



5th Dianalund International Conference on Epilepsy
7-8 April, 2022

Implementing Epilepsy Genetics In Clinical Practice

In the last years, a great wealth of studies have demonstrated that genetic factors have a strong role in the etiology of epilepsy. Mutations in genes encoding ion channels, neurotransmitters and other proteins involved in the neuronal biology have been recognized in different epileptic conditions. Genetic testing has become more and more an essential part of the clinical practice in epilepsy. It helps in reaching an etiological diagnosis, providing prognostic information, guiding more precisely therapy possibly targeting in the near future the underlying dysfunctional pathophysiological mechanisms, or also avoiding drugs that may worsen the seizures. In this conference, we intend to discuss the current role of genetics in areas of clinical practice where a debate is still ongoing and in which further knowledge is necessary. We will discuss the yields of genetic studies in adult epilepsy patients, and how this diagnostic tool can lead to better management and be relevant in the transition from pediatric to adult care. The methodology necessary to perform clinical studies in rare conditions and how these studies in limited cohort of subjects can be crucial for the development of targeted drugs will be illustrated. Moreover, the role in this endeavour of patients' associations, international epilepsy networks and registries will be debated. Finally, the evidences suggesting that genetics can have an impact in selecting epilepsy surgery candidates and can inform on post-surgical prognosis will be presented. Case-presentations from the audience will complete this conference. Ultimately, we aim to emphasize how the collaboration of epileptologists, geneticists and patients' representatives can be instrumental to further improve epilepsy diagnosis and management.

Guido Rubboli, Elena Gardella, Rikke S. Møller

Venue: Comwell Klarskovgaard, Korsør (Denmark)



7 April 2021, Day 1

8.45-9.00 Welcome from Filadelfias direction (Mads Henrik Ravnborg, Denmark)

9.00-9.15 Introduction to the topic (Guido Rubboli / Rikke Møller, Denmark)

Session 1 - *Health care in epilepsy from pediatric to adult*

9.15-9.40 Genetic testing in adulthood (Katrine Johannesen, Denmark)

9.40-10.05 Transition from pediatric to adulthood in epilepsy (Rima Nabbout, France)

10.05-10.30 Epilepsy transition guide for adulthood neurologists (Danielle Andrade, Canada)

10.30-10.45 The Danish transition program (Mads Ravnborg, Denmark)

10.45-11.15 Coffee Break

Session 2 - *Clinical trials readiness efforts*

11.15-11.45 Natural history studies: what they are and why we need them (Ann Poduri – to be confirmed, USA)

11.45-12.10 Natural history studies in practice – the example of SCN8A (Elena Gardella, Denmark)

12.10-12.35 The relevance of international registries (Renzo Guerrini, Italy)

12.35-13.00 The role of Reference Network for rare and complex epilepsies (Alexis Arzimanoglou, France)

13.00-14.15 Lunch

Session 3 – *Molecular therapeutic board*

14.15-14.35 Introduction to precision medicine and perspectives of n-of-1 trials (Holger Lerche, Germany)

14.35 -15.00 The n-of-1 clinical trials (Victoria Defelippe, The Netherlands)

15.00-15.45 Case presentations (n=3)

15.45-17.00 Coffee Break / Poster Session

17.00-19.00 Case presentations (n=7)

20.00 - Dinner

8 April 2021, Day 2

Session 1 - *Genetic Testing for Epilepsy Surgery*

8.30-8.55 Epilepsy surgery: state-of-the-art and outcome predictors (Philippe Ryvlin, Switzerland)

8.55-9:20 The ENIGMA consortium: a bridge between neuroimaging and genetics (TBA)

9:20-9.45 Genotype-phenotype correlations of Focal Cortical Dysplasias (Eleonora Aronica, The Netherlands)

9.45-10.00 Break

10.00-10.25 Should genetics be part of standard presurgical evaluation? (Kees Braun, The Netherlands)

10.25-10.50 Biomarkers Based Stratification of Human Epilepsies (Reetta Kälviäinen, Finland)

Session 2 - *Bridging the gaps*

10:50-11.15 Patients associations and epilepsy clinical/research community: time to strengthen the alliance (Francesca Sofia, Italy)

11.15-11.30 Patients and health care providers: challenges in bridging the gaps (Torie Robinson, UK)

11.30-12.40 Coffee Break

Session 3

12.40-13:00 Late breaking news (n=5)

13.00-13.15 Concluding remarks

13.15-14.30 Farewell lunch