Patient Workshop - Thursday 12 December

9:00 Opening - **Maria Puiu, Anne Hugon**

9:10 Added value of ERN ITHACA to engage patient organisation at national level Rare Diseases Roumania, state of art - **Dorica Dan**

9:20 Activity report and added value from PAB activities in ERN ITHACA - **Dorica Dan, Anne Hugon**

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

9:30 Introduction - **Tanja Zdolšek Draksler**

9:35 *Clinics and Patient care*

1. NoRo in community or how to upscale your resources? - **Dorica Dan**
2. Multidisciplinary clinic for patients with macroglossia in BWS - **Monica Bertoletti**
3. PKS Clinic: a multidimensional approach to Pallister-Killian Syndrome care - **Samantha Carletti**
4. Care coordination for all rare conditions in Ireland – A comparison between 22q11 Deletion Syndrome and 2q24 Deletion Syndrome - **Lyndsey Walsh**, **Anne Lawlor**

10:30 **Coffee break**

Patient Workshop - Thursday 12 December (PM)

11:00 *Global collaboration and guidelines*

5. The Value of WaihonaPedia for your community, guide for community leaders - **Gerritjan Koekkoek**
6. Parent co-led initiative in creating the first global Clinical Practice Guideline (CPG) for a rare neurodevelopmental disorder called SATB2-Associated Syndrome (SAS) - **Erika Stariha**

11:30 *Data collection and registries*

7. GASR - Global Angelman Syndrome eRegistry - **Ellen Koekoeckx**
8. Building a research portal for rare disease collaboration: CHAMP1 Approach - **Daniele Palumbo**
9. Solving rare diseases: How global collaboration and data are advancing Kleefstra Syndrome research - **Tanja Zdolšek Draksler**

12:10 General discussion - **Tanja Zdolšek Draksler, Dorica Dan**

12:40 **Lunch**

Part 2: The Patient Journey — Navigating Diagnosis, Grief, and Mental Health

13:30 Mental health and wellbeing and rare conditions - **Kristen Johnson**

Session 1: Diagnosis

14:00 Role of genetic counseling in managing the impact of a diagnosis - **Sofia Douzgou Houge, Adela Chirita**

14:15 Communicating diagnosis: Addressing the needs of patients, families, and doctors - **Eduardo Tizzano**

14:30 Diagnostic journey of a rare disease patient, a parent perspective - **Erika Stariha**

14:45 Patient advisory board feedback on communicating diagnosis, discussion - **Ellen Koekoeckx**

Session 2: Parallel sessions

	Group 1 Identifying and mapping resources to boost family resilience	**Group 2** Different faces of grief
15:30	Introduction: **Ioana Streata**, **Ammi Sundqvist Andersson** Moderation and participation: **Dorica Dan**, **Monica Bertoletti**	Introduction: **Andrada Ciuca** Moderation and participation: **Kasia Katarzyna Świeczkowska**, **Marie-Christine Rousseau**, **Esther Szabo**, **Samantha Carletti**, **Gerritjan Koekkoek**

17:00 Summary of groups 1 and 2, results and final conclusions - **Dorica Dan, Tanja Zdolšek Draksler**

17:15 **Cocktail reception**

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ERN Board Meeting - Friday 13 December

9:00 Opening - **Nicolae Istudor, Maria Puiu, Adela Chirita Emandi**

Plenary 1 - **Alain Verloes**

9:30 ERN ITHACA Assessment of past year, workplan, upcoming activities - **Alain Verloes**

9:45 ERN ITHACA Activity report, budget overview; new grant; HCP performance review - **Klea Vyshka**

10:00 HCP performance scheme: Feedback and evaluation - **Alain Verloes**

10:15 WG Research & Innovation activity review - **Zeynep Tümer, Marco Tartaglia**

10:30 WG CPMS/Online Demo CPMS 2 - **Sofia Douzgou Houge, Clementina Radio**

10:45 **Coffee break**

Plenary 2 - **Klea Vyshka**

11:15 WG Guidelines activity review - **Agnies Van Eeghen**
Endorsement procedure - **Katalin Szakszon**
Prioritization criteria - **Mirthe Klein Haneveld**
Discussion and vote on priorization

11:45 WG NDD activity review - **Tjitske Kleefstra, Christiane Zweier**

12:00 Joint Action on Integration of ERNs into National Healthcare Systems - JARDIN - **Till Voigtländer**

12:30 WG ILIAD activity review - **Marije van der Geest**

12:45 General discussion

13:00 **Lunch**

Plenary 3 - **Anne Hugon**

14:15 GestaltMatcher - presenting the new app - **Peter Krawitz**

14:45 WG Education and Training activities - **Laurence Faivre, Giuseppe Zampino**

15:00 WG SBoD activity review - **Giovanni Mosiello, Jean-Marie Jouannic**

15:15 Patient Advisory Board activity review - **Tanja Zdolsek Draksler, Dorica Dan**

15:45 **Coffee Break**

16:30 ESHG Young Geneticists Committee: Achievements, new challenges - **Silvia Kalantari, Ana Raquel Silva**

16:45 WG Fetal Medicine activity review - **Andreas Dufke, Gijs Santen, Tania Attie**

17:00 WG Fetal Medicine: New taskforce on congenital malformations and neurodevelopmental disorders in MC twins - **Yves Ville**

17:15 Panel discussion: The future of ERN ITHACA - strengthening synergies and empowerment of HCPs

18:00 **Closing - Alain Verloes**

19:30 **Dinner at Caru' cu Bere**
Strada Stavropoleos 5, București 030081

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Research Workshop - Saturday 14 December

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

Joint session with ERN EYE

9:30 ERN EYE introduction - **Hélène Dollfus**

9:45 Bardet-Biedl syndrome improved diagnosis criteria and management: Inter-ERN consensus statement and recommendations - **Hélène Dollfus**

10:15 The role of the dark matter of the genome in rare diseases or multi-omics to tackle missing heritability in rare diseases - **Elfride De Baere**

10:45 **Coffee break**

Novel approaches

11:15 Disease modelling in cultured murine hippocampal neurons using multi-electrode array - **Geeske van Woerden**

11:45 Small RNAs, transcription factors, lipids and ID - **Alex Reymond**

12:15 Unravelling undiagnosed rare disease cases by HiFi long-read genome sequencing - **Lisenka Vissers**

12:45 **Lunch**

Collaborative research talks

13:45 Further delineation of RPL26-associated malformations - **Clemence Vanlerberghe**

14:05 Epigenomic and phenotypic characterization of DEGCAGS syndrome - **Sofia Douzgou**

14:25 Biallelic loss-of-function variants in ZNF142 are associated with a robust and distinctive DNA methylation signature - **Mathis Hildonen**

14:45 **Coffee Break**

15:00 XRCC4-related Microcephalic Dwarfism - **Silvestre Cuinat**

15:20 GTF2I and neurodevelopmental disorder

15:40 Closing - **Marco Tartaglia, Zeynep Tümer**

16:00 **Networking with drinks**

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