, world
15 min	9:05 – 9:20	Introduction to Massively Parallel Sequencing
40 min	9:20 – 10:00	Sequencing Platforms
15 min	10:00 – 10:15	Break
45 min	10:15 – 11:00	Library Preparation Methods
45 min	11:00 – 11:45	Data Analysis Software Overview
10 min	11:45 – 11:55	Break
35 min	11:55 – 12:30	STRNaming: STR Nomenclature and Compatibility
20 min	12:30 – 12:50	Online Resources: STRidER and STRSeq
10 min	12:50 – 13:00	Questions

Bioinformatics MPS: Advanced

This workshop will focus on employing the STRait Razor Suite and FDSTools software for analysis of Massively Parallel Sequencing (MPS) data. Attendees will be instructed on the usage of primary (STRAit Razor v3 and TSSV) and secondary analysis (STRAit Razor Online and FDSTools) software to analyze data from commercially available kits. Additionally, guidance will be given for modifying the default settings, developing configuration files for both primary and secondary analysis of laboratory-developed tests (e.g., in-house PCR multiplex assays), and considerations for further processing the output. The strengths of and rationale behind the different methods will be explored by examining the results. This way, attendees will gain a deeper insight into the powers and challenges (and solutions!) that arise from the application of MPS to forensic samples.

Attendees should be familiar with general constructs of DNA markers and formats of standard sequencing data storage file types (e.g., BED, FASTQ, SAM/BAM, etc.). While no formal familiarity with any programming languages (e.g., R, Python, etc.) is required, modification of open-source materials may require some basic understanding of computer programming.

Intended audience

- Having a general understanding of MPS raw output data (i.e., reads)
- Preferably having prior experience using first-party analysis software (e.g., ForenSeq UAS or Ion Torrent Browser)
- Understanding of the format and function of MPS workflow file types (e.g., BED, FASTQ)
- General comfort or willingness to utilize command line interface to install and execute commands

Learning outcome

- Get hands-on experience analyzing MPS data with open-source software
- Learn the process of alignment, bracketing of repeats, and visualization of reads
- Learn how to analyze data generated with an in-house developed kit
- Learn how command-line software can be used to automate data analysis workflows

Preliminary program

Time schedule (CEST)		Subject
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5 min	14:00 – 14:05	Hello, world
10 min	14:05 – 14:15	Workshop Overview and Onboarding
30 min	14:15 – 14:45	FDSTools Primary Analysis (TSSV), Nomenclature, and Sample Visualization
45 min	14:45 – 15:30	FDSTools Secondary Analysis: Noise Correction, Interpretation, Automation
15 min	15:30 – 15:45	Break
30 min	15:45 – 16:15	STRait Razor v3 Primary Analysis
45 min	16:15 – 17:00	STRait Razor Online Secondary Analysis: UI Walkthrough
10 min	17:00 – 17:10	Break
25 min	17:10 – 17:35	Creating Your Own FDSTools Noise Correction Database
25 min	17:35 – 18:00	Custom Analysis: Laboratory-Developed Assay Recommendations
15 min	18:00 – 18:15	Questions