FILADELFIA

6th Dianalund International Conference on Epilepsy

Overlapping clinical phenotypes in monogenic epilepsies – common molecular pathways?

EpiCARE

UNIVERSITY OF COPENHAGEN

SDU University of Southern Denmark

DANSK EPILEPSI SELSKAB Danish Epilepsy Society

IBE International Bureau for Epilepsy

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REGISTRATION:

conferencemanager.dk/dice2024

MAY 02 - 03, 2024

Køge, Denmark

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Dear participants

It is our great pleasure to invite you to the 6th Dianalund International Conference on Epilepsy. The topic of the conference is:

"**Overlapping clinical phenotypes in monogenic epilepsies – common molecular pathways?"**

We hope that you'll be able to join us, and we are looking forward welcoming you to Denmark.

Guido Rubboli

Elena Gardella

Rikke S. Møller

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Introduction

Amongst many genetic epilepsies and epileptic encephalopathies, the phenotypic spectrum can be broad (even in patients with identical genetic alterations) and genetic modifiers are typically implicated. Despite their different genetic etiologies, patients can have seemingly similar clinical presentations. For example, SCN1A-related disorders can clinically overlap with other genetic diseases such as those which are GABAA-receptor-related. Then there are various comorbidities – for instance, movement disorders – which can also have features which overlap with those of other conditions. It has been suggested that the comorbidities of distinct phenotypes may reflect an overlap of both the causative genes and the involvement of similar molecular processes for these disorders. It is clear that the study paradigms required to successfully address these questions are lacking and therefore require further research.

At the conference

We shall review current knowledge on the phenotypic expressions, their overlapping features, and the genotype-phenotype correlations of some epileptic disorders and epileptic encephalopathies. With an aim to guide the discovery and development of effective targeted treatments, we shall discuss possible underlying and shared pathophysiological mechanisms and molecular substrates and then present both the emerging concepts in the field of precision medicine and the yields of the most advanced research strategies.

The conference will conclude with evidence and case presentations from the audience which, based on the understanding of underlying genetic anomalies, support the role of novel treatments.

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Agenda

Wednesday 1 May

19:00 - 21:00 Welcome reception

Thursday 2 May

Session 1 **Overlapping clinical phenotypes – common molecular pathways?**

09:00 - 09:25 **Andreas Brunklaus** (Glasgow): The spectrum of gain of function SCN1A disorders

09:25 - 09:50 **Elena Gardella** (Dianalund): Sodium channelopathies: clinical commonalities and differences

09:50 - 10:15 **Sebastian Ortiz** (Dianalund): Distinct clinical phenotypes associated with LOF vs GOF GABAA-receptor variants

Break Coffee break

Session 1 **- continued**

10:45 - 11:10 **Carla Marini** (Ancona): PRRT2 variants in self-limiting epilepsy, paroxysmal dyskinesia, and hemiplegic migraine

11:10 - 11:35 **Steffen Syrbe** (Heidelberg): The spectrum of CACNA1A-related disorders

11:35 - 12:00 **Robert Lauerer-Braun** (Tübingen): CACNA1Evariants in Developmental and Epileptic Encephalopathy with contractures, macrocephaly, and dyskinesias

12:00 - 12:25 **Johannes Lemke** (Leipzig) GRIN-related disorders:diversification of inheritance pattern, phenotypic spectrum, and treatment approaches

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Lunch

12:25 - 13:30 Lunch

Session 1 - continued

13:20 - 14:05 **Kevin Bender (San Francisco) & Stephan Sanders** (Oxford): Similar clinical phenotypes - shared genetic mechanisms?

14:05 - 14:30 **Panel discussion**

Break

14:30 - 14:45 Coffee break

Session 2: Clinical relevance of EEG biomarkers in monogenic epilepsies

14:45 - 15:15 **Roberta Cilio** (Brussels): EEG biomarkers in neonatal onset epilepsies

15:15 - 15:40 **Guido Rubboli** (Dianalund): Any yields from EEG in distinguishing LOF from GOF diseases?

15:40 - 15:55 **Alberto Cossu** (Verona): Quantitative EEG biomarkers for STXBP1-related disorders

15:55 - 16:25 The usefulness of EEG biomarkers in clinical trials - **TBA**

16:25 - 17:00 **Panel discussion**

Break

17:00 - 18:00 Coffee break and poster session

Session 3: Late Breaking News

18:00 - 19:00 Late Breaking News

Evening

19:45 Dinner

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Friday 3 May

Session 4: Precision Medicine in Genetic Epilepsies: Concepts and Research Strategies

08:30 - 09:00	**Rikke Møller** (Dianalund): Precision medicine in genetic epilepsies
09:00 - 09:30	**Maurizio Taglialatela** (Naples): How can drug repurposing inform us of dysfunctional mechanisms? Yields and challenges
09:30 - 10:00	**Snezana Maljevic** (Melbourne): Leveraging iPSC-derived disease models to propel precision medicine treatments for Developmental and Epileptic Encephalopathy
10:00 - 10:30	**Massimo Mantegazza** (Nice): Experimental models of genetic epilepsy in a precision medicine framework
10:30 - 10:45	**Panel discussion**

Break

10:45 - 11:15	Coffee break

Session 5: Molecular therapeutic Board

Chair: Holger Lerche

11:15 - 13:15	Case presentations of precision medicine in genetic epilepsies
13:15 - 13:45	**Matthew C. Walker** (London): Therapies of the future
13:45 - 14:00	Concluding remarks

Farewell

14:00	Lunch

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SOLD OUT!!

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