Cover photograph: Purkinje neurons. Golgi apparatus © Jcalvo | Dreamstime.com
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WELCOME MESSAGES

Welcome Message From The Local Organisers

We welcome you to the 8th Annual Scientific Meeting of the Australia and New Zealand Child Neurology Society. The theme of this year’s conference is ‘Precision medicine and novel treatments in childhood neurological disorders’. Our distinguished international speakers - Prof Anne Connolly (USA), Prof John Sharon (USA) and Prof Francesco Pisani (Italy) along with an illustrious line-up of speakers from Australia and New Zealand will discuss the latest advances in epilepsy, neuromuscular medicine, neurogenetics and movement disorders. Posters and Platforms presentations will showcase research in childhood neurological disorders being led by researchers in Australia and New Zealand.

We also welcome your participation in the Neonatal EEG course, which will be led international and national experts in neonatal EEG.

The content of the sessions will be of interest to general paediatricians, sub-specialists with an interest in child neurology, and consultant and trainee child neurologists.

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

Manoj Menezes
ASM Organising Committee 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 2019 Annual Scientific Meeting. The Organising Committee has worked hard to arrange this event at The Sydney Children's Hospital. 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

Local Organizing Committee (LOC)
Manoj Menezes (Chair)
Hugo Sampaio
Michelle Farrar
Alexandra Johnson
Shekeeb Mohammad
Simone Ardern-Holmes

Abstract Review Committee
Simone Ardern-Holmes
Russell Dale
Michelle Farrar
Alexandra Johnson
Mark MacKay
Manoj Menezes
Shekeeb Mohammad
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
c/- Seed Events Pty Ltd, PO Box 2137, Glenelg SA 5045, Australia
e: admin@anzcns.org.au
m: +61 423 827 488

The Conference Venue

Sydney Children’s Hospital at Randwick
Level 1, John Beveridge Lecture Theatre
The Sydney Children’s Hospital, Randwick, is located in High Street, Randwick.

Parking
- The car park is open 24 hours a day, 365 days of the year
- Entry to the car park is via the roundabout on Barker Street, Randwick
- Once you have parked your car, take the lifts located at the back of the car park to Level 0
- Enter the main doors of Prince of Wales Campus Centre, then follow the signs to Sydney Children’s Hospital, Randwick

The car park is managed by Metro Parking. For more information, please call (02) 9382 3400.

Parking rates (effective 1 July 2018)
- 0.0 – 0.5hrs $5.00
- 0.5 – 1.0hrs $10.00
- 1.0 – 1.5hrs $15.00
- 1.5 – 2.0hrs $20.00
- 2.0 – 2.5hrs $25.00
- 2.5 + hrs $30.00
- 5-day visitor pass $90.00

Restricted parking
Parking spaces with the signage ‘PERMIT HOLDERS ONLY’ are restricted to STAFF permit holders ONLY.
Registration Desk

The registration desk is located on Level 1 of the Sydney Children’s Hospital. Please note that name badges must be clearly displayed at all times.

Registration desk opening hours are:
Wednesday 4th September: 08:30-17:00
Thursday 5th September: 08:00-17:00
Friday 6th September: 08:00-17:00

Exhibition

A trade exhibition is held in conjunction with the 8th 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 John Beveridge Lecture Theatre Foyer, Level 1, 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 the Foyer. Any delegates with particular catering needs should have notified these on their online registration form and will have their special meals labelled.

Mobile/cell phones

Mobile/cell phones must be on silent or 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 8th ANZCNS Annual Scientific Meeting is primarily for health professionals. Community and consumer support groups have been welcomed to the 8th 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 8th 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

The Conference Dinner will be held at the Centennial Homestead, Wisteria Room.

Date:  Thursday 5th September
Time:  7.00pm for pre-dinner drinks
Dinner: 7.30 - 10.30pm

Transport to and from the dinner venue will be provided. Transfer schedule is as below;
Pick up from the Crowne Plaza Coogee Beach Sydney Hotel at 6.30pm
Pick up from Centennial Homestead to the Crowne Plaza Coogee Beach Sydney Hotel at 10.30pm
The Australia and New Zealand Child Neurology Society wish to extend their thanks to our major sponsors who have supported this event:

MAJOR SPONSORS

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### INTERNATIONAL SPEAKER SPONSOR

![Biogen Logo]

### KEYNOTE SPEAKER SPONSOR

![Cerebral Palsy Alliance Logo]

### EXHIBITORS

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INTERNATIONAL SPEAKERS

We welcome our International Speakers:

Anne M. Connolly, MD, is chief of the Division of Neurology at Nationwide Children’s Hospital, professor of Pediatrics at The Ohio State University College of Medicine, and a member of the Center for Gene Therapy in The Research Institute at Nationwide Children's. An internationally recognized expert in pediatric neuromuscular disease and neuroimmunology, Dr. Connolly's early research examined the relationship between auto-antibodies and childhood neurological disorders. She has also help develop outcome measures for infants and boys with Duchenne muscular dystrophy (DMD) and has led or been a collaborator in numerous pivotal clinical trials in children with neuromuscular disorders. She has served as a national thought leader for multiple foundation and government agency efforts and has specifically contributed to efforts to achieve newborn screening for neuromuscular disorders including SMA and DMD. She is an American Academy of Neurology Fellow, and member of the Child Neurology and the World Muscle Society. Named among the “Best Doctors in America,” Dr. Connolly has also participated in numerous national and international work groups for the development of standards of care for SMA, DMD, and CMD.

John Svaren, Ph.D. is a Professor at the University of Wisconsin-Madison and a member/core-director in the Waisman Center, which is dedicated to research in developmental disabilities. His laboratory focuses on gene regulation of peripheral nerve myelination and translational research for the heritable neuropathy Charcot-Marie-Tooth Disease (CMT). The latter studies include development of drug screening assays, biomarker development, and genomic/epigenomic studies of peripheral neuropathy, with a focus on the most prevalent form of CMT (CMT1A). He is a member of the board of the Charcot-Marie-Tooth Association, and chair of its scientific advisory board.

Francesco Pisani is Associate Professor and head of the Child Neuropsychiatry Unit at the University of Parma Hospital. Francesco's training and experience in Child Neurology has been in Italy and the UK. He is the author of more than a hundred publications. Prof Pisani's research interest covers a spectrum of childhood neurological disorders: he is internationally acclaimed and recognised for his work on neonatal seizures and neonatal neurology. Prof Pisani is also active in teaching neonatal neurology and neonatal EEGs across Europe.
Every child is born with 100 billion neurons in their brain.

Let’s put them to work and

 CPA Early Childhood Intervention

Every child is born with 100 billion neurons in their brain.

100 billion... that’s a galaxy worth of nerve cells ready to start exchanging electrical impulses and creating neural pathways. And the earlier they start doing that, the stronger the foundations for a child’s learning and development.

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1300 888 378  ask@cerebralpalsy.org.au
 cerebralfpalsy.org.au
# WEDNESDAY 4TH SEPTEMBER

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<td><strong>ANZCNS Board Meeting</strong></td>
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<td><strong>ANZCNS Trainee Breakfast Meeting</strong></td>
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<td><strong>Trainee Lecture</strong></td>
<td>Annie Bye</td>
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<td><strong>Symposium - Epilepsy and Neurodevelopment - Chair: Stephen Malone</strong></td>
<td>Ingrid Scheffer</td>
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<td>Precision medicine in focal epilepsies: Hype or reality?</td>
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<td>Instructive cases of childhood focal epilepsy</td>
<td>Deepak Gill &amp; Sachin Gupta</td>
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<td><strong>Lunch</strong></td>
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<td><strong>ANZCNS Paediatric Neurology Trainee meeting</strong></td>
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<td>Abusive Head Trauma</td>
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<td><strong>Symposium - Epilepsy and Neurodevelopment (continued) - Chair: Claire Pridmore</strong></td>
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<td>Inflammation and neurodevelopment</td>
<td>Russell Dale</td>
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<td>Localising focal epilepsies using seizure semiology</td>
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<td>15:45</td>
<td><strong>Platform Session 1 - Epilepsy and Neurodevelopment - Chair: Claire Spooner</strong></td>
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<td>17:00</td>
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08:30 REGISTRATION DESK OPEN
09:00 ANZCNS BOARD MEETING
09:00 ANZCNS TRAINEE BREAKFAST MEETING
09:30 CONFERENCE OFFICIAL OPENING (John Beveridge Lecture Theatre)
10:00 TRAINEE LECTURE
Prof Annie Bye, Sydney Children’s Hospital, Randwick
10:45-12:15 SYMPOSIUM - EPILEPSY AND NEURODEVELOPMENT
10:45 Precision medicine in focal epilepsies: Hype or reality?
Prof Ingrid Scheffer, The University of Melbourne
11:30 Instructive cases of childhood focal epilepsy
Drs Deepak Gill & Sachin Gupta, The Children’s Hospital at Westmead
12:15-13:00 LUNCH
13:00 SCH GRAND ROUNDS
Abusive Head Trauma
Dr Lydia Garside, Sydney Children’s Hospital, Randwick
14:15-15:15 SYMPOSIUM - EPILEPSY AND NEURODEVELOPMENT (continued)
14:15 Inflammation and neurodevelopment
Prof Russell Dale, The Children’s Hospital at Westmead
14:45 Localising focal epilepsies using seizure semiology
Dr Chong Wong, The Children’s Hospital at Westmead
15:15-15:45 AFTERNOON TEA
15:45-17:00 PLATFORM SESSION 1: EPILEPSY AND NEURODEVELOPMENT
15:45 The likelihood and timing of seizures during prolonged continuous video-electroencephalography (cEEG) in neonates at risk of seizures
Emma Macdonald-Laurs 1,2, Cia Sharpe 3, Mark Nespeca 3,4, Nelly Rismanchi 3,4, Jeff Gold 3,4, Sonja Wang 3,4, Rachael Kuperman 5, Andrew Mower 6, Richard Haas 3,4, Peter Reed 7, Suzanne L Davis 8, Minh Le 8
(1) Paediatric Neurology, Royal Children’s Hospital, Parkville, Melbourne, VIC, Australia; (2) Paediatric Neurology, Starship Children’s Hospital, Auckland, New Zealand; (3) Department of Neurosciences, University of California, San Diego, San Diego, CA, USA; (4) Rady Children’s Hospital, San Diego, USA; (5) Paediatric Neurology, UCFS Benioff Children’s Hospital, San Francisco, CA, USA; (6) Department of Neurology, Children’s Hospital Orange County, Orange, CA, USA; (7) Statistice, Starship Children’s Hospital, Auckland, New Zealand; (8) Department of Neurosciences, Sharp Mary Birch Hospital for Women and Newborns, San Diego, CA, USA

Background: While continuous video-electroencephalography (cEEG) monitoring for 24 hours is recommended as standard-of-care for neonates at risk of seizures, reviewing this continuously in order to ascertain when to start treatment is not always feasible. We hypothesise that, similar to older children in the ICU, risk of seizure decreases with increased time seizure-free, with the possible exception of neonates.
receiving cooling for hypoxic ischemic encephalopathy (HIE).4.

Methods: 268 term neonates identified as being at-risk of seizures, including 173 who had HIE, were enrolled in the NEOLEV2 trial. Time to first seizure was identified and Kaplan-Meier survival analyses with log-rank testing were used to analyse time to seizure for all-comers, and neonates HIE with and without cooling.

Results: 100/268 (37%) had seizures. Seizures occurred during the first hour in 56/100 (56%), and within 24 hours in 95/100 (95%) for all-comers vs. 23/53 (44%) and 51/53 (96%) for HIE. Median time to seizure was 1.08 (IQR=0.08-5.7) hours for all-comers and 2.33 (IQR=0.05-9.7) hours for HIE. The risk of seizure decreased with increasing time seizure-free for both groups, however neonates with HIE remained at risk longer than those without (Log-Rank 13.2, p=0.0003). This trend was even more apparent among neonates who were cooled (Log-Rank 36.3, p<0.0001).

Conclusion: Seizure-risk decreases with duration of time seizure-free. Clinicians should scrutinise the first hour of cEEG carefully as >50% of neonates who seized did so during this time. While the majority of seizures occurred within 24 hours, neonates with HIE, particularly those receiving cooling, may require longer periods of monitoring to detect the first seizure.

References:

15:55

Treatment response to dose escalation of prednisolone in infantile spasms

Winston Dzau ¹, Sally Cheng ², Penny Snell ³,⁴ Michael Fahey ⁵, Ingrid Scheffer ²,³,⁶,⁷,⁸, Simon Harvey ²,³,⁶, Katherine Howell ²,³

(¹) The University of Melbourne, Melbourne, VIC, Australia; (²) Neurology, The Royal Children’s Hospital, Melbourne; (³) Neuroscience Research Group, Murdoch Children’s Research Institute, Melbourne, VIC, Australia; (⁴) Victorian Severe Epilepsy of Infancy Study Group, Melbourne, VIC, Australia; (⁵) Neurology, Monash Health, Melbourne, VIC, Australia; (⁶) Paediatrics, The University of Melbourne, Melbourne, VIC, Australia; (⁷) Austin Health, Melbourne, VIC, Australia; (⁸) Florey Institute of Neurosciences and Mental Health, Melbourne, VIC, Australia

Background: Outcomes in children with infantile spasms (IS) can be improved by treatment which achieves early, sustained spasm
cessation. The widely-used United Kingdom Infantile Spasm Study (UKISS) treatment protocol begins with prednisolone 40mg/day (PNL40), escalating to PNL60mg/day (PNL60) and then vigabatrin (VGB) if spasms continue. The incremental benefit of PNL60 over PNL40 is unknown. If this is minimal, substituting PNL60 with VGB in the treatment pathway may be justified. We investigated the response to PNL60 and hypothesised it would be inferior to that of VGB in the UKISS protocol.

Methods: We conducted a retrospective population-based study of infants with IS and without tuberous sclerosis complex (TSC). The primary outcome was treatment response to PNL40, PNL60 and VGB, defined as spasm freedom at day 42 after treatment commenced. Secondary outcomes were side effects requiring intervention, and outcomes at age two years (spasm and seizure freedom, EEG findings and development).

Results: 123 infants commenced the UKISS protocol. Treatment response to PNL40 (71/123 [58%, CI 49-67%]), PNL60 (12/32 [38%, CI 21-56%]) and VGB (13/30 [43%, CI 26-63%]) were similar. The rate of side effects requiring intervention was comparable between PNL40 (11/123 [8.9%, CI 4.5-15%]), PNL60 (5/32 [16%, CI 5.3-33%]) and VGB (2/30 [6.7%, CI 0.8-22%]). Outcomes at age two years were similar between PNL40, PNL60 and VGB responders.

Conclusion: Treatment response to PNL40, PNL60 and VGB were similar, demonstrating an appreciable benefit of each step of the UKISS protocol. These findings do not support substituting PNL60 with VGB in the IS treatment pathway.

16:05  
CT angiogram in the evaluation of suspected arterial ischaemic stroke

Romain Briest 1, Ian Andrews 1

(1) Sydney Children’s Hospital, Maroubra, NSW, Australia

Background: Thrombolysis and endovascular clot retrieval (ECR) are realistic, beneficial therapeutic options for children with acute ischaemic stroke (AIS). Timely diagnosis is crucial to delivery of these treatments, and depends upon timely, accurate imaging. Given MRI can be difficult to obtain urgently, we have explored CT angiogram (CTA) as the initial imaging for children with suggestive clinical scenarios.

Methods: We prospectively collected demographic and diagnostic data on children with clinical scenarios suggestive of AIS discussed with SCH Neurology from October 2017 to June 2019 for whom CTA was the initial diagnostic imaging modality.

Results: 15 patients had urgent CTA for suspected AIS. 12 presented to external sites and 3 to SCH. Age at presentation ranged from 6 weeks to 16 years. Final diagnoses included: 5 (33%) AIS, 5 first hemiplegic migraine, 1 other complicated migraine, 1 focal status and non-accidental injury, 1 first focal seizure, 1 (presumed) channelopathy and 1 polio-like, acute flaccid paresis. Median time to CTA was 183 minutes (range 81-990). CTA accurately identified large or medium vessel occlusion in
4/5 patients with AIS. For one patient the CTA was initially considered normal, but review revealed carotid dissection. CTA was normal in the non-stroke patients. One stroke patient was treated with tPA and ECR, 2 were treated with ECR alone and one was treated with arterial stenting.

Conclusions: CTA facilitated prompt recognition of patients with occluded large and medium arteries causing AIS. Timely diagnosis allowed acute treatment with thrombolysis and/or endovascular therapy for 4/5 stroke patients.

16:15

Endovascular clot retrieval for acute ischaemic stroke due to basilar artery occlusion in childhood: a case series and literature review

Emma Macdonald-Laurs 1,2, Jason Wenderoth 1,3, Michael Cardamone 1, Hugo Sampaio 1, Ian Andrews 1

(1) Sydney Children's Hospital, Randwick, NSW, Australia; (2) Royal Children's Hospital, Parkville, Melbourne, VIC, Australia; (3) Department of Radiology, Prince of Wales Hospital, Randwick, NSW, Australia

Background: Acute ischaemic stroke (AIS) due to basilar artery occlusion (BAO) is a rare but life-threatening condition1, although the natural history may be more favourable in children2. Endovascular clot retrieval (ECR) is standard of care in large vessel occlusions in the adult population and is increasingly used in children.

Methods: We report four children with AIS due to BAO treated with ECR from 2010-18. We reviewed the literature to characterise a “natural history” cohort of paediatric patients with AIS due to BAO who were not treated with thrombolysis or ECR, and compared them with our patients and 28 in the literature who were treated with ECR.

Results: 3 of 4 children treated with ECR had a good outcome (modified Rankin score (mRS) 0-2). Despite significant delays in diagnosis (median 1245 minutes), ECR achieved recanalisation in all, however one patient with leukaemia suffered recurrent basilar occlusion and died. Literature review identified 112 patients exhibiting the natural history of AIS due to BAO, among whom 44% had a good outcome (mRS 0-2), 46% had significant residual disability (mRS 3-5) and 10% died. Among 34 paediatric patients treated ECR with outcome recorded, 28 (79%) had a good outcome (mRS 0-2), 5 (15%) had significant residual disability (mRS 3-5) and one (3%) died. Recognising the limitations of the paediatric literature, these data suggest significantly improved good outcome when ECR is performed (p=0.0001).

Conclusion: The limited data suggests improved outcome for AIS due to BAO in children treated with ECR, despite considerable delay to intervention.

References:


16:25  
**Early experience of the first paediatric stroke code at an Australian Children's Hospital**

*Micheala Waak 1, Natalie Deuble 1, Louise Sparkes 1, Katrina Ronlund 1, Adriane Sinclair 1*  
(1) Queensland Children's Hospital, South Brisbane, QLD, Australia

**Background:** Arterial ischaemic stroke (AIS) is a time critical neurological emergency. In-hospital delay to diagnosis in children is common. A stroke code was developed aiming to improve time to diagnosis and standardisation of management. Our objective is to describe the first 7 months of stroke code experience with comparison to 24 months pre-code implementation.

**Methods:** An auditing process was established at the time of code implementation. Stroke code and AIS data from January 2019 to July 2019 were reviewed and compared with data from AIS patients between January 2017 and December 2018.

**Results:** Ten stroke codes were activated. Two patients had AIS, 7 had migraine and 1 had autoimmune encephalitis. Four additional patients were diagnosed with AIS but did not meet criteria for code activation (presentation > 24 hours post symptom onset, age). All code patients were triaged in <10 minutes and were assessed by an emergency senior medical officer within 20 minutes. Time to magnetic resonance imaging (MRI) was 20 – 180 minutes (mean 56 min). In all patients MRI was diagnostic and impacted treatment decision. Pre-code 16 outpatients were diagnosed with AIS state-wide however only 3 (< 50% of potentially eligible patients) would have activated a stroke code on arrival to our institution. Two patients received reperfusion therapies.

**Conclusion:** Our early data indicates that stroke code implementation has resulted in rapid access to medical assessment, diagnostic imaging and treatment for AIS. Comparison to pre-code AIS data suggests that code implementation has also improved state-wide access to comprehensive stroke care.

**References:**  

16:35  
**Tortured teens and toxic screen time? Correlation between screen time and headache in an adolescent cohort**

*Gideon Richard Budiono 1, Yi Lynn Leong 1, Hugo Sampaio 1,2, Ian Andrews 1,2, Alexandra Johnson 1,2*  
(1) University of New South Wales, Sydney Children's Hospital, Sydney, NSW Australia;  
(2) Neurology, Sydney Children's Hospital, Sydney, NSW, Australia

**Background:** Headache is common in older children and teenagers and causes significant pain and disability. We hypothesized electronic device usage was associated with headache prevalence.

**Methods:** A cross-sectional study of 43 adolescents (10-18 years old) attending outpatient clinics was conducted. Demographic data, headache type and frequency, and screen exposure was collected, with
validated questionnaires examining sleep quality (Epworth Sleepiness Scale, Pittsburgh Sleep Quality Index), activity levels (Sedentary Behaviour Questionnaire, Patient-centred Assessment & Counselling for Exercise), and quality of life (self-reported EQ-5D-Y). Descriptive and non-parametric statistics were utilised. Ethics was granted.

Results: Headaches affected 29 adolescents (67%) with a median severity of 6/10 and a median frequency of three per month (range 0–31). Headache severity correlated with duration of schoolwork using electronic devices per day ($r=0.503; p=0.006$), duration of texting per day ($r = 0.393; p = 0.039$), and duration of active laptop usage ($r=0.510; p=0.006$). Texting positively correlated with frequency of headache per month ($r=0.388; p=0.038$). Headache frequency was unchanged with longer gaming duration ($p=0.11$) and significantly reduced with higher total device usage ($p=0.0031$). Additional features including poor sleep and lower activity levels were not associated with headache severity or frequency.

Conclusion: Specific device usage (texting, laptop use) may be a risk factor for increased headache in adolescents. This requires further exploration in larger studies. We hypothesized this may be due to head and neck positioning in both activities, which would also require further confirmation with consideration of educational recommendations for device usage in learning.

16:45

**Forme Fruste Neuronal Ceroid Lipofuscinosis**

*Lauren Taylor*¹,², *Emma MacDonald-Laurs*², *Nicola Fearn*², *Victoria Rodriguez-Casero*², *Ian Woodcock*¹,², *Gabriel Dabscheck*²

¹Neuroscience, Murdoch Children’s Research Institute, Parkville, VIC, Australia; ²The Royal Children’s Hospital, Parkville, VIC, Australia

**Background:** Neuronal ceroid lipofuscinosis type 2 (CLN2) is caused by a variant in the tripeptidyl-peptidase-1 (TPP1) gene causing low TPP1 enzyme. CLN2 presents with early-onset language delay, epilepsy and ataxia followed by blindness and neurodegeneration. Treatment with enzyme replacement therapy (ceroliponase-alpha) has become available for CLN2. Low activity of TPP1 enzyme has also been reported in spinocerebellar ataxia type-7 (SCAR7), a slowly progressive ataxia. We describe a case of an “intermediate-phenotype” between CLN2 and SCAR7, who presented as an eight-year-old with four years of progressive ataxia and neuro-cognitive decline. Investigations showed borderline-low leucocyte TPP1 enzyme. Whole-exome sequence revealed two variants of uncertain significance in the TPP1 gene, occurring in trans-configuration.

**Methods:** We reviewed the literature of described cases of CLN2 and SCAR7, and compared our patient’s presentation and investigation findings with those described in the literature.

**Results:** Neither the clinical nor investigative features of our case were classical of CLN2 or SCAR7. Our patient had late but progressive neurodegeneration and microcephaly which would not be expected in SCAR7, however she had normal vision and no seizures thus her
clinical phenotype was milder than classical CLN2. Additionally, EEG, MRI, enzymatic and genetic testing were supportive but not diagnostic of CLN2 suggesting our patient had diminished rather than abolished TPP1 activity.

Conclusions: We contend that this child has an intermediate form of TPP1 deficiency, neither classic of CLN2 nor SCAR7, and would benefit from enzyme replacement therapy. As there is now a treatment available, diagnosis of these atypical phenotypes is essential.

17:00 ANZCNS AGM
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<td>Early Morning Session</td>
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<td>08:30</td>
<td><strong>Symposium - Neuromuscular Medicine: The changing aspects of diagnosis and treatment - Chair: Michelle Farrar</strong></td>
<td>Josh Burns, Ian Alexander, Anne Connolly</td>
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<td>Non-pharmacological treatment of in Charcot-Marie-Tooth Disease</td>
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<td>Gene therapy approaches for precision medicine in neurological disease</td>
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<td>International perspective: Newborn screening and therapies for neuromuscular disorders</td>
<td>Anne Connolly</td>
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<td><strong>Symposium - Neuromuscular Medicine (continued) - Chair: Monique Ryan</strong></td>
<td>Didu Kariyawasam, John Svaren</td>
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<td>NSW perspective: Newborn screening for spinal muscular atrophy</td>
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<td>Therapeutics for Charcot-Marie-Tooth Disease: Clinical Trials and Prospects in Development</td>
<td>John Svaren</td>
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<td><strong>Symposium - Neurogenetics - Chair: Simone Ardern-Holmes</strong></td>
<td>Lynette Sadleir</td>
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<td>Developmental and Epileptic Encephalopathies: Evolving Knowledge</td>
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<td>Choices in genetic testing in the Epilepsy Clinic</td>
<td>Rani Sachdev</td>
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<td>Utility of whole genome versus whole exome genetic testing in developmental and epileptic encephalopathies</td>
<td>Emma Palmer</td>
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<td>Australian Genomic Health Alliance Acute Care Flagship: Neurology Cases</td>
<td>Jason Pinner</td>
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<td>Australian Genomic Health Alliance: Intellectual Disability</td>
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<td>Mackenzie’s Mission</td>
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<td><strong>Platform Session 2 - Neuromuscular medicine and Neurogenetics - Chair: Anita Cairns</strong></td>
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<td><strong>ANZCNS Conference drinks &amp; dinner</strong> - Centennial Homestead</td>
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08:00       REGISTRATION DESK OPEN
08:30-10:35 SYMPOSIUM - NEUROMUSCULAR MEDICINE: THE CHANGING ASPECTS OF DIAGNOSIS AND TREATMENT
08:30       Non-pharmacological treatment of in Charcot-Marie-Tooth Disease
            Prof Josh Burns, The Children's Hospital at Westmead
09:05       Gene therapy approaches for precision medicine in neurological disease
            Prof Ian Alexander, The Children's Hospital at Westmead
09:45       International perspective: Newborn screening and therapies for neuromuscular disorders
            Prof Anne Connolly, Nationwide Children's Hospital, USA
            (International Speaker Sponsored by Biogen)
10:35-11:05 MORNING TEA
11:05-12:15 SYMPOSIUM - NEUROMUSCULAR MEDICINE (continued)
11:05       NSW perspective: Newborn screening for spinal muscular atrophy
            Dr Didu Kariyawasam, Sydney Children’s Hospital, Randwick
11:35       Therapeutics for Charcot-Marie-Tooth Disease: Clinical Trials and Prospects in Development
            Prof John Svaren, University of Wisconsin-Madison
12:15-13:00 LUNCH
13:00-15:30 SYMPOSIUM - FOETAL NEUROLOGY
13:00       Developmental and Epileptic Encephalopathies: Evolving Knowledge
            Prof Lynette Sadleir, University of Otago, Wellington, New Zealand
13:40       Choices in genetic testing in the Epilepsy Clinic
            Dr Rani Sachdev, Sydney Children’s Hospital
14:00       Utility of whole genome versus whole exome genetic testing in developmental and epileptic encephalopathies
            Dr Emma Palmer, Sydney Children’s Hospital
14:20       Australian Genomic Health Alliance Acute Care Flagship: Neurology Cases
            Dr Jason Pinner, Sydney Children’s Hospital
14:40       Australian Genomic Health Alliance: Intellectual Disability
            A/Prof Tony Roscioli, Sydney Children’s Hospital
15:00       Mackenzie’s Mission
            Prof Edwin Kirk, Sydney Children’s Hospital
15:30-16:00 AFTERNOON TEA
16:00-17:00  PLATFORM SESSION 2: NEUROMUSCULAR MEDICINE AND NEUROGENETICS

16:00  Onasemnogene abeparvovec gene-replacement therapy (GRT) in presymptomatic spinal muscular atrophy (SMA): SPR1NT study update


(1) Department of Neurology, Sydney Children’s Hospital Network, Randwick, NSW, Australia; (2) Clinic for Special Children, Strasburg, PA, USA; (3) Department of Neurology, Massachusetts General Hospital, Boston, MA, USA; (4) Department of Pediatrics, Children’s Hospital of Eastern Ontario, Ottawa, Canada; (5) Department of Pediatrics, University of Colorado School of Medicine, Aurora, CO, USA; (6) Department of Neurology, Helen DeVos Children’s Hospital, Grand Rapids, MI, USA; (7) Department of Pediatrics, University of Texas Southwestern Medical Center, Dallas, TX, USA; (8) Division of Pediatric Neurology, Columbia University Medical Center, New York, NY, USA; (9) Department of Neurology, University of Wisconsin School of Medicine and Public Health, Madison, WI, USA; (10) Institute of Medical Genetics, Tokyo Women’s Medical University, Tokyo, Japan; (11) The Dubowitz Neuromuscular Centre, University College London, Great Ormond Street Institute of Child Health & Great Ormond Street Hospital, London, UK; (12) Gene Therapy Research Unit, Children’s Medical Research Institute and The Children’s Hospital at Westmead, Sydney, NSW, Australia; (13) AveXis, Inc., Bannockburn, IL, USA; (14) National Institute of Health Research, Great Ormond Street Hospital Biomedical Research Centre, London, UK

**Background:** Onasemnogene abeparvovec (formerly AVXS-101) treats the genetic root cause of SMA, biallelic survival motor neuron 1 gene (SMN1) deletion/mutation. This study evaluates onasemnogene abeparvovec in presymptomatic SMA patients.

**Methods:** SPR1NT is a multicenter, open-label, phase 3 study. Asymptomatic SMA patients (2–3xSMN2) ≤6 weeks of age receive a one-time intravenous onasemnogene abeparvovec infusion. Safety and efficacy are assessed through study end (2xSMN2: 18 months; 3xSMN2: 24 months). Primary outcomes include independent sitting ≥30 seconds (2xSMN2) and standing unassisted (3xSMN2). Exploratory outcomes include motor function improvement (CHOP INTEND).

**Results:** As of 8 March 2019, 18 infants were dosed (8–40 days of age [mean: 23]; 2xSMN2, n=8; 3xSMN2, n=9; 4xSMN2, n=1). Amongst the 8 patients with 2xSMN2, mean baseline CHOP INTEND score was 44.0 points (maximum 64), which increased by a mean of 14.4 (n=7) points at 3 months post-dosing; 6 patients scored ≥60 points; 3 reached maximum score; 50% (4/8) sat without support ≥30 seconds; 12.5% (1/8) stood with assistance. All patients achieving milestones were younger than 9.1 months (mean [range]: 6.1 [1.7–9.1]) and within the WHO range of the sitting milestone (1st–99th percentile: 3.8–9.2 months); no child older than 9.1 months failed to achieve sitting. Treatment-emergent adverse events of special interest were not associated with clinical signs and symptoms; all resolved without sequelae.
Conclusions: Preliminary data from SPR1NT show age-appropriate motor development, intact swallowing, and lack of ventilatory support in presymptomatic SMA patients dosed with onasemnogene abeparvovec, underlining the importance of early treatment.

16:10

**Nusinersen in infants who initiate treatment in a presymptomatic stage of spinal muscular atrophy (SMA): interim results from the phase 2 NURTURE study**


1. Royal Children's Hospital, University of Melbourne and Murdoch Children's Research Institute, Melbourne, VIC, Australia; 2. Children's Hospital Colorado, University of Colorado, Aurora, CO, USA; 3. Departments of Neurology and Pediatrics, Columbia University Irving Medical Center, New York, NY, USA; 4. Unit of Neuromuscular and Neurodegenerative Disorders, Post-Graduate Bambino Gesù Children's Research Hospital, Rome, Italy; 5. Departments of Medical Genetics and Pediatrics, National Taiwan University Hospital, Taipei, Taiwan; 6. Department of Neurology, Johns Hopkins University School of Medicine, Baltimore, MD, USA; 7. Department of Neurology, Center for Genomic Medicine, Massachusetts General Hospital, Boston, MA, USA; 8. Division of Neurology, Department of Pediatrics, Nemours Children's Hospital, Orlando, FL, USA; 9. University Hospital Bonn, Bonn, Germany; 10. Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, USA; 11. University of Utah, Department of Pediatrics and Neurology, Salt Lake City, UT, USA; 12. Department of Pediatric Neurology, Hacettepe University, Ankara, Turkey; 13. Sidra Medicine, Department of Pediatrics, Qatar Foundation, Doha, Qatar; 14. Division of Clinical and Metabolic Genetics, Department of Pediatrics, Hamad Medical Corporation, Doha, Qatar; 15. NEMO Clinical Center – NEuroMuscular Omniscience, Milan, Italy; 16. Department of Biomedical Sciences for Health, Università degli Studi di Milano, Milan, Italy; 17. Graduate Institute of Clinical Medicine, College of Medicine, Kaohsiung Medical University; Departments of Pediatrics and Laboratory Medicine, Kaohsiung Medical University Hospital, Kaohsiung Medical University, Kaohsiung, Taiwan; 18. Department of Neurology, David Geffen School of Medicine at UCLA, Los Angeles, CA, USA; 19. Biogen, Cambridge, MA, USA; 20. Biogen, Maidenhead, Berkshire, UK

**Background:** Nusinersen is the first approved treatment for spinal muscular atrophy (SMA). We present interim results from the ongoing NURTURE study (NCT02386553) examining efficacy and safety of intrathecal nusinersen initiated in presymptomatic infants with 2 or 3 SMN2 copies.

**Methods:** Enrolled infants were age ≤6 weeks at first dose, clinically presymptomatic, and genetically diagnosed with SMA. Primary endpoint is time to death or respiratory intervention (≥6 hours/day continuously for ≥7 days or tracheostomy).

**Results:** Twenty-five infants (2 copies SMN2, n=15; 3 copies, n=10) were enrolled in NURTURE. As of 29 March 2019, median age at last visit was 34.8 (range 25.7–45.4) months. All infants were alive and none required permanent ventilation. Median time to death or respiratory
intervention could not be estimated because of too few events. Four infants (all with 2 SMN2 copies) required respiratory intervention for ≥6 hours/day continuously for ≥7 days, with all cases initiated during acute, reversible illness. All 25 (100%) infants achieved the WHO motor milestone sitting without support and 23/25 (92%) achieved walking with assistance; 22/25 (88%) were walking alone. Nearly all children reached the maximum score on the CHOP INTEND scale. Phosphorylated neurofilament heavy chain levels rapidly declined during the loading phase of nusinersen and then stabilized. No new safety concerns were identified. Results from additional assessments from the 29 March 2019 interim analysis will be presented.

Conclusions: There was continued benefit to infants who initiated nusinersen before symptom onset, emphasizing the value of early treatment and newborn screening.

16:20
Health professionals’ experiences of wheelchair prescription for children with neuromuscular disorders
Sarah-Grace Paguinto 1,2, Michelle Farrar 1, Nadine Kasparian 3,4, Paula Bray 5
(1) Occupational Therapy, Sydney Children’s Hospital, Randwick, NSW, Australia; (2) School of Women and Children’s Health, University of New South Wales, Sydney, NSW, Australia; (3) Harvard Medical School, Harvard University, Boston, USA; (4) Department of Cardiology, Boston Children’s Hospital, Boston, USA; (5) Faculty of Health Sciences, University of Sydney, Sydney, NSW, Australia

Introduction: Standards of care for neuromuscular disorders (NMDs) recommend timely provision of wheelchair equipment. The transition to wheelchair use is a complex process. In our previous study, parents reported varying factors related to their readiness to engage in wheelchair prescription for their child. These factors include emotional responses, the importance of walking, psychological support, and a collaborative approach to decision-making. Exploration of health professionals’ experiences regarding wheelchair prescription for children with NMDs is required for further insight into the barriers and enablers to this complex process.

Methods: Twenty multi-disciplinary health professionals who have experience related to wheelchair prescription for children with NMDs will be interviewed. A phenomenological approach, incorporating qualitative data analysis strategies from Miles et al and Bazeley are utilised for data analysis, with data managed using NVivo 12 software.

Results: Recruitment is ongoing, with over ten professionals interviewed from Australia and USA. Health professions include occupational therapist, physiotherapist, social work, neurologist, clinical nurse consultant and psychologist. Perceived barriers to parent engagement in wheelchair prescription commonly related to emotional responses and timely access to appropriate equipment. Perceived enablers to parent engagement include ready access to appropriate equipment for trial, framing wheelchairs as a tool for child participation, access to psychosocial support, and collaborative decision-making.
Conclusion: Collaborative multi-disciplinary supports are required to optimise parents’ receptivity to wheelchair prescription, and facilitate timely wheelchair provision for children with NMDs. This has implications for current health and disability systems.

References:

16:30
Clinical phenotypes and response to mexileine in childhood non-dystrophic myotonia
Tariq Gubara 1, Jayne Antony 1, Shekeeb Mohammad 1,2, Richard Webster 1, Helen Young 1,2, Manoj Menezes 1,2
(1) TY Nelson Department of Neurology and Neurosurgery, The Children's Hospital at Westmead, Sydney, NSW, Australia; (2) The University of Sydney, Sydney, NSW, Australia

Introduction: Non-dystrophic myotonias (NDM) are a phenotypically heterogeneous group of skeletal muscle channelopathies characterised by altered membrane excitability. Dramatic response to mexiletine has been described in adult cohorts but has not been well characterised in children.

Methods: We reviewed the medical records of nine children (8 families) who presented to a tertiary paediatric neuromuscular clinic with NDM.

Results: Clinical phenotypes included myotonia congenita (MC, five CLCN1 mutation, one SCN4A mutation), paramyotonia congenita (PMC, one genetically unclassified) and Shwartz-Jampel syndrome (SJS, one HSPG2 mutation, one genetically unclassified). Individuals presented between the age of five and 13 years, with symptom onset between 12 months and six years of age. Muscle stiffness was prominent, seen in all cases and severe enough to interfere with daily activities. Individuals with MC had generalised muscle hypertrophy, clinical myotonia, and warm-up phenomenon on exercise. The individual with SCN4A mutation also had cold-triggered eyelid myotonia and gait difficulty. The individual with PMC had exercise-triggered gait difficulty and showed decrement of CMAPs post-cooling. Individuals with SJS had characteristic facial features and myotonia. All probands demonstrated myotonic discharges on needle EMG. All individuals with MC had favourable response to mexiletine (100-200 mg twice daily). In contrast, one individual each with PMC and SJS showed no benefit. There were no side-effects warranting cessation of therapy.

Conclusions: We characterise the unique clinical phenotype and response to mexiletine in NDM with videos. Early diagnosis and genetic characterisations enables treatment with mexiletine which is well tolerated and effective in individuals with MC.
Influence of Body Mass Index on disability in children with Charcot-Marie-Tooth Disease

Gabrielle A Donlevy 1,2, Sarah Garnett 1,2, Kayla Cornett 3, Marnee McKay 1, Jennifer Baldwin 1, Joshua Burns 1,2, Manoj Menezes 1,2

(1) The University of Sydney, Sydney, NSW, Australia; (2) The Children’s Hospital at Westmead, Sydney, NSW, Australia; (3) Department of Neurology, Columbia University Irving Medical Centre, New York, NY, USA; (4) School of Clinical Sciences, Auckland University of Technology, Auckland, New Zealand

Background: Growth and body mass influence disability in childhood neuromuscular disorders. This study examined the relationship between Body Mass Index (BMI) and disability in children with Charcot-Marie-Tooth Disease (CMT).

Methods: We conducted a cross sectional analysis of 477 patients with CMT aged 3-20 years from the Inherited Neuropathies Consortium, and 316 age-and-sex matched controls from the 1000 Norms Project. BMI was categorised according to the International Obesity Task Force (IOTF), and compared with scores on the CMT Pediatric Scale (CMTPedS). ITOF categories were collapsed into five age-and-sex equivalent BMI groups: severely underweight, underweight, healthy weight, overweight, obese.

Results: Compared to controls, there was a significantly higher proportion of children with CMT categorised as severely underweight (5.6% vs 0.3%), underweight (10.4% vs 5.1%), and obese (7.6 vs 3.8%) (p<0.05). There was fewer children categorised as healthy weight (61.2% vs 74.4%) (p<0.05); the distribution of overweight (15.1% vs 16.5%) between groups was comparable. Mean CMTPedS scores for groups were: severely underweight (27 ±9), underweight (20 ±8), healthy weight (17± 9), overweight (17± 9) and obese (22 ±10).

Compared to healthy weight children with CMT, being severely underweight with CMT was significantly more disabling (p< 0.0001), as was being obese (p=0.015).

Conclusions: There is a higher frequency of underweight and obese children with CMT compared to age-and-sex matched healthy children. Underweight and obese children with CMT are more disabled than children of healthy weight. A longitudinal study is required to determine the need for specific nutritional intervention to reduce the burden of CMT.

Personalised ankle-foot orthoses using 3D printing for children and adolescents with Charcot-Marie-Tooth disease

Elizabeth Wojciechowski 1,2, Sean Hogan 1, Manoj P Menezes 1,2, Tegan L Cheng 1,2, David Little 1,2, Joshua Burns 1,2

(1) Sydney Children’s Hospital Network, Westmead, NSW, Australia; (2) The University of Sydney, Sydney, NSW, Australia

Background: Children with Charcot-Marie-Tooth disease (CMT) are often prescribed ankle-foot orthoses (AFO) to manage walking difficulty. They are handmade using thermoplastic vacuum forming which provides limited design options, is labour-intensive and
associated with long wait times. 3D printing has the potential to transform how AFOs are designed, manufactured and delivered. The aim of this study is to evaluate personalised 3D printed AFOs vs. traditional handmade AFOs on walking ability for children and adolescents with CMT.

Methods: Three AFO designs were evaluated including traditional AFO, replicated 3D printed AFO (same design as traditional AFO) and optimised 3D printed AFO (novel design), compared to a shoe only condition. Traditional AFOs were handmade by an Orthotist from a plaster cast of the lower limb. Computer aided models of AFOs were generated from a 3D surface scan of the positive mould and 3D printed in Nylon 12. Patient-reported outcomes and 3D gait analysis, including in-shoe plantar pressure data, were collected to assess differences in gait between conditions.

Results: Twelve children with varying types CMT were recruited (11±3.8 years). The traditional AFOs significantly improved ankle position at initial contact compared to shoes only. The 3D printed AFOs produced equivalent gait correction to the traditional AFOs. The optimised designs reduced the weight of the AFO by 36 percent compared to the replicated AFOs.

Conclusion: 3D printing personalised AFOs is feasible pipeline for the design, manufacture and delivery of AFOs and has many potential benefits including improved biomechanical function, comfort and fit, patient satisfaction and delivery times.

19:00 ANZCNS CONFERENCE DRINKS & DINNER
Centennial Homestead

Pick up from the Crowne Plaza Coogee Beach Sydney Hotel at 6.30pm
Pick up from Centennial Homestead to the Crowne Plaza Coogee Beach Sydney Hotel at 10.30pm
## FRIDAY 6TH SEPTEMBER

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<td><strong>Symposium - Movement Disorders - Chair: Neil Mahant &amp; Russell Dale</strong></td>
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<td>Dystonia - Phenomenology, classification and recent updates</td>
<td>Victor Fung</td>
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<td>Common childhood movement disorders, genetic movement disorders</td>
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<td>The rate and severity of Cerebral Palsy have decreased in Australia. So what’s next?</td>
<td>Nadia Badawi</td>
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<td><strong>Platform Session 3: Movement Disorders and Miscellaneous - Chair: Richard Webster</strong></td>
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<td><strong>Neonatal EEG Course - Chair: Lakshmi Nagarajan</strong></td>
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<td>Francesco Pisani, Lakshmi Nagarajan, Sue Davis &amp; Cynthia Sharpe</td>
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<td><strong>Neonatal EEG Course (continued) - Chair: Lakshmi Nagarajan</strong></td>
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08:00 REGISTRATION DESK OPEN

08:30-10:30 SYMPOSIUM - MOVEMENT DISORDERS

08:30 Dystonia - Phenomenology, classification and recent updates
A/Prof Victor Fung, Westmead Hospital

CASE BASED SESSION: Common and emerging themes in childhood onset movement disorders

09:15 Tics and Stereotypies Russell Dale
09:35 Deep Brain stimulation in monogenic movement disorders
Shekeeb Mohammad
09:55 The movement disorder/epilepsy interface - Paroxysmal movement disorders Hannah Jones

10:15 Basal ganglia abnormalities on neuroimaging - a pattern recognition approach Shekeeb Mohammad

10:30-11:00 MORNING TEA

11:00-11:30 SYMPOSIUM - MOVEMENT DISORDERS (continued)

11:00 KEYNOTE LECTURE: The rate and severity of Cerebral Palsy have decreased in Australia. So what’s next?
Prof Nadia Badawi, The Children’s Hospital at Westmead (Sponsored by the Cerebral Palsy Alliance)

11:30-12:30 PLATFORM SESSION 3: MOVEMENT DISORDERS AND MISCELLANEOUS

11:30 The Cerebral Palsy Early diagnostic clinic 16 months on: outcome after implementation of the 2017 guidelines
Esther Tantsis 1, Anna te Velde 2, Cathy MORGAN 2, Ronda Shehata 2, Prue Golland 2, Johanna Korkalainen 2, Jane Berry 2, Robyn McMurdo 2, Iona Novak 2, Nadia Badawi 1
(1) Children’s Hospital at Westmead, Westmead, NSW, Australia;
(2) Cerebral Palsy Alliance, Sydney, NSW, Australia

Background: The diagnosis of cerebral palsy (CP) has historically been made at 2 years of age when a child’s motor abilities could be predicted accurately. The emergence of motor assessment tools for neonates and infants was fundamental in the development of international guidelines for the early diagnosis of CP. A multidisciplinary clinic was established in 2018 to implement the guidelines suggested diagnostic pathway. We review the first 16 months of clinic and compare the rate, age and presentation of children diagnosed with CP.

Methods: We performed a chart review of patient data collected prospectively in the 16 months since the clinic was established. We compared the risk factors, age of diagnosis and limiting factors for CP diagnosis.

Results: Sixty-three children were assessed; 27/63 (43%) were diagnosed with CP at an average age of 8.2 months. 15 (24%) were diagnosed as being at high risk of CP at an average age of 6.7 months. 21 (33%) did not fulfil criteria for CP.

Conclusions: Early diagnosis of CP is achievable with implementation of the 2017 guidelines. The diagnosis of cerebral palsy or high risk of CP was established in 42 children (67%) at an average age of 7.8 months.
which is well below the average age of CP diagnosis across Australia. To further improve the diagnostic pathway, access to neuroimaging and identifying biological markers for children at high risk will become increasingly important.

References:

11:40

The pooled diagnostic accuracy of three tests for diagnosing cerebral palsy early in high risk infants: a case control study

Cathy Morgan 1, Domenico Romeo 2, Iona Novak 1, Olena Chorna 3, Claire Galea 1,4, Sabrina Del Secco 3, Andrea Guzzetta 3,5

(1) Cerebral Palsy Alliance Research Institute, Brain Mind Research Centre, University of Sydney, Camperdown, NSW, Australia; (2) Fondazione Policlinico A. Gemelli, IRCCS, Rome, Italy; (3) Dept. of Developmental Neuroscience, Stella Maris Scientific Institute, Pisa, Italy; (4) Grace Centre for Newborn Care, Children’s Hospital at Westmead, Sydney, NSW, Australia; (5) Dept. of Clinical and Experimental Medicine, University of Pisa, Pisa, Italy

Background and Objectives: International Clinical guidelines recommend the use of neuroimaging, Prechtl’s General Movements Assessment (GMA) and the Hammersmith Infant Neurological Examination (HINE) to diagnose cerebral palsy early in infancy1. The aim of this study was to examine the predictive power of the combined use of GMA, neuroimaging and HINE in the early diagnosis of cerebral palsy.

Methods: We conducted a retrospective case-control study. Participants were 441 high risk infants who were NICU graduates born between 2003 and 2014, from three hospitals in Italy. Three groups of infants with either a normal outcome, mild disability or cerebral palsy at two years, were matched for birth year, gender and gestational age. Three-month HINE scores, fidgety GMA and neuroimaging reports (CUS and/or MRI), were retrieved from medical records. Logistic regression was conducted with log-likelihood used to determine model fit and Area Under the Curve (AUC) for accuracy.

Results: Sensitivity and specificity for detecting cerebral palsy were respectively, 88% and 62% for three-month HINE (cut-off score 57), 95% and 97% for absent fidgety GMs and 79% and 99% for neuroimaging. The pooled diagnostic accuracy of all 3 assessments gave sensitivity and specificity values of 97.86% and 99.22% respectively (PPV 98.56% and NPV 98.84%).

Conclusions and Significance: Cerebral palsy can be detected early and accurately in high risk infants when findings from these three tests triangulate. Clinical implementation of these tools is likely to reduce the average age when cerebral palsy is diagnosed, and intervention is started.

References:
The Psychiatry of Paediatric Movement Disorders

Michelle S Lorentzos ¹, Isobel Heyman ², Anna E Coughtrey ³, Andrew McWilliams ², Joshua Burns ⁴, Manoj P Menezes ⁴, Shekeeb S Mohammad ⁴, Ruth Evans ¹, Mary-Clare Waugh ¹, David Dossettor ¹, Padraic Grattan-Smith ¹, Robert Goodman ⁵, Manju A Kurian ⁵, Russell C Dale ⁶

¹ The Children’s Hospital at Westmead, Sydney, NSW, Australia; ² UCL Great Ormond Street Institute of Child Health, Great Ormond Street Institute of Child Health, London, UK; ³ Great Ormond Street Hospital for Children NHS Foundation Trust and Great Ormond Street Institute of Child Health, London, UK; ⁴ The University of Sydney & The Children’s Hospital at Westmead, Sydney, NSW, Australia; ⁵ Developmental Neurosciences, Great Ormond Street Institute of Child Health and Great Ormond Street Hospital for Children NHS Foundation Trust, University College, London, UK; ⁶ Kids Neuroscience Centre at Kids Research, Brain and Mind Centre, Children’s Hospital at Westmead, Faculty of Medicine and Health, University of Sydney, Sydney, NSW, Australia

Background: Psychiatric co-morbidities have been well described in children with Tourette Syndrome. There is, however, little known about the psychiatric problems that affect children with a broader range of non-tic movement disorders. This study compared the rate of psychiatric comorbidity in children with non-tic movement disorders to children with tics and TS, as well as other control groups.

Method: Children between the ages of 5 and 16 years were recruited at The Children’s Hospital at Westmead, Australia and Great Ormond Street Hospital, United Kingdom, with tic movement disorders (n=158) and non-tic movement disorders (n=102). Control groups included Emergency department control cohort (n=100), neurology control cohort with peripheral neuropathy or epilepsy (n=37), and normative data from UK community cohort (n=10,438). All patients were assessed using a validated psychiatric tool, the DAWBA, which generates DSMV criteria.

Results: There was no difference in the presence of any psychiatric comorbidity in the non-tic cohort (39.2%) compared with the Tic cohort (41.8%). Psychiatric comorbidity in the non-tic cohort was greater than the emergency control group (18%, p<0.0001) and the community cohort (9.5%, p<0.00001). 45.5% and 43.9% of the tic cohort with psychiatric comorbidity were receiving medical psychiatric treatment or psychology respectively, compared to only 22.5% and 15.0% of the non-tic cohort with psychiatric comorbidity.

Conclusion: This study demonstrates that children with non-tic movement disorders are just as vulnerable to psychiatric comorbidities as children with tics and Tourette syndrome. These psychiatric comorbidities appear to be under-recognised and under-treated.

References:


12:00  
**Acquired Ptosis in Children: A Case Series and Diagnostic Algorithm**  
Wui-Kwan Wong 1, Christopher L Troedson 1, Manoj Menezes 1,2, Shekeeb S Mohammad 1,3, Simone M Ardern-Holmes 1,2  
(1) T Y Nelson Department of Neurology and Neurosurgery, The Children’s Hospital at Westmead, Westmead, NSW, Australia; (2) The University of Sydney, Sydney, NSW, Australia; (3) Kids Research and The Children’s Hospital at Westmead Clinical School, The University of Sydney, Sydney, NSW, Australia

*Background:* Blepharoptosis, or ptosis, is the abnormal drooping of the upper eyelid. This can be present at birth (congenital) or develop later in life (acquired). There are a myriad of possible diagnoses for acquired ptosis. It may occur as an isolated abnormality or be part of a systemic disorder, including potentially life-threatening, but treatable, illnesses such as infantile botulism, tick paralysis or congenital myasthenia. Furthermore, uncorrected ptosis in early life can result in amblyopia, strabismus and psychological impacts due to cosmetic concerns. Thus, early and accurate diagnosis is integral in managing the paediatric patient presenting with ptosis.

*Methods:* The cases of five paediatric patients presenting with acquired ptosis to a tertiary neurology service were reviewed. All five patients had different diagnoses: anti-MuSK (muscle specific kinase) and anti-acetylcholine antibody myasthenia gravis, chronic inflammatory polyneuropathy, infant botulism and ophthalmoplegic migraine. A literature search was performed in order to develop a diagnostic algorithm for acquired ptosis.

*Results:* Clinical evaluation of a child with ptosis should encompass a thorough history, examination (eye, neurological and developmental) and targeted investigations for efficient and accurate diagnosis. A diagnostic algorithm has been developed for a child who presents with ptosis to assist clinicians in everyday practice.

*Conclusions:* A child presenting with acquired ptosis is not uncommon. There are a number of possible diagnoses, including systemic disorders, which may be managed with either medical or surgical treatments. In order to provide the best care to these patients and optimise patient outcomes, a timely, accurate diagnosis is crucial.

*References:*  

12:10  
**Paroxysmal extreme pain treated with novel sodium channel therapies in a neonate with a rare SCN9A mutation**  
Claire Murray 1, Adriane Sinclair 1, Sarfaraz Rahiman 1, Michaela Waak 1, Anthony Herbert 1, Mark Alcock 1  
(1) Queensland Children’s Hospital, South Brisbane, QLD, Australia

*Background:* Gain of function mutations of SCN9A encoded Nav1.7
cause two distinct inherited disorders of pain and autonomic dysfunction; inherited erythromelalgia (IEM) and paroxysmal extreme pain disorder (PEPD). These disorders are typically resistant to pharmacological therapy although sodium channel blockers may have some effectiveness.

Case Report: A neonate presented with severe and very frequent paroxysmal events characterised by apnoea, bradycardia, tonic stiffening and harlequin rash. Events were provoked by variable stimuli including bowel movements. Unprovoked events also occurred. Cardiopulmonary resuscitation was required on multiple occasions. The ictal electroencephalography pattern was not consistent with seizures. Feeding difficulties co-existed. Magnetic resonance brain imaging was normal.

Direct sequencing analysis of the SCN9A gene identified a heterozygous mutation (A1632E). This had been reported in a patient exhibiting features of both PEPD and IEM with functional studies indicating change in function of the Nav1.7 channel.

Carbamazepine, oxcarbazepine and mexiletine were trialed with no response and were subsequently withdrawn. Introduction of ranolazine, a novel sodium channel blocker, was followed by mild clinical improvement. Further mild improvement was seen after introduction of lacosamide. Non-pharmacological therapies including cooling of extremities were not consistently effective.

Conclusion: SCN9A disorders can present in the neonatal period with severe paroxysmal events characterized by autonomic symptoms and provoked by bowel movements. Genetic diagnosis allows for targeted therapy with sodium channel blockers. The use of novel sodium channel blockers is an important consideration in this rare disorder where life threatening events can occur during infancy and debilitating episodic pain can continue throughout life.

References:
From royal madness to somatoform pain: misdirection on the path to a rash diagnosis (a case and review)
Alexandra M Johnson 1,2,3, Hugh Allen 1
(1) Royal North Shore Hospital, Sydney, NSW, Australia; (2) University of New South Wales, Sydney, NSW, Australia; (3) Sydney Children's Hospital, Randwick, NSW, Australia

Background: Porphyrias are rare and have protean symptomatology, leading to their reputation as ‘great pretenders’. Sufferers may be mislabelled as having functional disorders and have a lengthy odyssey to diagnosis.

Methods: An eight year old girl presented with symptoms suggestive of erythromelalgia, with pain in feet and hands relieved by sleeping with her extremities submerged in water. Lichenification of her hands was noted. She was dismissed from her local emergency and the children’s hospital on multiple occasions. On review, an association with light exposure was noted and elevated blood porphyrin was diagnostic of erythropoietic protoporphyria (EPP). Case history and diagnostic path were reviewed. The rate of misdiagnosis and historical misdirection were reviewed to understand causes for protracted diagnosis in EPP.

Results: On literature review, children with EPP were often misdiagnosed as hysterical or malingering. Previous studies demonstrate lag to diagnosis of 11-21 years1-3, with multiple consultations (five or more) prior to diagnosis in 38%1. Furthermore, popular perceptions of porphyria symptomatology can be incorrect or restricted to subtype. Chronic psychosis may be falsely attributed to porphyria (madness of King George). Non-blistering forms of porphyria occur, including EPP which can present without skin changes4. Furthermore, urine porphyrins are not elevated in EPP, so classical urinary colour will not occur5.

Conclusions: EPP is the commonest porphyria in children but often has a lengthy time to diagnosis due to a lack of awareness and non-classical symptoms. Improved knowledge of EPP may help to improve time to diagnosis and management avoiding further pain and complications.

References:
Cryptococcus gattii meningitis in immunocompetent children: varied presentations and outcome

Lakshmi B Kalband¹, Joshua Davis², Christina I Miteff¹, Gopinath M Subramanian¹  
¹John Hunter Children’s Hospital, New Lambton Heights, NSW, Australia; ²Infectious Diseases, John Hunter Hospital, New Lambton, NSW, Australia

Background: Recent epidemiological data from Hunter region indicate an incidence of C gattii infection higher than the Australian nationwide incidence. Though literature is replete with information on cryptococcal meningitis in immunocompromised adults, there is scant data about the same in immunocompetent children. We describe two immunocompetent children with C gattii meningitis in the last 3 years treated at our centre.

Subjects and results: Two children (M:F =1:1, 13y) were treated for C gattii meningitis. The time to final diagnosis was varied (2 weeks vs 3 months). Following the diagnosis, cerebrospinal fluid (CSF) sterilization was achieved successfully within weeks of antifungal therapy. Immune Reconstitution Inflammatory Syndrome (IRIS) occurred in both patients requiring use of steroids. Both children required serial general anaesthetic assisted lumbar punctures and drainage to control the intracranial hypertension; however, both eventually required a ventriculoperitoneal (VP) shunt for hydrocephalus. Both children were successfully treated without neurological deficits in the short term. The long term outcome in the first patient has been excellent and second continues to be under follow up care, with nil sequelae so far.

Conclusion: This report highlights the varied clinical presentation of Cryptococcus gattii meningitis and the challenges to diagnosis in an immunocompetent setting. IRIS requires prompt recognition and treatment. Management of raised intracranial pressure with serial lumbar puncture may be effective initially but VP shunt may be inevitable in the long term.
POSTERS

1. The Australian Neuromuscular Disease Registry: recent progress and future directions
   Monique Ryan, Robin Forbes, Belinda Burns, Kristen Nowak, Leanne Lamont

2. Hypothalamic hamartoma: epilepsy and neurodevelopmental profiles
   Clair Pridmore, Felice D’Arco, Varsha Siyani, Leah Bull, Hanna Richardson

3. TANGO2-related disease in Australasian children: a clinical spectrum
   Ellen Hurley, Carolyn Bursle, Drago Bratkovic, David Coman, Emma Glamuzina, Thomas Hurley, Anita Inwood, Maina Kava, Michelle Lipke, Jim McGill, Heidi Peters, Nicholas Smith

4. Parent’s experience of hospital-based care of their child with cerebral palsy: An integrative literature review
   Josefina Natividad Villanueva, Suzanne Sheppard-Law, Karen Walker, Cathrine Fowler

5. Respiratory and nutritional outcomes in SMA type 1 patients treated with nusinersen
   Kerrie-Anne Chen, Michelle Farrar, John Widger, Dominic Fitzgerald

6. Stopping the dance of eyes and feet: A therapeutic challenge
   Lakshmi B Kalband, Emma Turton, Rani Bhatia, Christina I Miteff

7. Onasemnogene abeparvovec gene-replacement therapy (GRT) for spinal muscular atrophy type 1 (SMA1): STR1VE phase 3 study update

8. An Aboriginal girl with Acute Rheumatic Fever and anti-N-Methyl-D-Aspartate receptor encephalitis
   Eleanor Ng, Snehal Shah, Peter W Rowe

9. Osmotic Demyelination Syndrome
   Najm Khan, Rahul Lakshmanan, Snehal Shah, Lakshmi Nagarajan

10. Coma from unknown origin in one healthy boy: ammonia blood level as one small step for doctors and a giant leap for patients. Would you think about non-hepatic hyperammonemia as a cause?
    Egeu Bosse, Evandro Souto, Marcelly Balducci
11. Interim report on the safety and efficacy of longer-term treatment with nusinersen in infantile-onset spinal muscular atrophy (SMA): updated results from the SHINE study
   Michelle A Farrar, Richard S Finkel, Diana Castro, Mar Tulinius, Kristin J Krosschell, Kayoko Saito, Sandra P Reyna, Giulia Gambino, Richard Foster, Ishir Bhan, Janice Wong, Wildon Farwell

12. Idiopathic intracranial hypertension in young patients: if one size does not fit all, can we customize first-line option based on patient’s profile?
   Egeu Bosse, Adriana Espindola, Thiago Scopetta, Roberto Mendonça, Mario Conti

13. Further update on status epilepticus in children. Delays in management in tertiary and non-tertiary hospitals
   Preena Uppal, Michael Cardamone, John Lawson

14. Newborn Screening for Spinal Muscular Atrophy – the perspective of Australian families
   Didu ST Kariyawasam, Arlene D’Silva, Janine Vetsch, Veronica Wiley, Michelle A Farrar

15. Introducing a “Stroke Code” in a Quaternary Australian Paediatric Hospital – triage tool development and assessment of the impact on Emergency Department Capacity Standards
   Natalie Deuble, Michaela Waak, Adriane Sinclair
See you in Melbourne for the 9th ANZCNS Annual Scientific Meeting September 2020