Welcome to the 2018 Family Conference on Genetic Epilepsy

Genetic Epilepsy Team Australia (GETA) hope you are informed and inspired and get an opportunity to connect with other families with children with genetic epilepsy.

During the conference:

- Access conference livestream at: GETA2018.5stream.com
- Use our hashtag on social media: #GETA2018

After the conference:

- Keep in touch via social media:
  - Facebook: @genetic.epilepsy.team.australia
  - Twitter: @geneticepilepsy
- Access recordings of sessions via: geneticepilepsyteam.com.au/conference

The 2018 Family Conference is Supported By:

Program
Genetic Epilepsy Family Conference 2018

9:00am – 9:45am	Professor Annapurna Poduri	Precision Medicine for Genetic Epilepsy – Small Steps Toward a Big Vision
9:45am – 9:50am	Scotty Simms - American KCNQ2 Foundation President	
9:50am – 10:35am	Professor Ingrid Scheffer	New Discoveries in Epilepsy Genetics Transform Clinical Care
10-:35 – 10:40am	Claire Audibert-Legué - KCNQ2 Parent	
10:40am – 11:00am	Morning tea	
11:00am – 11:45am	Dr Snezana Maljevic	Brain in a Dish: Modelling Epileptic Encephalopathy Using Human Stem Cells
11:45am – 11:50am	Monica Weldon – American Syngap Foundation President	
11:50am – 12:35pm	Professor Gavin Rumbaugh	Development of Targeted Therapies for Genetically-Defined Neurodevelopmental Disorders
12:35pm – 1:30pm	Lunch and group photo	
1:30pm – 2:15pm	Professor Steve Petrou	Therapeutic Approaches in Neurogenetic Disease
2:15pm – 2:20pm	Kimberley Hoffman – Dravet Sibling	
2:20pm – 3:05pm	Professor Glenn King	How Can Venomous Animals Help us Understand and Treat Genetic Epilepsies?
3:05pm – 3:30pm	Afternoon tea	
3:30pm – 3:35pm	Will Pierce – SCN2a Champion	
3:35pm – 4:30pm	Panel Discussion	Sara James leads the panel of Professors

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Professor Annapurna Poduri

Professor Annapurna Poduri is an Associate Professor of Neurology at Harvard Medical School and Associate in Neurology at the Epilepsy Genetic Program, Boston Children's Hospital. Dr. Poduri received her BA in Biology from Harvard University, her MD from the University of Pennsylvania School of Medicine, and her MPH from the Harvard School of Public Health. She completed her paediatric training at Boston Children's Hospital, child neurology residency at the Children's Hospital of Philadelphia, and returned to Boston for a fellowship in clinical neurophysiology at Boston Children's Hospital. She went on to pursue training in neurogenetics in the clinic and through a post-doctoral fellowship with Dr. Christopher Walsh. Dr. Poduri began her independent research program at Boston Children's Hospital in 2013 focusing on the genetics of epilepsy. She has been awarded the prestigious Dreifuss-Penry Epilepsy Award from the American Academy of Neurology and the Derek Denny-Brown Young Neurological Scholar Award from the American Neurological Association in 2015.

Professor Poduri's profile page from Boston Children's Hospital

Topic for Genetic Epilepsy Conference 2018:

Precision Medicine for Genetic Epilepsy – Small Steps Toward a Big Vision

As the keynote speaker, Anna will review a modern vision of precision medicine for epilepsy. After a brief review of epilepsy genetics, she will discuss a framework for precision medicine in epilepsy that thrives on partnerships among physicians, researchers, and parent-led organizations. Some of the key issues to consider include identifying clinical endpoints, the choice of models in the laboratory setting, and the clinical systems in which we will try to implement novel, precision therapies. The talk will attempt to combine aspirational and practical issues, challenging the status quo but also being realistic about what we still need to accomplish to work toward precision medicine.

Professor Ingrid Scheffer

Laureate Professor Ingrid Scheffer has pioneered and led the fields of epilepsy genetics and epilepsy classification over 25 years in her work as a paediatric neurologist and epileptologist at the University of Melbourne, Austin Health and the Royal Children's Hospital. With Professor Samuel Berkovic and molecular geneticists, she identified the first gene for epilepsy and subsequently discovered many more. Professor Scheffer was the first to describe multiple new epilepsy syndromes and refined our understanding of others. Now she is building on her work defining the genetic basis of epilepsy to develop precision treatments for these serious diseases.

She recently led the first major reclassification of the epilepsies in thirty years for the International League Against Epilepsy (ILAE). She has published more than 400 peer-reviewed papers, and in 2017 she co-authored the first study to show that medicinal cannabis is effective in epilepsy, published in the New England Journal of Medicine. She also leads research into the genetics of speech disorders, autism spectrum disorders, cortical malformations and intellectual disability.

Professor Scheffer has received a range of awards including the 2007 American Epilepsy Society Clinical Research Recognition Award, 2009 Royal Australasian College of Physicians Eric Susman Prize, ILAE Ambassador for Epilepsy Award, 2013 Australian Neuroscience Medallion, and was the L'Oréal-UNESCO Women in Science Laureate for the Asia-Pacific region for 2012. In 2014, she was elected as a Fellow of the Australian Academy of Science and also elected as the inaugural Vice-President and Foundation Fellow of the Australian Academy of Health and Medical Sciences. Professor Scheffer sits on the National Health and Medical Research Council and is Director of Paediatrics at Austin Health. In 2014, she received the Prime Minister's Prize for Science and was made an Officer of the Order of Australia "for distinguished service to medicine as a clinician, academic and mentor".

Professor Scheffer's website has more information on her research and achievements.

Topic for Genetic Epilepsy Conference 2018:

New discoveries in epilepsy genetics transform clinical care

In the last year, there have been seminal discoveries in our understanding of the genetics of the epilepsies. Ingrid will discuss what we mean by developmental and epileptic encephalopathies and why finding the cause is essential for the development of precision therapies. New genetic discoveries will be presented including the issue of mosaicism that is critical for families who would like more children and the steps that need to be taken so that families can make more informed decisions. She will also talk about new insights into hidden mutations in what used to be called 'rubbish' DNA, which we now know holds promising answers to the cause of specific forms of epilepsy. These exciting times in the genetics of epilepsy mean that the field is fast-paced and scientific discoveries are changing the way we approach these disorders.

Dr Snezana Maljevic

Dr Maljevic is a Research Fellow at the Florey Institute for Neuroscience and Mental Health. She is a neuroscientist working in the epilepsy research. Dr Maljevic's Master's degree focused in Molecular biology and physiology from the University of Belgrade. This was followed by a PhD degree from the University of Ulm, Germany. Dr Maljevic is skilled in Life Sciences and Disease Neurobiology.

Links to some of Dr Maljevic's research papers can be found here.

Topic for Genetic Epilepsy Conference 2018:

Brain in a Dish: Modelling Epileptic Encephalopathy Using Human Stem Cells.

This talk will provide insights into the rationale, methodology and use of induced pluripotent stem cells to study epilepsy and develop new treatments. This exciting technology has emerged in the recent years as one of the most promising tool in disease research and we will look into the opportunities and challenges specific to the modelling of epileptic encephalopathies.

Associate Professor Gavin Rumbaugh

Gavin is a tenured Associate Professor at The Scripps Research Institute, in Jupiter, Florida. Gavin's lab has identified several genes that are critical regulators of synapse biology, excitatory balance and cognitive function. These genes also increase the risk for developing neurodevelopmental disorders. Current studies in the lab are aimed at understanding how risk genes disrupt synaptic properties during developmentally sensitive periods and how this process triggers behavioural impairment and seizure. The lab's overarching goal is to apply knowledge gained from biological studies of genetic risk factors to accelerate the development of therapeutic agents to treat impaired brain excitability and behavioural dysfunction.

Links to some of Assoc Prof Rumbaugh's research papers can be found here.

Topic for Genetic Epilepsy Conference 2018:

Development of Targeted Therapies for Genetically-Defined Neurodevelopmental Disorders.

Rare genetic variants are known to cause epilepsy and related neurodevelopmental disorders. When the causal link is a single gene, it is possible to create highly specialized experimental systems for the purpose of producing tailored-made therapies for a given patient group. My talk will provide an overview of our efforts to create cutting-edge infrastructure and model systems that enable the identification and validation of drug-like compounds that target a genetically-defined neurodevelopmental disorder with epilepsy.

Professor Steven Petrou

Professor Steven Petrou is Deputy Director of the Florey Institute for Neuroscience and Mental Health and Head of the Florey's Division of Epilepsy. He heads the Laboratory of Ion Channels and Human Disease, a multidisciplinary team of researchers with a focus on revealing fundamental mechanisms of disease genesis in the central nervous system. Current major areas of investigation centre on the development and characterisation of genetically engineered mice models for the study of human familial epilepsy. He works closely with industry and has several patents for his discoveries. In addition to his many roles within the Florey Institutes and the University of Melbourne, he serves on the editorial board of the Journal Neurobiology of Disease and the Investigators Workshop Committee for the American Epilepsy Society.

Some of Professor Petrou's research papers can be found here.

Topic for Genetic Epilepsy Conference 2018:

Therapeutic Approaches in Neurogenetic Disease

Knowledge of the genetic underpinnings of a disease provides opportunities to develop therapies that precisely target disease mechanisms. The talk will introduce three major modalities that can be used to deliver on the promise of "precision medicine". In particular the promise of RNA based therapies that can precisely address many of the common genetic disease mechanisms.

Professor Glenn King

Professor Glenn King is a biochemist and structural biologist whose expertise lies in translating venom-derived peptides into human drugs and bio-insecticides. His lab maintains the most extensive collection of venoms in the world, which includes venoms from more than 600 species of venomous spiders, scorpions, centipedes and assassin bugs.

Professor King's primary focus is on the development of drugs to treat three pervasive nervous system disorders: chronic pain, epilepsy, and stroke. His lab is working closely with several pharmaceutical companies to develop drugs for clinical use.

Professor King's profile page at The University of Queensland

Topic for Genetic Epilepsy 2018:

How Can Venomous Animals Help us Understand and Treat Genetic Epilepsies?

Many epilepsies arise from genetic mutations in ion channels. Venoms are the best natural source of ion channel modulators and therefore they provide unique tools to help us understand and treat genetic epilepsies.

Panel Discussion

Chair: Sara James

Panellists: Prof Annapurna Poduri, Prof Ingrid Scheffer, Dr Snezana Maljevic, Assoc Prof Gavin Rumbaugh, Prof Steven Petrou, Prof Glenn King