



DIANALUND INTERNATIONAL CONFERENCE on EPILEPSY

FOCUS ON PROGRESSIVE MYOCLONUS EPILEPSIES

14-15 April, 2016

Sorø (Denmark)

Progressive myoclonus epilepsies (PME) encompass a wide spectrum of clinically and genetically heterogeneous epileptic conditions, whose core features are action myoclonus, generalized seizures and progressive neurological decline. Clinical diagnosis of specific forms of PME can be challenging, particularly at disease onset, because of phenotypic similarities and overlap of symptoms with other epileptic and degenerative diseases. Recent advances in molecular genetics have helped to diagnose several forms of PME and to achieve a better understanding of the different disorders that cause them. In addition, in recent years new types of PME have been described and genetically characterized, identifying new genes, such as for instance SCARB2, GOSR2, ASAH, KCNC1 whose mutations can cause a PME. The main aims of this conference are to provide an updated overview of the currently recognized forms of PMEs, to illustrate their genetic background, with a particular focus on genotype-phenotype correlations, and to discuss the proper diagnostic process and therapeutic approaches.

Scientific Committee

Guido Rubboli

Helle Hjalgrim

Rikke Møller

Elena Gardella

Anna-Elina Lehesjoki

Organizing Committee

Alice Bøjlund Lyseen

Margarethe Kölmel

Faculty

S. Berkovic (Australia), L Canafoglia (Italy), L Dibbens (Australia), S Franceschetti (Italy), E Gardella (Denmark), P Genton (France), H Hjalgrim (Denmark), R Kalviainen (Finland), D Kasteleijn-Nolst Trenite' (The Netherlands); A-E Lehesjoki (Finland), H Lerche (Germany), E. Mervaala (Finland), R Michelucci (Italy), S Mole (UK), M Muona (Finland), R Møller (Denmark), M Nikanorova (Denmark), K Oliver (Australia), G Rubboli (Denmark), J Serratosa (Spain), CA Tassinari (Italy).

14 April 2016

8:45- 9:00 Presentation of the conference: *H Hjalgrim (Denmark), G Rubboli (Denmark)*

Clinical aspects and diagnosis Chairs: *G Rubboli (Denmark), S Beniczky (Denmark)*

9:00 - 9:45 **Lecture:** Clinical approach to PME: Historical overview and state of the art in 2016 *S Berkovic (Australia)*

9:45 - 10:15 Video session on clinical features *E Gardella (Denmark)*

10:15 - 10:45 The neurophysiology of PME: EEG and polygraphic findings
G Rubboli (Denmark)

10:45-11:00: *Coffee break*

11:00-11:30 Action myoclonus: clinical and neurophysiological features
CA Tassinari (Italy)

11:30-12:00 Cortical excitability in PMEs *E Mervaala (Finland)*

12:00-12:30 Photosensitivity in PME *D Kasteleijn Nolst-Trenite' (The Netherlands)*

12:30-13:30 *Lunch*

14 April 2016 cont.

Genetics and genotype-phenotype correlations Chairs: *R Møller (Denmark), J Serratos (Spain)*

13:30-14:15- **Lecture:** Progress on genetics of PME and implications for new treatments
A-E Lehesjoki, (Finland)

14:15-14:45 Large scale genetics studies in PMEs *M Muona (Finland)*

14:45-15:15 - Unverricht-Lundborg disease *R Kalviainen (Finland)*

15:15-15:45 - Myoclonus epilepsy and ataxia due to potassium channel mutation
K Oliver (Australia)

15:45-16:00 *Coffee break*

16:00-16:30 Sialidoses *S Franceschetti (Italy)*

16:30-17:00 The spectrum of PMEs associated with CLN6 mutations
L Canafoglia (Italy)

17:00- 17:30 Neuronal ceroid lipofuscinosis: genotype-phenotype correlations
S Mole (UK)

17:30-18:00 General Discussion - Chairs: *S Berkovic (Australia), A-E Lehesjoki E (Finland)*

15 April 2016

Genetics and genotype-phenotype correlations Chairs: *H Hjalgrim (Denmark),
R Kalviainen (Finland)*

8:30-9:00 Lafora disease *J Serratosa (Spain)*

9:00- 9:30 PME phenotypes associated with SCARB2 mutations *L Dibbens (Australia)*

9:30-10:00 PME caused by GOSR2 mutations *R Møller/ H Hjalgrim (Denmark)*

10:00-10:30 Other rare forms of PMEs *P Genton (France)*

10:30-10:45 *Coffee break*

Update on treatment of PMEs Chairs: *P Genton (France), R Michelucci (Italy)*

10:45-11:15 Medical treatment *R Michelucci (Italy)*

11:15-11:45 Other treatments, (i.e. diets, VNS, DBS) *M Nikanorova (Denmark)*

11:45-12:30 **Lecture:** Precision medicine in genetic epilepsies *H Lerche (Germany)*

12:30-13:00 General Discussion and end of the meeting - All chairs

GENERAL INFORMATION

The venue of the conference is Comwell Hotel which is located in Abildvej 100, Sorø, Denmark. The conference will start on April 14th, 2016 at 8:45 and will end on April 15th, 2016 at 13:00.

Comwell Hotel is about 4 km from Sorø railway station. Sorø can be reached by train from Copenhagen (about 50 minutes). Sorø is also directly connected by train to nearby cities, Slagelse (about 15 minutes) and Ringsted (about 15 minutes). Public bus is also available.

Highway E20 connects Sorø to Copenhagen (about 1 hour drive), Slagelse (about 20 minutes drive), Ringsted (about 20 minutes drive).

A limited number of rooms are available for the participants at a discounted price at Comwell Hotel. For reservation contact Mette Anker Hansen e-mail: mette.hansen@comwell.dk, specifying that the reservation is in connection with the conference.

Registration fee: 150 euro (1100 DKK) to be paid on bank account:
Danske Bank

Address: Torvet 6, 4100 Ringsted (Denmark)

Account n. 4343 0006406319 IBAN: DK45 3000 0006 4063 19 SWIFT-BIC: DABADKKK.

Please report in the payment your name and the title of the conference.

Please send the filled registration form to Alice Bøjlund Lyseen: ally@filadelfia.dk

Further information, registration form and the program are be available at the website:

<http://www.filadelfia.dk/filadelfia/aktuelt/nyheder/2015/12/10/conference-on-epilepsy#Information>

