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6th Dianalund International Conference on Epilepsy

Overlapping clinical phenotypes in monogenic epilepsies – common molecular pathways?

UNIVERSITY OF COPENHAGEN

SDU University of Southern Denmark

DANSK EPILEPSI SELSKAB Danish Epilepsy Society

EpiCARE

ILAE International League Against Epilepsy

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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 Møller

Secretariats: Line Overgaard, Torie Robinson

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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- 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

16:30 - 19:00 Pre-conference meeting

19:30 - 21:00 Welcome reception

Thursday 2 May

08:45 - 09:00 **Introduction to conference**

Session 1 **Overlapping clinical phenotypes – common molecular pathways?**
Chairs: Carla Marini, Reetta Kälviäinen

09:00 - 09:25 **Andreas Brunklaus** (Glasgow, UK): The extended spectrum of *SCN1A*-related disorders: how does function relate to the clinic?

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 

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Thursday 2 May (continued)

Session 1 **- continued**
Chairs: Roberta Cilio, Rikke Møller

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

Lunch

12:25 - 13:30 Lunch

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**

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Thursday 2 May (continued)

Break

14:35 - 15:00 Coffee break

Session 2: Clinical relevance of EEG biomarkers in monogenic epilepsies

Chairs: Eleni Panagiotakaki, Elena Gardella

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, FRA): The usefulness of EEG biomarkers in clinical trials

16:40 - 17:00 **Panel discussion**

Break

17:00 - 18:00 Coffee break & Poster Session

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Thursday 2 May (continued)

Session 3: Late Breaking News
Chairs: Kevin Bender, Dick Lindhout

18:00 - 19:00	Late Breaking News
18:00 - 18:15	**Lorenz Kiwull** (Salzburg, AUT): Overlapping molecular pathways – common clinical phenotypes? Standard procedure for common data elements in groups of rare diseases using the example of *SYNGAP1* and classical RASopathies
18:15 - 18:30	**Philip K. Ahring** (Sydney, AUS): Can paralogous epilepsy-associated GABAA receptor variants be used as indicators for clinical outcomes?
18:30 - 18:45	**Christopher Reid** (Melbourne, AUS): Enhanced inhibition in hippocampal pyramidal neurons in a gain-of-function *GABRB3* mouse model of epilepsy
18:45 - 19:00	**Jean-Francois Perrier** (Copenhagen, DK): Understanding and treating *STXBP1* neurodevelopmental encephalopathy

Evening

19:45 **Conference dinner**

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Friday 3 May

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

Chairs: Mary Chebib, Christopher Reid

08:30 - 08:55	**Rikke Møller** (Dianalund, DK): Precision medicine in genetic epilepsies
08:55 - 09:20	**Maurizio Taglialatela** (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, FRA): The role of mouse models of genetic epilepsies in precision medicine
10:10 - 10:20	**Rami Aqeilan** (Jerusalem, ISR): Modelling and Characterizing WOREE Syndrome: from Basic Science to Translational Medicine
10:20 - 10:45	**Panel discussion**

Break

10:45 - 11:10 Coffee break 

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Friday 3 May (continued)

Session 5: **Molecular therapeutic Board**

Chairs: Holger Lerche, Guido Rubboli, Stéphane Auvin

11:10 - 13:10 **Case presentations of precision medicine in genetic epilepsies**

11:10 - 11:25 **Matthias De Wachter** (Antwerp/Dianalund, BEL/DK): *KCNA2*-GOF and GOF-LOF Developmental and Epileptic Encephalopathy: an update on treatment response of 4-Aminopyridine

11:25 - 11:40 **Illona Krey** (Leipzig/Atlanta, GER/USA): Precision medicine approaches in GRIN-related disorders – what do we know and what's next?

11:40 - 11:55 **Pierandrea Muglia** (Brussels, BEL): Radiprodil, a NR2B-NMDA negative allosteric modulator in clinical development for GRIN related disorders

11:55 - 12:10 **Cathrine Gjerulfsen** (Dianalund, DK): Cenobamate as add-on treatment in SCN8A related developmental and epileptic encephalopathy

12:10 - 12:25 **Laia Nou Fontanet** (Barcelona, SP): *AFG2A*-related Encephalopathy: Clinical phenotype and ketogenic diet effect (in vivo and in vitro effect)

12:25 - 12:40 **Vivian Liao** (Sydney, AUS): Vinpocetine improves epilepsy and comorbidity outcomes in patients with GABAA receptor loss-of-function variants

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Friday 3 May (continued)

Session 5 - **continued**

12:40 - 12:50 **Steve Petrou** (Boston, USA): Revolutionizing Treatment for Early-Onset Developmental and Epileptic Encephalopathy: A First Look at ASO Therapy for *SCN2A*

12:50 - 13:00 **Evelina Carapancea** (Brussels, BEL): Everolimus precision therapy in a patient with NPRL3-related epilepsy

13:00 - 13:10 **Ida Cursio** (Ancona, IT): Generalized epilepsy, intellectual disability, behavioral disorder and familiar tremor due to compound heterozigosity of the *CAD* gene

13:10 - 13:45 **Matthew Walker** (London, UK): Therapies of the future

13:45 - 14:00 **Concluding remarks**
Chairs: Guido Rubboli, Elena Gardella, Rikke Møller

Farewell

14:00 Farewell lunch

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WELCOME

Location & directions

Join us at Comwell Køge Strand

Ways to get there (approx. 45 km from Copenhagen)

Train
Travel to **Køge** (***not*** Køge Nord!) **train station** from which the venue is a 20 minute walk or 10 minute bus ride

Road
Via highway E47 and E20

Register via www.conferencemanager.dk/dice2024

Accommodation
Available for participants at Comwell Køge Strand and can be included in the registration.

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