



### 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/>

Registration runs till January 31th!

## Draft 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 **Touch and Tuberous Sclerosis Complex: what human and animal studies are teaching us**  
Ewelina Knapska (PL)

9:15 – 9:45 **Brain malformations phenotypes as determined by neuroimaging and neuropathology.**  
Renzo Guerrini (IT)

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 (PO)

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)



**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** (PO)

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***

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

14:30 – 17:30 ***Break-out sessions***

- Educational sessions and roundtable topics are currently in construction
- Begin February participants will be invited to express their preference for educational session and/or roundtable table discussion.

14:30 – 17:30 **Roundtable discussions & Educational sessions**

Supervised by event coordination team

14:30 – 15:45 First round

Educational sessions (max 3 in a row of max 50 persons)

Table topics .... (max 5 in a row of max 25 persons)

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

16:15 – 17:30 Second round

Educational sessions (max 3 in a row of max 50 persons)

Table topics .... (max 5 in a row of max 25 persons)

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 [coordination@ern-ithaca.eu](mailto:coordination@ern-ithaca.eu).

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 – 10:45 **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.**

Niklinka 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 **The Undiagnosed Hackathon Model**

Helene Cedderoth (SE)

10:45 – 11:15 ***Coffee break***

11:15 – 12:30 **Plenary session 3:**

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

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

11:15 – 11:30 Psychometric properties of a patient reported outcome measure (PROM) set for genetic intellectual disability

**Alannah Hijlkema** (NL)

11:30 - 11:45 Pain Assessment in Individuals with Profound Intellectual and Multiple Disabilities (PIMD/Polyhandicap): Evidence, Expert Opinion, and Caregiver Perspectives

**Ilse Zaal-Schuller** (NL)

11:45 - 12:00 A study on caregiver burden and quality of life of informal caregivers of children with metachromatic leukodystrophy in Poland

**Karolina Śledzińska** (PO)

12:00 - 12:15 The use of AAC in genetic syndromes; lessons learned during 10 years clinical practice

**Maartje Radstaake** (NL)

12:15 - 12:30 People with Profound Intellectual and Multiple Disabilities/Polyhandicap, Families, and Institutional Caregivers: The French EVAL-PLH Cohort. Results and Perspectives

**Marie Christine Rousseau** (FR)





12:30 – 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 – 15:45 **Break-out sessions**

- Educational sessions and roundtable are currently in construction.
- Begin February participants will be invited to express their preference for educational session and/or roundtable table discussion.

14:30 – 15:45 **Roundtable discussions & Educational sessions**

Supervised by event coordination team

Educational sessions (max 3 in a row of max 50 persons)

Table topics .... (max 5 in a row of max 25 persons)

15:45 – 16:00 **Final words**

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