



1. Understanding Neurobiology

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

2. From Molecular Diagnostics 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 Systems & Data

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



Stay tuned on our website

<https://ern-ithaca.eu/events-news/eurondd/>



Program on Thursday April 9

8:00 – 8:30 **Registration**

8:30 – 8:45 **Welcome & technical information**

8:45 – 10:45 **Plenary session 1:**

From understanding the neurobiology towards personalized interventions

Chairs: Christiane Zweier (CH) & Tjitske Kleefstra (NL)

Invited speakers

8:45 – 9:15 **The Undiagnosed Hackathon Model**

Helene Cederoth (SE)

9:15 – 9:45 **Touch and Tuberous Sclerosis Complex: what human and animal studies are teaching us**

Ewelina Knapska (PL)

9:45 – 10:15 **Precision medicine for monogenic epilepsies**

Gaetan Lesca (FR)

10:15 – 10:45 **"The road from diagnosis to intervention:**

15 years of ENCORE Expertise Care for Angelman Syndrome"

Karen Bindels de Heus (NL)

10:45 – 11:15 ***Coffee break***

11:15 – 12:30 **Parallel session 1 – Understanding the neurobiology**

Chairs: Zeynep Tümer (DK) & Agnieszka Madej Pilarzyk (PL)

11:15 – 11:30 NARS1 Variants in Neurodevelopmental Disorders Using, *Xenopus tropicalis*, *Drosophila* and Molecular Dynamics Simulations

Sehime Temel (TR)

11:30 – 11:45 De novo variants in LDB1 are linked to distinct neurodevelopmental disorders depending on variant location and consequences

Anne Gregor (CH)

11:45 – 12:00 Biological parameters in 100 girls with Rett syndrome: toward a better pathophysiological understanding and new therapeutic perspectives

Béatrice Desnous (FR)

12:00 – 12:15 Expanding knowledge of ultra-rare CACNA2D2-related developmental and epileptic encephalopathy: insights from 33 individuals

Myriam Essid (FR)

12:15 – 12:30 Biallelic rare variants in fat mass and obesity-associated (FTO) cause a variable developmental phenotype: A genotype-phenotype correlation

Radhakrishnan Periyasamy (CH)



EuroNDD Program overview

11:15 – 12:15 Parallel session 2 – From molecular diagnostics to interventions

Chairs: Gaetan Lesca (FR) & Marco Tartiglia

11:15 – 11:30 B2B-RARE: Bench to bedside in rare diseases

Andreas Roos (GER)

11:30 – 11:45 Modelling Tubulinopathies using human iPSCs-derived motor neurons

Ilaria Svezia (IT)

11:45 – 12:00 PURA Syndrome as Congenital Myasthenic Disorder. Evidence from Muscle Morphology, Proteomics and Blood Biomarkers

Magdalena Mroczek (PL)

12:00 – 12:15 Dissecting the Epigenetic Basis of USP7-Related Neurodevelopmental Disorders: From DNA Methylation Signatures to Chromatin Dysregulation

Manasa Kalya Purushothama (NL)

12:30 – 13:30 **Lunch**

13:30 – 14:15 **A guided poster tour**

A bootcamp format: starting at multiple spots organized around clustered topics per theme & move clock wise to the next presenter. In-depth discussions are facilitated during break-out sessions

14:15 – 14:30 **Transition to the break-out sessions**

14:30 – 17:30 **Parallel break-out sessions**

Supervised by event coordination team

14:30 – 15:45 **Topics of the first round**

1. Solving the undiagnosed: How recent insights translate into optimized workflows.
Katrin Ounap (EE), Helene Cederroth (SE), Zeynep Tümer (Dk), Tjitske Kleefstra (NL)
2. Pain in people with intellectual disabilities: methodical and interdisciplinary collaboration (an educational session). **Leendert Sneep & Nanda de Knegt (NL)**
3. From Guideline to Practice - Implementing ERN ITHACA Guidelines (an educational session) **Mirthe Klein Haneveld (NL), Agnies van Eeghen (NL) & co-workers cross 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 (NL)**



15:45 – 16:15 ***Exchange break with drinks & snacks***

16:15 – 17:30 **Second round topics**

1. Towards Precision Medicine for NDD. **Geatan Lesca (FR), Beatrice Desnous (FR), Adreas Roos (GER), Kasia Kotulska (PL)**
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 (UK), Maartje ten Hooven-Radstaake (NL), Cindy Navis (NL), Paulina Rutka (PL), Elżbieta Dawidek (PL)**
3. Building Patient Registries under the GDPR – The Good, the Bad and the Ugly (an educational session followed by a roundtable discussion). **Christian Gebhard (GER), David Townend (UK)**
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 (NL)**
5. From Networks to Norms: Developing First European Recommendations and Care Principles for PIMD / Polyhandicap. **Sylvia Huisman (NL), Annette van der Putten (NL), Marie-Christine Rousseau (FR), Ilse H. Zaal-Schuller (NL)**

17:30 – 18:30 ***Optional time to discuss other matters in the available rooms.***

If you would like to do so, please let us know via info@in-act.nl

20:00 – 22:00 **Dinner. On registration only!**





Draft Program on Friday April 10

8:30 – 8:45 **Welcome & reflection on day 1**

8:45 – 10:15 **Plenary session 2:**

From holistic (health)care networks towards systematic data capture

Chairs: Christiane Zweier (CH) & Tjitske Kleefstra (NL)

Invited speakers

8:45 – 9:15 **Early interventions in intellectual disabilities associated with epilepsy: lessons from TSC**
Katarzyna Kotulska (PL)

9:15 – 9:45 **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 (BG)

9:45 – 10:15 **Rare disease discovery: from molecular diagnosis to clinical intervention and care. Experience in Estonia**

Katrin Ounap (EE)

10:15 – 10:45 ***Coffee break***

10:45 – 12:00 **Plenary session 3:**

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

Chairs: Laura de Graaff (NL) & Dorica Dan (RO)

10:45 – 11:00 Psychometric properties of a patient reported outcome measure (PROM) set for genetic intellectual disability

Alannah Hijlkema (NL)

11:00 - 11:15 Pain Assessment in Individuals with Profound Intellectual and Multiple Disabilities (PIMD/Polyhandicap): Evidence, Expert Opinion, and Caregiver Perspectives

Ilse Zaal-Schuller (NL)

11:15 – 11:30 A study on caregiver burden and quality of life of informal caregivers of children with metachromatic leukodystrophy in Poland

Karolina Śledzińska (PL)

11:30 - 11:45 The use of AAC in genetic syndromes; lessons learned during 10 years clinical practice

Maartje Radstaake (NL)

11:45 - 12:00 People with Profound Intellectual and Multiple Disabilities/Polyhandicap, Families, and Institutional Caregivers: The French EVAL-PLH Cohort. Results and Perspectives

Marie Christine Rousseau (FR)





12:00 – 13:00 **Lunch**

13:00 – 14:00 **A guided poster tour**

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14:00 – 14:15 **Transition to the break-out sessions**

14:15 – 15:30 **Parallel break-out sessions**

Supervised by event coordination team

14:15 – 15:30 **Third round topics**

1. Measuring what matters in genetic neurodevelopmental disorders: trials and care.
Agnies van Eeghen & Ellen Elsman (NL)
2. Transition of Care for Adults with Intellectual Disabilities in Genetic Syndromes.
Nikolinka Yordanova(BU), Laura de Graaff (NL), Kasia Swieczkowska (PL)
3. Communication support for individuals with neurodevelopmental disorders and complex communication needs: sharing best practices (a round table discussion). **Gillian Townend (UK), Maartje ten Hooven-Radstaake (NL), Cindy Navis (NL), Paulina Rutka (PL), Elżbieta Dawidek (PL)**
4. Pain in people with intellectual disabilities: implementation and network formation (a round table discussion). **Leendert Sneep & Nanda de Knecht (NL)**

15:30 – 15:45 **Final words**

Prof. Tjitske Kleefstra (NL), Prof. Christiane Zweier (CH), Prof. Alain Verloes (FR)

