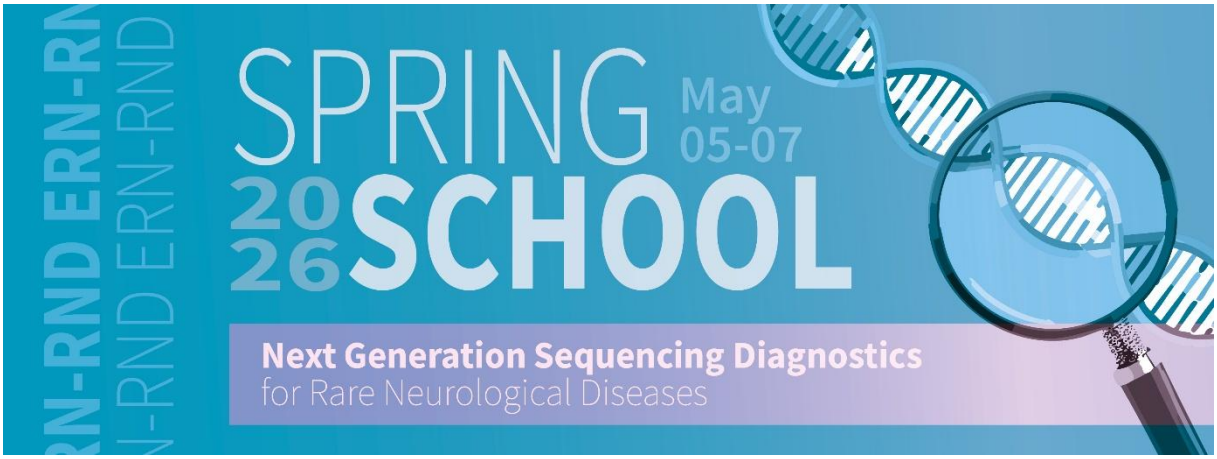

PROGRAMME - To **register** click here.

05 May 2026 NGS Technologies

TIME (CET)	**TOPIC**	**SPEAKER**
9:00-9:45	**Bioinformatic Tools** for Diagnosing Rare Diseases	Christian Gilissen Genome bioinformatics, Radboud University Medical Center, Nijmegen, The Netherlands
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	**RNAseq in RD**	Diana Baralle Human Development and Health, Faculty of Medicine, University of Southampton, United Kingdom
11:15-11:30	*Coffee Break*	
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

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	*Coffee break*	
11:15-13:00	**Case presentations**	Panel

07 May 2025 Application of NGS to a Specific Disease Group

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

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