anzcns

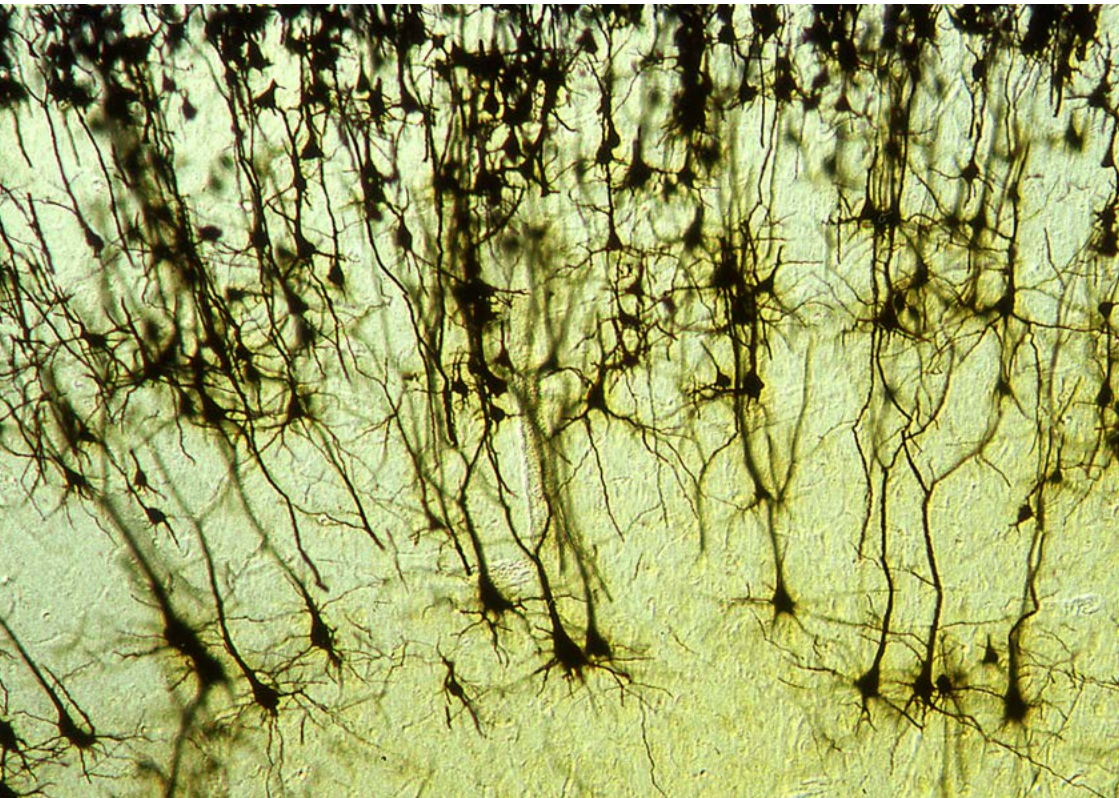
THE AUSTRALIA AND NEW ZEALAND
CHILD NEUROLOGY SOCIETY

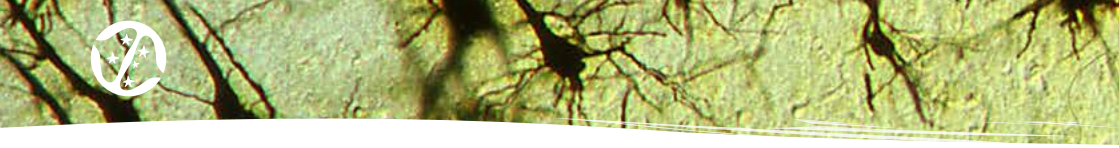
11th Annual Scientific Meeting & Trainee Day

13th - 16th September 2023

Adelaide Convention Centre

Adelaide, South Australia





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WELCOME MESSAGES

Welcome Message from the Local Organisers

We extend a warm welcome to the 11th Annual Scientific Meeting of the Australia and New Zealand Child Neurology Society in Adelaide.

The theme of this year's conference is 'Growing Older and Old Before Their Time'. It has been a privilege to create an innovative program, showcasing the latest in neurocritical care, neurooncology, headache and intellectual disability, adolescent neurology and dementia. We will hear from expert international and local speakers, including neurologists, geneticists, metabolists, psychologists and researchers.

Symposia and platform and poster sessions will provide opportunities for ANZCNS members and trainees to present their research and illuminate the challenges and opportunities in their fields.

The content and the cross-disciplinary focus of many of the presentations will be of interest to paediatricians, sub-specialists with an interest in child neurology, allied health professionals, and consultant and trainee child neurologists.

We look forward to seeing you in Adelaide and hope this meeting will inspire.

Clair Pridmore, Kerrie-Anne Chen & **Romain Briest**
ASM Organising Committee Co-chairs

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 2023 Annual Scientific Meeting. The Organising Committee has worked hard to arrange this event which has talks from Australia-based international experts in childhood neurological disorders, and will showcase local research. I look forward to seeing you all at the Meeting.

Manoj Menezes
President, Australia and New Zealand Child Neurology Society 2023

About Australia and New Zealand Child Neurology Society

The Australia and New Zealand Child Neurology Society (ANZCNS) is a collaborative group of approximately 140 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

Organising Committee (OC)

Clair Pridmore, Co-Chair and Board Member
Romain Briest, Co-Chair
Kerrie-Anne Chen, Co-Chair
Erik Andersen
Annie Bye
Mark Mackay, Board Member

GENERAL CONFERENCE INFORMATION

Secretariat Office

For queries, please contact:

Australia and New Zealand Child Neurology Society Ltd
c/- Seed Events Pty Ltd, PO Box 2137, Glenelg SA 5045, Australia

e: admin@anzcns.org.au **m:** +61 423 827 488

Conference Venue

Adelaide Convention Centre

The Centre is conveniently located at the heart of the City. To the North, the Centre overlooks the River Torrens and surrounding parklands, while to the South, the Centre sits on North Terrace, one of four major terraces that bind Adelaide's City Centre.

The Adelaide Riverbank Precinct is a hive of activity and where the City's cultural, sporting, education, medical, conference and entertainment zones merge, making it Australia's best connected business events precinct. In addition to the Centre, Adelaide Oval and Adelaide SkyCity Casino, the Riverbank Precinct is home to the world-leading BioMed City – the largest life and health sciences cluster in the Southern Hemisphere – which is located right next door.

Parking

If you are driving to the Centre, they offer two undercover car parking options, North Terrace and Riverbank car parks. The Riverbank car park is accessible from Morphett Street / Festival Drive. Both car parks are located directly under the Centre and operate 24 hours a day, 7 days a week. Each is fitted with video surveillance camera systems and security patrols both areas.

Clearance is 2.0 metres and automated payment stations accept payment via card only (major credit cards, debit cards and mobile pay). Please note that car parks can reach capacity early in the day during busy events.

0-1 HOUR	1-2 HOURS	2-3 HOURS	3-4 HOURS	EARLY BIRD *
$8.50	$13.00	$17.00	$19.00	$18.00

**Early Bird rate available 7 days for entry between 5.30am-9.30am and exit between 2.30pm-6.30pm*

An additional $1.00 charged for each additional hour to a maximum of $33.00 over a 24-hour period.

Adelaide Convention Centre

Registration Desk

The registration desk is located in the Riverbank Foyer, on Lower Ground level at the Adelaide Convention Centre. Please note that name badges must be clearly displayed at all times.

Registration desk opening hours are:

Wednesday 13th September:	07:30 - 17:00
Thursday 14th September:	08:30 - 17:00
Friday 15th September:	08:30 - 17:00
Saturday 16th September	08:30 - 16:45 (Trainee Day)

Speakers' Information

Speakers must submit their final presentation to the audiovisual technician in the main plenary room (Riverbank Rooms 6&7) **at least 2 hours prior to their session start time**, unless you are presenting in the first session, in which case we ask that you submit it the day prior.

Exhibition

A trade exhibition is held in conjunction with the 11th ANZCNS Annual Scientific Meeting. This is an integral part of the Meeting 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 in the Riverbank Foyer, on Lower Ground level, 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 Riverbank Foyer. Any delegates with particular catering needs should have noted these on their online registration form. Please make sure you see the venue catering staff on the day.

Mobile phones

Mobile 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 11th ANZCNS Annual Scientific Meeting is primarily for health professionals. Community and consumer support groups have been welcomed to the 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 11th 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 meeting communications. In registering and attending the meeting 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 meetings (including but not limited to: use within newsletters, presentations or other media that recounts the events that occurred during this meeting and/or in the promotion of future meetings). Delegates should inform the Australia and New Zealand Child Neurology Society (by email) in advance of or during the meeting 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.

MEETING DINNER

The dinner will be held at Magill Estate Kitchen which is situated at the home of Penfolds. Magill Estate Kitchen combines a relaxed dining experience with breathtaking views of the historic Penfolds vineyard and Adelaide city. Located adjacent to the Penfolds Magill Estate Cellar Door, Magill Estate Kitchen centers around the casual enjoyment of exceptional food, wine and sharing.

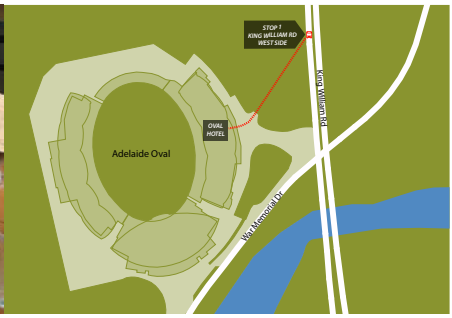
Venue: Magill Estate Kitchen,78 Penfold Road, Magill
Date: Thursday 14th September
Time: 7.00 - 10.30pm

A coach will depart from The Oval Hotel at 6.30pm and depart the Magill Estate Kitchen at 10.45pm returning guests back to the hotel.

**Please note the coach will be positioned at Stop 1 King William Rd, West Side, a 200 metre walk to/from the Oval Hotel. Please refer to the map below.*

MAGILL ESTATE

KITCHEN



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Alexion	7
Aspen Australia	5
Biogen	10
BioMarin	2
Clinect	12
LivaNova	9
Novartis	4
Nutricia Ketogenic	13
PTC Therapeutics Australia	1
Seer Medical	8
Tilray Medical Australia New Zealand	6
UCB	3

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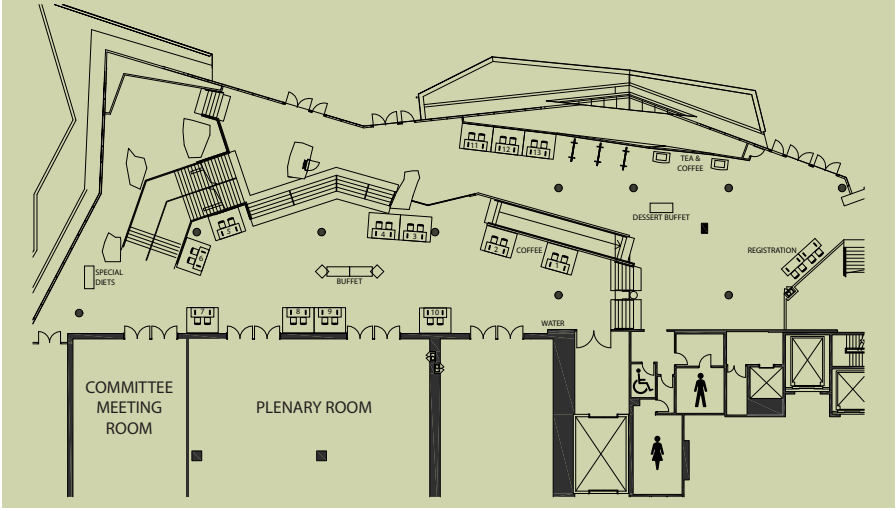

INTERNATIONAL SPEAKER

We are delighted to welcome our international speaker:

Dr. Nicholas Abend is a Professor of Neurology, Pediatrics, and Anesthesia and Critical Care at the University of Pennsylvania and the Children's Hospital of Philadelphia. He is the Senior Medical Director for Neuroscience, the Medical Director of Clinical Neurophysiology, and an Associate Scholar in the Center for Epidemiology and Biostatistics. His clinical practice focuses on seizure management and EEG interpretation in critically ill children. His research focuses on the significance, pathophysiology, and appropriate management of electrographic seizures in critically ill children and neonates, the treatment of status epilepticus, and the utility of EEG as a prognostic biomarker.

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[†]After 13 weeks of treatment vs. placebo; p <0.05

Before prescribing please review approved Product Information by clicking here.

PBS Information: This product is not listed on the PBS.

References: 1. Slenyto Product Information. Last amended June 2021. **2.** Gringras P, et al. J Am Acad Child Adolesc Psychiatry 2017;56(11):948–957. **3.** Maras A, et al. J Child Adolesc Psychopharmacology 2018; 28 (10);699 –710. **4.** Schroder CM, et al. J Autism Devel Disorders 2019; 49:3218–3230.
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DAY 1	WEDNESDAY 13TH SEPTEMBER	
Time	**Event**	**Speaker**
08:45 - 09:00	**Welcome**	*Manoj Menezes*
09:00 - 11:00	**Symposium: Neurocritical Care** Chairs: Clair Pridmore & Annie Bye	
09:00	EEG Monitoring and Seizures in Critically Ill Children	*Nicholas Abend* LivaNova Health innovation that matters
09:45	EEG indications, pitfalls and what really matters for PICU patients - thinking beyond seizures!	*Michaela Waak*
10:15	Paediatric post-traumatic epilepsy	*Bridgette Semple*
10:45	Panel Q+A	
11:00 - 11:30	**Morning Tea**	
11:30 - 12:30	**Platform Session 1 - Epilepsy** Chairs: Snehal Shah & Eleanor Ng	
11:30	Uridine Responsive Developmental and Epileptic Encephalopathy *(p12)*	*Jia Yi Leong*
11:45	A case of Cardio-Facio-Cutaneous syndrome and temporal lobe epilepsy with hippocampal sclerosis: Insights from epilepsy surgery and role of BRAF *(p13)*	*Sameer Dal*
12:00	Eadweard Muybridge and the Nineteenth-century roots of Gait Lab *(p14)*	*Kathryn Irving*
12:15	Cannabidiol (CBD) for treatment resistant epilepsy of childhood (TREC) - experience from Western Australia *(p15)*	*Najm Khan*
12:30 - 13:30	**Lunch & Poster viewing**	
13:30 - 15:30	**Symposium: Neuro-oncology** Chairs: Michael Hayman & Erik Andersen	
13:30	Advances in neuro-oncology	*David Ziegler*
14:00	Neurological Sequelae of Chemotherapy – The Old and The New	*Tej Kandula*
14:30	NF Update	*Gabriel Dabschek*
15:00	Australian Proton Radiotherapy - An Update	*Jordan Hansford*
15:30 - 16:00	**Afternoon Tea**	

16:00 - 17:00	**Platform Session 2 - Neuromuscular** Chairs: Suzanna MacLennan & Gina O'Grady	
16:00	Safety and efficacy of ataluren in nmDMD patients from Study 041, and pooled analysis of results from three randomized, double-blind, placebo-controlled trials *(p16 & 17)*	*Eppie Yiu*
16:15	Epidemiology and clinical features of Duchenne and Becker Muscular Dystrophy in Australia - Findings from the Australian Neuromuscular Disease Registry *(p18)*	*Chien Yen Poh*
16:30	Improving the transition journey for neuromuscular patients in Queensland *(p19)*	*Rebecca Leung*
16:45	Retrospective review of clinical presentation, neuroimaging features and treatment responsiveness of paediatric IIH presenting at tertiary care hospital *(p20)*	*Snehal Shah*
17:00 - 18:30	**ANZCNS Annual General Meeting**	

PLATFORM ABSTRACTS - SESSION 1: EPILEPSY

11:30 **Uridine Responsive Developmental and Epileptic Encephalopathy**

Jia Yi Leong [3,2,1], *Cameron Dickson* [1], *Paul Shillito* [1]

[1] *Paediatric Neurology, Canterbury, New Zealand*
[2] *Paediatrics, Canterbury, New Zealand*
[3] *Adelaide, SA, Australia*

This is a case presentation of 2 cases of a DEE responsive to Uridine therapy.

Background: We present two patients with mutations in the CAD gene presenting with a DEE responsive to Uridine therapy. To our knowledge, these are the first two cases diagnosed in New Zealand. **The distinct recognizable clinical phenotype and availability of a safe treatment option makes it important**. CAD deficiency is described as a cause of infantile epileptic encephalopathy characterised by global developmental delay, progressive epileptic encephalopathy refractory to treatment, loss of skills, anaemia with anisopoikilocytosis and cerebellar atrophy. Whilst this is a disorder of pyrimidine pathways with secondary effect on glycosylation, testing for the biomarkers of these disorders is normal. The diagnosis is made via genetic testing.

P1 (14 years) and P2 (5 years) presented with difficult to control epilepsy. P1 had developmental regression and associated anisopoikilocytosis and pan-cerebellar atrophy; P2 had epilepsy and developmental delay without neuroimaging or haematological changes, which we hypothesize is due to being younger (family member of P1). Both patients were started on uridine and had significant improvement in their seizure control and functioning, with no side effects

We also identified that P1 was deficient in folate and required supplementation while on uridine which has not been described. We hypothesize that this is due to competitive absorption and increased folate metabolism due to increased pyrimidine formation.

Conclusions: Consider CAD mutations in patients with an epileptic encephalopathy, developmental delay and anaemia with anisopoikilocytosis. Excellent treatment options exist including Uridine supplementation.

11:45

A case of Cardio-Facio-Cutaneous syndrome and temporal lobe epilepsy with hippocampal sclerosis: Insights from epilepsy surgery and role of BRAF

Sameer Dal [1], Emma Macdonald-Laurs [1], A. Simon Harvey [1]

[1] The Royal Children's Hospital, Melbourne, Parkville, VIC, Australia

Cardio-Facio-Cutaneous syndrome (CFCS) is a genetic disorder caused by an activating *de novo BRAF*mutation, resulting in upregulation of the RAS-MAPK pathway[1,2]. In addition to characteristic dysmorphic, cardiac and skeletal anomalies, CFCS is associated with drug-resistant focal epilepsy (DRFE) in 35%-38%, many having temporal lobe epilepsy (TLE) with hippocampal sclerosis (HS)[2,5]. Somatic *BRAF* variants are also reported in low-grade epilepsy associated tumours and in mesial TLE with and without HS, suggesting activation of RAS-MAPK signalling in the pathogenesis of DRFE[3].

We report a 16-year-old male, with CFCS and a pathogenic variant in *BRAF* (exon 6: c.784c → A. heterozygous; p.Q262K). He had frequent focal seizures from age 12 years associated with left temporal slowing, interictal epileptiform discharges and ictal rhythms on EEG, left HS on MRI, and left temporal hypometabolism on FDG-PET. He underwent left anterior temporal lobectomy and amygdalohippocampectomy, with hippocampal spiking recorded on ECoG and HS confirmed on histopathology. Postoperatively, seizures persisted unchanged and EEG showed bilateral interictal epileptiform activity and ictal rhythms.

This case suggests epilepsy surgery is ineffective in CFCS, despite the impression of unilateral temporal lobe seizures and pathology. The pathogenesis of seizures and HS in CFCS is uncertain[4,5]. HS may be a consequence of genetically-based epilepsy, rather than the cause, and might ultimately become bilateral. This patient's resected tissue is being examined for additional somatic variants ("two-hit") as a possible mechanism.

References:

[1] *Sran, S., & Bedrosian, T. A. (2023). RAS pathway: The new frontier of brain mosaicism in epilepsy. Neurobiology of Disease, 180, 106074. https://doi.org/10.1016/j.nbd.2023.106074*

[2] *Rauen, K. A. (2023, February 9). Cardiofaciocutaneous Syndrome. GeneReviews® - NCBI Bookshelf. https://www.ncbi.nlm.nih.gov/books/NBK1186/*

[3] *Contribution of Somatic Ras/Raf/Mitogen-Activated Protein Kinase Variants in the Hippocampus in Drug-Resistant Mesial Temporal Lobe Epilepsy. JAMA Neurology, 80(6), 578*

[4] *Battaglia, D., Gambardella, M. L., Veltri, S., Contaldo, I., Chillemi, G., Veredice, C., Quintiliani, M., Leoni, C., Onesimo, R., Verdolotti, T., Radio, F. C., Martinelli, D., Trivisano, M., Specchio, N., Dravet, C., Tartaglia, M., & Zampino, G. (2021). Epilepsy and BRAF Mutations: Phenotypes, Natural History and Genotype-Phenotype Correlations. Genes, 12(9), 1316*

[5] *Pierpont, E. I., Kenney-Jung, D. L., Shanley, R., Zatkalik, A. L., Whitmarsh, A., Kroening, S. J., Roberts, A. E., & Zenker, M. (2022). Neurologic and neurodevelopmental complications in cardiofaciocutaneous syndrome are associated with genotype: A multinational cohort study. Genetics in Medicine, 24(7), 1556–1566*

12:00 **Eadweard Muybridge and the Nineteenth-century roots of Gait Lab**

Kathryn B Irving [1]

[1] *Royal Children's Hospital, Melbourne, Parkville, VIC, Australia*

In 1878, pioneering photographer Eadweard Muybridge won a bet for millionaire Leland Stanford by proving that his prize stallion had four limbs off the ground when galloping, using a new technology that recorded the horse's movement frame by frame. A decade later, Muybridge collaborated with the University of Pennsylvania and neurologist Francis Dercum to produce "Animal Locomotion." Muybridge's masterpiece used chronophotography to record his human subjects, largely nude or scantily clad, involved in various physical activities, including walking, running, climbing and throwing.

In addition to Muybridge's "normal" subjects, he included examples of "abnormal" adults and children, with conditions such as hemiparesis, movement disorders, and limb deformity. Some of the adult subjects have recently been identified in casebooks from Penn, but the identity of the children remains unknown. The technology allowed neurologists and musculoskeletal experts to visualise the precise components of normal and abnormal gait, with a degree of detail difficult to recognise with the naked eye. As such, it was the precursor to modern "gait-lab" analysis.

This paper highlights some of Muybridge's "abnormal" subjects, and considers their images from various angles—as works of art, as products of emerging photographic technologies, and as key moments in the history of neurology. Importantly, the photographs also record glimpses of the subjects' personalities and adaptive strategies, that were perhaps unintended by Muybridge and Dercum. For the modern viewer, reconciling these different perspectives allows us to approach historical medical data in an ethically-sensitive manner.

12:15

Cannabidiol (CBD) for treatment resistant epilepsy of childhood (TREC) - experience from Western Australia

Najm Khan [1], Snehal Shah [1,2], Simon Williams [1], Jon Silberstein [1], Lakshmi Nagarajan [1,3,2]

[1] Perth Children's Hospital, Nadlands, WA, Australia
[2] Telethon Kids, Perth Children's Hospital, Perth, WA, Australia
[3] Medicine, University of Western Australia, Perth, WA, Australia

Objective: To assess whether CBD was beneficial and safe in our cohort of children with Treatment Resistant Epilepsy of Childhood (TREC).

Methods: This retrospective cohort study (1/2/2021- 31/5/2021) involved 41 children treated with CBD for TREC. Primary outcomes were effect of CBD on seizures and safety. Secondary outcomes were impact on sleep interruption, school function, disruption of family life, and change in overall condition of the child (using Caregiver Global Impression of Change (CGIC) scale). Pre and post CBD outcomes were compared using Wilcoxon signed rank test.

Results: At least 50% reduction in weekly seizure frequency occurred in 30/41 (73%) patients. Seizure duration, rescue medication use, emergency visits, status epilepticus episodes and hospital admissions decreased in 30 (73%), 13 (31.7%), 13 (31.7%), 11 (26.8%), and 10 (24.4%) of the cases respectively. One patient became seizure free. Ten patients experienced minor adverse events. At least 50% improvement in sleep interruption and school function score was reported in 18 (43.9%) patients. Disruption of family life scores improved in 22 (53.6%) cases. Overall condition of the child improved in 34 (83%) children on CGIC scale. Post CBD there was a statistically significant improvement in median weekly seizure frequency, sleep interruption, school function and disruption of family life scores. Other reported benefits included improvement in speech in 9 (21.9%) children, alertness in 6 (14.6%), and behaviour in 6 (14.6%).

Conclusions: CBD was beneficial and safe in reducing seizure burden, with positive impact on child and family function in our cohort of TREC patients.

PLATFORM ABSTRACTS - SESSION 2: NEUROMUSCULAR

16:00

Safety and efficacy of ataluren in nmDMD patients from Study 041, a phase 3, randomized, double-blind, placebo-controlled trial

Eppie Yiu [1], Anita Cairns [2], Michelle Lorentzos [3], Ian Woodcock [1], Maina Kava [4], Craig McDonald [5]

[1] Royal Children's Hospital, Parkville, VIC, Australia
[2] Queensland Children's Hospital, Brisbane, QLD, Australia
[3] The Children's Hospital at Westmead, Sydney, NSW, Australia
[4] Perth Children's Hospital, Perth, WA, Australia
[5] University of California Davis School of Medicine, Davis, California, USA

Background: Study 041 (NCT03179631) is an international, phase 3, randomised, double-blind, placebo-controlled 72-week ataluren trial in patients with nonsense mutation Duchenne muscular dystrophy (nmDMD) followed by a 72-week open-label period. We describe efficacy and safety results from the placebo-controlled phase.

Methods: Boys with nmDMD aged ≥5 years, on corticosteroids, and with a 6-minute walk distance (6MWD) ≥150m were eligible. The primary objective was to determine ataluren's effect on ambulatory function, assessed by the 6-minute walk test. Boys were randomised 1:1 to ataluren:placebo. The intention-to-treat (ITT) population comprised randomised boys who received ≥1 dose of study treatment. Predefined subgroups included boys with ≥300m 6MWD and ≥5s stand from supine (primary) and those with 300-400m 6MWD.

Results: Ataluren and placebo groups in the ITT population and key subgroups were balanced according to age, baseline 6MWD, corticosteroid use and time to stand from supine. Significant differences in mean 6MWD change from baseline and rate of change favoured ataluren in the ITT population (14.4m; 0.20m/week; p=0.0248) and 300-400m 6MWD subgroup (24.2m; 0.34m/week; p=0.0310), representing a 21% and 30% slowing of the decline rate in 6MWD respectively. There were significant treatment benefits in time to 10% worsening of 6MWD. The number of ITT patients who lost ambulation receiving placebo was almost double that of those receiving ataluren. Ataluren was well tolerated, had no probable drug-related serious adverse events (AEs), and AE frequency (85.3%) was similar to placebo (84.7%).

Conclusions: Study 041 confirms ataluren's favourable risk-benefit as shown in previous clinical and real- world evidence studies.

16:00

Ataluren preserves muscle function in nmDMD patients: a pooled analysis of results from three randomized, double-blind, placebo-controlled trials

Eppie Yiu [1], Kristi Jones [2], Anita Cairns [3], Michelle Lorentzos [2], Ian Woodcock [1], Maina Kava [4], Craig M McDonald [5]

[1] The Royal Children's Hospital, Melbourne, VIC, Australia
[2] Sydney Children's Hospital Network, Sydney, NSW, Australia
[3] Queensland Children's Hospital, Brisbane, QLD, Australia
[4] Perth Children's Hospital, Perth, WA, Australia
[5] University of California Davis School of Medicine, Davis, California, USA

Introduction: Study 041 (NCT03179631) is a phase 3, randomized, placebo-controlled 72-week ataluren trial in nonsense mutation Duchenne muscular dystrophy (nmDMD) patients.

Objectives: To describe results of a pooled analysis of ataluren muscle function efficacy results from Study 041 and two randomized, placebo-controlled 48-week ataluren trials (Study 007 [phase 2b] and Study 020 [phase 3]).

Method: Patients were eligible if they were male, had phenotypic evidence of DMD, and had an nmDMD diagnosis confirmed by genetic testing. Patients were randomized 1:1 (ataluren:placebo).

Pooled efficacy results for 48-week change in 6-minute walk distance (6MWD), North Star Ambulatory Assessment (NSAA) total and linear scores (where available), and timed function tests (TFTs; 10m walk/run, 4-stair ascent and 4-stair descent) are described for the overall pooled study population; and 48-week change in 6MWD for a key subgroup with baseline 6MWD 300–400m.

Results: The pooled study population included 354 patients receiving ataluren and 347 patients receiving placebo. Treatment with ataluren significantly reduced mean change from baseline in all measures vs placebo for the overall population (6MWD: 19.3m, p=0.0002; NSAA total score: 1.07, p=0.0010; NSAA linear score: 2.70, p=0.0031; 10m walk/run time: -1.31s, p=0.0001; 4-stair ascent time: -1.45s (p=0.0003); 4-stair descent time: -1.54s, p=0.0003). The pooled subgroup with baseline 6MWD 300–400m included 155 patients receiving ataluren and 157 patients receiving placebo. Ataluren preserved 32.1m of 6MWD in this subgroup vs placebo (p=0.0005).

Conclusion: Pooled placebo-controlled clinical trial data from 701 patients demonstrate that ataluren preserves muscle function, assessed by clinical meaningful endpoints, in nmDMD patients.

16:15

Epidemiology and clinical features of Duchenne and Becker Muscular Dystrophy in Australia - Findings from the Australian Neuromuscular Disease Registry

Chien Yen Poh [1,2], Robin Forbes [1], Justine Adams [1,2], Anita Cairns [3], Damian R Clark [4], Michelle A Farrar [5,6], Kathryn Irving [2], Kristi Jones [7], Andrew Kornberg [2], Kristen Nowak [8], Cullen O'Gorman [9], Anna O'Malley [1], Peter Rowe [10], Georgina Walter [1], Ian Woodcock [1,2], Eppie M Yiu [1,2]

[1] *Murdoch Children's Research Institute, Melbourne, VIC, Australia*
[2] *Royal Children's Hospital, Melbourne, VIC, Australia*
[3] *Queensland Children's Hospital, Brisbane, QLD, Australia*
[4] *Women's and Children's Hospital, Adelaide, SA, Australia*
[5] *Sydney Children's Hospital, Sydney, NSW, Australia*
[6] *School of Clinical Medicine, UNSW Sydney, Sydney, NSW, Australia*
[7] *Discipline of Child and Adolescent Health, University of Sydney, Sydney, NSW, Australia*
[8] *Western Australian Department of Health, East Perth, WA, Australia*
[9] *Princess Alexandra Hospital, Brisbane, QLD, Australia*
[10] *Perth Children's Hospital, Perth, WA, Australia*

Background: An understanding of the current clinical and therapeutic landscape of Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) in Australia is essential to improve clinical outcomes, healthcare provision and access to therapeutic.

Aims: To describe the epidemiology, clinical features and treatment practices of DMD/BMD in Australia.

Method: This was a cross-sectional study of children and adults with DMD/ BMD enrolled in the Australian Neuromuscular Disease Registry (ANMDR) from February 2020 to June 2023. Demographic, clinical and genetic data was analysed using descriptive statistics.

Results: As of June 2023, a total of 315 individuals with DMD/BMD are enrolled in the ANMDR, 270 (85.7%) with DMD and 45 (14.3%) with BMD. Dystrophin pathogenic variants include 209 deletions (65.9%), 64 point mutations (20.2%), and 38 duplications (11.9%). 201 (74.4%) individuals with DMD are currently treated with corticosteroids and 17 (6.3%) are on a disease modifying therapy. 152 (56.3%) participants with DMD are independently ambulant. Non-invasive ventilatory support is used by 55 (20.4%) participants with DMD compared to 2 (4.4%) with BMD. Data on other medical comorbidities, outcome measures and health service utilisation will also be presented.

Conclusion: In the context of a rapidly evolving treatment landscape, the ANMDR represents a powerful tool to improve our understanding of the healthcare needs of people with DMD and BMD in Australia. Findings from the registry can identify clinical outcomes, healthcare needs and inform provision of future therapeutics.

16:30

Improving the transition journey for neuromuscular patients in Queensland

Rebecca M Leung [1], *Kate Munro* [1], *Anita Cairns* [1]

[1] *Queensland Children's Hospital, South Brisbane, QLD, Australia*

Background: Children with neuromuscular disease are now able to live into adulthood thanks to advancements in medical care and this has created the need for adult specialists for conditions originating in childhood. Currently, there aren't any published guidelines to help this population's transition from paediatric to adult services. The transition phase, representing the process of moving from adolescence to adulthood, can be severely affected by growing up with a neuromuscular disorder, with significant impact on the patient and family's quality of life.

Methods: To evaluate our current neuromuscular transition model, we developed an online survey that has been distributed to clinicians, young people, and parents.

Results: Our study found that all parties involved in the transition process, noted problems with the current transition process. Patients found the adult-oriented clinicians, clinics and hospitals difficult to navigate and reported feeling unsupported when they left the paediatric setting. Parents found that transition at times was addressed too late, leaving them with little time to consider all the needs of the transition process. Some of the factors that were thought to have positively impacted on transition was the quality of transition preparation, longer duration of follow-up and availability of the paediatric neurologist and stable medical condition prior to transition.

Conclusion: This data will be utilised in conjunction with a working group to re-design the current transition pathway and ensure the right support for our patients with neuromuscular disease in Queensland.

16:45

Retrospective review of clinical presentation, neuroimaging features and treatment responsiveness of paediatric IIH presenting at tertiary care hospital

Aleks Khan [1], Snehal Shah [2, 3]

[1] School of Medicine, University of Western Australia, Nedlands, WA, Australia
[2] Lecturer, University of Western Australia, Nedlands, WA, Australia
[3] Department of Neurology, Perth Children's Hospital, Nedlands, WA, Australia

Background: Idiopathic intracranial hypertension (IIH) poses a diagnostic challenge in the paediatric population. Managing IIH in children is challenging due to limited medical options, the need for frequent lumbar punctures, and the possibility of ventriculoperitoneal (VP) shunt in refractory cases. There is a paucity of literature specific to paediatric IIH, necessitating a comprehensive evaluation of paediatric IIH.

Aim: The aim was to assess the common clinical and neuroimaging finding and treatment responsiveness in a cohort of patients with IIH.

Methods: Using the International Classification of Diseases (ICD) diagnostic criteria for IIH, we identified a cohort IIH over the past 5 years. Comprehensive clinical information was collected by analysing medical charts.

Results: A total of 50 children were included. There was female predominance ≥ 12 years cohort. The commonest clinical features were headache (90%) and visual symptoms (blurring/diplopia) (55%). Notably, 33% of children were referred due to incidentally detected papilledema. 85% ≥ 12 yrs and 55 % < 12 yrs were overweight. The predominant MRI features were optic nerve sheath enlargement (75%). The average number of lumbar punctures was 1.6 for those ≥ 12 and 2.4 for < 12 yrs. Medications used: Acetazolamide (94%), Glycerol (27%)/Topiramate (6%). 5 patients required VP shunt. The resolution of IIH was in 92% and headache in 90%. 35% experienced relapse of IIH.

Conclusion: Headache and visual symptoms were commonest clinical presentation. Overweight was significant co-morbidity in both pre and post pubertal children. The resolution of IIH and headache were achieved in majority.

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References:
1. Elliott, RE., et al,. (2011) Epilepsy & Behavior. 20; 478–483.
2. Orosz, I., et al. (2014) Epilepsia 55(10):1576-84.
3. Helmers, et al. (2011) Epilepsy Behav; 22:370-5.
4. Ben-Menachem, EJ., (2001) Clin Neurophysiol 18(5):415-8.
5. Morris, GL., 3rd et al., (1999) Neurology 53(8):1731-5.

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LivaNova Epilepsy

DAY 2	THURSDAY 14TH SEPTEMBER	
Time	**Event**	**Speaker**
09:00 - 11:00	**Symposium: Headache and Intellectual Disability** Chairs: Ian Andrews & Sophie Calvert	
09:00	Challenging headaches	*Jon Silberstein*
09:30	IIH - A developmental compliance disorder?	*Tyson Ware*
10:00	Genomic medicine of early onset neurodiversity	*Jozef Gecz*
10:30	Treatable metabolic causes of ID	*Drago Bratkovic*
11:00 - 11:05	**Kate Sinclair Memorial Scholarship Award 2023 Presentation** - Gina O'Grady	
11:05 - 11:30	**Morning Tea & Poster viewing**	
11:30 - 12:30	**Learning Health Systems in relation to Epilepsy** Chairs: Adriane Sinclair & Kerrie-Anne Chen	
11:30	Quality Improvement and Learning Health system approaches to epilepsy	*Nicholas Abend* LivaNova Health innovation that matters
12:10	Work smart and not harder: Increasing eMR documentation by improving workflow	*Hooi Ling Teoh*
12:30 - 13:30	**Lunch & Poster viewing**	
13:30 - 15:30	**Symposium: Adolescent Neurology** Chairs: Christina Miteff & Melinda Nolan	
13:30	Improving outcomes for adolescents with multiple sclerosis	*Eppie Yiu*
14:00	Transition in epilepsy	*Annie Bye*
14:30	Adolescent functional neurological disorders - moving from moral judgement to modern medical care	*Kate Riney*
15:00	More than a team of FND Champions: A pragmatic overview of what it takes to have an evidence-informed multidimensional rehabilitation approach	*Mark Pertini*
15:30 - 16:00	**Afternoon Tea & Poster viewing**	
16:00 - 16:45	**Platform Session 3** Chairs: Anita Cairns & Damian Clark	
16:00	Establishing a Gene-CP score to guide genetic testing in cerebral palsy *(p23)*	*Esther Tantsis*
16:15	Outcomes of Neonates with Hypoxic Ischemic Encephalopathy Treated with Magnesium Sulphate: A Systematic Review with Meta-Analysis *(p24)*	*Bhanu Gowda*
16:30	Kate Sinclair Memorial Scholarship Research Presentation	*Hannah Jones*
19:00 - 23:00	**Meeting Dinner, *Magill Estate Kitchen***	

PLATFORM ABSTRACTS - SESSION 3:

16:00 **Establishing a Gene-CP score to guide genetic testing in cerebral palsy**

Esther Tantsis [1,2], Michael Fahey [3], Yisselle Virella [1], Shekeeb Mohammad [1], Simon Paget [1], Angie Morrow [1], Michelle Farrar [4], Elizabeth Palmer [5], Mary-Clare Waugh [1], Yana Wilson [2], Maria Kyriagis [4], Chaya Goldman [3], Kavitha Kothur [1], Richard Webster [1], Sachin Gupta [1], Cathy Morgan [2], Anna te Velde [2], Rod Hunt [3], Gladys Ho [1], Bruce Bennetts [1], Jozef Gecz [6], Russell Dale [1,7]

[1] *CHW, Westmead, NSW, Australia*
[2] *Cerebral Palsy Alliance, Sydney, NSW, Australia*
[3] *Monash Children's Hospital, Melbourne, VIC, Australia*
[4] *Sydney Children's Hospital, Randwick, NSW, Australia*
[5] *University of NSW, Randwick, NSW, Australia*
[6] *Adelaide Medical School, The University of Adelaide, Adelaide, SA, Australia*
[7] *Paediatrics and Child health, University of Sydney, Sydney, NSW, Australia*

Background: In some children with cerebral palsy (CP) the cause is monogenic. A standardised approach to determine which children with CP should be tested doesn't exist. We aimed to establish a gene-CP score using 2 projects.

Methods: A DELPHI survey determined which factors were considered predictive of a monogenic cause of CP by experts. 26 experts (national/international) participated in the survey. The factors extracted were tested in a retrospective cohort of 100 CP patients (n=22 genetic, n=78 non-genetic).

Results: CP experts thought 10 factors predict a monogenic cause of CP; response to Levodopa, family history of similar presentation, persistent lactic acidosis, congenital anomalies, dysmorphic features, regression of skills, a normal MRI >2 years of age with or without epilepsy and Parkinsonism. History of birth asphyxia, extreme prematurity, MRI demonstrating typical changes of HIE, intraventricular haemorrhage, periventricular leukomalacia or ischaemic stroke were negatively associated with genetic CP.

In the retrospective review; dystonia in newborn (OR 5.82, CI1.8-18.81), intellectual disability (OR 5.33, CI 1.42-20.0), Dysmorphology(OR 4.29, CI 1.12-16.52), small head circumference <3rd (OR 4.09, CI 1.41-11.85), hypotonia (OR 3.54-1.04-11.97), malformation cortical development (OR 3.54, CI 1.04-11.97), regression of skills (OR 3.23, CI 1.18-8.81) were significant in the genetic CP group. Factors against a genetic cause were hemiplegic phenotype (OR 0.23, CI-0.05-0.7), normal intellect (OR 0.19, CI 0.05-0.70) and MRI compatible with clinical picture (Or 0.12, CI 0.04-0.36).

Conclusions: There is no standardised approach to genetic testing in CP, we have established predictive factors which will help create a gene-CP score.

16:15

Outcomes of Neonates with Hypoxic Ischemic Encephalopathy Treated with Magnesium Sulphate: A Systematic Review with Meta-Analysis

Bhanu B Gowda [1], Chandra Rath [2], Saravanan Muthusamy [3], Lakshmi Nagarajan [1], Shripada Rao [2]

[1] Neurology, Perth Children's Hospital, Nedlands, WA, Australia
[2] Neonatology, Perth Children's Hospital, Nedlands, WA, Australia
[3] Neonatology, Joondalup Health campus, Joondalup, WA, Australia

Objective: To assess Magnesium Sulphate (MgSO4) as a neuroprotective agent in hypoxic-ischemic encephalopathy.

Study Design: For this Systematic Review, PubMed, EMBASE, Cochrane Library, EMCARE and MedNar were searched in November 2022. Meta-analysis was conducted using Stata 16.0 and RevMan 5.3.

Results: Twenty RCTs (n=1485) were included, of which 16 were from settings where therapeutic hypothermia (TH) was not offered. *MgSO4 in settings where TH was not offered*: Only one study evaluated composite outcome of "Death or disability" at ≥18 months and reported such poor outcome in 8 out of 14 control infants and 4 out of 8 in the MgSO4 group. MgSO4 was not associated with mortality [RR: 0.86, CI: 0.72-1.03, 13RCTs)] or hypotension (RR:1.02, CI:0.88-1.18, 5RCTs). Thirteen studies reported that MgSO4 improved in-hospital outcomes such as reduced seizure burden, and improved neurological status at discharge. MgSO4 reduced the risk of poor suck feeds (RR: 0.52, 95%CI: 0.40-0.68, 6RCTs), and abnormal EEG (RR: 0.64, CI: 0.45-0.93, 5RCTs). Certainty of Evidence (COE) was 'moderate' for mortality and 'low/very low' for other outcomes. *MgSO4 as an adjunct to TH*: None reported on "death or neurodevelopmental disability" at ≥18 months. MgSO4 was not associated with mortality (RR 0.65, CI: 0.34-1.27, 3RCTs) or hypotension (RR: 1.0, CI: 0.71-1.40, 3RCTs).

Conclusions: Evidence around long-term outcomes of MgSO4 when used with or without TH was scant. MgSO4 therapy may improve in-hospital neurological outcomes without affecting mortality in settings where TH is not offered. Well-designed RCTs are needed, especially in low-resource settings.

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DAY 3	FRIDAY, 15TH SEPTEMBER	
Time	**Event**	**Speaker**
09:00 - 11:00	**Symposium: Dementia** Chairs: Roman Briest & Michael Cardamone	
09:00	Shining a light on childhood dementia: a burden of illness study	*Kristina Elvidge*
09:30	Stop all the clocks – Clues to the diagnosis of childhood dementia	*Alex Johnson*
10:00	Developing Therapies for Childhood Dementia	*Nicholas Smith*
10:30	Gene Therapy in Paediatric Neurology: Just the Beginning…	*Ian Alexander*
11:00 - 11:30	**Morning Tea & Poster viewing**	
11:30 - 12:30	**Platform Session 4 - Autoimmune** Chairs: Rakesh Patel & Sekhar Pillai	
11:30	The neuroinflammatory profile of disease progression in subacute sclerosing panencephalitis *(p27)*	*Joanne Davidson*
11:45	Genetic causes of infection induced encephalitis – A case series *(p28)*	*Olivia-Paris Quinn*
12:00	Neurotransmitter-related disorders in an Australian cohort *(p29)*	*Rachel Wong*
12:15	Contemporary analysis of the clinical and genetic landscape of spinal muscular atrophy – a registry-based study *(p30)*	*Lakshmi Balaji*
12:30 - 12:45	**Awards and close**	
12:45 - 13:45	**Lunch**	

PLATFORM ABSTRACTS - SESSION 4: AUTOIMMUNE

11:30 **The neuroinflammatory profile of disease progression in subacute sclerosing panencephalitis**

Joanne Davidson [1], Richard Webster [1], Philip Britton [2], Abhinav Rajkumar [2], Simone Ardern-Holmes [1], Russell Dale [3, 4, 1]

[1] T.Y. Nelson Department of Neurology and Neurosurgery, The Children's Hospital at Westmead, Sydney, NSW, Australia
[2] Infectious Diseases and Microbiology, The Children's Hospital at Westmead, Sydney, NSW, Australia
[3] Clinical Neuroimmunology and Movement Disorders, Kids Neuroscience Centre, Westmead, NSW, Australia
[4] Faculty of Medicine and Health, University of Sydney, Sydney, NSW, Australia

Background: Subacute sclerosing panencephalitis (SSPE) is an insidious and fatal illness occurring years after a primary measles infection. Further insight is required regarding the complex interplay between neuroinflammation and targeted treatment of the invasive measles virus to find an effective cure.

Methods: The clinical course and outcome of a child with SSPE, treated at a tertiary Australian hospital is described. Regular cerebrospinal fluid (CSF) sampling was performed and the neuroinflammatory profile established through progression of the disease clinical states by measurements of neopterin, quinolonic acid, kynurenin and tryptophan.

Results: A 9-year-old girl presented with rapid onset refractory myoclonic atonic seizures and neurodevelopmental regression after measles infection at 8 months of age. The diagnosis of SSPE was fulfilled according to Dyken's modified criteria. She showed initial clinical improvement and remission of seizures with carbamazepine, IVIG, ribavirin, inosine and interferon alpha-2b but within months had a significant deterioration from Jabbour clinical stage I to IV. Despite further treatment with molnupirivir, ruxolitinib, remdesivir and steroids she succumbed to her illness. Analysis of her CSF showed that the clinical deterioration was associated with an exponential increase in neopterin, quinolinic acid and the kynurenine/tryptophan ratio upon a background of already significantly raised neopterin.

Conclusions: Clinical progression through the final stages of SSPE is associated with a significant neuroinflammatory response including activation of the kynurenine pathway. CSF biomarkers play an important role in monitoring disease progression, inform our understanding of SSPE pathogenesis and identify potential pharmacological targets.

11:45 **Genetic causes of infection induced encephalitis – A case series**

Olivia-Paris Quinn [1], Kate Riney [1]

[1] *Neurology Department, Queensland Children's Hospital, South Brisbane, QLD, Australia*

Introduction: Patients with encephalitis following a viral infection are often thought to have a para infectious, inflammatory, or autoimmune cause for their presentation. These diagnoses usually result in treatments with immunosuppressant therapies, often continued for some time and associated with high rates of detrimental side effects[1,2]. However, there is an increasing body of evidence demonstrating that patients can have a direct genetic cause mediating viral infection triggered encephalopathy, where inflammation is a secondary response. Examples include mutations in UNC-93B1, TLR3 and DBR1 which result in encephalopathy and acquired brain injury with viral infections that can include HSV, and mutations in RANBP2 which can cause acute necrotising encephalopathy after viral trigger[3,4]. However, the literature on genetic causes of viral induced encephalitis is growing[5-7].

Methods: A small case series of paediatric neurology patients (n < 5) from a single institution with infection induced encephalitis and underlying genetic cause will be presented. Patients with a confirmed genetic cause of infection induced encephalitis were identified and approached by their treating neurologist for inclusion in this case series.

Conclusion: The case series presented raises awareness of this group of disorders. Identification of these patients is important, as these patients benefit, not from immunosuppressant therapies, but from protection from infection, including dedicated immunisation programs and early antiviral therapies at times of infections. Identifying the underlying gene can have implications for prognosis (risk of recurring encephalitis, risk of progressive neurological deterioration) and is also important for assessment of siblings and family planning.

12:00

Neurotransmitter-related disorders in an Australian cohort

Rachel SH Wong [1, 2], Dianah BA Hadi [1], Bindu Parayil Sankaran [3, 4], Russell Dale [1, 4], Hugo Sampaio [3], Katherine Lewis [5], Kate Lichkus [5], Carolyn Ellaway [5], Shanti Balasubramanium [5], Kaustuv Bhattacharya [5], Shekeeb Mohammad [1, 4]

[1] *TY Nelson Department of Neurology and Neurosurgery, The Children's Hospital at Westmead, Sydney, NSW, Australia*
[2] *The Royal Children's Hospital, Parkville, VIC, Australia*
[3] *The Sydney Children's Hospital Network, Westmead, NSW, Australia*
[4] *University of Sydney Children's Hospital at Westmead Clinical School, Faculty of Medicine and Health, Sydney, NSW, Australia*
[5] *Genetic Metabolic Disorders Service, The Sydney Children's Hospital Network, Westmead, NSW, Australia*

Background: The International Working Group on Neurotransmitter Related Disorders (iNTD) set up an international registry in 2014. The Sydney Children's Hospital Network (SCHN) joined the INTD in 2021. This registry provides a common platform to document the diagnostic aspects, genotype, phenotype, therapeutic response and natural history of individuals with neurotransmitter-related disorders. Many of these disorders are treatable and span a variety of neurological syndromes, such as cerebral palsy or epileptic encephalopathies. Methods: This cohort study recruited retrospective and prospectively diagnosed children with neurotransmitter disorders across SCHN, recording their biochemical and genomic diagnostic information, clinical features, therapeutic details and responses. We compared treatment approaches in our centre to consensus guidelines where available. Results: 16 patients were enrolled in the iNTD registry. Of these entries, 13 patients had disorders of tetrahydrobiopterin metabolism. The remainder had disorders in GABA-related and glycine metabolism. There were no patients with primary disorders of biogenic amines, serine deficiencies or cerebral folate deficiencies recruited. All diagnoses were confirmed either by genotype or a combination of specific biochemical tests in CSF, urine and blood. Time to diagnosis ranged from symptom onset to 5 years 8 months. None were diagnosed prenatally. Two patients remain clinically asymptomatic. One patient was noted to have intrauterine growth restriction. Postnatal outcomes include feeding problems, lethargy and hypotonia. Treatment involves a combination of dietary interventions, oral therapies, or none. Conclusions: This data provides the first insights into the phenotypic spectrum of these rare neurodevelopmental disorders and current therapeutic strategies in an Australian cohort.

12:15

Contemporary analysis of the clinical and genetic landscape of spinal muscular atrophy – a registry-based study

Lakshmi Balaji [1,2], Robin Forbes [3], Michelle A Farrar [1,2], Anita Cairns [4], Andrew Kornberg [5], Lauren Sanders [6,7], Phillipa Lamont [8], Christina Liang [9], Kristi Jones [10,11], Kristen Nowak [12], Cullen O'Gorman [13,14], Ian Woodcock [15], Eppie Yiu [15,3], Hugo Sampaio [16], Didu S T Kariyawasam [2,16]

[1] Neurology, Sydney Children's Hospital, Randwick, NSW, Australia
[2] University of New South Wales, Kensington, NSW, Australia
[3] Australian Neuromuscular Disease Registry, Murdoch Children's Research Institute, Melbourne, VIC, Australia
[4] Neurosciences, Royal Brisbane And Women's Hospital, Herston, QLD, Australia
[5] Neurology, The Royal Children's Hospital Melbourne, Parkville, VIC, Australia
[6] The University of Melbourne, Melbourne, VIC, Australia
[7] Neurology, St Vincent's Hospital, Melbourne, VIC, Australia
[8] Neurology, Royal Perth Hospital, Subiaco, WA, Australia
[9] Neurology, Royal North Shore Hospital, St Leonards, NSW, Australia
[10] Neurology, Children's Hospital at Westmead, Westmead, NSW, Australia
[11] Discipline of Child and Adolescent Health, University of Sydney, Sydney, NSW, Australia
[12] University of Western Australia, Perth, WA, Australia
[13] University of Queensland, Brisbane, QLD, Australia
[14] Princess Alexandra Hospital, Brisbane, QLD, Australia
[15] Neurology, Royal Children's Hospital, Parkville, VIC, Australia
[16] Neurology, Sydney Children's Hospital, Randwick, NSW, Australia

Background: Disease modifying therapies (DMTs) for spinal muscular atrophy (SMA) have significantly changed disease course and management. Within this evolving landscape, registries are a critical tool to increase knowledge, monitor interventions and support translational research to improve healthcare quality and guide service provision(1).

Methods: Cross-sectional study of individuals with genetically diagnosed 5qSMA enrolled in the Australian Neuromuscular Disease Registry (ANMDR) from 2020 to 2023. Demographic, clinical and genetic data was collated and analysed using descriptive statistics.

Results: As of June 2023, 195 individuals with SMA had been enrolled in the ANMDR (56% children, 44% adults; age range 0.6-75 years, 51% female). Five were deceased. Minimum Australian prevalence of SMA was 0.73/100,000 individuals and reduced with increased age. There was a phenotypic spectrum, with 1 SMA0(0.5%), 45 SMA1(23%), 76 SMA2(40%), 65 SMA3(33%), 2 SMA4(1%) and 6 presymptomatic/clinically silent(3%). Diagnosis was through newborn screening(5.1%), family screening(2.6%), prenatal screening(1%) or following symptoms/clinical assessment(91.8%). *SMN1* pathogenic variants included homozygous deletions of exon7(91.7%), compound heterozygous deletion and sequence changes(7.1%) and biallelic sequence changes(1.2%). Comorbidities related to weakness (scoliosis, contractures, impaired mobility) were frequent, and most prevalent in type 2 SMA. DMTs were prescribed for 96% children and 63%

adults. Access to and engagement with multidisciplinary care were significantly higher for children than adults (paediatric 85%, adults <10%).

Conclusions: The evolving diagnostic and therapeutic Australian landscape encompasses a varied population, with substantial morbidities and healthcare needs. Registry data identify the adult population of SMA to be less served under the multidisciplinary umbrella, representing an unmet need.

References:

[1] *Nathalie Jette, Megan Johnston, Tamara Pringsheim, Lawrence Korngut. The Case for Neurological Registry Best Practice Guidelines in Canada. Can J Neurol Sci. 2013 Jul;40(4 Suppl 2):S1-3. doi: 10.1017/s031716710001711x*

POSTERS

#2931 **Familial Epilepsy-Aphasia with SCN1A variant in a Father and Daughter**

Saranya Subbaraman [1], Nathan Haywood [2], Himanshu Goel [3], Sachin Gupta [4], Janis Brown [1], Laurence Roddick [1], Ian Wilkinson [1], Robert Smith [1]

[1] John Hunter Children's Hospital, New Lambton Heights, NSW, Australia
[2] HNEKids Rehab, Newcastle, NSW, Australia
[3] Hunter Genetics, Waratah, NSW, Australia
[4] The Children's Hospital at Westmead, Westmead, NSW, Australia

Background: Epilepsy-Aphasia syndromes present with variable speech/language impairment +/- seizures. They include Developmental Epileptic Encephalopathy with Spike Wave Activation in Sleep (DEE-SWAS) also known as a Continuous Spike and Wave during Slow Wave Sleep/CSWS; Electrical Status Epilepticus during Slow Wave Sleep/ESES and Landau-Kleffner Syndrome. There is a recognised genetic association with *GRIN2A* variants. We describe a familial SCN1A mutation segregated with aphasia in a father and daughter and also epilepsy with SWAS in the daughter.

Methods: Case reports.

Results: The father was diagnosed with Landau-Kleffner Syndrome at the age of 8 after rapid onset of aphasia. Available awake EEG results documented Left Centrotemporal spikes. He was treated with Carbamazepine/Acetazolamide and speech therapy with improvement. His EEG was normalised over 3 years.

His daughter presented at age 4.5 with rapid regression in her already delayed speech and language. Awake EEG revealed generalised spike-wave epileptic discharges and significant spike-wave activation in sleep. Seizures were consistent with atypical absence seizures. She was treated with pulse methylprednisolone, clobazam and sulthiame. At 11, she is in remission with EEG normalisation and she had no treatment for the past year. Speech and Language are stable.

Genetic testing identified a likely pathogenic heterozygous variant, c.2293C>A; p.[(Val765Met)]: in *SCN1A* in father and daughter. This was a novel variant that was not previously associated with Epilepsy-aphasia.

Conclusion: SCN1A variant in both the father and daughter adds to our knowledge of genetic links within the Epilepsy-Aphasia spectrum disorders. It also widens the phenotype of *SCN1A* associated epilepsies.

#2861

HaNDL in children: Case reports of two children with HaNDL and literature Review

Bhanu B Gowda [1], Najm Khan [1], Soumya Ghosh [1,2], Lakshmi Nagarajan [1]

[1] Neurology, Perth Children's Hospital, Nedlands, WA, Australia
[2] Neurology, Sir Charles Gairdner Hospital, Nedlands, WA, Australia

Introduction: The syndrome of Transient Headache and Neurologic Deficit with Cerebrospinal Fluid Lymphocytosis (HaNDL) is rare. We present two children with HaNDL.

Case1: A ten-year-old girl was admitted with a 2-week history of headaches, confusion, dysphasia, paraesthesia, and abdominal pain. She reported 3 similar episodes in the previous months. MRI Brain was normal. Papilledema was observed. There were no neurological deficits. CSF opening pressure was 48cm of H2O; there was normal protein and sugar, and 63 mononuclear cells. Infective, inflammatory, and autoimmune workup was unremarkable. There was asymmetric slowing on EEG. Her treatment included analgesics, Chlorpromazine, Cyproheptadine, Steroids and Topiramate. She was discharged 11 days later. Her medications were weaned over 6-8 weeks. She made a full recovery without recurrence at 24-month follow-up.

Case2: An 11-year-old boy presented with an episode of acute confusion, dysphasia, and fluctuating level of consciousness following a 1-day history of bifrontal headache and right-sided paraesthesia. He was afebrile. Neuroimaging and examination were normal. Inflammatory and autoimmune workup was unremarkable. EEG showed asymmetric slowing. Serum was positive for Mycoplasma IgM. CSF showed marginally elevated protein and sugar. CSF showed neutrophilic pleocytosis which is atypical for the ICDH-3:7.3.5 classification of HaNDL. He made a full recovery without recurrence at 12-month follow-up. The clinical picture and outcome fits best for HaNDL.

Conclusion: We present 2 cases of HaNDL that posed diagnostic challenges. Although HaNDL is rare in children, it's worth considering in atypical headache presentations.

#2863

A novel rapamycin cream formulation improves facial angiofibromas associated with tuberous sclerosis complex: a double-blinded, randomized, placebo-controlled trial

Phillip Aitken [1], Ioana Stanescu [1], Laura Boddington [1], Caroline Mahon [2], Andras Fogarasi [3], Yi-Hua Liao [4], Marta Ivars [5], Ester Moreno-Artero [5], Doris Trauner [6], Steven T DeRoos [7], Jasna Jancic [8], Milos Nikolic [9], Patricia Balazova [10], Harper N Price [11], Kinga Hadzsiev [12], Kate Riney [13], Stacie Stapleton [14], Megha M Tollefson [15], Derek Bauer [16], Blanka Pinkova [17], Hartley Atkinson [1]

[1] *AFT Pharmaceuticals Ltd, Auckland, New Zealand*
[2] *Dermatology Department, Christchurch Hospital, Christchurch, New Zealand*
[3] *Bethesda Children's Hospital, Budapest, Hungary, Hungary*
[4] *Department of Dermatology, National Taiwan University Hospital and National Taiwan University College of Medicine, Taipei, Taiwan*
[5] *Dermatology Department, Clínica Universidad de Navarra, Madrid, Spain*
[6] *Department of Neurosciences, University of California San Diego Health Sciences, San Diego, CA, USA*
[7] *Helen DeVos Children's Hospital, Grand Rapids, MI, USA*
[8] *Clinic of Neurology and Psychiatry for Children and Youth, University of Belgrade, Belgrade, Serbia*
[9] *Department of Dermatovenereology, University of Belgrade School of Medicine, Belgrade, Serbia*
[10] *Department of Pediatric Neurology, Comenius University, National Institute of Children's Diseases, Bratislava, Slovakia*
[11] *Division of Dermatology, Phoenix Children's Hospital, Phoenix, AZ, USA*
[12] *Department of Medical Genetics, University of Pécs, Hungary*
[13] *Neurosciences Unit, Queensland Children's Hospital, Queensland, Australia*
[14] *Johns Hopkins All Children's Hospital, St Petersburg, FL, USA*
[15] *Departments of Dermatology and Pediatrics, Mayo Clinic and Mayo Clinic Children's Center, Rochester, MN, USA*
[16] *University of Virginia, Charlottesville, VA, USA*
[17] *Department of Paediatric Dermatology, Faculty Hospital, Brno, Czech Republic*

Background: Tuberous Sclerosis Complex (TSC) is a rare genetic disorder which causes non-cancerous tumours in the brain, kidneys, and skin. Topical rapamycin can successfully treat Facial Angiofibroma (FA) in patients with TSC. Few topical formulations are available and compounded formulations are inadequate since rapamycin rapidly degrades. A novel stabilized topical cream which protects rapamycin from oxidation has been developed in 0.5% and 1% concentrations.

Methods: This multicenter, double-blind, randomized, placebo-controlled, dose-response phase II/III study with a parallel design aimed to assess the efficacy and safety of the topical cream. 107 participants aged 6-65 years with mild-to-moderate FA associated with TSC were randomized to receive topical rapamycin 0.5% (n=36), 1% (n=33), or placebo (n=38) once daily for 26 weeks. Efficacy was assessed using an Investigator Global Assessment (IGA) scale, the facial angiofibroma severity index, subject and clinician-reported percentage-based scales, and categorical improvement.

Results: Both strengths of cream led to statistically significant and clinically

meaningful improvements across multiple efficacy assessments at 26 weeks. 60.6% and 55.6% of patients treated with 1% and 0.5% rapamycin cream, respectively, achieved a measurable improvement on the IGA scale. 71.4% and 90% of patients reported they were "Moderately" or "Significantly" better following treatment with 1% or 0.5% rapamycin cream, respectively. Both strengths were well tolerated, with mild application site irritation the most common adverse event.

Conclusions: This novel, stable, rapamycin cream is effective at reducing angiofibromas and is well-tolerated.

#2865 **Hypertrophic Polyneuropathy in CMV-Related Guillain-Barré Syndrome: A Case Study with MRI Findings**

Bhanu B Gowda [1], Snehal Shah [1], Rahul Lakshmanan [2], Jonathan Silberstein [1]

[1] Neurology, Perth Children's Hospital, Nedlands, WA, Australia
[2] Radiology, Perth Children's Hospital, Nedlands, WA, Australia

We present a unique instance of hypertrophic polyneuropathy in a patient with CMV-related Guillain-Barré Syndrome (GBS) as depicted on MRI. The patient, a previously healthy five-year-old girl, exhibited a three-week history of bilateral leg pain, gradually worsening ataxia, and painful spasms in her paraspinal muscles. Clinical examination revealed bilateral proximal lower limb weakness, depressed deep tendon reflexes, and an ataxic gait. MRI findings demonstrated diffuse thickening of multiple cranial nerves, cervical and cauda equina nerve roots, as well as the brachial and lumbosacral plexuses. Extensive diagnostic workup, including metabolic, autoimmune, and infection evaluations, was conducted due to the striking MRI findings. Cytomegalovirus (CMV) IgM and IgG were positive in viral serology, and CMV was detected in the urine through PCR. The patient also tested positive for anti-ganglioside antibodies to GM-2. Treatment with intravenous immunoglobulin resulted in rapid symptom improvement, with a return to baseline function within six weeks. Follow-up MRI after three months demonstrated persistent enhancement and thickening of cranial nerves and plexus. This case highlights the rare occurrence of hypertrophic polyneuropathy in CMV-related GBS and emphasizes the favourable prognosis observed in the paediatric age group.

References:

(1) Zhu J, Zhang Y, Li R, et al. Anti-ganglioside Antibodies in Guillain-Barre Syndrome: A Novel Immunoblotting-Panel Assay. Front Neurol. 2021;12:760889. Published 2021 Nov 25. doi:10.3389/fneur.2021.760889 2. Khadilkar SV, Yadav RS, Soni G. A practical approach to enlargement of nerves, plexuses and roots. Pract Neurol. 2015;15(2):105-115. doi:10.1136/practneurol-2014-001004

#2899

Prediction of fractures in Duchenne muscular dystrophy: a tertiary paediatric hospital cross sectional study

Ellen West [1]

[1] *Sydney Children's Hospital, Randwick, NSW, Australia*

Background: Duchenne Muscular Dystrophy (DMD) is associated with an increased risk of long bone and vertebral fractures, with resultant pain and reduced mobility. A retrospective chart review of boys with DMD treated through the Queensland Children's Hospital (QCH) between 2015-2019 was undertaken to identify fracture rates and predictive factors for the development of fractures.

Methods: 103 patients were identified with 45 boys sustaining at least one life-time fracture (44%) and 23 sustaining at least one fracture (51%).

Results: The initial fracture was symptomatic vertebral compression fracture in 22 patients (49%) and long bone in 23 patients (51%) with the average age of initial fracture 11.2 years (5.7-13.8; SD 2.5) for vertebral fracture and 8.1 years (2.7 – 17.5; SD 4.2) for long bone fractures. Mean duration of corticosteroid treatment prior to fracture occurrence was 5.2 years (0-12.5 years; SD 3.8), with vertebral fractures occurring after 6.3 years (prednisolone 3.4 years, deflazacort 2.9 years) and long bone fractures 4.4 years (prednisolone 3.5 years, deflazacort 1.2 years). Mean bone mineral density (BMD) dual energy X-ray absorptiometry (DXA) age adjusted Z-scores at the time of fracture were -1.56, -3.43 and -2.73 for spine, hip and total body less head (TBLH), respectively. The mean North Star Ambulatory Assessment (NSAA) score of mobility at the time of fracture occurrence was 16 compared to 29 in boys without fracture.

Conclusion: No routine screening investigations show a clear correlation to fracture occurrence in DMD. Preventative interventions are required before 8 years of age for fracture risk minimisation.

#2902 ***GNB*-Related Neurodevelopmental Disorders: Case Series of Two Unrelated Families**

Sameer Dal [1], Janis Brown [1], Christina Miteff [1], Gopinath Subramanian [1], Robert Smith [1], Himanshu Goel [2]

[1] The John Hunter Children's Hospital, Newcastle, NSW, Australia
[2] Hunter Genetics, Waratah, NSW, Australia

Neurodevelopmental disorders encompass a spectrum of neurological & behavioural symptoms. While the aetiology of most cases remains elusive, there has been a rapid surge in the identification of underlying monogenic factors. One such family of genes (*GNB1* to *GNB5*) is related to beta subunits of G-protein signalling pathways that are initiated by G-protein coupled receptors (GPCRs). G-proteins, present in abundance in brain, eye and cardiac tissues transmit signals from various molecules and governs synaptic transmission, photoreception, and cellular growth[1-2].

We report two unrelated families with previously undiagnosed syndrome comprising global developmental delay, epilepsy, bradycardia, and neurodevelopmental disorders (NDD). One autosomal dominant family had 5 members with a novel *GNB1* variant. This variant was segregating well in the family with probability of 1/32. This family cohort was consisting of four affected children aged 3 to 12 years and their affected mother. They presented with a spectrum of intellectual disability (ID) from moderate to severe and distinctive language disorder. Additionally, comorbid conditions including autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) were prevalent among all.

The other family had an affected female with biallelic novel variant in *GNB5*. This is a known autosomal recessive condition. Our patient had previously described features, intellectual disability, epilepsy and sinus bradycardia in *GNB5* related NDD[2].

Conclusion: This report highlights the significance of G-protein signalling pathways and genetic variations in the manifestation of neurodevelopmental disorders. Further research is warranted to elucidate the underlying mechanisms and develop targeted interventions for individuals affected by these conditions[3].

References:

[1] Malerba N, De Nittis P, Merla G. The Emerging Role of Gβ Subunits in Human Genetic Diseases. Cells [Internet]. 2019 Dec 4 [cited 2020 Sep 15]; 8(12):1567
[2] Poke G, Sadleir LG, Merla G, Valles-Ibáñez G de, Skinner JR. GNB5-Related Neurodevelopmental Disorder [Internet]. www.ncbi.nlm.nih.gov. University of Washington, Seattle; 2021 [cited 2023 Jul 6]
[3] Kamato D, Thach L, Bernard R, Chan V, Zheng W, Kaur H, et al. Structure, Function, Pharmacology, and Therapeutic Potential of the G Protein, Gî±/q,11. Frontiers in Cardiovascular Medicine [Internet]. 2015 Mar 24;2

	TRAINEE DAY - FRIDAY, 15TH SEPTEMBER	
Time	**Event**	**Speaker**
13:45 - 15:15	Chairs: Romain Briest & Hooi Ling Teoh	
13:45	HIE. Neonatal encephalopathy	*Rod Hunt*
14:15	CP	*Ray Russo*
14:45	Congenital and neonatal infectons (HSV, CMV)	*Nan Vasilunas*
15:15 - 15:45	**Afternoon Tea**	
15:45 - 16:40	Chairs: Manoj Menezes & Tejaswi Kandula	
15:45	Small fibres, big problems: inherited and acquired autonomic neuropathies of childhood	*Manoj Menezes*
16:10	Recognising and Responding to Child Maltreatment	*Jane Edwards*
	TRAINEE DAY - SATURDAY, 16TH SEPTEMBER	
09:00 - 10:20	Chair: Alexandra Johnson	
09:00	Delivering bad news - a parents perspective	*Kate Beattie*
09:40	Ethical discussion about end of life decisions	*Nicholas Mills*
10:20 - 10:50	**Morning Tea**	
10:50 - 12:30	Chairs: Michael Cardamone & Christina Miteff	
10:50	Speech therapy: bulbar function, feeding difficulties	*Jane Fong*
11:30	Physio and OT therapies: how to make the most out of it	*Kirstie Morgan & Ashleigh Olsen*
12:30 - 13:10	**Lunch**	
13:10 - 15:00	Chair: Clair Pridmore	
13:10	An update on the hereditary ataxias	*Kishore Kumar*
13:50	Approach to Pediatric Leukodystrophies	*Ajay Taranath*
14:30	Approach to Prescribing Psychiatric medication	*Mohammed Usman*
15:00 - 15:15	**Afternoon Tea**	
15:15 - 16:45	Chair: Ellen Hurley	
15:15	Cardiology approach to POTS	*Dennis Lau*
15:45	Bone health in neurology (NM disease, AED, non ambulant patient): clinical approach	*Jenny Harrington*
16:15	Bad to the Bone: orthopaedic issues in neurology patients	*Nicole Williams*
16:45	**Close**	

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