



## PROGRAMME - To register click [here](#).

**05 May 2026**      **NGS Technologies**  
 Chair                Holm Graessner, Tübingen (Germany)

TIME (CET)	TOPIC	SPEAKER
9:00-9:45	RNAseq in RD	Diana Baralle Human Development and Health, Faculty of Medicine, University of Southampton, United Kingdom
9:45-10:30	Episignatures in Diagnostics of Rare Diseases	Camille Charbonnier Cancer and Brain Genomics, Inserm UMR1245, Université de Rouen Normandie, France
10:30-11:15	Bioinformatic Tools for Diagnosing Rare Diseases	Christian Gilissen Genome bioinformatics, Radboud University Medical Center, Nijmegen, The Netherlands
11:15-11:30	<i>Coffee Break</i>	
11:30-12:15	Non-coding Variants in Rare Diseases	Nicola Whiffin Big Data Institute, Wellcome Centre for Human Genetics, Oxford, United Kingdom
12:15-13:00	snRNA Genes in Neurodevelopmental Disorders	Christel Depienne Institute for Human Genetics, University Hospital Essen, Germany

**06 May 2026**      **External Quality Assessment Scheme, Recommendations and Cases**  
 Chair                tbc

TIME (CET)	TOPIC	SPEAKER
9:00-9:45	Quality Insurance	Katja Lohmann Institute of Neurogenetics, University of Lübeck, Germany Ales Maver Centre for Mendelian Genomics, University Medical Centre Ljubljana, Slovenia
9:45-10:30	External Quality Assessment Scheme	Katja Lohmann Ales Maver
10:30-11:00	Recommendations	Katja Lohmann Ales Maver
11:00-11:15	<i>Coffee break</i>	
11:15-13:00	Case presentations	Panel

## 07 May 2025 Application of NGS to a Specific Disease Group

Chair Elisabetta Indelicato (Innsbruck, Austria)

TIME (CET)	TOPIC	SPEAKER
9:00-9:45	NGS for <b>Ataxia and HSP</b>	Erik-Jan Kamsteeg Radboudumc, Nijmegen, The Netherlands
9:45-10:30	NGS for <b>Atypical Parkinsonian Syndromes</b>	Giorgos Koutsis 1st Department of Neurology, Eginition Hospital, National and Kapodistrian University of Athens, Greece
10:30-11:15	NGS for <b>Frontotemporal Dementia</b>	Laura Donker Kaat Erasmus Medical Center Rotterdam, The Netherlands
11:15-11:30	<i>Coffee Break</i>	
11:30-12:15	NGS for <b>HD and Chorea</b>	Larissa Arning-Bünder Ruhr University Bochum, Germany
12:15-13:00	NGS for <b>Dystonia, Paroxysmal Disorders and NBIA</b>	Michael Zech Institute of Human Genetics, University Hospital Rechts der Isar, Technical University of Munich, Germany
13:00-13:45	NGS for <b>Leukoencephalopathies</b>	Fanny Mochel Paris Brain Institute, Sorbonne Université, AP-HP, Paris, France
	Closing Remarks	

For further information please contact Sophie Ripp ([Sophie.ripp@med.uni-tuebingen.de](mailto:Sophie.ripp@med.uni-tuebingen.de))

### PROGRAMME COMMITTEE

- **Elisabetta Indelicato**, (Medical University of Innsbruck, Austria)
- **Hoa Huu Phuc Nguyen**, (Ruhr University Bochum, Germany)
- **Alexander Hoischen**, (Radboudumc Nijmegen, The Netherlands)
- **Holm Graessner**, Tübingen (ERN-RND, Tübingen, Germany)