# Summer School (EIII): General information

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| Place: | Skovskolen (UCPH conference location), Fredensborg, Denmark |
| When: | 13-17 September 2021 |
| Length: | 5 days (5 days compulsory for ESRs; Days 1 compulsory for all beneficiaries and partners) |
| Attendants: | ESRs, Beneficiaries, Partners, Guest speakers (around 30-40 people) |
| Venue: | Campus for accommodation with breakfast and dinner; Seminar room/Lecture Hall for training activities and lunch at facilities from the University of Copenhagen |

# Program Summer School (EIII)

## Day 1 (compulsory for all ESRs, beneficiaries and partners)

### *Morning session (9:00-12:30)*

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| 9:00 – 9:15 | Welcome |
| 9:15 - 10:15 | Seminar: Stephan Mundlos |
| 10:15 – 10:45 | Coffee Break |
| 10:45 – 12.45 | ESR poster session |

### *Lunch Break (12:45-14:00)*

Lunch at the campus

### *Afternoon session (14:00 – 18:00)*

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| 14:00 - 16.30 | ESR poster session |
| 16:30 – 17:00 | Coffee Break |
| 17:00 – 18:00 | Management Meeting for PIs |

### *Dinner (19:30-21:00)*

Welcome Dinner

## Day 2 (compulsory for all ESRs and participants involved in training)

### *Morning session (8:30-12:30)*

Networking time

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| 10:15 – 10:45 | Coffee Break |

### *Lunch Break (12:30-14:00)*

### *Afternoon session (14:00-17:45)* Medical genetics II (Jorge Ferrer, Contributors: Sebastian Waszak, Victor Sanchez-Gaya, Torben Hansen, Ernest Turro)

From GWAS and whole-genome sequencing to causal genetic variants: *Overview of experimental and computational tools available to identify non-coding disease-causative genetic variants; Calling SNVs and structural variants in whole genome sequence data; Machine learning approaches to prioritise pathogenic mutations in enhancers; Statistical genetic analysis of common regulatory variants, eQTLS.*

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| 14:00 – 15:00 | Sebastian Waszak, noncoding genome variation in cancer |
| 15:00 - 15:30 | Prediction of pathogenic structural variants (Victor Sanchez-Gaya, University of Cantabria, Santander, Spain) |
| 15:30 – 16:00 | Coffee Break |
| 16:00 – 17:00 | Torben Hansen (Copenhagen) Genetic discoveries in isolated human populations |
| 17:00 - 17:45 | Ernest Turro (university of Columbia, NY, note time difference). Rare disease diagnosis with whole genome sequencing. The 100 thousand genomes experience. |

### *Dinner (19:30 – 21:00)*

## Day 3 (compulsory for all ESRs and participants involved in training)

### *Morning session (8:30-12:30)* Technologies II (Wouter de Laat; Contributors: Jop Kind, Alexander van Oudenaarden)

Topological control of gene expression and enhancer function: *Introduction to 3C-related technologies (e.g. 4C-seq, Hi-C, ChIA-PET, HiChIP, C-HiC, MC-4C);*

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| 8:30 – 9:45 | Wouter de Laat: C-technologies and microscopy methods for 3D genome measurements |
| 9:45 - 10:15 | Coffee Break |
| 10:15 - 11:15 | Jop Kind (Hubrecht Institute) |
| 11:15 - 12:15 | Alexander van Oudenaarden |

***Lunch Break (12:30-14:00)***

## *Afternoon session (14:00-?)* Communication I – Theory (Gioacchino Natoli; Potential Contributors: Dariusz Plewczynski, Jean- Mehdi Grangeon, Meritxell Alberich-Jorda)

Presenting scientific work: *A critical transferable skill in science is based on the ability to present our data to the scientific community. In this workshop, ESRs will be presented with several tools to improve their oral and poster presentation skills.*

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| 15:30 – 16:00 | Coffee Break |

***Dinner (19:30-21:00)***

## Day 4 (compulsory for all ESRs and participants involved in training)

## *Morning session (8:30-12:00)* Bioinformatics II – Practice (Dariusz Plewczynski; Contributors: Pelin Sahlén, Amin Allahyar)

Computational tools for 3C-related data: *processing, statistical analysing modelling and visualization of 4C-seq, Hi-C, ChIA-PET, HiChIP, Capture-HiC. Theoretical overview and practical exercises*

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| 8:30 - 9:30 | Hi-C and adapted technologies (Amin Allahyar post-doc at Hubrecht Institute, Pelin Sahlén) |
| 9:30 - 10:30 | Multi-contact 4C, digital 4C (Amin Allahyar post-doc at Hubrecht Institute) |
| 10:30 - 11:00 | Coffee Break |
| 11:00 - 12:00 | Capture Hi-C (Pelin Sahlén) |

***Lunch Break (12:30-14:00)***

### *Afternoon session (14:00-17:00)* Bioinformatics II – Theory (Dariusz Plewczynski; Contributors: Pelin Sahlén, Amin Allahyar)

Computational tools for 3C-related data: *processing, statistical analysing modelling and visualization of 4C-seq, Hi-C, ChIA-PET, HiChIP, Capture-HiC. Theoretical overview and practical exercises*

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| 14:00 - 15:30 | 4C data analysis (Amin Allahyar post-doc at Hubrecht Institute, Pelin Sahlén) |
| 15:30 - 16:00 | Coffee Break |
| 15:00 - 17:00 | Capture Hi-C data analysis (Pelin Sahlén) |

### *Dinner (19:30-22:00)*

## Day 5 (compulsory for all ESRs and participants involved in training)

### *Morning session (8:30-12:30)* Medical genetics III (Terranova (Novartis), Contributor: Giorgio Galli, Kathleen Sprouffske, Felix Lohmann)

Pharmacogenomics and gene-environmental interaction

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| 8:30 – 8:50 | NIBR partner introduction and overview of potential hosts (Remi Terranova) |
| 8:50 – 9:30 | New therapeutic target identification and characterization - Functional epigenomics of transcriptional regulatory elements (Giorgio Galli, Kathleen Sprouffske) |
| 9:30 – 10:00 | Coffee Break |
| 9:45 – 10:30 | Targeting the enhancer landscape in regenerative medicine - BET bromodomain inhibition in skin wound healing (Felix Lohmann) – 45 min incl QA |
| 10:30 – 11:15 | Epigenomics and regulatory genome – impact for translational drug safety (Remi Terranova) - 45 min incl QA |