HELSE BERGEN
Haukeland universitetssjukehus

ITHACA

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ITHACA

Board Meeting

3-5 December 2025

Bergen, Norway

Patient Workshop - Wednesday 3 December

9:00 Opening - **Dorica Dan, Anne Hugon**

9:10 Engaging with patient organisations in the field of Rare Genetic/Genomic Disease - **Sofia Douzgou Houge**

9:30 Flash talks - Introduction: “Learning from each other” - **Tanja Zdolšek Draksler**

Part 1: Learning from each other workshop - Best practice sharing

9:35 *Clinics and Patient care, global collaboration*

1. From Model Transfer to Sustainable Impact: The NoRo–Frambu Collaboration for Integrated Rare Disease Care in Romania - **Dorica Dan**
2. How to shift from being perceived as a burden by professionals to being recognized as symbiotic partners - **Daniele Palumbo**
3. AlfaSAAC Project: Enhancing communication in children with Phelan-McDermid - **Rui Barbosa Guedes**
4. Children need to be aware - **Monica Bertoletti**

10:30 **Coffee break**

Patient Workshop - Wednesday 3 December (PM)

11:00 *Data collection and registries*

5. Building Patient-Driven Natural History Studies for Rare Neurodevelopmental Disorders - **Verena Schmeder**
6. Angelman caregiver survey - **Ellen Koekoeckx**

11:30 *Extra communication*

A Paternalistic Barrier: The Fight for Patient Information Access in Rare Disease Care in Hungary - **Eszter Szabo**

11:40 Joint Debate: Takeaways from the Learning from each other workshop - **Tanja Zdolšek Draksler, Dorica Dan**

12:00 **Lunch**

Part 2: Innovative Approaches in rare diseases

13:00 Round Table: Co-Creating Care, Patients and Clinicians in Guideline Development - **Antje Enekwe, Sue Routledge, Mirthe Klein Haneveld**

13:40 (genetically) Undiagnosed communities: a Norwegian approach - **Sofia Douzgou Houge**

14:00 Norway Holistic views (Frambu, Rare resources net) - **Eirik Hovlan, Heidi Nag**

14:20 Dealing with marginalised families and populations within society - **Sally Ann Lynch**

14:40 From Pathways to Well-Being: How Inter-ERN WG SBoD from Care pathway, Center Mapping, and Quality of Life for Children - **Benoit Fourcroy, Giovanni Mosiello, Jean Marie Jouannic**

15:00 Discussion, Wrap-up

15:30 **Coffee break**

Part 3: Innovative Approaches in rare diseases

16:00

Group 1

Natural History Studies (NHS) - **Erika Stariha, SATB2 Europe**

In this discussion-based session, we will revisit why Natural History Studies (NHS) are essential, explore the different types and typical components of NHS for rare neurodevelopmental disorders (NDD), and share a few examples of ongoing initiatives. Together, we will reflect on the main challenges in establishing NHS (especially for very rare NDD) and consider how we might develop a more standardised and feasible approach going forward.

Group 2

School in the Circles of support - student/family-centred education model - **Katarzyna Świeczkowska, Polish Association for Persons with Intellectual Disability Gdańsk**

The objective is to discuss the challenges associated with educating a child with NDD and present an innovative model of child and family centred education for children with disabilities, including intellectual disability.

17:00 **Closing**

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ERN Board Meeting - Thursday 4 December

9:00 Opening - **Eivind Hansen, Gunnar Douzgos Houge**

9:15 National coordination Unit for Rare diseases in Norway - **Martin Aker**

Plenary 1 - Chair: Alain Verloes & Sally Ann Lynch

9:35 ERN-ITHACA 2026 news - **Alain Verloes**

- Welcome to our coordinator-elect - **Sally Ann Lynch**
- The future of ERNs, Plans for 2026-27

10:15 ERN-ITHACA Activity report, Budget overview; Grant monitoring; HCP performance review - **Klea Vyshka**

10:30 **Coffee break**

Plenary 2 - Chair: Klea Vyshka

11:00 Screen4Care - **Alessandra Ferlini**

11:15 WG Research & Innovation activity review - **Zeynep Tümer, Marco Tartaglia**

11:30 WG CPMS 2.00. What can we do? - **Sofia Douzgou Houge**

11:45 Round table: the Future of CPMS for ERN-ITHACA

12:00 **Lunch**

Plenary 3 - Chair: Anne Hugon

13:00 WG Education and Training activities - **Laurence Faivre, Giuseppe Zampino**

13:15 Genetics Half-Hour - **Dagmar Wieczorek**

13:20 APOGeE Update - **Alain Verloes, Nicholas Szeto**

13:25 Patient Advisory Board activities, Key messages Patient Workshop - **Dorica Dan, Tanja Zdolšek Draksler**

13:45 WG Fetal Medicine - **Gijs Santen**

14:00 WG ILIAD - **Gerieke Been, Marije Van Der Geest**

14:15 ERN Liaison group with ESHG - **Holm Graessner, Carla Oliveira**

14:45 **Coffee break**

15:15 National and International Solvatons in the Jardin Project - **Katrin Õunap**

15:30 Achievement new challenges, Young Geneticists Committee - **ESHG-Y: Silvia Kalantari, Mathys Weber**

15:45 WG SBoD activity review - **Giovanni Mosiello, Jean-Marie Jouannic**

16:00 WG NDD activity review - **Tjitske Kleefstra, Christiane Zweier**

16:15 EuroNDD2026 programme - **Jolanda Van Golde**

16:25 WG Guidelines activity review - **Agnies Van Eeghen, Katalin Szakszon**

16:45 PROMs - **Agnies Van Eeghen, Jolanda Van Golde**

17:00 Panel discussion: The future of ERN-ITHACA - strengthening synergies and empowerment of HCPs

18:00 **Closing - Alain Verloes**

20:00 **Dinner at Scandic Bergen City**

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Research Workshop - Friday 5 December

8:55 Welcome - **Marco Tartaglia, Zeynep Tümer**

ERDERA Diagnostic Research Workstreams

9:00 Diagnostic Research Workstreams in ERDERA: data submission processes and data rediness - **Leslie Matalonga**

9:30 Diagnostic Research Workstreams in ERDERA: DATF analyses - **Sergi Beltran**

10:00 Diagnostic Research Workstreams in ERDERA: DITF and data interpretation - **Lisenka Vissers**

10:30 **Coffee break**

ERN-ITHACA meets Genturis

11:00 Introduction to GENTURIS and genomic instability in cancer and diseases - **Marjolijn Ligtenberg**

11:30 The concept of mosaic RASopathies /neurofibromatosis and schwannomatosis - **Hilde Brems**

12:00 **Lunch**

13:00 Telomere biology and its link to developmental disorders - **Kleoniki Roka**

Collaborative research talks

13:30 Defining Myhre syndrome in Adulthood - **Bert Callewaert**

13:45 AUTS2 syndrome: defining genotype-phenotype correlation - **Alessandra Renieri**

14:00 Genotype-phenotype characterization of YWHAG-related disorders - **Maria Eugenia Amato**

14:15 **Break**

14:30 Further delineation of BRSK2-associated NDDs - **Christiane Zweier**

14:45 Clinical, genetic and neuroimaging features of biallelic NOTCH3 disorders - **Pablo Iruzubieta Agudo**

15:00 Houge-Janssens syndrome: Clinical overview and novel pathogenic mechanisms - **Gunnar Douzgos Houge**

15:15 **Closing - Marco Tartaglia, Zeynep Tümer**

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