



EuroNDD 2026

Third European Workshop for a Multidisciplinary view on
Rare Genetic Neurodevelopmental Disorders

9-10 April 2026 | Warsaw, Poland



Full programme



Let's create learning waves together

1. Understanding Neurobiology

Understanding genes, disease, mechanisms, model systems, and genotype-phenotype correlations

2. From Molecular Diagnosis to Intervention

From molecular diagnostics to precision therapies

3. Holistic Care

Multidisciplinary diagnostics and interventions translated into a holistic (health)care network

4. Health Information System & Data

Data availability and the ethical, legal and biopsychosocial aspects of reuse

Thursday 9 April (AM)

8:00 Registration

8:30 Welcome - **Marek Migdal**, Director of Children's Memorial Health Institute

Plenary 1 - Chairs: Christiane Zweier, Tjitske Kleefstra

From understanding the neurobiology towards personalized interventions

8:45 The Undiagnosed Hackathon Model - **Helene Cederroth**

9:15 Touch and Tuberous Sclerosis Complex: what human and animal studies are teaching us - **Ewelina Knapska**

9:45 Precision medicine for monogenic epilepsies - **Gaëtan Lesca**

10:15 "The road from diagnosis to intervention: 15 years of ENCORE Expertise Care for Angelman Syndrome" - **Karen Bindels de Heus**

10:45 **Coffee break**

Parallel session 1 –

Understanding the neurobiology

Chairs: Zeynep Tümer, Agnieszka Madej Pilarzyk

11:15 NARS1 variants in neurodevelopmental disorders using, *Xenopus tropicalis*, *Drosophila* and molecular dynamics simulations
Sehime Temel

11:30 De novo variants in LDB1 are linked to distinct neurodevelopmental disorders depending on variant location and consequences
Anne Gregor

11:45 Biological parameters in 100 girls with Rett syndrome: toward a better pathophysiological understanding and new therapeutic perspectives
Béatrice Desnous

12:00 Expanding knowledge of ultra-rare CACNA2D2-related developmental and epilepticencephalopathy: insights from 33 individuals
Myriam Essid

12:15 Biallelic rare variants in fat mass and obesity-associated (FTO) cause a variable developmental phenotype: A genotype-phenotype correlation
Radhakrishnan Periyasamy

Parallel session 2 –

From molecular diagnostics to interventions

Chairs: Gaëtan Lesca, Marco Tartaglia

B2B-RARE: Bench to bedside in rare diseases

Andreas Roos

Modelling Tubulinopathies using human iPSCs-derived motor neurons
Ilaria Svezia

PURA syndrome as congenital myasthenic disorder. Evidence from muscle morphology, proteomics and blood biomarkers

Magdalena Mroczek

Dissecting the epigenetic basis of USP7-related neurodevelopmental disorders: From DNA methylation signatures to chromatin dysregulation

Manasa Kalya Purushothama

A study on caregiver burden and quality of life of informal caregivers of children with metachromatic leukodystrophy in Poland -

Karolina Śledzińska

Thursday 9 April (PM)

12:30 Lunch

13:30 Guided poster tour

14:15 Proceed to break-out sessions

Parallel break-out sessions

14:30 First round of parallel break-out sessions

1. Solving the undiagnosed: How recent insights translate into optimized workflows - **Katrin Ounap, Helene Cederroth, Zeynep Tümer, Tjitske Kleefstra**
 2. Pain in people with intellectual disabilities: Methodical and interdisciplinary collaboration (an educational session) - **Leendert Sneep, Nanda de Knegt**
 3. From Guideline to Practice - Implementing ERN ITHACA Guidelines (an educational session) - **Mirthe Klein Haneveld, Agnies van Eeghen, co-workers across Europe**
 4. From Principles to Practice: Interprofessional Collaboration in Rare Genetic Intellectual Disability Syndromes – A European Round Table - **Mana Nasori, Kim Oostrom, Lotte Haverman, Sylvia Huisman**
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15:45 Exchange break with drinks & snacks

16:15 Second round of parallel break-out sessions

1. Towards Precision Medicine for NDD - **Geatan Lesca, Beatrice Desnous, Adreas Roos, Kasia Kotulska**
2. From training skills to enabling interactions: A new paradigm in AAC for individuals with neurodevelopmental disorders and complex communication needs (an educational session) - **Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek**
3. Building Patient Registries under the GDPR – The Good, the Bad and the Ugly (an educational session & roundtable) - **Christian Gebhard, David Townend**
4. Neuropsychological insights: Cognition as a bridge between brain and behaviour in rare genetic syndromes? - **Anja Bos-Roubos, Jennifer Kramer, Carmen Oldenboom, Ellen Wingbermhühle, Jos Egger**
5. From Networks to Norms: Developing First European Recommendations and Care Principles for PIMD / Polyhandicap - **Sylvia Huisman, Annette van der Putten, Marie-Christine Rousseau, Ilse H. Zaal-Schuller**

17:30 Optional time to discuss other matters in the available rooms

20:00 Dinner - *by registration only*

Café Mozaika

Puławska 53, 02-508 Warszawa

Friday 10 April

8:30 Welcome & reflection on day 1

Plenary 2 - Chairs: Christiane Zweier & Tjitske Kleefstra

From holistic (health)care networks towards systematic data capture

8:45 Early interventions in intellectual disabilities associated with epilepsy: lessons from TSC - **Katarzyna Kotulska**

9:15 The transition process among (para)medics involved in the care network surrounding children with rare genetic neurodevelopmental disorders: the Single Bulgarian Expert centre expertise - **Nikolinka Yordanova**

9:45 Rare disease discovery: From molecular diagnosis to clinical intervention and care. Experience in Estonia - **Katrin Ounap**

10:15 **Coffee break**

Plenary 3 - Chairs: Laura de Graaff, Dorica Dan

Multidisciplinary diagnostics & interventions translated into a holistic (health) care network

10:45 Psychometric properties of a patient reported outcome measure (PROM) set for genetic intellectual disability - **Alannah Hijlkema**

11:00 Pain Assessment in Individuals with Profound Intellectual and Multiple Disabilities (PIMD/Polyhandicap): Evidence, Expert Opinion, and Caregiver Perspectives - **Ilse Zaal-Schuller**

11:15 The use of AAC in genetic syndromes; lessons learned during 10 years clinical practice - **Maartje Radstaake**

11:30 People with Profound Intellectual and Multiple Disabilities/Polyhandicap, Families, and Institutional Caregivers: The French EVAL-PLH Cohort. Results and Perspectives - **Marie Christine Rousseau**

11:45 Panel discussion

12:00 **Lunch**

13:00 **Guided poster tour**

14:00 **Proceed to break-out sessions**

Parallel break-out sessions

14:15 **Third round of parallel break-out sessions**

1. Measuring what matters in genetic neurodevelopmental disorders: trials and care - **Agnies van Eeghen & Ellen Elsman**
2. Transition of care for adults with intellectual disabilities in genetic syndromes - **Nikolinka Yordanova, Laura de Graaff, Kasia Swieczkowska**
3. Communication support for individuals with neurodevelopmental disorders and complex communication needs: sharing best practices (a round table discussion) - **Gillian Townend, Maartje ten Hooven-Radstaake, Cindy Navis, Paulina Rutka, Elżbieta Dawidek**

15:45 **Closing - Tjitske Kleefstra, Christiane Zweier, Alain Verloes**