



AGENDA

For the kick-off 2025

16th -17th of April 2025

Wilhelmstrasse 3
72074 Tübingen

Germany

<https://med-uni-tuebingen-de.zoom-x.de/j/65225341517?pwd=ktVGgCXPPjuw35MJh8z7f6GeBapiEJ.1>

if something urgent arises:

Prof. Olaf Rieß

+49 173 3014957

16th of April 2025

Time	Topic	Further Info
12.00h	Welcome and introduction by Olaf Rieß	
12.30h	Start of session one: chaired by Olaf Rieß	
12.30h-12.40h	Short introduction of Institute of Life Sciences, HES-SO Valais-Wallis	Alexandre Kuhn
12.45h-12.55h	Short introduction of Aarhus University Hospital	Ebbe Bak & Ole Larsen
13.00h-13.45h	Lunch break	
13.45h-13.55h	Short introduction of University Hospital Aachen	Florian Kraft
14.00h-14.10h	Short introduction of Genomics Research and Development Institute from Bucharest	Octavian Bucur (ONLINE)
14.15h-14.25h	Short Introduction of KU Leuven	Joris Vermeesch
14.25h-14.45h	Coffee/Tea break	
14.45h	Start session two: chaired by Barbara Bizjan	
14.45h-15.10h	Complex structural variants and repeat expansions in long read sequencing 15min, 10min discussion	German Demidov IMGAG Tübingen
15.10h-15.35h	The Future of Oxford Nanopore Sequencing in Genetic Disease Testing 15min, 10min discussion	Cerissa French Oxford Nanopore Technologies
15.35h-15.45h	Introduction to bioinformatics of IrGS 10min, 5min discussion	Stephan Ossowski IMGAG Tübingen
15.45h-16.10h	Nanopore Data Analysis - Technology Update 15min, 10min discussion	Andreas Klingenhoff Oxford Nanopore Technologies
16.10h-16.35h	Benchmarking SV Calling for Fast and Comprehensive Chromosomal Rearrangement Characterization via Nanopore Sequencing 15min, 10min discussion	Quentin Testard (ONLINE) University Hospital of Lyon
16.35h-17.00	(TBA)	Uwe Samer NVIDIA
17.00h	End of the day	
18.00h	Social event: Stocherkahn; Wöhrdstraße 25 (follow the path to the water)	
19.30h	Social event: dinner at Neckawa Tübingen, Wöhrdstraße 25 72072 Tübingen	

17th of April 2025

Time	Topic	Further Info
8.30h-8.45h	Arrival and Coffee/Tea	
8.45h	Start session one: chaired by Ramya Potabattula	
8.45h-9.00h	Introduction WG Wet-lab 10min, 5min discussion	Ebbe Bak Aarhus University Hospital
9.00h - 9.25h	The Quest for the Longest Read: From Sample to Data 15min, 10min discussion	Jernej Kovac University Medical Centre Ljubljana
9.25h - 9.50h	Implementing IrGS in diagnostics 15min, 10min discussion	Tjakko van Ham Erasmus MC Rotterdam
9.50h - 10.15h	Fetal methylation signatures using ONT: first steps 15min, 10min discussion	Gijs Santen (ONLINE) Leiden University Medical Center
10.15h - 10.40h	Solve the unsolved 15min, 10min discussion	Laura Ebner University of Applied Sciences and Arts, Switzerland
10.40h-11.00h	Coffee/Tea Break, picture taken	
11.00h	Start session two	
11.00h - 11.25h	Imprinting disorders in germline and cancer 15min, 10min discussion	Florian Kraft University Hospital RWTH Aachen
11.25h - 11.50h	Long-Read Sequencing makes diagnostic testing of pseudohypoparathyroidism a one-step analysis 15min, 10min discussion	Ole Larson Aarhus University Hospital
11.50h - 12.15h	Long-Read Sequencing in the Annotation of Genome Diversity in Eastern European Populations: Insights from Ukraine's T1D Initiative 15min, 10min discussion	Taras Oleksyk Oakland University
12.15h - 12.40h	T2T sequencing of human genome on ONT platform 15min, 10min discussion	Ramya Potabattula IMGAG Tübingen
12.40h - 13.05h	De novo assembly of the H9 cell line 15min, 10min discussion	Erika Souche KU Leuven
13.05h	Closing remarks by Olaf Rieß	
13.15h	End of kick-off	