

Day 1

WORKSHOP SESSION

09:00 WORKSHOP OPENING

DIAGNOSIS AND PHENOTYPE-GENOTYPE CORRELATION

Location: Auditori Sant Joan de Déu

Chairs: Alexis Arzimanoglou (France) – Emilio Fernandez Alvarez (Spain)

09:10 SYNaPS program: Synaptopathies and Paroxysmal Syndromes Study - Henry Houlden (United Kingdom)

09:30 Genotype-phenotype correlations in SCN8A-related disorders reveal prognostic and therapeutic implications. Rikke Moller (Denmark)

09:50 SCN1A variants with movement disorder phenotypes. Andreas Brunklaus (United Kingdom)

10:10 GNAO1-related neurodevelopmental disorder: phenotypic and genotypic characterization of Spanish patients. Juan Dario Ortigoza-Escobar (Spain)

10:30 Discussion

11:10 COFFEE BREAK

TRANSLATIONAL RESEARCH & TREATMENT FOR MOVEMENT DISORDERS - I

Location: Auditori Sant Joan de Déu

Chairs : Eleni Panagiotakaki (France) – Mohamad Mikati (USA)

11:50 Drug repositioning in ATP1A3. Francesco Danilo Tiziano (Italy), Elisa De Grandis (Italy), Carmen Fons (Spain), Eleni Panagiotakaki (France)

12:10 A translational approach to childhood-onset forms of hereditary spastic paraplegia. Darius Ebrahimi-Fakhari (United States)

12:30 Discussion

13:10 Lunch

14:10 ROUND TABLE – SENIOR AND YOUNG RESEARCHERS

TRANSLATIONAL RESEARCH & TREATMENT FOR MOVEMENT DISORDERS - II

Location: Auditori Sant Joan de Déu

Chairs: Renzo Guerrini (Italy) – Jaume Campistol (Spain)

15:00 Development of AAV-mediated gene therapy for GNAO1 encephalopathy – Maryana Bardina (Russia)

15:20 AADC gene therapy. Agathe Roubertie (France)

15:40 Experimental models in Tyrosine Hydroxylase deficiency. Alba Tristán* (Spain)

16:00 Gene therapy for infantile parkinsonism - Manju Kurian (United Kingdom)

16:20 Discussion

17:00 COFFEE BREAK

NEW CHALLENGES IN EPILEPTIC ENCEPHALOPATHIES

Location: Auditori Sant Joan de Déu

Chairs: Gaetan Lesca (France) – Elena Gardella (Denmark)

17:30 The role of novel therapies in genetic syndromes with movement disorders and epilepsy-clinician perspective - Galina Stevanović (Serbia)

17:50 Challenges in diagnostics of epilepsy and movement disorders treatable with novel therapies in Bulgaria. Petia Dimova and Slavena Hirstova* (Bulgaria)

18:10 Discussion

18:50 End of sessions

20:00 DINNER

Day 2

WORKSHOP SESSION

NEW MEDICAL TREATMENT AND REPOSITIONING DRUGS FOR EPILEPTIC ENCEPHALOPATHIES

Location: Auditori Sant Joan de Déu

Chairs: Helen Cross (UK) – Masa Malenica (Croatia)

08:30 The role of new medical treatments for the management of developmental and epileptic encephalopathies: Novel concepts and results. Cecilie Johannessen Landmark (Norway)

08:50 Repositioning of drugs in epileptic encephalopathies. Sandra Acosta (Spain)

09:10 Discussion

09:50 Coffee break

EPILEPTIC ENCEPHALOPATHIES: TRANSLATIONAL MEDICINE AND TREATMENT

Location: Auditori Sant Joan de Déu

Chairs: Nicola Specchio (Italy) – Guido Rubboli (Denmark)

10:20 Therapeutic advances in SCN8A Developmental and Epileptic Encephalopathy Syndrome - Anna Keleman (Hungary)

10:40 L-Serine dietary supplementation in loss-of-function GRIN2B-related pediatric encephalopathy - Natalia Julia* (Spain)

11:00 Radiprodil, a NR2B (GRIN) negative allosteric modulator, from bench to bedside in infantile spasm syndrome - Stephane Auvin (France)

11:20 Progressive myoclonus epilepsy, EPM1 - towards targeted treatments. Anna-Elina Lehesjoki (Finland)

11:40 DBS in Lennox-Gastaut syndrome. Marios Kaliakatsos (United Kingdom)

12:00 Discussion

12:40 LUNCH

13:40 ROUND TABLE - Patient advocacy groups

CLINICAL CASES PRESENTATIONS

CHAIRS: CARMEN FONS (SPAIN) – FLOOR JANSEN (THE NETHERLANDS)

14:40 SCN1A responsive to Carbamazepine/Phenytoin / Hyperekplexia-like SCN1A patient - ESTONIA / ROMANIA

15:00 FOXP1 BIAYNA SUKHUDYAN, (ARMENIA)

15:20 ADVANCES IN THE MANAGEMENT OF DRAVET SYNDROME. DANA CRAIU (ROMANIA)

15:40 Discussion

16:20 WORKSHOP CLOSURE

16:30 DEBRIEFING SESSION

(CHAIR: A. ARZIMANOGLU, ERN EPICARE COORDINATOR; ORGANIZERS; LEADERS OF INVOLVED RESEARCH TEAMS, PATIENT ADVOCACY REPRESENTATIVES AND OTHER STAKEHOLDERS)

*Young researcher