**EJP RD**

European Joint Programme on Rare Diseases

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Rare Disease European Joint Programme Cofund

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**Variant Prioritization and Enrichment for Rare Diseases Workshop**

20.09.2023 – 22.09.2023

**Istanbul**

The connections links are inserted in the Program (at a glance and per day).

Please click on the “Sessions” title (or text) inside each box of the program to get directed to the online meeting Room.

Simple, step by step and quick **“Connection Instructions & User-Guide”** are provided page 48.

Please read them before the start of the meeting

This online meeting is **recorded**. If you do not wish to have your image appear, please make sure to turn off the camera before joining the sessions (see instructions below).

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# Proposed dates and attendees

Dates: 20.09.2023 – 22.09.2023

Format: Face to Face

Attendees: ERNs, EJP RD

Audience: ERNs, EJP RD

# Aims

Participants will learn the fundamentals of Whole Exome Sequencing (WES) including terminology, pre and post-alignment processing, variant calling using Genome Analysis Toolkit (GATK) pipeline and annotation/filtration/interpretation of the variants. Moreover, Polygenic Risk Score (PRS) Analysis, predatoR: Pathogenicity Prediction tool, Wikipathways, Multi-omic Data Analysis and Pathway Enrichment Methods will be covered.

# Agenda

**Day 1**

|  |  |
| --- | --- |
| **Time** | **Topic** |
| 09:00-09:15 | Opening Remarks |
| 09:15-10:30 | Basics of Whole Exome Sequencing (WES) |
| 10:30-11:00 | *Coffee break* |
| 11:30-12:30 | Pre-alignment Processing and Alignment |
| 12:30-13:30 | *Lunch* |
| 13:30-15:00 | Post-alignment Processing |
| 15:00-15:30 | *Coffee break* |
| 15:30-17:00 | Variant Calling and Genotyping |

**Day 2**

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| --- | --- |
| **Time** | **Topic** |
| 09:00-10:30 | Annotation of Variants |
| 10:30-11:00 | *Coffee break* |
| 11:30-12:30 | Filtration, Variant Prioritization and Further Analysis |
| 12:30-13:30 | *Lunch* |
| 13:30-15:00 | Polygenic Risk Score (PRS) Analysis, predatoR: Pathogenicity Prediction Algorithm |
| 15:00- | Istanbul Tour |

**Day 3**

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| --- | --- |
| **Time** | **Topic** |
| 09:00-10:30 | Introduction to Wikipathways |
| 10:30-10:45 | *Coffee break* |
| 10:45-12:45 | Multi-omic Data Analysis and Pathway Enrichment Methods |
| 12:45-13:30 | *Lunch* |
| 13:30-15:00 | Hands-on session |
| 15:00-15:30 | *Coffee break* |
| 15:30-17:00 | Hands-on session |
| 17:00-17:15 | Closing Remarks |

# Program per day

20.09.2023- Day 1

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| 09:00 | Morning session | 09:00-09:15  Opening Remarks | 09:15-10:30  Basics of Whole Exome Sequencing (WES) | 10:30-11:00  *Coffee break* | 11:00-12:30  Pre-alignment Processing and Alignment |
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| 12:30 |

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|  | Lunch |  | | | |
| 13:30 | Afternoon session | 13:30-15:00  Post-alignment Processing | 15:00-15:30  *Coffee break* | 15:30-17:00  Variant Calling and Genotyping |
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| 17:00 |

21.09.2023- Day 2

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| 09:00 | Morning session | 09:00-10:30  Annotation of Variants | 10:30-11:00  *Coffee break* | 11:00-12:30  Filtration, Variant Prioritization and Further Analysis |
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| 12:30 |
|  | Lunch |  | | | |
| 13:30 | Afternoon session | 13:30-15:00  Polygenic Risk Score (PRS) Analysis, predatoR: Pathogenicity Prediction Algorithm | 15:00-  Istanbul Tour |
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| 15:00 |

22.09.2023- Day 3

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| 09:00 | Morning session | 09:00-10:30  Introduction to Wikipathways | 10:30-10:45  *Coffee break* | 10:45-12:45  Multi-omic Data Analysis and Pathway Enrichment Methods |
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| 12:45 |

Lunch

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| --- | --- | --- | --- | --- | --- |
| 13:30 | Afternoon session | 13:30-15:00  Hands-on session | 15:00-15:30  *Coffee break* | 15:30-17:00  Hands-on session | 17:00-17:15  Closing Remarks |
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| 17:15 |