# Program on Thursday April 4

8:00 – 8:30 **Registration**

**Grande Auditorio**

8:30 – 8:45 **Welcome on behalf of the Organising Committee & technical information**
Tjitske Kleefstra (NL) and Christiane Zweier (CH)

8:45 – 10:15 **Plenary session 1**
Chairs: Christiane Zweier (CH) and Tjitske Kleefstra (NL)

**Invited speakers**

8:45 – 9:15 **Unravelling autism and neurodevelopmental disorders: a comprehensive and ecological approach**
Susana Mouga (PO)

9:15 – 9:45 **Psychiatric Genetics and the Neurodevelopmental Continuum**
Michael Owen (UK)

9:45 – 10:15 **Emergence of neocortical function in heterotopic neurons**
Denis Jabaudon (CH)

10:15 – 10:45 ***Exchange break with drinks and small snack***

**Grande Auditorio**

10:45 – 12:30 **Parallel session 1 – Genes and pathways**
Chairs: Zeynep Tümer (DK) and Gaëtan Lesca (FR)

**Invited speaker**

10:45 – 11:15 **Identification of new genes**
Hülya Kayserili (TR)

**Selected abstracts**

11:15 – 11:30 **ANO4 is a novel gene causing developmental and epileptic encephalopathy or inherited epilepsies**
Anais Begemann (CH)

11:30 – 11:40 **Towards Personalized Treatments for Synapthopathies using CAMK2 disorders as proof of concept**
Danielle Veenma (NL)

11:40 – 11:50 **Genetic heterogeneity of anomalies of the corpus callosum: lessons from a cohort of 350 individuals**
Solveig Heide (FR)

11:50 – 12:00 **Kleefstra syndrome: Beyond EHMT1 haploinsufficiency, beyond typical phenotype**
Dmitrijs Rots (NL)

12:00 – 12:30 Panel discussion

**Auditorio B2-03**

10:45 – 12:30 **Parallel session 2 – Ethical, legal and Psycho-social aspects**
Chairs: Katarzyna Swieczkowska (PL) and Susana Mouga (PO)

**Invited speaker**

10:45 – 11:15 **Multiple exemplar learning from rare neurodevelopment disease in the postmaster curricula for psychological and medical specialists**
Jos Egger (NL)

**Selected abstracts**

11:15 – 11:30 **'Mind the gap' – An ERN-ITHACA survey on the transition to adult healthcare**
Mirthe Klein Haneveld (NL)

11:30 – 11:40 **A longitudinal study of challenging behaviour and autistic symptoms Smith-Magenis'syndrome**
Monica Stolen Dønnum (NO)

11:40 – 11:50 **Navigating neurodevelopmental disorders: insights from a Romanian cohort and empowering patients through education**
Alexandra-Aurora Dumitra (RO)

11:50 – 12:00 **Integrated care for patients with NDD and rare diseases in Romania**
Dorica Dan (RO)

12:00 – 12:30 Panel discussion

12:30 – 14:00 ***Lunch & poster tour***
For detailed poster tour information see separate flyer.

**Auditorio B2-03**

14:00 – 15:40 **Parallel session 3 – Applied & Emerging Therapies**
Chairs: Alain Verloes (FR) and Hülya Kayserili (TR)

**Invited speakers**

14:00 – 14:20 **New horizons: Gonadotropin-Releasing Hormone, Trisomy 21 and Cognition**
Vincent Prevot (FR)

14:20 - 14:40 **Understanding Down syndrome as an interferonopathy: mechanisms and therapeutic opportunities**
Joaquin Espinosa (USA)

**Selected abstracts**

14:40 – 14:55 **AMFR dysfunction causes spastic paraplegia amenable to statin treatment in a preclinical model**
Stefan Barakat (NL)

14:55 – 15:05 **Eating behaviour and issues in rare genetic disorders**
Heidi Elisabeth Nag (NO)

15:05 – 15:15 **Individualized antisense oligonucleotide therapies for patients with rare neurological disorders**
Marlen Lauffer (NL)

15:15 – 15:25 **Patient-named elesclomol-copper therapy in a child with menkes disease**
Francesc Palau (ES)

15:25 – 15:40 Panel discussion

**Grande Auditorio**

14:00 – 15:40 **Parallel session 4 – Diagnostics**
Chairs: Stephanie Miot (FR) and Claudine Laurent-Levinson (FR)

**Invited speakers**

14:00 – 14:40 **What is aging – and why is it different in patients with rare genetic syndromes?**
Francesco Mattace Raso (NL) and Laura de Graaff (NL)

**Selected abstracts**

14:40 – 14:55 **Cognitive trajectories and dementia biomarkers in adults with ID and epilepsy**
Hadassa Kwetsie (NL)

14:55 – 15:05 **Genome sequencing supports the role of scn1a and pcdh19 in patients with undiagnosed dravet syndrome and related disorders**
Miriam Essid (FR)

15:05 – 15:15 **The natural history of adults with KBG syndrome: a physician's reported experience**
Allan Bayat (DK)

15:15 – 15:25 **Mapping the trajectory of syt1-associated neurodevelopmental disorder (baker-gordon syndrome)**
Sam Norwitz (UK)

15:25 – 15:40 Panel discussion

15:40 – 16:30 ***Exchange break with drinks and small snack***

16:30 – 18:45 **Roundtable discussions**
For detailed information see separate flyer.

16:30 – 17:30 First round

17:45 – 18:45 Second round

18:45 – 19:00 ***Wrap up - Reflection on the round table discussions by the chairs***

20:00 – 22:30 **Dinner at the Restaurant of the Casa Do Alentejo, a historical venue located in a 17th Century palace.**
***On registration only !***

## Program on Friday April 5

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

**Grande Auditorio**

8:45 – 10:15 **Plenary session 2**
Chairs: Christiane Zweier (CH) and Tjitske Kleefstra (NL)

**Invited speakers**

8:45 – 9:15 **Nutritional management for individuals with rare genetic neurodevelopmental disorders**
Marianne Nordstrøm (NO)

9:15 – 9:45 **Enhancing care for Children and Adolescents with profound intellectual and multiple disabilities (PIMD): A Holistic Approach through the French National Network.**
Béatrice Desnous (FR)

9:45 – 10:15 **Insights on the potential of NLRP3 Inflammasome inhibition in Epilepsy – an ex vivo approach**
Claudia Valente (PO)

10:15 – 10:45 ***Exchange break with drinks and small snack***

**Grande Auditorio**

10:45 – 12: 00 **Parallel session 5 - Mechanisms of diseases, model systems & translational pre-clinical work**
Chairs: Marco Tartaglia (IT) and Gaëtan Lesca (FR)

**Invited speaker**

10:45 – 11:15 **Functional testing in potassium channelopathies related to epilepsy & NDD and perspectives for targeted therapies**
Maurizio Taglialatela (IT)

**Selected abstracts**

11:15 – 11:30 **Advanced brain organoid modeling and transcriptomic investigation of Rett syndrome**
Pelin Saglam Metiner (TR)

11:30 – 11:40 **Neuronal phenotypes associated with FBXO11-deficiency can be alleviated with chemical activation of the proteasome**
Anne Gregor (CH)

11:40 – 11:50 **Deleterious ZNRF3 germline variants as a novel cause of neurodevelopmental disorders with mirror brain phenotypes due to distinct domain-specific effects on Wnt/β-catenin signaling**
Paranchai Boonsawat (CH)

11:50 – 12:00 **In Vivo Xenotransplantation of Patient-Derived Neurons in MECP2 Neurodevelopmental Disorders: Exploring the Cellular and Molecular Landscape**
Nona Merckx (BE)

12:00 – 12:15 Panel discussion

**Auditorio B2-03**

10:45 – 12: 00 **Parallel session 6 – Polyhandicap**

Chairs: Marie Christine Rousseau (FR) and Sylvia Huisman (NL)

**Invited speaker**

10:45 – 11:15 **Let's get together; supporting persons with profound intellectual and multiple disabilities from an interdisciplinary perspective.**

Annette van der Putten (NL)

**Selected abstracts**

11:15 – 11:30 **Burnout among institutional healthcare workers caring for patients with polyhandicap**

Houria El Ouazzani (FR)

11:30 – 11:40 **Development and initial validation of the polyhandicap severity scale**

Marie Christine Rousseau (FR)

11:40 – 11:50 **How can we improve outpatient care for children with PIMD? Insight into experiences and preferences of parents and healthcare professionals**

Catelijne Coppens (NL)

11:50 – 12:00 **Gaps in transitional care for adolescents with profound intellectual and multiple disabilities in the Netherlands: experiences of health care professionals and parents.**

Ilse Ooms (NL)

12:00 – 12:15 Panel discussion

**Grande Auditorio**

12:15 – 12:30 ***Final words***

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