GENETIC SYNDROMES
WITH
MOVEMENT DISORDERS
AND
EPILEPSY

FEBRUARY 6-7
2020
BARCELONA

PROGRAM

UPDATED VERSION

Chairmen
Alexis ARZIMANOGLOU (France)
Jaume CAMPISTOL (Spain)
Emilio FERNANDEZ-ALVAREZ (Spain)
Renzo GUERRINI (Italy)
Nardo NARDOCCI (Italy)

www.paedmovdissymposium.com

Barcelona

We have the pleasure to inform you that the 1st International Symposium on Genetic Syndromes with Movement Disorders and Epilepsy will be held in Barcelona, Spain on February 6-7, 2020. This first edition is organized in continuity of the International Symposium on Child Movement Disorders, which gathered every other year more than 300 professionals from 30 different countries since 2004. The best qualified experts involved in pediatric movement disorders took part in this biannual meeting.

The main topics proposed for the 2020 meeting include:

- Co-occurrence of Movement Disorders and Epilepsy: an historical overview;
- The hunting of causative genes;
- Basic cellular mechanisms involved in movement disorders and epilepsy;
- Pathways involved in the co-occurrence of movement disorders and epilepsy;
- Disorders related to: GNAO1; ATP1A3; SCN1A; FOXG1; TBC1D24; Other;
- Principles of current and future management: medical treatments of the epilepsies, the movement disorders; deep brain stimulation; Gene therapy and other innovative treatments

We remain at your disposal for any further information you might like.

Best regards,

The organizing committee
Alexis Arzimanoglou (France), Jaume Campistol (Spain), Emilio Fernandez-Alvarez (Spain), Renzo Guerrini (Italy), Nardo Nardocci (Italy)

Barcelona

CHAIRMEN

Alexis ARZIMANOGLOU (France)
Jaume CAMPISTOL (Spain)
Emilio FERNANDEZ-ALVAREZ (Spain)
Renzo GUERRINI (Italy)
Nardo NARDOCCI (Italy)

CONGRESS SECRETARIAT

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MANY THANKS TO OUR PARTNERS FOR THEIR CONTINUING SUPPORT

SJD Education Sant Joan de Déu

International Parkinson and Movement Disorder Society

PTC THERAPEUTICS Translating Science - Transforming Lives

BIOCODEX

ABSTRACTS WILL BE AVAILABLE ONLINE:

www.paedmovdissymposium.com

CONGRESS VENUE

Auditorio Hospital Sant Joan de Déu
Aula Pediatria
C/Santa Rosa, 39
080950 Esplugues de Llobregat
Barcelona

WIFI NETWORK : PUBLIC
No Password

ACCESS

From the airport:
15 min by taxi
Taxi Groc: +34933222222
Radio Taxi Miramar: +34933033033
50 min by public transportation
Bus line A1 - Stop «Plaça Espanya»
Subway Line L3 - Stop «Zona Universitaria»
Bus Line JM to «PL Montfalcone» - Stop «Pg. Sant Joan de Dèu - Santa Rosa»

From the train station «Barcelona Sants»
15 min by taxi
30 min by public transportation
Subway Line L3 from «Plaça del Centre» near the train station - Stop «Zona Universitaria»
Bus Line JM to «PL Montfalcone» - Stop «Pg. Sant Joan de Dèu - Santa Rosa»

OPENING HOURS

Congress Registration desk - Level 0
February 6th, 2020 - from 09.00 to 20.45
February 7th, 2020 - from 07.30 to 18.15

CATERING - Level 1

Lunches & coffee breaks are included in your registration and will be held near the plenary session room & posters exhibition.

POSTERS - Level 1

Posters will be listed by abstract number. Fixing supplies will be handed out to you onsite at the symposium welcome desk.

February 6th, 2020

10h00 –11h30	**Session 1: Background-1**
11h30 –12h00	Coffee Break
12h00 –13h30	**Session 2: Background-2**
13h30 – 15h00	Lunch & Posters
15h00 – 16h00	**Video session-1**
16h00 – 17h30	**Session 3: Clinical spectrum- 1**
17h30 – 18h00	Coffee break
18h00 – 19h45	**Session 4: Clinical spectrum- 2**
19h45 – 20h45	**Video session-2**

February 7th, 2020

07h45 – 08h45	PTC Therapeutics Industry-sponsored breakfast symposium
08h45 – 10h15	**Session 5: Clinical spectrum- 3**
10h15 – 10h45	Coffee Break
10h45 – 12h15	**Session 6: Clinical spectrum- 4**
12h15 – 13h15	**Video session-3**
13h15 – 14h45	Lunch & Posters
14h45 – 17h15	**Session 7: Therapeutics**
17h15 – 18h15	**Video session-4**
18h15	**Closing ceremony**

Session 1: Background-1 *Chair: Birgit Assmann (Germany)*	
10h00 – 10h30	The hunting of causative genes *Manju Kurian (London, UK)*
10h30 – 11h00	Co-occurrence of movement disorders and epilepsy: an historical overview - *Renzo Guerrini (Firenze, Italy)*
11h00 – 11h30	Discussion
11h30 –12h00	Coffee Break
Session 2: Background-2 *Chair: Mohammad Shekeeb (Australia)*	
12h00 – 12h30	Basic cellular mechanisms involved in movement disorders and epilepsy *Jonathan Mink (Rochester, USA)*
12h30 – 13h00	Pathways involved in the co-occurrence of movement disorders and epilepsy *Philippe Kahane (Grenoble, France)*
13h00 – 13h30	Discussion
13h30 – 15h00	Lunch & Posters
15h00 – 16h00	Video session-1 *Chair : José Obeso (Spain)*
15H00 - 15H10	Epilepsy, dyskinesia and ASD in an infant with probably ALG1 mutation (CDG-Ik) – a case presentation – *Mihaela Vintan (Romania)*
15h10 – 15h20	Co-occurence of epilepsy and paroxysmal dyskinesia - case report – *Galina Stevanovic (Serbia)*
15h20 – 15h30	NCAM2 deletion in a boy with neurodevelopmental disorder, epilepsy and subtle movement disorder – *Dina Amrom (Luxembourg)*

15h30 – 15h40	Epilepsy and hyperkinetic movement disorders in a patient with Williams-Beuren syndrome *Mario Mastrangelo (Italy)*
15h40 – 15h50	Acyl-CoA-binding domain-containing protein 6 (ACBD6) loss of function leads to GSMDE *Rauan Kaiyrzhanov (UK)*
15h50 – 16h00	Long-term follow-up of two siblings with Succinic Semialdehyde Dehydrogenase Deficiency *Anna Commone (Italy)*
Session 3: Clinical spectrum- 1 *Chair: Mostafa Awadh (Egypt)*	
16h00 – 16h30	Complex hyperkinetic movement disorders as a core feature of GSMDE. ADCY5, GNAO1 and PDE10A related diseases - *Miryam Carecchio (Padova, Italy)*
16h30 – 17h00	TBC1D24 related disorders *Renzo Guerrini (Firenze, Italy)*
17h00 – 17h30	Discussion
17h30 - 18h00	Coffee break
Session 4: Clinical spectrum- 2 *Chair: Monica Troncoso (Chile)*	
18h00 – 18h45	Movement disorders associated with different genetic, developmental and epileptic encephalopathies. FOXG1 and STXBP1 related diseases – *Ingrid Scheffer (Melbourne, Australia)*
18h45 – 19h15	GSMDE due to loss- of-function mutations. FRRS1L and other genetic related diseases *Michael Kruer (Phoenix, USA)*
19h15 – 19h45	Discussion

19h45 – 20h45	**Video session-2** *Chair: Warren Marks (USA)*
19h45 – 19h55	Successful treatment of refractory chorea in a patient with a common gain-of-function GNAO1 variant by folinic acid – *Ching Wan Lam (China)*
19h55 – 20h05	Ocular movements and other visual function in children with GNAO1 Syndrome *Domenica Battaglia (Italy)*
20h05 – 20h15	Sandhoff disease & sensory trick: when myoclonus and dystonia meet at the cortical-subcortical boundary – *Giorgia Olivieri (Italy)*
20h15 – 20h25	Expanding the spectrum of Segawa syndrome: more than dopa-responsive dystonia *Sara Vila Bedmar (Spain)*
20h25 – 20h35	Ataxia, verbal apraxia and late onset myoclonia: a long journey and a still mysterious diagnosis *Marine Jequier Gygax (Switzerland)*

PTC Therapeutics Industry-sponsored breakfast symposium
Beyond movement disorders and epilepsy
Understanding AADC deficiency

7h45 - 7h50	Welcome and introduction *Prof. Angeles Garcia Cazorla (Spain)*
7h50 - 8h05	The basics of AADC deficiency: Genetics and symptoms - *Dr. Nastassja Himmelreich (Germany)*
8h05 - 8h20	A challenging clinical case: Differential diagnosis of AADC deficiency *Prof. Angeles Garcia Cazorla (Spain)*
8h20 - 8h35	Managing AADC deficiency: Current options and future outlook - *Prof. Bruria Ben-Zeev (Israel)*
8h35 - 8h45	Q&A session - *Prof. Angeles Garcia Cazorla (Spain)*

Session 5: Clinical spectrum- 3
Chair: Carlotta Canavese (Italy)

8h45 – 9h15	When the movement disorder is the dominant feature in GSMDE – *Diane Doummar (Paris, France)*
9h15 – 9h45	When epilepsy is the dominant feature in GSMDE - *Ingrid Scheffer (Melbourne, Australia)*
9h45 – 10h15	Discussion
10h15 – 10h45	Coffee Break

Session 6: Clinical spectrum- 4
Chair: Wang-Tso Lee (Taiwan)

10h45 – 11h15	When neurodevelopmental deficit is in the frontline in GSMDE – *Vincenzo Leuzzi (Rome, Italy)*
11h15 – 11h45	GSMDE with parkinsonism as main sign *Roser Pons (Athens, Greece)*

11h45 – 12h15	Discussion
12h15 – 13h15	**Video session-3** *Chair: Toni Pearson (USA)*
12h15 – 12h25	Diagnostic journey of patient with paroxysmal jerks and seizures: video-presentation *Vera Fominykh (Russia)*
12h25 – 12h35	A girl with a de novo heterozygous mutation in NALCN gene and pyridoxine-dependent seizures and movement disorder – *Eve Õiglane-Šlik (Estonia)*
12h35 – 12h45	Expanding the clinical phenotype associated with KCNC1 – related disorders *Patricia Lipari Pinto (Portugal)*
12h45 – 12h55	Phenotypic spectrum of POLR1C leukodystrophy – *Susana Roldán (Spain)*
12h55 – 13h05	Dyskinesia and pharmacorefractory epilepsy in a child with a homozygous PCDH12 mutation *Anne Koy (Germany)*
13h05 – 13h15	Hyperkinesis associated with dystonia responds to pallidal deep brain stimulation *Warren Marks (United States)*
13h15 - 14h45	Lunch & Posters
Session 7: Therapeutics *Chair: Jaume Campistol (Spain)*	
14h45 – 15h15	Medical treatment of the epilepsy in GSMDE *Alexis Arzimanoglou (Lyon, France)*
15h15 – 15h45	Medical Treatment of the movement disorder - *Giovanna Zorzi (Milano, Italy)*

15h45 – 16h15	The place of deep brain stimulation in GSMDE – *Jean-Pierre Lin (London, UK)*
16h15 – 16h45	Innovative treatment perspectives *Manju Kurian (London, UK)*
16h45 – 17h15	Discussion
17h15 – 18h15	**Video session-4** *Chair: Eleni Panagiotakaki (France)*
17h15 – 17h25	ADCY5-related movement disorder with paroxysmal events-video case report from infancy to adolescence – *Oliver Maier (Switzerland)*
17h25 – 17h35	KGD4 biallelic variants in two siblings with bilateral striatal necrosis: a new gene of Krebs cycle associated with Leigh syndrome *Serena Galosi (Italy)*
17h35 – 17h45	The GRIA3 c.2477G>A variant causes a new and distinctive phenotype of early-onset multifocal myoclonus, generalized chorea and exaggerated startle reflex – *Juliette Piard (France)*
17h45 – 17h55	Episodic axial hyperextension – a case of GRIN2B Encephalopathy – *Ralf Eberhard (Switzerland)*
17h55 – 18h05	Non-paroxysmal movement disorders in patients with Alternating Hemiplegia of Childhood: “soft” and “stiff” – *Eleni Panagiotakaki (France)*
18h05 – 18h15	Atypical presentation of Subacute Sclerosing Panencephalitis: periodic hyperkinetic movements and EEG as key features in case of with unusual MRI – *Leticia Pias-Peleteiro (Spain)*
18h15	**Closing ceremony**

P1 - Epilepsy patients with TSC gene mutation: clinical and scalp EEG feature analysis - *HE Jing (China)*

P2 - A novel mutation of GRIN1 in a patient with hyperkinetic movements, infantile spasm and cortical visual impairment - *YOUNG OK Kim (Republic of Korea)*

P3 - Expanding the phenotype of chromosome 4 duplication with dystonia and central non-ictal apnoea. - *CHENG Elliott (United Kingdom)*

P4 - Successful treatment of recurrent hemiparesis caused by a heterozygote ATP1A3 variant with oxcarbazepine - *NASRELDIEN Ali (United Arab Emirates)*

P5 - RHOBTB2 variant in a case of epileptic encephalopathy associated to paroxysmal movement disorder - *SOLIANI Luca (Italy)*

P6 - A distinctive SCN1A-related phenotype: multiple contractures, early onset epileptic encephalopathy and hyperkinetic movement disorder - *CAMACHO Ana (Spain)*

P7 - Movement Disorders and Epilepsy in Four Forms of Neuronal Ceroid Lipofuscinosis (NCL) - *MINK Jonathan (USA)*

P8 - The case of severe infantile epileptic encephalopathy caused by bi-allelic mutation in the UGDH-gene. - *APANASENKO Olena (Ukraine)*

P9 - KCNH1 related syndrome presenting with oculomotor apraxia and ataxia: a case report - *CANAVESE Carlotta (Italy)*

P10 - Recognizing the phenotype of "GNAO1-associated encephalopathy": when to suspect this genetic condition - *VILA BEDMAR Sara (Spain)*

P11 - Early infantile SCN1A epileptic encephalopathy: a combination of epileptic encephalopathy and hyperkinetic movement disorder - *SOLIANI Luca (Italy)*

P12 - Long term survival in a patient with GF1M gene mutation - *SOLIANI Luca (Italy)*

P13 - Pantothenate Kinase-Associated Neurodegeneration (PKAN) In Saudi: A Phenotypic Characterization and Novel Mutations - *ALDOSSARY Hussein (Saudi Arabia)*

P14 - Movement disorders in early onset epileptic encephalopathies: a comprehensive literature review - *MARTINEZ-ESTEVE MELNIKOVA Anastasia (Spain)*

P15 - Biallelic variants in CSTB cause a developmental and epileptic encephalopathy with dyskinesia - *CALAME Daniel (USA)*

P16 - GNAO1-assocoated movement disorders in Hong Kong: a case series of 5 families - *LAM Ching Wan (China)*

P17 - PEDiDBS 2020: Update on the international registry of pediatric patients undergoing deep brain stimulation - *MARKS Warren (USA)*

P18 - Expanding the phenotypic spectrum of FOXG1 syndrome - *WONG Leechin (Taiwan)*

P19 - Eye movement disorders in children with epilepsy: study from one University Hospital in Taiwan - *LEE Wang-Tso (Taiwan)*

P20 - Action myoclonus and mental deficiency in a patient with a SCN8A splicing mutation - *CANAFOGLIA Laura (Italy)*

P21 - Genetic Syndromes with Movement Disorders and Epilepsy: a single-centre experience - *SOLAZZI Roberta (Italy)*

P22 - Movement disorders as a leading clinical symptoms in a patient with a RHOBTB2 mutation described in epileptic encephalopathy - *POPOVICH Sophia (Russia)*

P23 - A case of Early-Infantile Epileptic Encephalopathy associated with compound heterozygosity for mutations in TBC1D24 - *MARCOTULLI Daniele (Italy)*

P24 - PLA2G6, a phenotypic continuum with major clinical implications - *BUSTUCHINA VLAICU Mihaela (France)*

P25 - Phenotypic Characteristics of a Mutation in the ADP-Ribosylation Factor Guanine Nucleotide-Exchange Factor 2 (ARFGEF2): A Report of Two Siblings From a Saudi Family - *ALMAIRI Mohammed (Saudi Arabia)*

P26 - De novo deletion of CTNND2 presenting as progressive hyperkinetic movement disorder - *DI MARZIO Giulia Maria (Italy)*

P27 - Combination of chelation therapy and low-manganese diet in a patient with SLC39A14-associated hypermanganesemia and dystonia - *HEIM Jennifer (USA)*

P28 - A case of DYT1 dystonia initially misdiagnosed as focal epilepsy - *HANNA Jang (Republic of Korea)*

P29 - GRIN related disorders - an expanding group with epilepsy and movement disorders with a personalized therapy strategy - *LIPARI PINTO Patrícia (Portugal)*

P30 - ATP1A3-related epilepsy: report of seven cases and literature-based analysis of treatment response - *VON STÜLPNAGEL Celina (Germany)*

P31 - The expanded phenotypic spectrum of BRAT1 gene mutations: a new compound heterozygous case - *BAGLIONI Valentina (Italy)*

P32 - A Rare Case of Infantile Spasm: An Unexpected Mutation - *ALDOSSARY Hussein (Saudi Arabia)*

P33 - Bilateral striatal lesions, coreoathetosis and epilepsy: possible association with a PDE10A variant - *TORRES Maria Angeles (United Kingdom)*

P34 - Hyperekplexia in patient with GNAO1-related syndrome. - *ZAWADZKA Marta (Poland)*

P35 - Movement Disorder Childhood Rating Scale for clinical management and follow up in FOXG1 related syndrome - *SCALISE Roberta (Italy)*

P36 - Bardet–Biedl syndrome with Seizures: A Novel Mutation Report - *ALDOSSARY Hussein (Saudi Arabia)*

P37 - Movement disorders in PIGA-related DEE - *TRIVISANO Marina (Italy)*

P38 - Neuropsychological evaluation in GNAO1 encephalopathy using eye tracker - *GRAZIOLA Frederica (Italy)*

BEYOND MOVEMENT DISORDERS AND EPILEPSY

Understanding AADC deficiency

PTC Therapeutics Symposium at the 1st International Symposium on Genetic Syndromes with Movement Disorders and Epilepsy (GSMDE)

Friday, 7 February 2020, 7:45 – 8:45 AM

Auditorio Hospital Sant Joan de Déu, Barcelona, Spain

It is our great pleasure to invite you to the symposium "Beyond movement disorders and epilepsy – Understanding AADC deficiency"

Join our expert faculty in a peer-to-peer interactive discussion and share your point of view on the challenges associated with the differential diagnosis and management of aromatic l-amino acid decarboxylase (AADC) deficiency.

Expert faculty members

Prof Angeles García Cazorla
Neurometabolic Diseases Unit
Neurology Department
Hospital Sant Joan de Déu,
Barcelona, Spain

Dr Nastasja Himmelreich
Center for Child and
Adolescent Medicine
University of Heidelberg,
Germany

Prof Bruria Ben-Zeev
Paediatric Neurology Dept
The Edmond and Lily Safra
Children's Hospital
Sheba Medical Center, Israel

Symposium organised and funded by PTC Therapeutics

MED-ES-AADC-2000022 January 2020