ERN ITHACA is pleased to announce that the First European Workshop **for a multidisciplinary view on 'rare genetic neurodevelopmental disorders (NDD)'** will be organized in Amsterdam, April 20 – 21, 2023.

Thursday April 20, 2023

12:30 – 13:00 **Registration**

13:00 – 13:30 **Welcome on behalf of the Organising Committee**
Prof. Tjitske Kleefstra (NL) and Prof. Christiane Zweier (CH)

13:30 – 15:00 **Plenary session 1**
Chair: Prof. Tjitske Kleefstra (NL) & Prof. Christiane Zweier (CH)
Invited speakers
13:30 – 14:00 **What can ERNs do for patients with NDD**
Prof. Alain Verloes (ITHACA Coordinator, FR)
14:00 – 14:30 **Navigating the uncertainties of next-generation sequencing in the genetics clinic**
Prof. Hilde Van Esch (BE)
14:30 – 15:00 **Precision diagnostics in NDD - next to next-generation-sequencing**
Prof. Zeynep Tümer (DK)

15:00 – 15:45 ***Networking break / poster viewing***

15:45 – 17:15 **Parallel session 1 - Applied & Emerging Therapies**
Chairs: Prof. Marco Tartaglia (IT) & Dr Agnies van Eeghen (NL)
Invited speaker
15:45 – 16:15 **Personal healthcare through braincells on a chip**
Prof. Nael Nadif Kasri (NL)

Selected abstracts
16:15 – 16:30 **Making sense of junk: functional enhancers with medical relevance identified by computational analysis and CHIP-STARR-SEQ in neural cell models enable prioritizing non-coding variants from patient whole genome sequencing studies**
Dr. Stefan Barakat (NL)
16:30 – 16:45 **Antisense oligonucleotide therapies for PLP1-associated hypomyelination of early myelinating structures**
Dr. Bianca Zardetto (NL)
16:45 – 17:00 **Efficacy and tolerance of cannabidiol in the epilepsy treatment in patients with RETT Syndrome: experience in a single center cohort**
Dr. Béatrice Desnous (FR)
17:00 – 17:15 **Glutamate / GABA modulation as a novel therapeutic target for psychotic and cognitive symptoms in 22Q11 deletion Syndrome**
Prof. Therese van Amelsvoort (NL)

15:45 – 17:30 **Parallel session 2 - Data collection, database, registries, use of AI**
Chair: Dr. Franziska Degenhardt (DE)

Invited speakers

15:45 – 16:15 **Phenotypic effects of genetic variants associated with autism beyond diagnosis**
Prof. Thomas Bourgeron (FR)

16:15 – 16:45 **SysNDD and overview on the landscape of NDD**
Dr. Bernt Popp (DE)

Selected abstracts

16:45 – 17:00 **Development of a guideline on the Kleefstra Syndrome within the framework of the European Reference Network ITHACA**
Arianne Bouman, MD (NL)

17:00 – 17:15 **Real time analysis of patient META_COHORT for neurodevelopmental disorders: an innovative platform proposal**
Prof. Natália Oliva-Teles (PT) & Adrian Harwood (UK)

17:15 – 17:30 **The genetic landscape of neurodevelopmental disorders in a large cohort of multiplex consanguineous families from Turkey**
Prof. André Reis (DE)

17:30 – 18:15 ***Networking break / poster viewing***

18:15 – 19:15 **Keynote lecture followed by a panel discussion**
Chair: Dr. Sofia Douzgou (NO)

18:15 – 18:45 **Why we have the diseases we have**
Prof. Han Brunner (NL)

18:45 – 19:15 **Panel discussion**

19:30 – 21:30 ***Dinner Cocktail (on registration only!)***

Friday April 21, 2023

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

8:45 – 10:15 **Plenary session 2**
Chair: Prof. Christiane Zweier (CH) & Prof. Tjitske Kleefstra (NL)
Invited speakers
8:45 – 9:15 **Big Data & Artificial Intelligence**
Prof. Wiro Niessen (NL)
9:15 – 9:45 **Transition and adult care**
Dr. Laura De Graaff (NL)
9:45 – 10:15 **GestaltMatcher, a deep convolutional neural network for the analysis of medical imaging data**
Prof. Peter Krawitz (DE)

10:15 – 10:45 ***Coffee break***

10:45 – 12: 00 **Parallel session 3 – Profound and multiple learning disability**
Chair: Dr. Marie Christine Rousseau (FR) and Dr. Sylvia Huisman (NL)
Invited speakers
10:45 – 11:15 **Communication in Neurodevelopmental Diseases: the importance of developing guidelines**
Dr. Gill Townend (UK)
11:15 – 11:45 **Polyhandicap and Neurodevelopmental Diseases**
Prof. Thierry Billette de Villemeur (FR)

Selected abstract
11:45 – 12:00 **Genetic (re-)evaluation to optimize the care of adults with intellectual disability**
Dr. Cordula Knopp (DE)

10:45 – 12:00 **Parallel session 4 - Mechanisms of diseases, model systems & translational pre-clinical work**
Chair: Prof. Alain Verloes (FR) and Dr. Sofia Douzgou Houge (NO)
Invited speakers
10:45 – 11:15 **PRISM screen: a tool to screen for functional importance of new genetic variants**
Dr. Geeske van Woerden (NL)
11:15 – 11:45 **Personalized gene therapy using CRISPR/Cas9 technology**
Prof. Alessandra Renieri (IT)

Selected abstract
11:45 – 12:00 **DNA Methylation episignature of valproate embryopathy**
Prof. Patrick Edery (FR)

12:00 -12:15 ***Break***

12:15 – 13:30 **Paralel session 5 - Ethical, legal and Psycho-social aspects**
Chair: Dr. Claudine Laurent (FR) and Dr. Laura de Graaff (NL)
Invited speakers
12:15 – 12:45 **Case management for rare diseases and rare disabilities in Europe**
Ms Dorica Dan (RO)
12:45 -13:15 **NDD adults management: Overview in Europe and new challenges**
Dr. Stephanie Miot (FR)

Selected abstract
13:15 – 13:30 **The impact of Sex Chromosome Trisomies (XXX, XXY, XYY) on early social functioning: social attention, affect recognition and Autism Spectrum Disorders symptoms**
Dr. Nienke Bouw (NL)

12:15 – 13:30 **Parallel session 6 - Genes and pathways**
Chair: Prof. Zeynep Tümer (DK)
Invited speaker
12:15 – 12:45 **Novel genetic mechanisms underlying RASopathy spectrum disorders**
Prof. Marco Tartaglia (IT)

Selected abstracts
12:45 – 13:00 **SEMA6B variants cause intellectual disability and alter dendritic spine density and axon guidance**
Prof. Annick Toutain (FR)
13:00 – 13:15 **LHX2 loss of function causes neurodevelopmental deficits in humans and flies**
Dr. Anne Gregor (CH)
13:15 – 13:30 **Biallelic KDM8 variants in two sibs with severe failure to thrive, intellectual disability and peculiar facial dysmorphism**
Prof. Katrin Ounap (EE)

13:30 – 14:30 ***Lunch***

14:30 – 16:45 **Plenary Session: Update on most frequent syndromes & wrap up with panel of speakers**
Chair: Prof. Hilde Van Esch (BE) & Dr. Sylvia Huisman (NL)

Selected abstracts
14:30 - 14:45 **Update on adults with 22q11.2 deletion syndrome**
Dr. Eric Boot (NL)
14:45 – 15:00 **Comparison of behavioural and socio-communicative capacities in school-aged children with 16p11.2 deletion and their siblings**
Drs. Jente Verbesselt (BE)
15:00 – 15:15 **Clinical and molecular spectrum of SMARCC2-associated Coffin-Siris syndrome**
Dr. Georgia Vasileiou (DE)
15:15 – 15:30 **Developmental characteristics of children with Helsmoortel-Van der Aa syndrome**
Dr. Anke Van Dijck (BE)
15:30 – 15:45 **Building in vivo human neuronal models for MECP2-related disorders**
Drs. Nona Merckx (BE)

15:45 – 16:00 **MED13L knockout in Cerabral organoids leads to a shifted developmental program through abnormal CIS-regulatory element activation**
Dr. Jamal Ghoumid (FR)

16:00 – 16:15 **Menke-Hennekam Syndrome; delineation of domain-specific subtypes with distinct clinical and DNA methylation profiles**
Dr. Leonie Menke (NL)

16:15 – 16:45 **Wrap up with panel of speakers**

16:45 – 17:15 **Final words**
Prof. Tjitske Kleefstra (NL), Prof. Christiane Zweier (CH), Prof. Alain Verloes (FR)