ANZCNS 7th Annual Scientific Meeting
12th – 14th September 2018
Pan Pacific Perth Hotel
Perth, Western Australia
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WELCOME MESSAGES

Welcome Message From The Local Organisers

We welcome you to the 7th Annual Scientific Meeting of the Australia and New Zealand Child Neurology Society. Our distinguished international speakers - Shinichi Hirose (Japan), Raman Shankar (USA), Nicola Specchio (Italy) and Tanuja Chitnis (USA) will discuss the latest advances in epilepsy, neurogenetics, neuroimmunology and neurocognitive disorders. Australian presenters will complement the international talent, covering fetal, child and adolescent neurology. Posters and Platforms will demonstrate the breadth of research in paediatric neurology.

We welcome your participation in an exciting hypothetical entitled “Your health dollar: who deserves it and who decides?”, in the context of expensive new therapies in child neurology.

Attendees will have the opportunity to present “Diagnostic dilemmas” during the last session of the meeting and obtain solutions with fun and flair.

We look forward to you joining us in Perth and hope you enjoy the meeting.

N. Lakshmi Nagarajan
ASM Chair

Welcome Message From The Australia And New Zealand Child Neurology Society

On behalf of the Board of the Australia and New Zealand Child Neurology Society Ltd, I would like to welcome you to our 2018 Annual Scientific Meeting. The Organising Committee has worked hard to arrange this event at Pan Pacific Perth Hotel, WA, which is known for its exceptional conference support. We are delighted to welcome international invited speakers and look forward to the camaraderie, discussions and celebrations.

Mark MacKay
Chief Executive Officer, Australia and New Zealand Child Neurology Society

About Australia and New Zealand Child Neurology Society

The Australia and New Zealand Child Neurology Society (ANZCNS) is a collaborative group of approximately 100 Paediatric Neurologists, Paediatric Neurology Trainees and Allied Specialities from all over Australia and New Zealand. The organisation aims to promote the best possible care for children with neurological disease by fostering education and training of those who care for children with neurological diseases, advancing neuro-paediatric research and promoting improved standards of care for children with neurological disease. More information about ANZCNS is available on the ANZCNS website: www.anzcns.org.au
COMMITTEES

Organizing Committee
Lakshmi Nagarajan (Chair)
Jon Silberstein (Co-Chair)
Phillipa Lamont
Peter Walsh

Abstract Review Committee
Anita Cairns
Maina Kava
Mark MacKay
Lakshmi Nagarajan
Kate Riney
Snehal Shah
Ubaid Shah

Platform Judges
Annie Bye
Ubaid Shah
Manoj Menezes
GENERAL CONFERENCE INFORMATION

Secretariat Office
Members of the Committee can be contacted via the Registration Desk.
For queries arising after the congress, please contact:
Australia and New Zealand Child Neurology Society
PO Box 8446, Woolloongabba, QLD 4102, Australia
e: admin@anzcns.org.au
m: +61 423 827 488

The Conference Venue
Pan Pacific Perth Hotel
Pan Pacific Perth Hotel, 207 Adelaide Terrace, Perth WA 6000.

Parking
Valet Parking
Available 24 hours a day for AUD35 per night including unlimited entries and exits.

Self-Parking
Self-parking in our undercover car park is available at the following competitive rates:
- 0 to 1 hour at A$6
- Day rate 6:00am to 6:00pm at A$18
- Night rate 6:00pm to 6:00am at A$12
- 24 hours at A$25*

*Please note our carpark has a maximum clearance height of 1.9 metres and is subject to availability
Registration Desk

The registration desk is located on Level 1 of the Pan Pacific Perth Hotel. Please note that name badges must be clearly displayed at all times.

Registration desk opening hours are:

Wednesday 12th September: 08:30-17:00
Thursday 13th September: 07:30-17:00
Friday 14th September: 08:00-15:00

Registration Entitlements

- Entry to all conference sessions
- Morning teas, lunches, afternoon teas
- Conference satchel and delegate satchel material
- Printed Handbook and final program

Speakers’ Information

Speakers must submit their final presentation to the audiovisual technician in the main plenary room (Grand River Ballroom) on Convention Floor, Level C at least 2 hours prior to their session start time.

Exhibition

A trade exhibition is held in conjunction with the 7th ANZCNS Annual Scientific Meeting. This is an integral part of the event and allows delegates the opportunity to be updated on the latest developments in products and services in the field of child neurology. The exhibition area is located outside Grand River Ballroom in Foyer 3, and is open at meal break times.

Continuing Medical Education

Fellows and trainees of the Royal Australasian College of Physicians can apply for one credit for each hour attended of the academic program of the conference.

Certificate of Attendance

A certificate of attendance will be emailed out to you after the meeting.

Catering

Lunches and morning and afternoon teas are served in Foyer 3. Any delegates with particular catering needs should have notified these on their online registration form and need to see the venue staff to collect their special meal.

Dress Code

The 7th ANZCNS Annual Scientific Meeting promotes casual business attire for the duration of the conference.

Mobile/cell phones

Mobile/cell phones must be turned off or to vibrate mode during all sessions so as not to disturb speakers or other delegates. Pagers should be on vibrate mode.

Community and Consumer Delegates

The 7th ANZCNS Annual Scientific Meeting is primarily for health professionals. Community and consumer support groups have been welcomed to the 7th ANZCNS Annual Scientific Meeting to promote partnerships with health professionals towards a common goal. Such delegates should understand that the presentations will contain medical jargon and may contain information and images that some might find distressing. Questions from the open floor during sessions are invited primarily from health professionals.
Photography

The 7th ANZCNS Annual Scientific Meeting strictly prohibits all photography (ash, digital, or otherwise), audio and/or videotaping during the conference except by authorised photographers. There will be official ANZCNS photographers who will be capturing candid photos of the conference. These photographs will be used in congress communications. In registering and attending this conference you accept that your likeness may be taken in the form of photographs taken by photographers approved by Australia and New Zealand Child Neurology Society for activities related to this and future conferences (including but not limited to: use within newsletters, presentations or other media that recounts the events that occurred during this conference and/or in the promotion of future conferences). Delegates should inform the Australia and New Zealand Child Neurology Society (by email) in advance of or during the conference week if they do not provide permission for this.

Liability and Insurance

The Australia and New Zealand Child Neurology Society will not accept any liability for personal injury or loss/damage to property/belongings of participants or accompanying persons, either during or following the conference or their stay. It is recommended that delegates arrange their own personal health, accident and travel insurance for themselves and their families.
CONFERENCE DINNER

Venue: Fraser’s Function Centre, Kings Park, Perth
Date: Thursday 13th September 2018
Time: 7.00pm for pre-dinner drinks
Dinner: 7.30 - 10.30pm

Fraser’s Restaurant opened in 1993, and is situated on Fraser Avenue in beautiful Kings Park. The avenue was named after Malcolm Fraser, the first Surveyor-General of Perth, and is lined with lemon-scented gums that form a majestic canopy, creating a spectacular entrance to the park.

Transport to and from the dinner venue will be provided. Transfer schedule is as below;

Pick up from the Pan Pacific Perth Hotel at 6.45pm
Pick up from Fraser’s Function Centre to the Pan Pacific Perth Hotel at 10.30pm.
SPONSORS

The Australia and New Zealand Child Neurology Society wish to extend their thanks to our major sponsors who have supported this event:

MAJOR SPONSORS

- Biogen
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- SANOFI GENZYME
- Inspired by patients. Driven by science.
INTERNATIONAL SPEAKER SPONSORS

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EXHIBITORS

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<td>UCB Australia</td>
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INTERNATIONAL SPEAKERS

We welcome our International Speakers:

**Shinichi Hirose**, MD, PhD is a Professor of Pediatrics, Head of both the Department of Pediatrics and the Research Center for Molecular Pathomechanisms of Epilepsy at Fukuoka University, and a member of the standing committee of the International Pediatric Association (IPA). His interests are in the molecular genetics of epilepsies. He has worked extensively on causative mutations and their molecular consequences in the neuroscience mechanisms underlying epilepsies, and has published extensively on the molecular pathomechanisms of epilepsies. He is the principal investigator on numerous clinical studies and has studied many childhood diseases including epilepsies, metabolic diseases, and inherited diseases.

**Raman Sankar**, MD, PhD, is Professor of Neurology and Pediatrics and Chief of Pediatric Neurology at the David Geffen School of Medicine at the University of California, Los Angeles. He holds the Rubin Brown Distinguished Chair in Pediatric Neurology. Dr. Sankar is a graduate of the University of Bombay, India. He obtained his PhD from the University of Washington in Medicinal Chemistry and his MD from Tulane Medical School. He trained in pediatrics at the Children’s Hospital of Los Angeles. He completed his training in neurology and pediatric neurology at UCLA. His laboratory research has addressed the mechanisms of seizure-induced injury and epileptogenicity in the developing brain. He has also undertaken studies to improve the throughput for screening compounds for antiepileptogenic action on the developing brain. Recent studies have demonstrated connections between the epileptic state, physiologic stress, inflammation and how these factors impact on the neurobehavioral comorbidities. He is a member of an active pediatric epilepsy program at UCLA that is well known internationally for many advances in pediatric epilepsy surgery. Dr. Sankar has authored more than 250 research articles, reviews and book chapters. He has also co-edited text books on pediatric epilepsy as well as the applications of basic science in epilepsy research. He is an elected Fellow of the American Academy of Neurology and an elected member of the American Pediatric society. Dr. Sankar is a member of the Professional Advisory Board of the Epilepsy Foundation. He has been a member of the Commission on Neurobiology of the International League Against Epilepsy and presently serves on the Commission’s Task Force for the Workshop on the Neurobiology of Epilepsy (WONOEP).
Nicola Specchio, MD, PhD is Head of the Epilepsy Unit in the Department of Neuroscience at Bambino Gesù Children’s Hospital, Rome, Italy, where within his role, he is responsible for the diagnosis and treatment of patients with paediatric epilepsy. This includes the pre surgical evaluation of patients with drug resistant epilepsy and the selection of patients with genetic epilepsies. Dr Specchio’s main interest lies with seizure semiology and the classification of epileptic seizures and syndromes. He has published papers in many international journals including Epilepsia, Epilepsy Research, Epilepsy and Behavior and is currently responsible for several clinical studies regarding the invasive monitoring of patients with epilepsy and the genetic aetiology of epileptic encephalopathy in the first three years of life. Dr Specchio is a representative of International League Against Epilepsy (ILAE) Commission on European Affairs and of the Italian Chapter of the ILAE.

Tanuja Chitnis, MD, FAAN is a neurologist specialising in multiple sclerosis (MS). She is an Associate Professor of Neurology at Harvard Medical School and has a research appointment as a Scientist within the Ann Romney Center for Neurologic Diseases at BWH. Her interest in children with MS led her to start the Partners Pediatric MS Center at MassGeneral Hospital for Children where she serves as the Director, and PI of several studies to understand the causes of MS in children. Since 2010, she has served as the elected Chair of the International Pediatric MS Study Group, where she has led several initiatives in the study of MS in children including the launch of the first clinical trials in this population. She also serves as the Medical Director of the CLIMB Natural History study at the Partners MS Center, Brigham and Women’s Hospital (BWH) which follows over 2400 MS patients longitudinally. Here she oversees a team of analysts and postdoctoral fellows working to understand the causes, heterogeneity and response to treatment in MS patients. In addition to her clinical roles, she has authored over 200 publications and reviews related to MS and demyelinating disorders, and serves on the advisory board and steering committee of several MS-related organisations and studies. Throughout her career, her primary scientific contributions have been in immune mechanisms in MS, paediatric MS research and clinical trials, the CLIMB observational cohort study, hormonal mechanism in MS, and Neuromyelitis Optica (NMO).
### WEDNESDAY 12TH SEPTEMBER

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<td>Early Morning Session</td>
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<td>09:00</td>
<td><strong>Registration Desk Open</strong></td>
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<td>09:30</td>
<td><strong>Arrival Tea and Coffee</strong></td>
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<td>10:00</td>
<td><strong>Conference Official Opening</strong> (Lakshmi Nagarajan, Co-convenor Local Organising Committee)</td>
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<td>10:15</td>
<td>Symposium - Focal Epilepsy: Enhance Your Awareness</td>
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<td>(Chair: Annie Bye &amp; Sonny Gubbay)</td>
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<td>Seizure Semiology in Focal epilepsy</td>
<td>Nicola Specchio</td>
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<td>Ictal onset and seizure propagation</td>
<td>Ubaid Shah</td>
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<td>Sleep and Focal Epilepsy</td>
<td>Lakshmi Nagarajan</td>
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<td>Genes in Focal Epilepsy</td>
<td>Shinichi Hirose</td>
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<td>12:15</td>
<td><strong>Lunch</strong></td>
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<td>12:30</td>
<td>ANZCNS Paediatric Neurology Trainee meeting</td>
<td>Mark Mackay</td>
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<td>13:30</td>
<td><strong>Paediatric Demyelinating Disorders</strong></td>
<td><strong>NOVARTIS</strong></td>
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<td>(Chair: Manoj Menezes &amp; Snehal Shah)</td>
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<td>Advances in Paediatric Demyelinating Disorders</td>
<td>Tanuja Chitnis</td>
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<td>14:15</td>
<td><strong>Platform Session 1 (Chair: Manoj Menezes &amp; Snehal Shah)</strong></td>
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<td>Etiology is the key determinant of neuroinflammation in epilepsy</td>
<td>Kavitha Kothur</td>
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<td>Role of Epilepsy Gene Panel in Childhood Epilepsy</td>
<td>Eleanor Ng</td>
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<td>Reliability of amplitude-integrated EEG for the detection of neonatal seizures</td>
<td>Abhijeet Rakhasbhuvankar</td>
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<td>15:00</td>
<td><strong>Afternoon tea</strong></td>
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<td>15:30</td>
<td>Maurice Spillane’s Hypothetical: Your health dollar: who deserves it and who decides?</td>
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<td>Panel discussion: Annie Bye, Victor Cheng, Christopher Etherton-Beer, Richard Webster, Chris Troedson, Maina Kava</td>
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<td>17:00</td>
<td><strong>ANZCNS Annual General Meeting</strong></td>
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08:30  REGISTRATION DESK OPEN
09:00  ANZCNS BOARD MEETING
09:00  ANZCNS TRAINEE BREAKFAST MEETING
09:30-10:00  ARRIVAL MORNING TEA
10:00  CONFERENCE OFFICIAL OPENING
10:15-12:15  SYMPOSIUM - FOCAL EPILEPSY: ENHANCE YOUR AWARENESS
10:15  Seizure Semiology in Focal epilepsy
       Nicola Specchio
11:00  Ictal onset and seizure propagation
       Ubaid Shah
11:25  Sleep and Focal Epilepsy
       Lakshmi Nagarajan
11:50  Genes in Focal Epilepsy
       Shiniche Hirose, Department of Pediatrics, School of Medicine and Central Research Institute for the Molecular Pathomechanisms of Epilepsy, Fukuoka University, Fukuoka, Japan
       Genetics for focal epilepsy remains underdeveloped. Nevertheless, germline mutations in several genes have been found in genetic focal epilepsies. Recent advancement in molecular biology has allowed identification of somatic mutations that are involved in focal cortical dysplasia as well as in nonlesional focal epilepsy. Further studies on genetics for focal epilepsy should uncover the underlying pathological mechanisms, and thereby develop novel therapeutics for epilepsy.
12:15-13:30  LUNCH
12:30  ANZCNS PAEDIATRIC NEUROLOGY TRAINEE MEETING
       Mark Mackay
13:30-14:15  PAEDIATRIC DEMYELINATING DISORDERS
13:30  Advances in the diagnosis and management of paediatric demyelinating disorders
       Tanuja Chitnis
       This presentation will review the clinical and radiological presentation of the spectrum of pediatric demyelinating disorders including pediatric multiple sclerosis, neuromyelitis optica spectrum disorders and acute disseminated encephalomyelitis, and MOG-antibody associated demyelination, as well as management strategies and recent therapeutic advances for these disorders in children.
14:15-15:00  PLATFORM SESSION 1

14:15  Etiology is the key determinant of neuroinflammation in epilepsy: The prominent elevation of cerebrospinal fluid cytokines and chemokines in FIRES and febrile status epilepticus

Kavitha Kothur 1, Sushil Bandodkar 2, Louise Wienholt 3, Stephanie Chu 4, Alun Pope 4, Deepak Gill 1, Russell C Dale 1

(1) Kids Neuroscience Centre, The Children’s Hospital at Westmead, Sydney, NSW; (2) The Children’s Hospital at Westmead, Sydney, NSW; (3) Royal Prince Alfred Hospital, Sydney, NSW; (4) The University of Sydney, NSW

Aim: To investigate the role of intrathecal inflammation using cerebrospinal fluid (CSF) cytokines and chemokines in paediatric epilepsy subgroup patients with frequent daily seizures.

Methods: We measured 34 cytokines/chemokines using multiplex immunoassay in CSF collected from paediatric patients with frequent daily seizures caused by febrile infection-related epilepsy syndrome (FIRES)/FIRES related disorders (FRD) (n=6), febrile status epilepticus (FSE) (n=8) and afebrile status epilepticus (ASE) (n=8)), new onset explosive focal seizures (NOEFS, n=10), infantile spasms (n=19), genetic epileptic encephalopathy (GEE, n=11) and genetic generalised epilepsy (GGE, n=10) and compared with encephalitis (n=43) and non-inflammatory neurological controls (n=20). We also performed serial CSF cytokine/chemokine studies in 3 cases with FIRES/FRD.

Results: The elevation of cytokine/chemokines was higher in FIRES followed by FSE and NOEFS when compared to non-inflammatory neurological controls and other epilepsy subgroups. Th1 associated cytokines (TNF alpha, CXCL9, CXCL10, CXCL11), Th17 (IL-6, IL-8) and CCL2, CCL19 and CXCL1 (p<0.05) were elevated in FIRES in contrast to the elevation of a broader network of cytokines/chemokines in encephalitis. The median concentrations of these cytokine/chemokines rapidly declined during the course of illness in all three FIRES/FRD cases. Infantile spasm, GGE and GEE generally lacked significant elevation of cytokines/chemokines despite frequent daily seizures.

Conclusion: The prominent elevation of CSF cytokines/chemokines in FIRES/FRD and to a lesser extent, FSE group compared to other epilepsy subgroups with frequent daily seizures highlights the importance of etiology in causing measurable CNS inflammation, rather than inflammation being secondary to the seizures themselves.

14:30  Role of Epilepsy Gene Panel in Childhood Epilepsy

Eleanor Ng 1, Dimitar Asmanov 2, Lakshmi Nagarajan 1

(1) Perth Children’s Hospital, Nedlands, WA; (2) PathWest Laboratory Medicine, Perth, WA

Objectives: To evaluate the role of an epilepsy gene panel in treatment refractory childhood epilepsy.

Methods: We reviewed the epilepsy gene panel tests performed at our hospital over 2 years between June 2015 and 2017. Thirty four patients had approval for epilepsy gene panel testing.

Massively parallel sequencing was performed on various instruments,
including Ion Proton (Life Technologies), Illumina MiSeq and Illumina NexSeq 550, by using either exome (Ion Ampliseq, Life Technologies) or sub-exomic gene panel (Illumina TruSight One) enrichment kits. Alignment and variant calling were performed using Torrent Suite (Life Technologies) or Illumina BWA Enrichment (v2.1.1) pipelines. Variants were analysed in Bench Lab NGS (Cartagenia) and classified according to ACMG guidelines. Potential cost savings if the epilepsy gene panel was undertaken earlier will be assessed.

Results: Mutations were found in 11 of 34 patients. Five had sodium channel mutations, one each of KCNQ2, GABRB3 mutation, CLN6, PRRT2, TBC1D24 and ARX mutation. Thirty out of thirty four had early onset epilepsy.

Conclusion: The search for a specific aetiological diagnosis may involve costly investigations such as repeated VEEG, MRI and metabolic work up. The epilepsy gene panel is likely to be cost effective. In this selected cohort, 32% had gene mutations relevant to their epilepsy. Epilepsy gene panel should be available early, for children with early onset and refractory epilepsy.

References:

14:45 Reliability of amplitude-integrated EEG for the detection of neonatal seizures
Abhijeet Rakshasbhuvankar 1, Deepika Wagh 2, Sam Athikarisamy 2, Jonathan Davis 2, Linda Palumbo 3, Soumya Ghosh 4, Lakshmi Nagarajan 5, Shripada Rao 2
(1) Neonatal Paediatrics, King Edward Memorial Hospital, Subiaco, WA; (2) Neonatal Paediatrics, Perth Children's hospital, Nedlands, WA; (3) Department of Neurology, Perth Children's Hospital, Nedlands, WA; (4) Centre for Neuromuscular and Neurological Disorders, Perron Institute, University of Western Australia, Perth, WA

Background: Reliability, as measured by inter-rater agreement, of a diagnostic test must be established before its use in clinical practice. Amplitude-integrated electroencephalogram (aEEG) is increasingly used for neonatal seizure detection. However, data regarding inter-rater agreement amongst neonatologists for the use of aEEG for neonatal seizures detection is limited.

Methods: The project was a part of a diagnostic accuracy study comparing aEEG with conventional video-electroencephalography (cEEG) for the detection of neonatal seizures. Twenty-four hour aEEG recordings of 35 infants at risk of seizures were interpreted independently by five neonatologists with experience in aEEG interpretation from four different neonatal intensive care units. Three out of the five neonatologists had also received training in neonatal aEEG interpretation. Inter-rater agreement between the five neonatologists was determined using intra-class correlation coefficient (ICC).

Results: Out of 35 infants, seven infants had seizures on cEEG with a total of 169 seizure episodes. ICC for the detection of “infants with seizures” by neonatologists using aEEG was 0.89 (95%CI: 0.82 - 0.94)
indicating good agreement. ICCs for the detection of "individual seizure episodes" and "duration of individual seizures" were 0.61 (95% CI: 0.54 - 0.67) and 0.60 (95%CI: 0.52 - 0.66) respectively, both indicating moderate degrees of agreement. Inter-rater agreement was good (ICC 0.87; 95%CI: 0.83 - 0.90) for true positive seizures.

Conclusion: aEEG has better reliability for the detection of "infant with seizures" than "individual seizure episodes". aEEG may not be a reliable method for the detection of "individual seizure episodes" in infants at risk of seizures.

15:00-15:30 AFTERNOON TEA

15:30 MAURICE SPILLANE’S HYPOTHETICAL: YOUR HEALTH DOLLAR: WHO DESERVES IT AND WHO DECIDES?
Panel discussion
Annie Bye, Victor Cheng, Christopher Etherton-Beer, Richard Webster, Chris Troedson, Maina Kava

17:00 ANZCNS AGM
## THURSDAY 13TH SEPTEMBER

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<td>07:45</td>
<td><strong>Breakfast Symposium</strong></td>
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<td>Past, present and future of management of Batten disease (CLN2)</td>
<td>Nicola Specchio</td>
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<td>09:00</td>
<td><strong>Symposium - Epilepsy, Brain &amp; Behaviour (Chair: Stephen Malone and Jon Silberstein)</strong></td>
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<td>Epilepsy, Inflammation and the Development of Comorbidities</td>
<td>Raman Sankar</td>
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<td>Non-Invasive Brain Stimulation in Epilepsy</td>
<td>Soumya Ghosh</td>
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<td>Advances in Autism</td>
<td>John Wray &amp; Andrew Whitehouse</td>
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<td>Juvenile Justice System and the Brain</td>
<td>Raewyn Mutch</td>
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<td>11:00</td>
<td><strong>Morning tea</strong></td>
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<td><strong>Late Morning Session</strong></td>
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<td>11:30</td>
<td><em>(Chair: Lakshmi Nagarajan &amp; Mark MacKay)</em></td>
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<td>Pathophysiology of Status Epilepticus</td>
<td>Raman Shankar</td>
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<td>Vagal Nerve Stimulation in Treatment Resistant Epilepsy in Childhood</td>
<td>Nicola Specchio</td>
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<td><strong>Lunch</strong></td>
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<td><strong>Symposium - Foetal Neurology (Chair: Hugo Sampaio &amp; Twinkle Ghia)</strong></td>
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<td>Prenatal counselling for Foetal Brain Malformations</td>
<td>Jan Dickinson</td>
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<td>MRI Imaging in Foetal Neurology</td>
<td>Laura Fender</td>
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<td>Foetal Cortical Malformation Disorders and Genes</td>
<td>Richard Leventer</td>
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<td>Causal Pathways in Brain Malformation Disorders</td>
<td>Julian Heng</td>
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<td><strong>Late Afternoon Session</strong></td>
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<td><strong>Platform Session 2 (Chair: Clair Pridmore &amp; Erik Andersen)</strong></td>
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<td>Treatment of late-infantile neuronal ceroid lipofuscinosis type 2 with cerliponase alfa; local experience</td>
<td>Sara Curnow</td>
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<td>Epilepsy and its management in CDKL5 Deficiency Disorder</td>
<td>Helen Leonard</td>
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<td>Language Outcomes in Children after Resective Brain Surgery for Childhood Epilepsy</td>
<td>Jean Bailey</td>
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<td>Can ESES be picked up on a standard outpatient EEG?</td>
<td>Snehal Shah</td>
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<td>19:00</td>
<td><strong>ANZCNS Conference drinks &amp; dinner</strong> - Fraser’s Function Centre, Kings Park</td>
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07:30  REGISTRATION DESK OPEN

07:45-08:30  BREAKFAST SYMPOSIUM

07:45  Past, present and future of management of Batten disease (CLN2)
   Nicola Specchio

09:00-11:00  SYMPOSIUM - EPILEPSY, BRAIN & BEHAVIOUR

09:00  Epilepsy, Inflammation and the Development of Comorbidities
   Raman Sankar

There is increasing recognition that the neuropsychiatric and neurocognitive comorbidities of epilepsy have a larger impact on the Quality of Life than the frequency of seizures per se. While socioeconomic and treatment-related factors influence these comorbidities substantially, there is also a strong neurobiological connection between epilepsy and its comorbidities, especially mood disorders and attention deficit/impulsivity disorders. By modeling these comorbidities in experimental rodent models of epilepsy, we show that inflammatory pathways involving interleukin signaling provide a crucial link. These studies also reveal potential novel targets for pharmacotherapy.

09:45  Non-Invasive Brain Stimulation in Epilepsy
   Soumya Ghosh

10:05  Advances in Autism
   John Wray & Andrew Whitehouse

10:30  Juvenile Justice System and the Brain
   Raewyn Mutch

11:00-11:30  MORNING TEA

11:30-12:30  SYMPOSIUM - FOETAL NEUROLOGY

11:30  Prenatal counselling for Foetal Brain Malformations
   Jan Dickinson

12:00  MRI Imaging in Foetal Neurology
   Laura Fender

14:00  Foetal Cortical Malformation Disorders and Genes
   Richard Leventer

14:00  Causal Pathways in Brain Malformation Disorders
   Julian Heng
Treatment of late-infantile neuronal ceroid lipofuscinosis type 2 with cerliponase alfa; local experience
Sarah R Curnow 1, Victoria M Rodriguez-Casero 1, Brian Lilley 1, Monique M Ryan 1
(1) Royal Children’s Hospital, Parkville, VIC

Background: Cerliponase alfa has recently been shown to be a safe and effective treatment for late-infantile neuronal ceroid lipofuscinosis (CLN2, Batten disease).1,2 This enzyme replacement therapy is administered fortnightly via the intracerebroventricular route. Although approved for use overseas, cerliponase alfa has not yet received regulatory approval, or national funding in Australia.3 We report the local experience of administration and tolerance of this agent to two Victorian children treated under the Therapeutic Goods Administration’s Special Access Scheme.

Methods: This is an observational review.

Results: Two boys, patient A (aged 5.5 years), and patient B (aged 3.75 years), with genetically proven CLN2 have been treated with cerliponase alfa via a Rickham reservoir at The Royal Children’s Hospital (RCH) for 10 and 7 months, respectively. Prior to treatment in Australia, Patient A had received this agent for 14 months in Italy through a compassionate access scheme. Since commencement of treatment at RCH, both children have shown slowed disease progression compared to natural history,4 with stable disease-specific severity scores and complete seizure control, as well as a small improvement in visual acuity for patient A. No significant infusion or device related complications have occurred. The agent itself is almost prohibitively expensive and requires significant hospital-based resources.

Conclusion: Early experience with use of cerliponase alfa for the treatment of CLN2 in Australia suggests that this is an extremely resource-intensive but effective treatment for an otherwise incurable neurodegenerative disease of early childhood.

References:
(3) Markham A. Cerliponase Alfa; First Global Approval. Drugs 2017; 77: 1247-1249
16:15

Epilepsy and its management in CDKL5 Deficiency Disorder: what have we learned from data contributed to the International CDKL5 Database

Helen Leonard 1, Jenny Downs 1, Kingsley Wong 1, Zhan Lim 1, Amy Epstein 1, Stephanie Smith 1, Keely Bebbington 1, Scott Demarest 2

(1) Telethon Kids Institute, West Perth, WA; (2) Child Neurology, University of Colorado, Denver, Colorado, USA

Background: The CDKL5 Deficiency disorder (CDD) is defined primarily by its genetic aetiology, a pathogenic mutation in the CDKL5 gene. Hallmark features include early-onset epilepsy and severe neurodevelopmental impairment. The refractory nature of the epilepsy and the burden on the individual and their family compared with other epileptic encephalopathies is considerable but not well documented in the literature.

Methods: Established in 2012, the International CDKL5 Disorder Database collects information from families/caregivers of a child with CDD through completion of online questionnaires. Data on epilepsy frequency and management will be presented on 240 individuals with a pathogenic mutation. Grouping of individual CDKL5 mutations was based on their predicted structural and functional consequences.

Results: The median age at seizure onset was 6 weeks (range 1 week – 1.5 years) and did not vary by mutation group. Almost two thirds had 1-5 seizures a day or more, and only 8% were seizure free with little improvement with age. Relationships between genotype and functional abilities and seizure frequency were observed. Most were on polypharmacy with almost 45% on three or more medications. Half had been on the ketogenic diet for a median duration of 17 months and 17% had had a vagal nerve stimulator (VNS) inserted.

Conclusions: The burden of epilepsy in CDD is significant. Cross-sectional data shows no improvement with age. Seizures appear to be refractory to anti-epileptic medications although the ketogenic diet and VNS may provide some benefit. There is an urgent need for new and novel therapies for epilepsy management.

16:30

Language Outcomes in Children after Resective Brain Surgery for Childhood Epilepsy

Jean Bailey 1, Lakshmi Nagarajan 1, Soumya Ghosh 1, Alison Buckland 2

(1) Perth Children’s Hospital, WA; (2) Sir Charles Gairdner Hospital, Perth, WA

Background: Epilepsy surgery, though a relatively safe and effective treatment for selected children, may be associated with neurological sequelae including language dysfunction. In this study we evaluated language function in 45 of 59 children who underwent epilepsy surgery at our centre between 1997 and 2016.

Methods: Children were aged between 18 months and 18 years at the time of surgery. We compared preoperative language function with postoperative language outcomes at intervals of between six months, two and four years postoperatively.
36 children, with Performance IQ greater than 70 (PIQ>70), underwent comprehensive standardised language assessment including the Clinical Evaluation of Language Fundamentals, Test of Language Competence or the Preschool Language Scales. Changes in performance by more than one standard deviation were considered meaningful. Single word receptive and single word expressive vocabulary assessment were administered to the 9 children with PIQ<70.

Results: There were 31 temporal, 7 frontal, 2 parietal, 1 parieto-occipital, 1 temporo-parietal resections, and 3 hemispherectomies.

Of the children with PIQ>70, 24 showed no change, 8 improved, and 4 (all temporal resections) showed a decline in language function postoperatively. Of the 4 children with decline in core language performance, 2 showed improvements in receptive language. Of the 9 children with PIQ<70, 7 remained unchanged and two had improved.

Conclusion: Speech-language remained stable or improved in the majority (91%) of children who underwent resective brain surgery for epilepsy.

16:45

Can ESES be picked up on a standard outpatient EEG?

Snehal Shah¹, Lakshmi Nagarajan¹, Peter Walsh¹, Jonathan Silberstein¹, Linda Palumbo¹, Soumya Ghosh³

¹Perth Children's Hospital, Floreat, WA

Introduction: Electrical Status Epilepticus in Sleep (ESES) was first described in 1971 and later classified by Tassinari et al. ESES is defined when 80% of slow wave sleep has epileptiform activity. The terms continuous spike wave in slow-wave sleep (CSWS) and Landau-Kleffner syndrome (LKS) describe the clinical epileptic syndromes seen with ESES. Although long-term outcome of epilepsy in ESES is favourable, the prognosis is guarded because of the persistence of severe neuropsychological and/or motor deficits in approximately one-half of patients. We aim to study if routine EEG will be a good screening tool to predict ESES in children.

Method: The database of EEG reports of patients from 2000-2014 was searched for terms “Electrical status epilepticus of sleep”, “ESES”, “CSSW” “continuous spike and wave during slow wave sleep”. The clinical and EEG profile of children identified were analysed.

Results: 25190 EEGs were undertaken during 2000-2014 in children of 1 to 18 years of age. Sleep recording was obtained in 19,396 (77%) of patients. 39 children with EEG suggestive of SES spectrum were identified. At the time of EEG that identified SES spectrum, 32/39 were on AEDs. Overnight V EEG monitoring was undertaken in 17 patients within 6 months of “SES spectrum” suspected on routine EEG. ESES was confirmed in all seventeen.

Conclusion: A routine EEG with sleep recording is a good screening tool for SES. An overnight study confirmed ESES in all the children in whom it was undertaken, after the routine EEG raised the possibility of SES spectrum.
19:00  ANZCNS CONFERENCE DINNER
Fraser’s Function Centre, Kings Park
Pick up from the Pan Pacific Perth Hotel at 6.45pm
Pick up from Fraser’s Function Centre to the Pan Pacific Perth Hotel at 10.30pm
# FRIDAY 14TH SEPTEMBER

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<td><strong>Symposium - Neurogenetics and Beyond (Chair: Richard Webster &amp; Peter Rowe)</strong></td>
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<td>Application of Genetics to Epilepsy after Gene-Hunting</td>
<td>Shinichi Hirose</td>
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<td>Application of Genetics to Neuromuscular disorders</td>
<td>Phillipa Lamont</td>
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<td>Arthrogryposis and Genes</td>
<td>Gina Ravenscroft</td>
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<td>A picture is worth a thousand genomes</td>
<td>Gareth Baynam</td>
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<td>Late Morning Session</td>
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<td><strong>Platform Session 3 (Chair: Harry Singh &amp; Maina Kava)</strong></td>
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<td>FaceMatch: a secure portal providing automated face-matching for individuals with undiagnosed intellectual disability</td>
<td>Tracy Dudding-Byth</td>
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<td>Extreme clinical variability in centronuclear myopathy with BIN1 mutations</td>
<td>Macarena Cabrera</td>
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<td>Abnormalities of fatty acid oxidation as potential candidate biomarker for spinal muscular atrophy (SMA)</td>
<td>Maina Kava</td>
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<td>Whole genome sequencing in persistently unsolved white matter disorders</td>
<td>Guy Helman</td>
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<td>13:30</td>
<td>(Chair: Snehal Shah &amp; Jon Silberstein)</td>
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<td>Stroke guidelines - the next step: Implementation</td>
<td>Mark Mackay</td>
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<td>Diagnostic Dioramas</td>
<td>Snehal Shah &amp; Jon Silberstein</td>
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<td>15:00</td>
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08:00 REGISTRATION DESK OPEN

08:30-10:30 SYMPOSIUM - NEUROGENETICS AND BEYOND

08:30 Application of Genetics to Epilepsy after Gene-Hunting
Shinichi Hirose, Department of Pediatrics, School of Medicine and Central Research Institute for the Molecular Pathomechanisms of Epilepsy, Fukuoka University, Fukuoka, Japan

In the last 20 years, genetics of epilepsy has been targeting to discover new genes associated with mostly monogenic epilepsies, i.e., “Gene Hunting”. Recently, however, the increment of the number of newly discovered genes has become sluggish. Conversely, current technologies, such as next generation sequencing, iPS cells, and gene editing, are rapidly making the genetic information applicable to diagnosis and developing novel rational therapies for epilepsy. Thus, genetics of epilepsy has entered a new era with the clinical application.

09:15 Application of Genetics to Neuromuscular disorders
Phillipa Lamont

09:40 Arthrogryposis and Genes
Gina Ravenscroft

10:05 A picture is worth a thousand genomes
Gareth Baynam

10:30-11:00 MORNING TEA

11:00-12:00 PLATFORM SESSION 3

11:00 FaceMatch: a secure portal providing automated face-matching for individuals with undiagnosed intellectual disability
Tracy E Dudding-Byth¹,²,³, Jackie Boyle³, John Attia³,²,³, Carlos Riveros², Anna Hackett³, Anne Baxter³, Sheridan O’Donnell³, Brian Lovell⁴
(¹) University of Newcastle, Newcastle, NSW; (²) Hunter Medical Research Institute, Newcastle, NSW; (³) Hunter New England Area Health Service, Newcastle, NSW; (⁴) University of Queensland, Brisbane, QLD

Background: Despite advances in genomic sequencing, 60-70% of individuals with moderate to severe intellectual disability [ID] remain undiagnosed. Many individuals with ID have facial features which can provide a clue to diagnosis, and the discovery of novel ID genes typically requires confirmation of candidate gene variants in multiple unrelated individuals with a similar phenotype. Historically, clinical geneticists manually presented images of undiagnosed patients to their colleagues. The matchmaker exchange initiative now enables sharing of Human Phenome Ontology [HPO] terms and relevant candidate gene information, but there is no ability to match on facial images.

Aim: Our aim was to establish an international portal providing automated face-matching for individuals with undiagnosed moderate to severe ID.

Methods: FaceMatch design and development was undertaken by our
multidisciplinary team with parent and doctor engagement.

**Outcome:** The parent empowering model offers dual parent/doctor participation, which can be initiated by a parent or a doctor. The secure child profile page contains images, medical history, and genetic information. Consent and email notifications are within the system. Using advanced IMAGUS face recognition technology, FaceMatch compares every new image automatically against images within the facebase, generating a matching score and ranking list. The FaceMatch team notify participating doctors of high-probability facial matches. We are also part of the international Minerva facial phenotype consortium.

**Conclusion:** FaceMatch [https://facematch.org.au/] is the first of its kind to provide automated face-matching for individuals who remain undiagnosed following exome or genome sequencing. Ongoing research includes the incorporation of HPO terms into the matching algorithm.

**Reference:**

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**11:15 Extreme clinical variability in centronuclear myopathy with BIN1 mutations**


(1) Harry Perkins Institute of Medical Research, Nedlands, WA; (2) Instituto de Biomedicina de Sevilla, Hospita Virgen del Rocio, Sevilla, Spain; (3) Anatomia Patologica, Hospital Virgen del Rocio, Sevilla, Spain; (4) Hospital Virgen de las Nieves, Granada, Spain; (5) Hospital 12 de Octubre, Madrid, Spain; (6) PathWest Laboratory Medicine, Perth, WA; (7) Universidad Pompeu Fabra, Barcelona, Spain; (8) University of Western Australia, Perth, WA

**Background:** Background: Centronuclear myopathy is a congenital myopathy named after its very specific pathological findings. Two different clinical presentations have been described; a severe congenital onset phenotype which is most commonly associated with MTM1 mutations known as Myotubular myopathy, and a milder phenotype with later onset during childhood or adulthood, frequently associated with BIN1, RYR1 or DNM2 mutations. Congenital presentation of BIN1 mutations is extremely rare.

**Methods:** We have screened all molecularly unresolved myotubular myopathy cases from two major neuromuscular reference centres in Spain for mutations in CNM genes using NGS and Sanger sequencing. DM1 has been excluded. Haplotype analysis was performed to study relatedness of patients carrying the same mutations.

**Results:** Eight cases were identified, fulfilling myotubular myopathy pathological criteria, and severe disease with early death. Seven carried MTM1 mutations including two missense and one splicing novel mutations. One female patient had compound heterozygous R234C/R145C BIN1 mutations. Interestingly, the same mutations had
previously been identified in an apparently unrelated adult CNM patient of Roma background, who presented in her childhood with mild proximal limb weakness and myotonia. Haplotype analyses demonstrated a common ancestor for each of the two mutations in both patients.

Conclusions: We have identified compound heterozygous BIN1 mutations presenting with extreme clinical variability in related patients, from a myotubular myopathy with catastrophic outcome to a late childhood onset disease with mild proximal weakness. It is possible that other genetic or non-genetic modifying factors are involved. BIN1 mutations should be considered in patients with myotubular myopathies without MTM1 mutations.

11:30 Abnormalities of fatty acid oxidation as potential candidate biomarker for spinal muscular atrophy (SMA)

Maina P Kava 1,2,3, Jiri Vajsar 4, Lianna Kyriakopoulou 5, Ingrid Tein 4

1) Metabolics and Rheumatology, Perth Children’s Hospital, Perth, WA; 2) School of Paediatrics and Child Health, University of Western Australia, Perth, WA; 3) Neurology, Perth Children’s Hospital, Perth, WA; 4) Neurology, The Hospital for Sick Children, Toronto, Ontario, Canada; 5) Metabolic Diseases Laboratory, The Hospital for Sick Children, Toronto, Ontario, Canada

Background: Abnormalities of fatty acid oxidation, such as dicarboxylic aciduria (DCA) and increased serum esterified carnitine (Cn), have been demonstrated in SMA children and may correlate with disease severity due to denervation.

Objective: To stratify SMA severity, and to test whether increased serum esterified Cn, decreased serum free Cn, and abnormalities in serum acylcarnitine profile, correlate with disease severity.

Methods: 13 SMA (1 SMA I, 10 SMA II, 2 SMA III; 9F, 4M) and 12 age-matched axonal Charcot Marie Tooth (CMT) patients (8F, 4M) were recruited. All underwent modified Hammersmith functional motor scale, neuromuscular disease QOL questionnaire, serum Cn (total and free), acylcarnitine profile and urine organic acids. SMA patients also had pulmonary functions and CMAP studies. Age- and sex-matched standardised control biochemical data were obtained. Comparisons were performed using t-test and ANOVA.

Results: Median age of SMA patients was ~8 years (13 months – 17 years). 6 SMA patients showed reduced serum free Cn and 3 in total Cn. 2 CMT patients showed reduced serum free and total Cn. All acylcarnitine profiles were normal except for increased C18:1 in one SMA II. One SMA I showed mild DCA, small 3-methylglutaconic acid, and 3-hydroxyisovaleric acid; 2 SMA II showed small to moderate 3-hydroxyisovaleric and trace methylglutaconic acids. All CMT patients showed normal organic acid profiles. None showed increased serum esterified Cn.

Conclusions: A larger cohort will be needed to determine whether serum Cn profile may prove a useful biomarker in SMA for disease severity and response to nusinersen.
Whole genome sequencing in persistently unsolved white matter disorders

Guy Helman ¹,², Bryan R Lajoie ³, Joanna Crawford ⁴, Amy Pizzino ⁴, Johanna Schmidt ⁴, Asako Takanohashi ⁴, Cas Simons ¹,², Ryan J Taft ³, Adeline Vanderver ⁴

(¹) Murdoch Children’s Research Institute, Parkville, VIC; (²) University of Queensland Institute for Molecular Biosciences, Brisbane, QLD; (³) Illumina, Inc., San Diego, CA, United States; (⁴) Department of Neurology, Children’s Hospital of Philadelphia, Philadelphia, PA, United States

Background: Next-generation sequencing (NGS) has become a mainstay in the diagnostic evaluation of a patient with a neurologic disorder of unknown origin. We performed a systematic study of the efficacy of genome sequencing (GS) on a series of persistently unresolved cases of genetic leukoencephalopathy with prior comprehensive clinical, biochemical, and molecular testing.

Methods: GS was used to interrogate 41 families (trio or greater! unresolved after previous exome sequencing (ES) and standard-of-care testing from an original cohort of 191 families. Sanger sequencing or microarray analysis was used to validate and perform segregation analysis of all candidate mutations.

Results: GS identified variants that enabled resolution of 14/41 (34%) families in our persistently unsolved cohort. Reanalysis in the context of improved variant curation and annotation (n=4) and novel disease-associated genes (n=4) resolved the majority of cases. The remainder were due to the identification of variants missed by other approaches including copy number variants (n=3), mitochondrial genome variants (n=1), and variants in technically difficult regions (n=2). 27 cases remain unsolved after GS, including three with potential candidate variants in novel disease genes. NGS approaches diagnosed 62% of cases (44/71).

Conclusion: Genetic white matter disorders have heterogeneous etiologies and overlapping clinical presentations. In most cases, a comprehensive approach provides the highest likelihood of diagnosis in prospective and historical cohorts. We present a genomic evaluation of a historical cohort of patients with genetic leukoencephalopathies, where all clinically-available approaches yielded a diagnostic success rate of 84%, well above historical norms of fewer than fifty percent of cases.
POSTERS

1. Evidence of Central Nervous System Inflammation with Elevated CSF Neopterin in Children with Severe Influenza-Associated Neurological Disease
   Emma Macdonald-Laurs, Phillip Britton, Archana Koirala, William Rawlinson, Chee Hiew, Jocelynne Mcrae, Rusell Dale, Cheryl Jones, Kristine Macartney

2. Vagus nerve stimulation for the treatment of refractory epilepsy in the CDKL5 Deficiency Disorder
   Zhan Lim, Kingsley Wong, Jenny Downs, Keely Bebbington, Scott Demarest, Helen Leonard

3. Interleukin-1 receptor antagonist (Anakinra) treatment in mitochondrial epilepsy: A case study of epilepsy partialis continua and super refractory status epilepticus
   Caroline Stewart, Emma Macdonald-Laurs, Katie Frith, Michael Cardamone

4. Clinical use, effectiveness and tolerability of rufinamide in children with treatment resistant epilepsy in childhood: an experience from an Australian centre
   Ellen MacKinnon, Lakshmi Nagarajan, Shalini Kassam

5. Djinn possession and exorcism of a teenage girl
   Kathryn Irving

6. Deafness Dystonia cerebral hypomyelination syndrome due to likely pathogenic homozygous mutation in BCAP31 gene
   Twinkle Ghia, Jonathon Silberstein, Jane Valentine, Tessa Dadd, Karen Carpenter, Ben Kamien

7. Anti HMG-Co A reductase antibodies associated with progressive necrotizing myositis and autophagosomes in a teenager
   Eleanor Ng, Maina Kava, Peter Rowe, Simon Williams, Phillipa Lamont, Rei Junckerstorff

8. Fatal neonatal lactic acidosis secondary to a novel defect in COX16 gene causing complex IV deficiency
   Maina Kava, Shanti Balasubramaniam, Barry Lewis, Richard Rodenburg

9. Pathway to genetic diagnosis: Characterising recessive MYH7-related myopathy in two families
   Sarah J Beecroft, Emily Oates, Martijn van de Locht, Coen Ottenheijm, Caroline Sewry, Shehla Mohammed, Monique Ryan, Ian Woodcock, Lauren Sanders, Rebecca Gooding, Mark Davis, Nigel Laing, Gianina Ravenscroft, Heinz Jungbluth, Catriona McLean

10. Lysine- restricted diet in a child with pyridoxine -dependant epilepsy prevents long term developmental delay
    Maina Kava, Leah Queit, Barry Lewis, Lawrence Greed, Peter W Rowe, Shanti Balasubramaniam

11. #1268 Summary Leigh-like syndrome due to homoplasmic m.8993T>G variant with hypocitrullinemia and unusual biochemical features suggestive of multiple carboxylase deficiency (MCD)
    Maina Kava, Barry Lewis, Lawrence Greed, Shanti Balasubramaniam

12. Presently, there are no clinically affective neuroprotective agents
    Neville Knuckey
13 Complete neurological recovery following severe primary pontine haemorrhage in a 3 year old
   Darshan Das, Deepak Gill, Brian Owler

14 PLEDs in children
   Snehal Shah, Lakshmi Nagarajan, Soumya Ghosh

15 Epidemiology of gastrostomy insertion for children and adolescents with intellectual disability
   Jenny Downs, Kingsley Wong, Helen Leonard, Emma Glasson, Lakshmi Nagarajan, David Forbes, Andrew Wilson, Madhur Ravikumara, Peter Jacoby, Jenny Bourke, Preeyaporn Srasuebkul, Julian Trollor

16 Clinical, MRI and electrographic characteristics of four children with HSV encephalitis and secondary immune activation
   Yeeshu Singh, Anita Cairns, Geoff Wallace, Kate Sinclair, Stephen Malone
See you in Sydney for the
8th ANZCNS Annual Scientific Meeting
August 2019