The MRC Centre for Neurodevelopmental Disorders' Symposium

Genetic Generalised Epilepsies: From Basic Science to Clinical Practice

PROGRAMME

08:30 **Registration**

09:00 Introduction and Welcome

Professor Mark Richardson and Professor Deb Pal, King's College London

SESSION 1: JUVENILE MYOCLONIC EPILEPSY

09:15 In the first person

Dr Timothy Counihan MD, University Hospital Galway, Ireland

09:30 Impulsivity in GGE – predictors and consequences

Dr Marte Syvertsen MD PhD, Drammen University Hospital, Norway

09:55 Structural and functional brain endophenotypes in GGE

Dr Jonny O'Muircheartaigh PhD, King's College London

10:20 The BIOJUME Study – Analysis of 600 JME deep phenotypes

Professor Deb Pal MA MSc PhD MRCP, King's College London

- COFFEE BREAK -

SESSION 2: SEIZURE PREDICTION IN GGE

11:00 In the second person

Patricia Westley, parent

11:25 Predicting or Preventing seizure transitions in mouse models

Professor Dr Gilles van Luijtelaar, Radboud University, Nijmegen

11:50 Pre-ictal brain state in human GGE

Dr Eugenio Abela MD, King's College London

12:10 How good are wearable seizure detection and prediction devices?

Dr Elisa Bruno MD, University of Catania

12:40 The Chronobiology of epilepsy

Dr Wendyl d'Souza MB ChB MPH PhD, St Vincent's Hospital, Melbourne

13:05 Lunch, posters and sponsors' Stalls

Includes charities SLC6A1 Connect and Epilepsy Action and genetic counsellor, Stephanie Oates MA LLB FHGSA, King's Health Partners

SESSION 3: ABSENCE EPILEPSIES

13:40 *Book launch:* Moment by Moment

Annie Page, parent

13:50 Genetic rescue of absence seizures

Professor Vincenzo Crunelli BSc PhD FMedSci, University of Cardiff

14:15 *To be confirmed*

14:40 The spectrum of *SLC6A1* phenotypes

Dr Katrine Johannesen, Danish National Epilepsy Centre

15:05 Myoclonic-astatic epilepsy is not what you thought it was

Dr Shan Tang MD MRCPCH PhD, King's College London

- COFFEE BREAK -

SESSION 4: CLINICAL PRACTICE

15:50 Oral presentations: best posters

16:15 The Valproate debate

Dr John Paul Leach MD FRCP, International League Against Epilepsy, UK President
Amanda Stoneman, Epilepsy Action

17:00 Unanswered questions and the future of clinical practice in GGE

Professor Mark Richardson MA MBBCh PhD FRCP, King's College London

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KING'S College LONDON

MRC | MRC Centre for Neurodevelopmental Disorders

Friday 21st June, 2019

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Further Information

Background

The clinical management of neurological disorders is changing dramatically as a result of rapid advances in basic science understanding ranging from genomics to neurophysiology, neural networks, artificial intelligence and high-resolution brain imaging. Of all the neurological disorders, epilepsy has the highest prevalence, affecting **1% of the population**.

This symposium is targeted to academics and **medical and nursing specialists** in the UK that manage people with epilepsy with particular encouragement for **neurologists in training** to attend.

Scholarly goals

Our l goal is to provide a **state-of-the-art clinical and basic science update on genetic generalised epilepsies (GGE**), a major category of epilepsy. We will achieve this through invited talks, a debate, invited scientific abstracts and patient-centred activities. The symposium is divided into four sessions over one day. Each session is opened by a patient or parent of a person with GGE in which the speaker talks about their lived experience of the condition. There then follow three to five interactive talks each lasting 17 minutes followed by 8 minutes of questions from the audience. The fourth session follows a slightly different format, starting with brief oral presentations of abstracts submitted by registered trainee attendees accompanied by prizegiving. There will then follow a moderated debate on a current hot topic in epilepsy practice lasting for 45 minutes. The session finishes with a keynote lecture on future translational challenges in epilepsy.

The **educational objectives** of the symposium are to:

Promote the **education of trainee and established neurologist**s in epilepsy.
Understand the specific **neuropsychiatric comorbidities** of different types of GGE.
Predict long term **adverse psychosocial outcomes** from early detected cognitive traits.
Define **endophenotypes** and learn behavioural and imaging examples in GGE.
Appraise the strengths and limitations of the **current, syndromic definitions of GGE**.
Correlate **mouse models** of seizure transition with human epilepsy data.
Elaborate a **circuit understanding** of seizures that overlaps with consciousness.
Evaluate the state-of-the-art in **wearable seizure detection devices**.
Identify hidden **temporal patterns** in seizure occurrence.
Follow the **translation** from successful preclinical models to precision medicine.
Articulate the arguments for and against **valproic acid** use in patient subgroups.
Recognise genetic phenotypes of GGE and indications for **genetic testing**.

Patient-Centred Activities

Each of the first three sessions will be opened by a patient or parent describing the **experience of living with the condition**. There will be representatives from the national patient charity **Epilepsy Action** and the international charity **SLC6A1 Connect** who will both host stalls at the lunch poster area. There will be a book launch by Annie Page of "Moment By Moment" on absence epilepsy which provides patients with information as well as being a great read in itself.

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