

European Reference Network for Rare Malformation Syndromes, Intellectual and Other Neurodevelopmental Disorders (ERN ITHACA)

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

Preliminary Program

(Version: 24 October 2022 with confirmed invited speakers and *tentative titles of their presentations; the selected abstract presentations in the different sessions will be added in January 2023!*)

Thursday 20 April 2023

12:30 – 13:00 Registration

13:00 – 13:30 **Welcome on behalf of the Scientific Committee**

Prof. Tjitske Kleefstra (NL) and Dr. Christiane Zweier (CH)

13:30 – 15:00 **Plenary session 1**

What can ERNs do for patients with NDD

Prof. Alain Verloes (ITHACA Coordinator FR)

Navigating the uncertainties of next-generation sequencing in the genetics clinic

Prof. Hilde Van Esch (BE)

Disability assessment and case management of NDD in Europe

Dr Stephanie Miot (FR)

Networking break / poster viewing

16:30 – 17:30 **Parallel session 1 Applied & Emerging Therapies**

Personal healthcare through braincells on a chip

Prof Nael Nadif Kasri (NL)

Restoring Gonadotropin-releasing hormone production to restore cognitive function

Dr. Vincent Prevot (Fr) & Prof. Nelly Pitteloud (CH)

Parallel session 2 Genetics of Neurodevelopmental Diseases

Multigenic score in autism

Prof. Thomas BOURGERON (FR)

SysNDD and broader NDD gene overview

Dr Bernt Popp (DE)

18:00 – 19:00 **Keynote lecture followed by a panel discussion**

A multidisciplinary view on rare genetic neurodevelopmental disorders

Prof. Han Brunner (NL)

19:00 – 21:00 **Networking buffet**

Friday 21 April 2023

8:45 – 9:00 Welcome & technical information

9:00 – 10:30 **Plenary session**

Big Data & Artificial Intelligence

Prof Wiro Niessen (NL)

Transition and adult care

ass. Prof. Laura De Graaff (NL)

Clinical Diagnostics with Semantic Similarity Searches in Ontologies

Prof. Peter Krawitz (DE)

Coffee break

11:00 – 12:15 **Parallel session 3 Profound and multiple learning disability**

Communication in Neurodevelopmental Diseases

Dr Gill Townend (NL)

Polyhandicap and Neurodevelopmental Diseases

Prof. Thierry Billette de Villemeur (FR)

Parallel session 4 Mechanisms of diseases, model systems & translational pre-clinical work

PRISM screen: a tool to screen for functional importance of new genetic variants

ass Prof Geeske van Woerden (NL)

Personalized gene therapy using CRISPR/Cas9 technology

Prof. Alessandra Renieri (IT)

12:15 – 13:30 **Parallel session 5 Ethical, legal, and Psycho-social aspects**

Disability assessment and case management of NDD in Europe

Mrs Dorica Dan (RO)

The burden of having a relative with a neurodevelopmental disorder

(Invited speaker will be announced soon)

Parallel session 6 Genes and pathways

Novel genetic mechanisms underlying RASopathy spectrum disorders

Dr Marco Tartaglia (IT)

Genetic and epigenetic mechanisms in disease development

Prof Zeynep Tümer (DK)

13:30 – 14:30 **Lunch**

14:30 – 17:00 **Plenary Session: Update on most frequent syndromes & wrap up with panel of speakers.**

Looking forward to learn from your perspectives on Rare Genetic Neurodevelopmental Disorders

Yours sincerely,

On behalf of the ERN-ITHACA *Chairs Organizing committee and Scientific committee*