



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

Secretariats: Line Overgaard, Torie Robinson

REGISTRATION:

Open from Jan 12, 2024

MAY 02 - 03, 2024

Køge, Denmark

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, UK): The spectrum of gain of function SCN1A disorders

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

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

Break

10:15 - 10:45 Coffee break

Session 1 - continued

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

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

11:35 - 12:00 **Robert Lauerer-Braun** (Tübingen, GER): CACNA1E variants in Developmental and Epileptic Encephalopathy with contractures, macrocephaly, and dyskinesias

12:00 - 12:25 **Johannes Lemke** (Leipzig, GER) 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

13:30 - 14:15 **Kevin Bender** (San Francisco, USA) & **Stephan Sanders** (Oxford, UK): Similar clinical phenotypes - shared genetic mechanisms?

14:15 - 14:35 **Panel discussion**

Break

14:35 - 15:00 Coffee break

Session 2: Clinical relevance of EEG biomarkers in monogenic epilepsies

15:00 - 15:25 **Roberta Cilio** (Brussels, BE): EEG biomarkers in neonatal onset epilepsies

15:25 - 15:50 **Guido Rubboli** (Dianalund, DK): EEG phenotyping: still a valuable tool for genotype-phenotype correlations?

15:50 - 16:15 **Alberto Cossu** (Verona, IT): Quantitative EEG biomarkers for STXBP1-related disorders

16:15 - 16:40 **Stéphane Auvin** (Paris, FR): The usefulness of EEG biomarkers in clinical trials

16:40 - 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 - 08:55 **Rikke Møller** (Dianalund, DK): Precision medicine in genetic epilepsies
- 08:55 - 09:20 **Maurizio Tagliatela** (Naples, IT): How can drug repurposing inform us of dysfunctional mechanisms? Yields and challenges
- 09:20 - 09:45 **Snezana Maljevic** (Melbourne, AUS): Leveraging iPSC-derived disease models to propel precision medicine treatments for Developmental and Epileptic Encephalopathy
- 09:45 - 10:10 **Massimo Mantegazza** (Nice, FR): The role of mouse models of genetic epilepsies in precision medicine
- 10:10 - 10:35 **Panel discussion**

Break

- 10:35 - 11:00 Coffee break

Session 5: Molecular therapeutic Board

- 11:00 - 13:00 Case presentations of precision medicine in genetic epilepsies
- 13:00 - 13:30 **Matthew C. Walker** (London, UK): Therapies of the future
- 13:30 - 13:45 Concluding remarks

Farewell

- 13:45 Farewell lunch

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