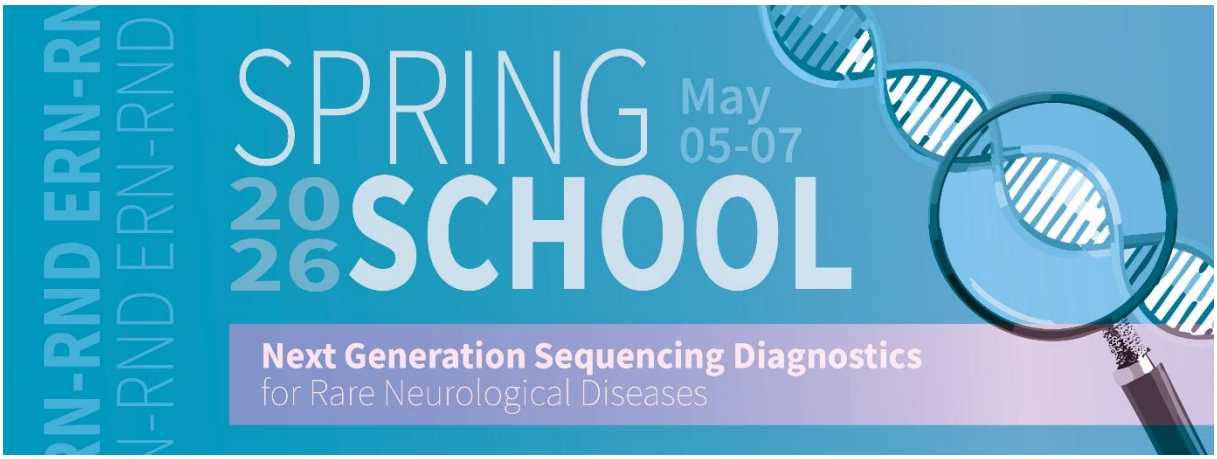
European Reference Network
for rare or low prevalence complex diseases
Network Neurological Diseases (ERN-RND)




Our 7th Winter School will focus on **"Next Generation Sequencing Diagnostics for Rare Neurological Diseases"** and is taking place on May 5–7 (online, free of charge)!

During this Spring School, participants will gain insights into:

- **NGS Technologies**
- **Quality Assurance, Recommendations, and Case Presentations**
- **Clinical Applications of NGS**

To **register** click here.

# PROGRAMME

## 05 May 2026 NGS Technologies

**CHAIR** --

| **TIME** (CET) | **TOPIC** | **SPEAKER** |
|---|---|---|
| **9:00-9:45** | **Bioinformatic Tools** for Diagnosing Rare Diseases | Christian Gilissen<br>Genome bioinformatics, Radboud University Medical Center, Nijmegen, The Netherlands |
| **9:45-10:30** | **Episignatures** in Diagnostics of Rare Diseases | Camille Charbonnier<br>Cancer and Brain Genomics, Inserm UMR1245, Université de Rouen Normandie, France |
| **10:30-11:15** | **RNAseq in RD** | Diana Baralle<br>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 | tbc |
| **12:15-13:00** | **snRNA Genes** in Neurodevelopmental Disorders | Christel Depienne<br>Institute for Human Genetics, University Hospital Essen, Germany |

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## 06 May 2026 External Quality Assessment Scheme, Recommendations and Cases

**CHAIR** --

| TIME (CET) | TOPIC | SPEAKER |
|---|---|---|
| **9:00-9:45** | **Quality Insurance** | Katja Lohmann<br>Institute of Neurogenetics, University of Lübeck, Germany<br>Ales Maver<br>Centre for Mendelian Genomics, University Medical Centre Ljubljana, Slovenia |
| **9:45-10:30** | **External** Quality Assessment Scheme | Katja Lohmann<br>Ales Maver |
| **10:30-11:00** | **Recommandations** | Katja Lohmann<br>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

**CHAIR** --

| TIME (CET) | TOPIC | SPEAKER |
|---|---|---|
| **9:00-9:45** | NGS for **Ataxia and HSP** | tbc |
| **9:45-10:30** | NGS for **Atypical Parkinsonian Syndromes** | tbc |
| **10:30-11:15** | NGS for **Frontotemporal Dementia** | Laura Donker Kaat<br>Erasmus Medical Center Rotterdam, The Netherlands |
| **11:15-11:30** | *Coffee Break* | |
| **11:30-12:15** | NGS for **HD and Chorea** | tbc |
| **12:15-13:00** | NGS for **Dystonia, Paroxysmal Disorders and NBIA** | Michael Zech<br>Institute of Human Genetics, University Hospital Rechts der Isar, Technical University of Munich, Germany |
| **13:00-13:45** | NGS for **Leukoencephalopathies** | Fanny Mochel<br>Paris Brain Institute, Sorbonne Université, AP-HP, Paris, France |
| | Closing Remarks | |

For further information please contact Sophie Ripp (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)