ITHACA

Board Meeting

1-3 December 2026

Fondation Biermans-Lapôtre,
Maison de la Belgique et du Luxembourg
9A Bd Jourdan, 75014

Paris, France



Patient & Clinician Workshop - Tuesday 1 December

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

9:20 French Rare Diseases Alliance: The Involvement of Rare Disease Patient Organisations in Research - **Lara Chappell**

Part 1: Learning from each other

9:40 Introduction - **Tanja Zdolšek Draksler**

9:45 SYNAPSE: Translating family caregivers' experiential knowledge into professional training through participatory action research in Angelman syndrome - **Lara Chappell**

10:00 Communication centres for persons with disabilities: Building a nationwide system of support for people with complex communication needs - **Daniele Palumbo**

10:15 Patient–professional collaboration in rare diseases: connecting community care, national expertise and European networks in Romania / Jardin project guideline for care coordination and case management - **Dorica Dan**

10:30 Discussion

10:45 **Coffee break**

11:15 Kabuki INSIGHTS: combining electronic health record and survey data in a patient organisation-led natural history registry - **Jessica Weatherstone & Clara Tang**

11:30 Partners in research and care: Collaboration between patient representatives and professionals in Kleefstra syndrome - **Tanja Zdolsek Draksler**

11:45 Access to information and paternalism in rare disease care: An expanded European follow-up analysis - **Eszter Szabóné Katona**

11:50 Discussion

12:15 **Lunch**

13:30 An example of a collaboration spanning more than a decade between clinicians, family members, and children with Mowat-Wilson syndrome, facilitated by the Italian Mowat-Wilson Association - **Livia Garavelli**

13:45 ALL IN ONE: Where Relationship Meets Science in BWSp - The therapeutic alliance led by AIBWS OdV in synergy with the ERN-ITHACA community - **Monica Bertoletti**

14:00 From Parent-Led Vision to Global Consensus: Lessons in Collaboration, Trust and Shared Commitment - **Erika Stariha**

14:15 Tuberous sclerosis complex: Making the invisible visible - **Mary Desmond Vasseghi**

14:30 Discussion

Part 2: Interactive presentations

14:45 ITHACA PROM set kick-off - **Agnies van Aeghen, Jolanda Van Golde**

15:15 **Coffee break**

Part 3: Round table

15:45 **Round table discussion: Shaping the Future of ERN ITHACA Together**

16:45 Key Outcomes, Added Value and Recommendations, Closing

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ERN Board Meeting - Wednesday 2 December

9:00 Opening - **Alain Verloes & Sally Ann Lynch**

Plenary 1 - Chair: Alain Verloes & Sally Ann Lynch

9:15 Activity report, Assessment of past year, Workplan, Activities to come - **Alain Verloes, Sally Lynch**

9:45 Budget overview; Grant monitoring; HCP performance review - **Mathias Falkenberg**

10:00 Building Bridges Across ERNs: Collaboration with MetabERN - **François Eyskens**

10:15 Online Inherited Metabolic Disorders (OIMD) – A comprehensive online guide to the diagnosis and treatment of IMDs - **Johannes Zschocke**

10:30 **Coffee break**

Plenary 2 - Chair: Mathias Falkenberg

11:00 WG Research & Innovation - **Zeynep Tümer & Marco Tartaglia**

11:15 WG CPMS - ITHACA Clinic - **Adela Chirita-Emandi**

11:30 WG Education and Training - **Laurence Faivre & Giuseppe Zampino**

11:45 WG Guidelines - **Agnies Van Eeghen & Katalin Szakszon**

12:00 **Lunch**

Plenary 3 - Chair: Anne Hugon

13:00 The future of ERNs: Insights from the French Ministry of Health and a BoMS Member - **Anne Sophie La Pointe**

13:20 Why parental perspectives matter in prenatal genetic counselling: CNVs and mosaicism - **Stina Lou**

13:40 WG Fetal Medicine - **Gijs Santen**

13:55 WG ILIAD - **Gerieke Been & Marije Van Der Geest**

14:10 The importance of exome/genome reanalysis in the UDP - **Katrin Õunap**

14:30 **Coffee break**

15:00 European university diploma in rare disease research: Exploring collaboration opportunities with ERN ITHACA - **Kaan Mert Güven**

15:20 Achievement new challenges, Young Geneticists Committee - **ESHG-Young: Silvia Kalantari & Mathys Weber**

15:35 WG SBoD - **Giovanni Mosiello & Jean-Marie Jouannic**

15:50 WG NDD - **Tjitske Kleefstra & Christiane Zweier**

16:05 Patient Advisory Board activities, Key messages from Patient Workshop - **Dorica Dan & Tanja Zdolsek Draksler**

16:20 Panel discussion: Empowering HCPs, building synergies, shaping the future: strategic priorities for the 2028–2029 ERN Bridging Grant

17:20 **Intermezzo - Sally Ann Lynch**

17:40 **Closing - Alain Verloes**

20:00 **Dinner at** 🍾🥂 **La Coupole** - 102 boulevard du Montparnasse, 75014 Paris

Research Workshop - Friday 5 December

9:00 Welcome - **Marco Tartaglia, Zeynep Tümer**

Session 1: MetabERN

9:15 Information on the learning & training programme of the MetabERN; Research initiatives of the subnetwork peroxisomal (Peroxisomal Biogenesis defects) and Lipid-related disorders (Smith Lemli Opitz syndrome) - **Francois Eyskens**

9:45 Beyond the genome: (Cellular) metabolomics and proteomics as functional readouts of genetic variation - **Irene Körver-Keularts**

10:15 Making sense of genetic variants: The role of fluxomics in functional diagnostics - **Marek Noga**

10:45 Metabolic disorders around the EU & Norway - **Sally Ann Lynch**

11:00 **Brunch**

Session 2A: Collaborative research

12:00 Shprintzen-Goldberg syndrome: Long term follow-up of the cardiovascular features in an international cohort of 29 patients - **Faivre Laurence**

12:15 Impact and natural history of metabolic and endocrine complications in a large RATARS cohort - **Francesca Clementina Radio**

12:30 Novel variants and DNA methylation alteration on pathologic tissues from a cohort of Italian patients with spinal dysraphisms - **Valeria Capra**

12:45 Delineation of the adult phenotype of Nicolaides-Baraitser, Coffin-Siris, Costello and CFC syndrome - **Dagmar Wieczorek**

13:00 **Break**

Session 2B: Collaborative research

13:30 Functional stratification of YWHAZ variants explains distinct neurodevelopmental disorders - **Marco Tartaglia**

13:45 Expanding the phenotypic and genotypic spectrum of TIMES syndrome, an LRRC8C-related disorder - **Livia Gravelli**

14:00 Vitamin D and calcium metabolism in adults with Williams-Beuren syndrome - **Laura de Graaff**

14:15 Expanding the phenotypic and genotyping spectrum of NAA15 related disorder - **Alessandra Renieri**

14:30 **Closing - Marco Tartaglia, Zeynep Tümer**

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