5th Australian & New Zealand Dravet & Genetic Epileptic Encephalopathies Family Conference

THE UNIVERSITY OF
MELBOURNE

SATURDAY 14 OCTOBER – MELBOURNE BRAIN CENTRE, PARKVILLE

Giving back to those who support our work at the Epilepsy Research Centre

Welcome

Prof Ingrid Scheffer
Paediatric Neurologist

We warmly welcome you to the 5th Australian and New Zealand Dravet and Genetic Epileptic Encephalopathies Family Conference. The previous four Family Conferences over the last 13 years have been focused on Dravet Syndrome, and we are now delighted to extend this to all the families whose children have epileptic encephalopathies. We are fortunate, on this occasion, to have a grant from The University of Melbourne which is supporting this event and providing live-streaming, filming of the talks, and sustenance for our in-person attendees. This is our first conference that is being live-streamed and we are excited that there are as many people registered for online participation as in-person participation. We are delighted that we have Australians and New Zealanders from far and wide travelling to be here. Our online participants are streaming from the USA, UK, Canada, Australia, New Zealand, India, Portugal, Central Africa, and Taiwan.

We start our program for the day with a general overview of the syndromes that come under the umbrella of the genetic epileptic encephalopathies. While Dravet Syndrome is the most well known of these diseases, there are many other diseases which fall under this umbrella. Affected children have a wide range of disability with some having many seizures per day or even per hour, while others may have less frequent seizures but they can be prolonged and associated with loss of skills. More than 60 genes are now implicated in causing these diseases. As such, each genetic disease has its own features. Nevertheless, there are many associated conditions (co-morbidities) that occur frequently and families are often dealing with similar issues. The aim of this conference is to focus on practical concerns that families face on a day-to-day basis. This includes sleep, behavioural problems, educational considerations, seizure monitoring and medication. In each session we have ensured there is time for questions. Please feel free to ask questions as often others will have similar concerns.

I extend our warmest thanks to speakers for preparing their talks and giving up their weekend time to share their knowledge and answer your questions. In particular, we are very grateful to Dr Deepak Gill who is joining us from Sydney to share his expertise with you. We are also indebted to our wonderful panel of parents chaired by ABC journalist Sara James, who will share their knowledge and insights into dealing with the day-to-day dilemmas of having children with these diseases. Our community is indeed fortunate to have such parents leading the way.

I would like to warmly thank Krysta Trevis who has co-ordinated and meticulously planned this conference, Professor Geoff Donnan (Director, The Florey Institute of Neuroscience and Mental Health) for allowing us to use this venue, 5Stream for streaming our event worldwide, and our team from the Epilepsy Research Centre at The University of Melbourne. Lastly, I want to thank you all for giving up your time to join us today and I look forward to hearing your feedback on our 5th Australian and New Zealand Dravet and Genetic Epileptic Encephalopathies Family Conference.

Laureate Professor Ingrid Scheffer AO MBBS FRACP PhD FAHMS FAA
Melbourne Laureate Professor, The University of Melbourne
Director of Paediatrics & Director of Children's Epilepsy Program, Austin Health
Neurologist, Royal Children's Hospital

Program

Dravet & Genetic Epileptic Encephalopathies Family Conference

9.15	Registration Open
MORNING SESSION: MAIN AUDITORIUM	
9.30	**Professor Ingrid Scheffer** – What are the genetic encephalopathies?
10:30	**Dr Deepak Gill** – Cannabidiol and other drugs: why drug trials matter
11:00	**Ms Samantha Turner** – Speech problems in epileptic encephalopathies
11.15	**Morning tea**
11.45	**Associate Professor Margot Davey** – Sleep
12:15	**Lisa Rath** – Seizure Monitoring

GENE SESSIONS

	Main Auditorium	Education Room (Ground floor)	Seminar Room (Level 5)
12:30	**Professor Ingrid Scheffer** – SCN1A **Ms Samantha Turner** – Speech in Dravet	**Dr Katherine Howell** – Specific Genetic Epileptic Encephalopathies	**Dr Deepak Gill** – What does it mean when the gene is not known?
1:15	**Lunch**		

AFTERNOON SESSION: MAIN AUDITORIUM

2:00	**Dr Chidambaram Prakash** – Managing challenging behaviour
2:40	**Helen Hatherly & Kris Baker**– Selecting the best school for my child
3:10	**Associate Professor Doug Crompton** – Mortality risk in epilepsy
3:30	Afternoon tea
4:00	**How families lead the way** – Panel members: Caterina Parisi, Tom Philbin, Danielle Williams and Joanna Hulland chaired by Sara James.
4:55	**Close** – Professor Ingrid Scheffer

2nd Australian and New Zealand Dravet Family Conference 2008

4th Australian and New Zealand Dravet Family Conference 2014

Speakers

PROFESSOR INGRID SCHEFFER AO
Paediatric Neurologist

Laureate Professor Ingrid Scheffer is a physician-scientist whose work as a paediatric neurologist and epileptologist at the University of Melbourne has led the field of epilepsy genetics over more than 25 years, in collaboration with Professor Samuel Berkovic and molecular geneticists. Together they identified the first epilepsy gene and many genes subsequently. Professor Scheffer has described many novel epilepsy syndromes and refined genotype–phenotype correlation of many disorders.

Her major interests are genetics of the epilepsies, epilepsy syndromology and classification, and translational research. She collaborates on research on the genetics of speech and language disorders, autism spectrum disorders, cortical malformations and intellectual disability. She led the first major reclassification of the epilepsies in three decades for the International League Against Epilepsy.

She has received many awards: 2007 American Epilepsy Society Clinical Research Recognition Award, ILAE Ambassador for Epilepsy Award, 2013 Australian Neuroscience Medallion, and the 2012 L'Oréal-UNESCO Women in Science Laureate for the Asia-Pacific region. In 2014, she was a co-recipient of the Prime Minister's Prize of Science, and awarded the Order of Australia. In 2014 she became the inaugural Vice-President of the Australian Academy of Health and Medical Sciences.

DR DEEPAK GILL
Paediatric Neurologist, The Children's Hospital at Westmead

Deepak Gill is a Senior Staff Specialist in paediatric neurology and is the Head of the Comprehensive Epilepsy Service at The Children's Hospital at Westmead, Sydney Children's Hospitals Network (SCHN), Sydney. Dr Gill underwent his undergraduate training in London, and postgraduate paediatric neurology training in London and Sydney.

Dr Gill has a broad experience in the full spectrum of early neurological disorders, and has an interest and expertise in the early diagnosis and treatment of childhood onset epilepsy and the epileptic encephalopathies. As part of the Westmead Epilepsy Surgery Program he has been exploring a new surgery technique of Stereo-EEG to find the region of onset of seizures in young children with intractable epilepsy. He is a Principal Investigator as part of the Cannabis Compassionate Access Scheme at SCHN and has embarked on clinical trials in the treatment of Dravet Syndrome.

Dr Gill is a Clinical Senior Lecturer at The University of Sydney, and has educated students both locally and overseas on all aspects of paediatric epileptology and EEG.

MS SAMANTHA TURNER
Speech Pathologist

Samantha is a paediatric speech pathologist and clinical researcher, and has recently completed her PhD at The University of Melbourne. She has worked in clinical research for 16 years, phenotyping families with various epilepsy syndromes, speech and language disorders, brain malformations and autism. This work has led to important and clinically relevant publications in journals such as Nature Genetics, Brain and Neurology.

She has a particular interest in Dravet syndrome, and examined speech and language skills of individuals with Dravet syndrome as part of her PhD.

ASSOCIATE PROFESSOR MARGOT DAVEY

Director, Melbourne Children's Sleep Centre, Monash Health

A/Prof Margot Davey has been Director of the Melbourne Children's Sleep Centre, based at Monash Children's Hospital, Clayton since 2003. This service includes outpatients, inpatient sleep studies, home oximetry and managing children who require non- invasive ventilation.

She is an Adjunct Senior Lecturer in the Ritchie Centre for Baby Health Research, Monash University, and is involved in an extensive research program evaluating the neurocognitive and cardiovascular effects of childhood sleep disordered breathing. She also has appointments at the Royal Children's Hospital, Epworth Sleep Centre and St Vincent's Hospital, East Melbourne.

LISA RATH

Epilepsy Support Worker, Epilepsy Foundation (Victoria)

Lisa has worked for the Epilepsy Foundation for 28 years, half the lifetime of the organisation. She gained a general nursing qualification at Royal Melbourne and Williamstown Hospitals in the late 1970's and subsequently gained an Associate Diploma of Welfare Studies at the Bendigo Regional Institute of TAFE before being employed by the Foundation as a part time regional worker based in Bendigo with responsibility for the Hume/Loddon Mallee areas. She moved to the Melbourne office in 2007.

Lisa is a strong advocate for people with epilepsy. She ensures that her clients receive the best services possible to maximize their opportunity to live well in the community despite their diagnosis of epilepsy. Lisa always places the person with epilepsy and their needs at the forefront of her work developing exemplary relationships with specialist epileptologists, paediatricians and community organisations. To this end she has the respect of the medical profession and her colleagues with whom she works so closely. Her clients see her as a supportive and reliable advocate in their lives.

DR KATHERINE HOWELL

Consultant Neurologist, Royal Children's Hospital, Melbourne

Dr Katherine Howell is a paediatric neurologist and epileptologist at the Royal Children's Hospital, Melbourne. Her research interests include epilepsy and neurogenetics.

She undertook a large population-based study of the incidence and causes of severe epilepsies of infancy for her PhD; this research is ongoing. She has been involved in national and international collaborative studies of genetic epilepsies, and is the state lead for the epileptic encephalopathies flagship of the Australian Genomic Health Alliance.

DR CHIDAMBARAM PRAKASH

Consultant Child & Adolescent Psychiatrist, Royal Children's Hospital Melbourne

Dr Prakash is the Principal Hospital Psychiatrist for The Royal Children's Hospital. A child and adolescent psychiatrist with many years' experience, Prakash has a special interest in the mental health problems of children and adolescents with intellectual disability, autism, neurological problems and chronic medical illness.

HELEN HATHERLY, OAM

Principal, Ashwood School

Helen Hatherly is the Principal of Ashwood School - a coeducational school for 210 students aged from 5 – 18 years who have mild intellectual disabilities. Students who graduate from Ashwood School leave with high self-esteem, a wide circle of friends and a knowledge and understanding that they can accept challenges and work with confidence at achieving their goals.

Helen commenced her teaching career in mainstream primary schools and has taught in specialist schools for over 25 years. She has been involved in both the establishment and implementation of a range of Victorian Government Department of Education and Training initiatives.

TBC

This is Helen's twelfth year as Principal of Ashwood School. She is also the President of Victorian Chapter of the Australian Association of Special Education.

KRIS BAKER

Assistant Principal, Frankston Special Development School

Kris Baker is assistant principal at Frankston Special developmental School, a coeducational school for around 130 students, 5 – 18 years with a moderate to severe intellectual disability. Key learning priorities of Frankston SDS is the development of communication, independence and social competencies in all students.

Kris is a member of the Principal's Association of Special Schools and was awarded a PASS Award for excellence for her role in the development of a comprehensive teaching and learning program around protective behaviours for students with a disability.

ASSOCIATE PROFESSOR DOUG CROMPTON

Neurologist, Northern Health

Dr Crompton is a Neurologist and Epilepsy specialist who trained in the UK, before completing a clinical fellowship in Epilepsy at Austin Health, 2006-2009. He then worked as a Senior Clinical Lecturer and Honorary Consultant Neurologist in Newcastle upon Tyne and Middlesbrough, UK before returning to Melbourne in 2012. He now heads Northern Health's expanding Neurology department. He also holds an Honorary Neurologist appointment at Austin Health, and is a University of Melbourne Associate Professor.

Dr Crompton's main research interest is in Epilepsy, particularly the Genetics of Epilepsy and Sudden Unexpected Death in Epilepsy (SUDEP). He has published in leading Neurology journals such as The Lancet Neurology, Annals of Neurology and Brain.

SARA JAMES

Sara James is an Emmy award-winning foreign correspondent who covers Australia for NBC News. She also reports on news related to the US for the ABC, including her fortnightly American in Oz segment on ABC News Breakfast. Sara co-anchored ABC News 24's coverage of the extraordinary 2016 US election and the 2017 Presidential Inauguration coverage. Now a featured ABC contributor, Sara will file more reports for other ABC programs coming up in 2017.

Prior to moving to Australia in 2008, Sara was a guest host on the news desk of NBC's Today Show. She travelled the world to report on stories including the Taliban, modern slavery in Sudan, the Bosnian War Crimes Tribunal, and 9/11 in her former hometown of New York. Sara has received numerous awards including the Overseas Press Club of America Citation for Excellence, the US National Press Club award, and an Emmy.

Sara has written two books – *The Best of Friends* (HarperCollins) and *An American in Oz* (Allen & Unwin) – and edited a third book, *An Extraordinary School* (ACER Press).

Parent panel

DANIELLE WILLIAMS

Danielle has worked in preventative healthcare in the corporate sector for 20 years. She travelled the diagnostic odyssey for 6 years and received a diagnosis for her daughters in 2016. Jaeli (8) and Dali (6) have a rare genetic epilepsy syndrome, Syngap. Since receiving the diagnosis, Danielle and husband Danny have advocated for whole genome sequencing in Australia with the Garvin Institute, connected with the global Syngap community, lobbied for a unified global Syngap community, ensuring an Australian presence within the American Syngap medical advisory board, ran and analysed the first International Syngap survey, partnered with The Epilepsy Foundation to create an online SynGAP research presence, launched the secret4syngap fundraising campaign, received community support and media coverage in syngap research fundraising efforts and along with 2 other families, formed GETA (Genetic Epilepsy Team Australia) with a mission to help world-leading Australian researchers cure Genetic Epilepsy.

Joanna & her husband Chris live in south west NSW, just over the border from Swan Hill, 4 hours north of Melbourne. They are farmers who own and operate a 7 day a week instant lawn business as well as produce sheep, cattle, & cereals, employing up to 8 staff. They have 2 children, Sarah 14 years old & Ryan 13 years old. Ryan has Dravet syndrome. Both children work on the farm, with Sarah driving trucks and tractors. Ryan's love is the sheep and cattle and is always involved if he is not at school. He would like to drive more of the machinery but we have restricted him due to his epilepsy.

JOANNA HULLAND

TOM PHILBIN

Originally from Liverpool in the North West of England, Tom Philbin met his partner, Sam Jackson, in London 16 years ago. They re-located to Sam's home town of Melbourne in 2003 and welcomed the birth of their son, Luke, in late 2005. Luke suffered his first seizure at 6 months of age and was diagnosed with Dravet Syndrome at the age of 1. Now almost 12 years old and despite many set backs, challenges and serious episodes, Luke continues to astound with his incredible resilience and killer smile. Tom likes nothing more than spending time with his inspirational partner, Sam, preferably over some fine wine and fare at any of Melbourne's fantastic eateries whenever they get the chance.

I'm from Melbourne and am a mum of two beautiful boys: Marco and Lucas aged 12 and 10. In hind sight- Marco suffered his first known seizure at 4 months of age, was diagnosed with severe, uncontrolled epilepsy at 2 and a half and diagnosed with an extremely rare genetic chromosome disorder at 3 and a half. In the beginning, Marco presented with similar traits to Dravet Syndrome, however the differences were such that further investigation confirmed an extremely rare genetic 2q chromosome disorder- called 2q23.1. In the early years, Marco suffered countless seizures and differing seizure types (including life threatening), and countless hospital admissions, however Marco now 12, is now on a medicine and seizure free journey.

Due to the rarity of the condition, not much is known, so we take it a day at the time. Marco attends a special school and enjoys his after school horse riding, swimming and gym activities and is his brothers biggest fan when watching him play basketball or soccer. He is an extremely resilient, energetic, fun loving little boy who is an absolute gun on his ipad. I like to spend time with my kids and husband – and love a well made latte.

CATERINA PARISI

Notes

Notes

Epilepsy Research Centre

The Epilepsy Research Centre is a large cohesive research group within The University of Melbourne Department of Medicine at the Austin Health campus. We study individuals and families with a history of seizures using cutting edge research methods and technologies. This includes characterising seizures, clinical trials of medications, and molecular genetic studies to find new genes and understand what they do. This is an important Australian-led international study.

Our research group has been responsible for the identification of several epilepsy syndromes and the discovery of many epilepsy genes, including the first gene in 1995. These discoveries continue to improve our understanding of the mechanisms underlying epilepsy, meaning the development of new treatments is becoming a reality. This work was recognized by the 2014 Prime Minister's Prize for Science awarded jointly to Professors Scheffer and Berkovic.

Some of our team are here today. If you have any questions, just let us know.


Amy
Schneider


Caitlin
Bennett


Georgie
Hollingsworth


Dr Danique Vlaskamp
visiting Doctor from the
Netherlands


Katja
Boysen


Olivia
Henry


Krysta
Trevis


Dr Hannah Stamberger
visiting Doctor from
Belgium

Contact us

Interested in staying involved with the Epilepsy Research Centre? Find us online at epilepsyresearch.org.au or get in touch at epilepsy-austin@unimelb.edu.au. Our research team are here today if you have any questions – keep an eye out for the blue name tags!

Proudly supported by:

THE UNIVERSITY OF MELBOURNE

EPILEPSY RESEARCH CENTRE
Melbourne, Australia

THE FLOREY
INSTITUTE OF NEUROSCIENCE & MENTAL HEALTH

Live stream: http://GeneticEpilepsy.5stream.com/

#GeneticEpilepsy