



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.

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
Katrín Ounap (EE)

10:15 – 10:45 **The Undiagnosed Hackathon Model**
Helene Cederroth (SE)

10:45 – 11:15 **Coffee break**

11:15 – 12:30 **Plenary session 3:**

Multidisciplinary diagnostics & interventions translated into a holistic (heath)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 Hijkema (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)