ANNUAL ERN ITHACA BOARD

MADRID, 9-11 DEC. 2021
LA PAZ University Hospital

9th PATIENT COUNCIL SESSION
From 3 PM to 5 PM

10th PLENARY BOARD, DAY 1
From 9 AM to 6 PM

11th PLENARY BOARD, DAY 2
From 9 AM to 1 PM

Co-funded by the Health Programme of the European Union

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Hospital Universitario La Paz

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THURSDAY 9TH PROGRAM

PATIENT COUNCIL
PRESENTED BY DORICA DAN AND GABOR POGANY

15 H 00 Opening session and overview of ERN ITHACA Patient Council - Speaker: Dorica Dan, PC Chair, Romanian Prader Willi, Romania ; Gabor Pogani, Co Chair,Hungarian Williams Syndrome, Hungary

15 H 15 Rare diseases Patient Organisation in Spain: the case of FEDER - Speaker: Alba Ancochea, President of FEDER, Italia

15 H 30 Learning from challenges, achievements and opportunities - Speaker: Sue Routledge, Pitt Hopkins Syndrome, UK ; Ioel Detton, Association Noonan France, FR ; Tomasz Grybek, Borys the Hero Foundation, PL; Katarzyna Swieczkowska, PSONI - Polish Association for People with ID, PL

16 H 00 *Discussion with clinicians* | Sharing expertise and expectations - Moderators: Ammi Andersson, Pitt Hopkins Syndrome, UK | SE ; Anne Hugon, ERN-ITHACA,FR

16 H 10 Bridging the gap between Centres of Expertise and Patient Groups - Speaker: Dorica Dan; Gerritjan Koekkoek, Cdls World federation, NL; Papatya Alkan, International Federation for Spina Bifida and Hydrocephalus, UK

16 H 40 *Discussion with clinicians* | Reinforcing our collaborations - Moderators: Ammi Andersson, Pitt Hopkins Syndrome, UK | SE; Annalisa Scopinaro, Uniamo FIMR Onlus – Federazione Italiana Malattie Rare, IT

17 H 00 Closing remark and vision for the next ITHACA 2022-2027 - Speaker: Dorica Dan, PC Chair, Romanian Prader Willi, Romania; Gabor Pogani Co Chair,Hungarian Williams Syndrome, Hungary

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FRIDAY 10TH PROGRAM

PLENARY BOARD - DAY 1

09 H 00	Welcome speeches - Speakers: Dr Juan José Ríos Blanco, Director of La Paz Hospital ; Dr Pablo Lapuninza, Spain ; Dr Alain Verloes, France
09 H 15	Overview of ITHACA Y4-Y5, Budget overview - Speakers: Coordination team
09 H 35	Second 5-year workplan propositions - Speakers: Coordination team
10 H 00	WG 4: Expert consensus statements - Speaker: Dr Raoul Hennekam, Netherlands
10 H 20	WG 6: ILIAD Registry - Speaker: Dr Morris Swertz, Germany
10 H 40	*Coffee break*
11 H 10	WG 7: Research & Innovation - Speaker: Dr Marco Tartaglia, Italia
11 H 30	WG 8: Teaching, Learning & Training - Speaker: Dr Guiseppe Zampino, Italia
11 H 50	WG 9: Neurodevelopmental Disorders - Speaker: Dr Tjitske Kleefstra, Netherlands
12 H 10	WG 10: Spina Bifida - Speaker: Dr Giovanni Mosiello, Italia
12 H 30	WG 13: Patient Council - Speaker: Dorica Dan, PC Chair, Romania
12 H 50	*Lunch break*
14 H 00	TALK| *Research strategies in EU* - Speaker: Hélène Le Borgne, European Commission Policy Officer
14 H 30	WG 5: Digital Health - Speaker: Dr Sofia Douzgou, Norway
14 H 45	WG 11: Fetal Medecine -Speaker: Dr Andreas Dufke, Germany
15 H 05	WG 12: APOGE - Speaker: Dr Alain Verloes, France
15 H 20	Final discussion and Vote on ITHACA's action plan for the next 5 years
15 H 40	TALK| 1+ Million Genome initiative/Beyond 1M Genome project overview - Speaker: Dr Serena Scollen, Head of Human Genomics and Translational Data at ELIXIR, UK
16 H 00	*Coffee break*
16 H 30	TALK| *Artificial intelligence* - Speaker: Pr. Tanja ZDOLSEK, Research Project Manager at Jozef Stefan Institute, Slovenia
17 H 00	TALK| *Gestalt matcher* - Speaker: Peter KRAWITZ, Head of the Institute for Genomic Statistics and Bioinformatics (W3), Bonn University, Germany
17 H 30	TALK| *Strengthening synergies between ITHACA and Conect4children* - Speaker: Mark TURNER, Professor of Neonatology and Research Delivery at University of Liverpool, UK
18 H 00	
18 H 30	Closing remarks
21 H 00	*Friendly Spanish dinner*

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SATURDAY 11TH PROGRAM

PLENARY BOARD - DAY 2

09 H 00 CIBERER and the Spanish Programs for Undiagnosed Rare Diseases - Speaker: Pr Luis Perez Jurado, Spain

09 H 30 TALK| A European Action Plan on Rare Diseases: How to get there and what to achieve - Speaker: Terkel Andersen, President of EURORDIS-Rare Diseases Europen, Denmark

10 H 00 5 years of ERNs - opportunities and challenges - Speaker: Dr. José Luis VALVERDE, European Commission Policy Officer, DG-SANTE

10 H 20 Coffee Break

10 H 50 TALK| Use of DNA methylation episignatures for diagnosis of hereditary neurodevelopmental disorders - Speaker: Bekim Sadikovic, Professor at the Department of Pathology and Lab Medicine, Western University; Director of the Verspeeten Clinical Genome Centre, London Health Sciences

11 H 40 TALK| Human Phenotype Ontology: extensions for prenatal phenotyping - Speaker: Pr. Peter Robinson, Professor at The Jackson Laboratory for Genomic Medicine, USA

12 H 20 Wrap up and conclusion

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