

# Advanced course on long-read sequencing and medical genomics

## PRELIMINARY AGENDA

## WELCOME

**08:30–08:50 - Registration**

**08:50–09:00 - Welcome and course objectives**

Joris Vermeesch, KU Leuven

*Head of Laboratory for Cytogenetics and Genome Research*

## BLOCK 1 – TECHNOLOGIES AND DATA ANALYSIS

**09:00–9:40 - Long-read sequencing: Technology update and applications**

Wouter Bossuyt, KU Leuven

*Manager of the Genomics Core Leuven, Center for Human Genetics*

**09:40–10:20 - Getting the most out of nanopore sequencing: From sample to library prep**

Mojca Strazisar, University of Antwerp

*Head of the Neuromics Support Facility, Center for Molecular Neurology, VIB*

**10:20–11:00 - Processing, QC and analysis of long-read RNA-seq data**

Kristin Köhler, Charité

*Bioinformatician and PhD Student at the Intelligent Imaging Group, Berlin Institute of Health*

**11:00–11:20 - Coffee break I (20 min)**

**11:20–12:00 - Handling and sharing long-read data**

Teresa D'Altri, CRG

*Scientific project manager and coordinator, EGA Team, Centre for Genomic Regulation*

## BLOCK 2 – APPLICATIONS IN MEDICAL GENOMICS

**12:00–12:40 - Long-read sequencing for rare kidney disease**

Janine Altmüller, MDC

*Head of Genomics Core Facility, Berlin Institute of Health at Charité and Max Delbrück Center for Molecular Medicine*

**12:40–13:30 - Lunch break (50 min)**

**13:30–14:10 - Long-read sequencing in neuromuscular disorders (African population)**

Pedro Rodríguez Cruz, CNAG

*La Caixa Junior Leader Fellow, Centro Nacional de Análisis Genómico*

**14:10–14:50 - Nanopore long-read whole-genome sequencing in developmental disorders**

Joris Vermeesch, KU Leuven

*Head of Laboratory for Cytogenetics and Genome Research*

**14:50–15:00 – FLASH TALK: CiFi long-read chromatin contact mapping to decode multi-way 3D genome interactions**

Nelson Martins, UGent

*UGent - PhD Student, Department of Biomolecular Medicine, Ghent University, Ghent & Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium*

**15:00–15:15 - Coffee break II (15 min)**

**15:15–15:55 - From cytogenetics to cytogenomics: Can we replace traditional cytogenetics with HiFi long-read sequencing?**

Robert Månsson-Welinder, NGI

*Head of Unit, National Genomics Infrastructure, KTH Royal Institute of Technology*

**15:55–16:35 - TBD**

**16:35–17:15 - Analysis of leukemia fusion gene expression at single-cell level**

Henrik Gezelius, Uppsala University

*Researcher, Department of Medical Sciences, Molecular Precision Medicine*

**17:15–17:25 – FLASH TALK 2**

**BLOCK 3 – ASK THE EXPERTS: YOUR QUESTIONS ON MEDICAL GENOMICS**

**17:25–17:55 - Panel Q&A – “Everything you wanted to ask about long-read sequencing”**

All Speakers

**17:55–18:00 - Closing Remarks and next steps**

Organising Committee