



Danish Epilepsy Centre



Comwell

**4th Dianalund International Conference on Epilepsy  
14-15th May, 2020 - Comwell, Korsør (Denmark)**

**Precision medicine in genetic epilepsies– where are we  
now, and where are we heading?**

Treatment of epilepsy remains largely empirical, and individual prescribing based on the mechanism of action is generally not possible. However, recent findings in genetic epilepsies have elucidated some mechanisms of epileptogenesis, unraveling the role of a number of genes with different functions, such as ion channels, proteins associated to the vesical synaptic cycle or involved in energy metabolism. The advent of Next Generation Sequencing is providing precision genetics enabling precision medicine in approximately one quarter of patients, illustrating the enormous utility of genetic testing for therapeutic decision-making. Although any patient with refractory epilepsy may benefit from genetic screening, such testing will be of most importance in patients with early-onset seizures (less than 3 years of age), a family history of seizures, associated neurological deficit, or learning intellectual disability. A major goal of the genetic studies is the identification of novel drug targets and tailored therapies based on the cause of disease. The discovery of specific genetic mutations has also helped us to repurpose drugs with specific actions which may have been used in entirely unrelated conditions. In this conference, clinicians, geneticists and basic scientists aim to provide an updated overview of the state-of-the art of precision medicine in those genetic epilepsies in which a precision medicine approach has been already implemented, or in which promising data are under evaluation. Within the next future, precision medicine will hopefully move within the reach of more patients, and as genetic technologies advance, a comprehensive approach informed also of the contribution of genetics in treatment choices will become an increasingly important part of the clinical management of the epilepsy patients.

**Preliminary Program**

- Precision medicine in the epilepsy clinic, state of the art 2020
- An old drug for a new indication: Repurposing Fenfluramine from anorexigen to AED
- *SCN2A*: From genotype-phenotype correlations to personalized treatment
- *SCN8A*: current and emerging treatments
- Predicting Functional Effects of Missense Variants in Voltage-Gated Sodium and Calcium Channels
- Clinical overview and treatment responsiveness in *KCNT1*-Related Epilepsy
- ASO therapies in genetic epilepsies: insights from *SCN2A* and *KCNT1*
- Targeted treatment for *KCNQ2* encephalopathy: recent developments and pitfalls
- Repurposing of drugs: insights from *KCNA2*
- Cellular models for drug screening - what can we learn?
- Precision Medicine -Lessons from Early N of 1 Experiences
- GRIN-related disorders – difficulties in translating promising treatment approaches into successful patient care
- The mTOR pathway in treatment of epilepsy
- Genetic findings in brain specimens: From Histology to an "Integrated" diagnostic approach
- Ganaxolone treatment in females with *PCDH19*-related epilepsy
- *CLN2*: enzyme replacement therapy
- New therapeutic strategies for the treatment of Lafora disease
- Towards ASO-therapy for Angelman syndrome
- Incidence, phenotypes and treatment consequences of childhood-onset genetic epilepsies Joseph Symonds (UK)
- Precision Medicine: What to gain in childhood?
- Precision Medicine: What to gain in adulthood
- The path forward

**Faculty**

Eleonora Aronica (The Netherlands), Berten Ceulemans (Belgium), Elena Gardella (Denmark), Renzo Guerrini (Italy), Henrike Heyne (Finland/USA), Christina Høi-Hansen (Denmark), Katrine M. Johannesen (Denmark), Johannes Lemke (Germany), Holger Lerche (Germany), Rikke S. Møller (Denmark), Marina Nikanorova (Denmark), Steven Petrou (Australia) Annapurna Poduri (USA), Guido Rubboli (Denmark), Jose' Serratosa (Spain), Sanjay Sisodiya (UK), Nicola Specchio (Italy) Joseph Symonds (UK), Steffen Syrbe (Germany), Maurizio Tagliatalata (Italy), Sarah Weckhuysen (Belgium)

**Scientific Committee**

*Rikke S Møller, Elena Gardella, Guido Rubboli*

Further information on the Conference at the website: <http://www.filadelfia.dk/filadelfia/aktuelt/ny-forskning-forside>



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