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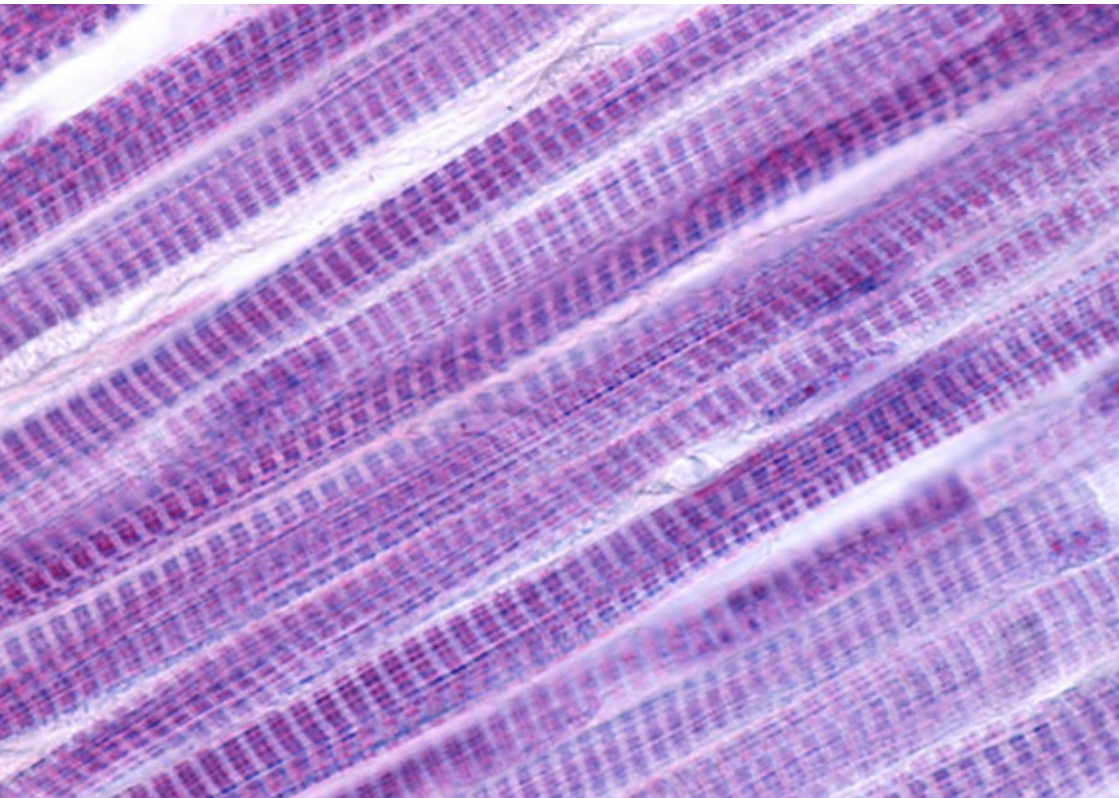
THE AUSTRALIA AND NEW ZEALAND
CHILD NEUROLOGY SOCIETY

13th Annual Scientific Meeting & Education Day

3th – 6th September 2025

Brisbane Convention and Exhibition Centre (BCEC)

South Bank, Brisbane, Queensland



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WELCOME MESSAGES

Welcome Message from the Local Organisers

On behalf of the organising committee and the ANZCNS executive, we welcome you to 13th Annual Scientific Meeting of the Australia and New Zealand Child Neurology Society in Meanjin/Brisbane.

As always, our Annual Scientific Meeting is dedicated to promoting the best possible care for children with neurological disease across Australia and New Zealand. This meeting brings together leading international and local experts to share knowledge, spark collaboration, and inspire innovation. Our guest international speaker this year is Dr Ana Ferreiro from the Myology Research Centre / Institute of Myology in Paris. We are looking forward to her sharing her knowledge and expertise with our group.

The programme features sessions on neuromuscular disorders, neuroinflammation, maternofetal medicine, neonatal neurology, and neuropathology, alongside timely updates in general neurology. We're proud to introduce the inaugural Wisdom of Our Elders session, celebrating the experience and insights of senior clinicians, and a stimulating ethics discussion that invites reflection on the evolving responsibilities in the era of precision medicine.

Platform presentations and poster sessions will showcase research from ANZCNS members and trainees, reinforcing our commitment to education, scientific excellence, and advocacy. We anticipate this meeting will be of interest to paediatricians, allied health professionals, and consultant and trainee child neurologists.

We hope you will join us for the conference dinner for the opportunity to reconnect with old friends and to make new ones. We also encourage you to explore Brisbane and stay on to experience Riverfire!

Anita Cairns
2025 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 2025 Annual Scientific Meeting. The Organising Committee has arranged an impressive line-up of overseas and Australia-based international experts in childhood neurological disorders who will showcase recent advances and local research in paediatric neuroscience. I look forward to seeing you all at the meeting.

Manoj Menezes
President, Australia and New Zealand Child Neurology Society 2025

About the Australia and New Zealand Child Neurology Society

The Australia and New Zealand Child Neurology Society (ANZCNS) is a collaborative group of approximately 150 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)

Anita Cairns, Chair
Matthew Lynch
Sophie Calvert
Snehal Shah
Stephen Malone
Sameer Dal

GENERAL CONFERENCE INFORMATION

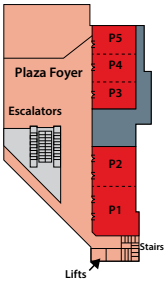
Secretariat Office

For queries arising after the congress, 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

Brisbane Convention and Exhibition Centre (BCEC)
Cnr Merivale and Glenelg Streets, South Bank, Brisbane
Plaza Rooms 1 to 4



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Parking

The car park has direct lift access to the Centre's multiple convention floors and exhibition facilities. Car park 1 is best for Merivale Street rooms, Car Park 2 & 3 are best for Exhibition Halls and Grey Street rooms via Russell Street Walkway. The car park is open 24 hours a day.

Casual parking rates for Monday – Sunday.

0-2 HOUR	2-3 HOURS	3-4 HOURS	4+ HOURS	EARLY BIRD *	AFTER DARK *
$15.00	$20.00	$25.00	$35.00	$16.00	$20.00

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**After Dark rate: Enter after 6pm and exit before 1am*
Max daily rate $35.00 - Note: 24-hour parking from time of entry to time of exit the following day
All car parks are cashless. Payment can only be made by credit or debit card

Brisbane / Meanjin

Registration Desk

The registration desk is located in the Plaza Foyer, at the BCEC.
Please note that name badges must be clearly displayed at all times.

Registration desk opening hours are:

Wednesday 3rd September:	07:30 - 17:00
Thursday 4th September:	08:00 - 17:00
Friday 5th September:	07:30 - 17:00
Saturday 6th September	08:00 - 13:00 (Education / Trainee Day)

Speakers' Information

Speakers must submit their final presentation to the audiovisual technician in the main plenary room (Plaza Rooms 3 & 4) **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 13th 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 Plaza Rooms 1 & 2, 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 Exhibition Area, Plaza Rooms 1 & 2. 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 13th 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 13th 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

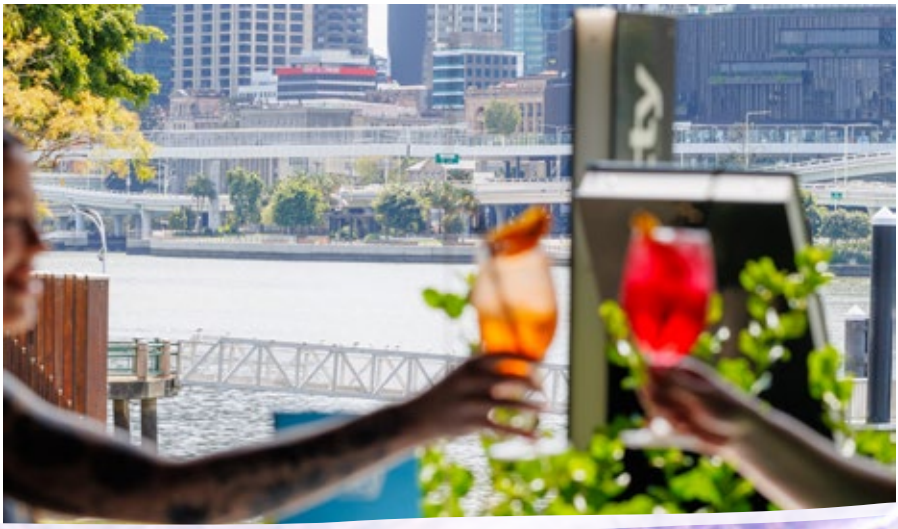
The Australia and New Zealand Child Neurology Society will not accept any liability for personal injury or loss/damage to property/belongings of participants or accompanying persons, either during or following the conference or their stay. It is recommended that delegates arrange their own personal health,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

MEETING DINNER

The Meeting dinner will be held at The Jetty South Bank, a 600 metre walk from the Mantra South Bank Hotel and 800m walk from the BCEC. Situated at the northern end of South Bank's River Quay dining precinct, The Jetty is nestled in a riverside nook with spectacular uninterrupted views across the Brisbane River to the CBD.

Venue: The Jetty South Bank, River Quay 4 Sidon Street, Brisbane
Date: Thursday 4th September
Time: 7.00 - 10.30pm

the jetty SOUTH BANK



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EXHIBITOR		Exhibition Space #
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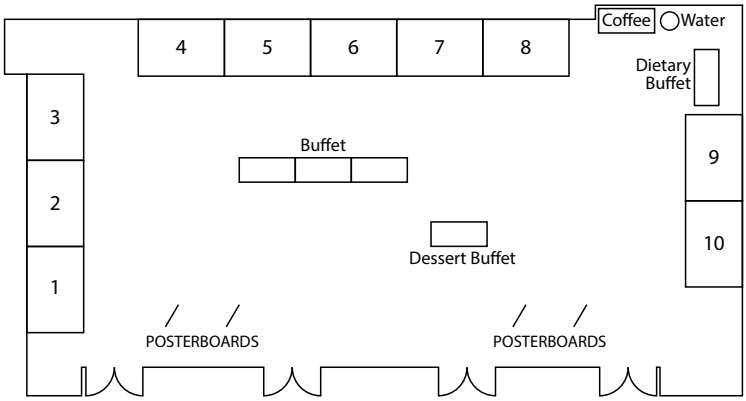
ucb Inspired by patients. Driven by science.	UCB	7 & 8

EXHIBITOR FLOORPLAN



INTERNATIONAL SPEAKER

We are delighted to welcome our international speaker:

Dr Ana Ferreiro is a Neuromuscular Neurologist at the Reference Center for Neuromuscular Diseases, Neuromyology Department and a translational scientist and Research Director at the Myology Research Centre / Institute of Myology, both in the Pitié-Salpêtrière University Hospital in Paris. For over 25 years, she has combined clinical care with basic, translational and therapeutic research on muscle diseases, with a particularly focus on congenital myopathies and desmin-related myopathies.
Her work led to the phenotypical and molecular description of several entities (i.e. congenital titinopathies, recessive RYR1-related myopathies and desminopathies, SELENON-related myopathy) and to the identification of pathophysiological pathways and potentially therapeutic drugs. Dr Ferreiro was for 6 years (2019-2024) the Research Director of the European Neuromuscular Center (ENMC) and serves currently as an Executive Associate Editor for Neuromuscular Disorders.

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Driven by **science.**

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DAY 1	WEDNESDAY 3RD SEPTEMBER	
Time	**Event**	**Speaker**
07:30 - 17:00	**Registration Desk open** Plaza Foyer	
08:45 - 09:00	**Welcome**	*Manoj Menezes*
09:00 - 11:00	**Symposium 1: Congenital neuromuscular disorders** Chairs: Manoj Menezes & Tyson Ware	
09:00	Congenital myopathy overview	*Ana Ferreiro* *Sponsored By* Inspired by patients. Driven by science.
09:40	Outcomes of SMA newborn screening	*Didu (Sandi) Kariyawasam*
10:10	Challenging genetic diagnoses	*Anita Cairns*
10:40	Respiratory complications of neuromuscular conditions	*Leanne Gauld*
11:00 - 11:30	**Morning Tea**	
11:30 - 13:00	**Platform Session 1 - Abstract Presentations** Chairs: Didu Kariyawasam & Claire Pridmore	
11:30	A systematic review of patient-reported and observer-reported outcome measures of fatigue in pediatric neuromuscular disorders *(p12)*	*Monica Marzouk*
11:42	Orthopaedic and bone health surveillance and management in a single paediatric tertiary centre *(p13)*	*Vanessa Hwee Ling Ng*
11:54	Assessment of respiratory morbidity, sleep dysfunction and fatigue in children with Charcot-Marie-Tooth Disease *(p14)*	*Jyoti Raghunandan*
12:06	Neuropalliative care within the Women's and Children's Hospital Network: A retrospective clinical audit of current models of care *(p15)*	*Aneka Larsen*
12:18	Challenges in the diagnosis of Guillain-Barré Syndrome in paediatrics *(p16)*	*Dev Seetharaman*
12:30	Consensus based clinical practice recommendations in the translation and implementation of advanced therapies and experimental neurotherapeutics in paediatric neurological disorders *(p17)*	*Natalie Lim*
12:42	Posterior Reversible Encephalopathy Syndrome with spinal cord involvement (PRES-SCI) in a child: A case report and literature review *(p18)*	*Jia Yi Leong*
13:00 - 14:10	**Lunch & Poster viewing**	

14:10 - 15:10	**Platform Session 2 - Abstract Presentations** Chairs: Christopher Troedson & Adriane Sinclair	
14:10	Clinical and radiological presentation of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS): two case reports from Western Australia *(p19)*	*Randal Moldrich*
14:22	Temporal Encephaloceles in Paediatric Temporal Lobe Epilepsy: Revisiting Negative MRI and FDG-PET Clues *(p20)*	*Halla Malik*
14:34	The Caregiver Burden of Developmental and Epileptic Encephalopathies: A Scoping Review *(p21)*	*Jeremy New*
14:46	Effectiveness and safety of an unregistered cannabidiol formulation in a compassionate access scheme for children with uncontrolled epilepsy *(p22)*	*Jia Yi Leong*
14:58	Does a treatment strategy for neonatal seizures stratified by seizure burden work prospectively? *(p23)*	*Cynthia Sharpe*
15:10 - 15:40	**Afternoon Tea**	
15:40 - 17:00	**Symposium 2: Maternofetal Medicine** Chairs: Sophie Calvert & Erik Andersen	
15:40	Imaging	*Samuel Manikkam*
16:00	Genetics	*Tony Roscioli*
16:20	Challenges in fetal counselling	*Michael Fahey*
16:40	Antenatal CMV treatment	*Renuka Sekar*
17:05 - 18:30	**ANZCNS Annual General Meeting**	

PLATFORM ABSTRACTS - SESSION 1:

11:30 **A systematic review of patient-reported and observer-reported outcome measures of fatigue in pediatric neuromuscular disorders**

Monica Marzouk [2, 1], Kayla Cornett [2, 3], Gabrielle Donlevy [2, 3], Carl Young [4], Joshua Burns [5], Manoj Menezes [2, 3], Marnee Mckay [2]

[1] Health Sciences, University of Sydney, Sydney, NSW, Australia
[2] Health Sciences, University of Sydney, Camperdown, NSW, Australia
[3] Paediatric Gait Analysis Service of New South Wales, Sydney Children's hospitals Network, Sydney, NSW, Australia
[4] Paracelsus Medical University, Nuremberg, Bavaria, Germany
[5] St. Jude Children's Research Hospital, Memphis, Tennessee, United States of America

Background: Fatigue is a complex symptom of neuromuscular disorders. Patient-reported outcome measures (PROMs) and observer-reported outcome measures (ObsROMs) are essential for understanding patient perspective of fatigue and providing patient-centred care. Understanding the range and quality of fatigue measures is fundamental for measuring and managing fatigue in neuromuscular disorders. This systematic review aims to summarise, compare and critically appraise studies that use PROMs and ObsROMs to measure fatigue in paediatric neuromuscular disorders.

Methods: Six electronic databases were searched. Primary investigations using PROMs and/or ObsROMs of fatigue in paediatric neuromuscular disorders were included using keywords and MeSH terms relating to 'fatigue', 'paediatric' and 'neuromuscular disorders'. The Newcastle-Ottawa Scale was used to assess risk of bias of included studies. The COSMIN risk of bias checklist was used to evaluate psychometric measurement properties of the fatigue PROMs identified in included studies.

Results: 5606 titles and abstracts were screened; of these 5373 were excluded based on having adult populations only, no neuromuscular disorder or no PROM or ObsROM of fatigue. 225 full texts were reviewed; of these 57 were included. 44 different fatigue PROMs and ObsROMs were used in addition to interviews and simple questions. 55% of included studies scored an unclear risk of bias on the Newcastle-Ottawa scale. Over 70% of the PROMs scored inadequately according to COSMIN ratings.

Conclusions: The results of this review indicate a lack of standardisation of fatigue measures, while highlighting the poor psychometric measurement properties of fatigue PROMs used in paediatric neuromuscular disorders.

11:42 **Orthopaedic and bone health surveillance and management in a single paediatric tertiary centre**

Vanessa Hwee Ling Ng [1], Anita Cairns [1], Craig Munns [2], Kate Munro [1], Nicole Thomas [1], Megan Reading [1]

[1] Paediatric neurology, Queensland Children's Hospital, South Brisbane, QLD, Australia
[2] Department of Endocrinology, Queensland Children's Hospital, South Brisbane, QLD, Australia

Background: Spinal muscular atrophy (SMA) is a neuromuscular condition characterised by progressive muscle atrophy and weakness. The interplay between progressive muscular atrophy, immobility and the role of SMN1 in bone metabolism puts SMA patients at high risk of musculoskeletal complications, osteoporosis, and fractures. The emergence of disease modifying therapies have improved mortality rate and motor function for patients. This highlights the importance of preventative measures in optimising co-morbidities such as bone and musculoskeletal health. While the 2017 International Standards of Care provide recommendations for surveillance and treatment, significant variability persists in clinical practice.

Methods: This is a retrospective cohort study conducted at Queensland Children's Hospital, including 51 children with SMA - Type 1(16), Type 2(21), and Type 3(14). Demographic and clinical data related to bone and orthopaedic management were analysed descriptively.

Preliminary results: Fifty-one children were included in the study (male=25). The median age was 9.05 years. Bone mineral density (BMD) assessments were conducted in 50/51 patients, with low BMD observed in 47/50. Long bone fractures were noted in 12/51, and 4/12 had commenced bisphosphonate therapy. Scoliosis was reported in 35/51 patients, of whom 9/35 underwent spinal surgery. Hip subluxation was identified in 27/51 patients. Hip dislocations were present in 9/51. Bilateral coxa valga was present in 27/51 patients. Hip surgery was performed in 3/51. Surveillance practices were inconsistent across the cohort.

Conclusion: Orthopaedic complications and poor bone health remain prevalent in SMA, with variability in assessment and management. Standardised surveillance protocols are needed to optimise long-term musculoskeletal outcomes.

11:54 **Assessment of respiratory morbidity, sleep dysfunction and fatigue in children with Charcot-Marie-Tooth Disease**

Jyoti Raghunandan 1, Manoj Menezes 1, Gabrielle Donlevy 1

[1] *Children's Hospital Westmead, Westmead, NSW, Australia*

Inherited neuropathies, and particularlyCMT (Charcot Marie Tooth Disease), are lifelong progressive conditions which significantly impact daily living and functioning. Understanding the prevalence and nature of the fatigue, respiratory and sleep dysfunction in children with these conditions will help in improving quality of life and patient experience.

Exploratory Prospective Cohort Study (Pediatric Focus)

- Design: Observational, prospective cohort study.
- Participants: 30 Children and adolescents (0–18 years) with CMT, attending the peripheral neuropathy Management clinic at the Children's Hospital Westmead
- Assessments:
  - Validated PROMs for sleep dysfunction (Pediatric Sleep Disturbance Scale), fatigue scales (Multidimensional fatigue scale).
  - CMT Pediatric Severity Score (e.g., CMTPedS).
- Analysis: Prevalence estimates and correlation/regression analysis for association with disease severity
- This project aims to bridge critical gaps in understanding and managing these under-recognized but impactful complications of CMT and support clinicians in optimizing care pathways for these children

12:06

Neuropalliative care within the Women's and Children's Hospital Network: A retrospective clinical audit of current models of care

Aneka Larsen [1], *Clair Pridmore* [1], *Mary Cossich* [2]

[1] *Neurology, Women's and Children's hospital, North Adelaide, SA, Australia*
[2] *Palliative Care, Women's and Children's hospital, North Adelaide, SA, Australia*

Background: International experts emphasise the need to integrate palliative care into Neurology, though guidance on implementation remains limited. We reviewed referral pathways at the Women's and Children's Hospital to identify gaps and inform guideline development.

Methods: We examined Paediatric Palliative Care (PPC) referrals for all current patients with neurological diagnoses and for deceased patients with neurological conditions over the past three years. Patients were grouped by disease type to compare referral patterns.

Results: Of the 96 patients currently managed by PPC, 44 had a neurological disease (45%). 66% of these diagnoses were neurodegenerative, 27% neuromuscular, 18% cerebral palsy, 18% refractory epilepsy and 4.5% hypoxic ischaemic encephalopathy (HIE). Of the 83 deceased cases, 31 had a neurological disease (37%). 29% were HIE, 23% neurodegenerative, 16% neuromuscular, 13% acute infective neurological disorders, 6% cerebral palsy, 6% neurometabolic (non-degenerative) and 3% refractory epilepsy. Among current patients, Neurology referred 58.3% of those with neuromuscular disease, 34.5% of neurodegenerative cases, 25% of refractory epilepsy and none with cerebral palsy or HIE. Among the deceased, Neurology referred 80% of neuromuscular disease cases, 43% of neurodegenerative patients, and none belonging to the other groups.

Conclusions: Neurological diagnoses account for a substantial portion of PPC referrals. Neurology frequently refers neuromuscular cases but less commonly initiates referrals for other groups. We believe these referral disparities could be explored further with a view to establishing clearer, disease-specific guidelines. Ongoing research will explore referral timing and caregiver experiences to further inform service integration and guideline development.

12:18 **Challenges in the diagnosis of Guillain-Barré Syndrome in paediatrics**

Dev Seetharaman [1], *Gopinath Subramanian* [1], *Christina Miteff* [1]

[1] *Paediatric Neurology, John Hunter Children's Hospital, Newcastle, NSW, Australia*

Background: Guillain-Barré Syndrome (GBS) remains largely a clinical diagnosis with wide clinical variability. Studies have previously shown that GBS may present atypically, particularly in younger children, leading to diagnostic delay [(1)]. This study aims to highlight the variation of diagnostic characteristics of GBS and the limitations of the diagnostic criteria in paediatrics.

Methods: A retrospective review of medical records of children (< 18 years) diagnosed with GBS at John Hunter Children's Hospital between 2009 and 2025 was conducted. The Brighton criteria was used as a guideline for the diagnosis.

Results: 10 cases (6 girls) of GBS with mean age of 7.1 years (range 1.1 – 14.0) were identified. Bilateral, symmetric limb weakness was present in all patients. Key clinical findings noted were pain (90%), areflexia (80%), cranial nerve involvement (20%) and younger children typically presented with refusal to weight bear (70%). An antecedent event was present in 9 cases with viral illness (60%), gastroenteritis (20%) and immunisation (10%) implicated. When performed, 6/7 (87%) patients had cytoalbuminologic dissociation, 6/6 (100%) had positive nerve conduction studies, 5/9 (56%) had nerve root enhancement on MRI, and 3/6 (50%) had positive ganglioside antibodies. The median time to disease nadir was 5 days. Only 30% fulfilled Brighton level 1, 20% level 2, 20% level 3 and 30% level 4 of diagnostic certainty.

Conclusions: We further highlight the variable nature of GBS presentation in paediatrics and the limitations of the Brighton criteria. GBS should be considered in non-weight bearing children particularly in the younger paediatric cohort.

References:

[(1)] *Roodbol J, de Wit MC, Walgaard C, de Hoog M, Catsman-Berrevoets CE, Jacobs BC. Recognizing Guillain-Barre syndrome in preschool children. Neurology. 2011;76(9):807-810. doi:10.1212/WNL.0b013e31820e7b62*

12:30 **Consensus based clinical practice recommendations in the translation and implementation of advanced therapies and experimental neurotherapeutics in paediatric neurological disorders**

Natalie Lim [1], Didu Kariyawasam [1,2], Christian Meagher [2], Michelle Farrar [1,2]

[1] Department of Neurology, Sydney Children's Hospital Network, Randwick, NSW, Australia

[2] School of Clinical Medicine, UNSW Medicine and Health, Randwick Clinical Campus, NSW, Australia

Background: Advanced therapies (ATs), including gene and stem cell therapy, and experimental neurotherapeutics hold great potential in paediatric neurology. However, their swift development poses significant data and implementation gaps. This modified Delphi study was conducted to inform a consensus-driven framework guiding the safe and effective integration of these therapies into the Australian health system.

Methods: The study uses a modified Delphi methodology with 19 purposively recruited experts in paediatric neurology, ethics, clinical research, governance, clinical trials and patient advocacy. A scoping review was presented to panel members through an online workshop generating 108 statements which were embedded in an electronic questionnaire (Q1). Using a 5-point Likert scale, the panel rated each of the statements and provided qualitative data through open text. Consensus was defined a priori as an agreement by at least 75% of the participants. Qualitative and descriptive data analysis of Q1 informed the second questionnaire (Q2) which included feasibility ratings.

Results: Four key domains were identified: (i) access pathways and responsibilities, (ii) communication and engagement with families, (iii) eligibility and patient selection, and (iv) evaluating the effects of trials or treatment.

Following Q2, 101 recommendations met consensus which were summarised and consolidated into a practical resource for clinicians and stakeholders. Respondents highlighted feasibility challenges relating to equity, sustainability and scalability.

Conclusions: These statements echo the call to action to build and equip a speciality and system that is ready for ATs. It provides a clinical perspective on the anticipated challenges and hopes these therapies hold for the future.

12:42 **Posterior Reversible Encephalopathy Syndrome with spinal cord involvement (PRES-SCI) in a child: A case report and literature review**

Jia Yi Leong [1], Sarah Curnow [1], Ahmed Kaki [1], Omar Alomar [2], Joshua Kausman [3, 4, 2], Simon Carter [2]

[1] *Department of Neurology, Royal Children's Hospital, Parkville, Melbourne, VIC, Australia*
[2] *Department of Nephrology, Royal Children's Hospital, Melbourne, VIC, Australia*
[3] *Department of Paediatrics, University of Melbourne Parkville, Melbourne, VIC, Australia*
[4] *Neurosciences, Murdoch Children's Research Institute, Parkville, Melbourne, VIC, Australia*

We report the rare entity of Posterior Reversible Encephalopathy Syndrome with spinal cord involvement (PRES-SCI) in a 14-year-old presenting with malignant hypertension and grade four hypertensive retinopathy causing near blindness, secondary to bilateral renal dysplasia. MRI brain showed diffuse T2 signal abnormalities involving the white matter, grey matter and MRI spine showed extensive, longitudinal expansile central spinal cord T2-hyperintensity from the cervical spine to the conus. She had no neurological signs referrable to this. She was admitted to ICU and was treated with IV antihypertensives before being transitioned onto 3 oral anti-hypertensives. The imaging changes completely resolved on follow-up neuroimaging once her hypertension was well-controlled.

A literature review revealed 15 other paediatric cases of PRES-SCI. 75% of this patient group had a renal cause for their presentation. Only 25% (4/16) presented with seizures. 31% (5/16) had symptoms referrable to the spinal cord lesion, 3 out of the 5 cases (60%) had a motor deficit.10/16 children (62%) had typical MRI changes in the bilateral parieto-occipital regions with spinal cord changes. 4/16 children (25%) had involvement of the entire spinal cord. 14/16 (87.5%) children achieved full recovery, and 100% had complete resolution of neuroimaging findings at follow up.

PRES-SCI should be considered as a differential diagnosis when patients present with extreme elevation in blood pressure, MRI changes that extend to the cervico-medullary junction or the whole spine with or without presence of neurological signs. Early recognition is vital to ensure timely treatment and to avoid unnecessary investigations.

PLATFORM ABSTRACTS - SESSION 2:

14:10 **Clinical and radiological presentation of autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS): two case reports from Western Australia**

Randal Moldrich [1], Lakshmanan Rahul [2], Jon Silberstein [1], Snehal Shah [1,3], Maina Kava [1,3,4]

[1] Paediatric Neurology, Perth Children's Hospital, Perth, WA, Australia
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Background: Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is an early-onset neurodegenerative disease caused by mutations in the SACS (Sacsin) gene. It is characterized by progressive cerebellar ataxia, pyramidal spasticity and peripheral neuropathy. The mean age at onset is approximately six years (range: 0-40 years).

Methods: Two unrelated children of non-consanguineous and non-Québécois origin were referred for delayed walking and frequent falls noted at 18 months and at 2 years of age. Both children had been seen by physiotherapists and other specialities before been seen by neurologists at 5 years of age.

Results: Case 1 had reduced lower limb reflexes with evolution of pyramidal signs and mild cerebellar signs including ataxia by 8 years of age. Case 2 had ataxia, early cerebellar signs, mild spasticity and decreased reflexes in the lower limbs. Metabolic workup was normal. MRI brain showed linear striated T2 hypointense signals within the pons, corticospinal tracts and thalami with cerebellar vermian volume loss. Genetic testing identified two pathogenic heterozygous mutations in the SACS gene in each child. In child 2 one mutation was somatic mosaic. All mutations are predicted to result in partial loss of Sacsin protein function.

Conclusions: A high index of clinical suspicion and early neuroimaging in children with delayed walking and mixed upper and lower motor neurone signs is needed for early diagnosis of ARSACS . Although no treatment is yet available, genetic family counselling can be provided in a timely fashion.

14:22

Temporal Encephaloceles in Paediatric Temporal Lobe Epilepsy: Revisiting Negative MRI and FDG-PET Clues

Halla Malik [1], Nolette Pereira [1], Amelia Jardim [1], Chris Ryan [2], Bruce Goodwin [1], Stephen Malone [1], Sameer DAL [1]

[1] Queensland Health, Brisbane City, QLD, Australia
[2] Queensland Health, Bundaberg Hospital, Bundaberg, QLD, Australia

Background: Temporal encephaloceles (TE) are an under-recognised, surgically remediable cause of drug-resistant temporal lobe epilepsy (TLE), infrequently seen in adults and rarely reported in children. We describe the electroclinical and radiological features of two paediatric cases.

Methods: Two children with TE and epilepsy were identified at the Queensland Children's Hospital. To our knowledge, these represent the only TE cases recorded at our centre, one of the largest paediatric epilepsy surgery units in Australia. Electroclinical and radiological features were examined.

Results: Two right-handed male children (aged 14 and 10 years) with TLE were diagnosed with TE after epilepsy onset (3 years in Patient A; 6 months in Patient B). Patient A experienced focal impaired consciousness (FIC) seizures and one focal-to-bilateral tonic-clonic seizure (FBTC). EEG showed background asymmetry, spike-slow wave and a seizure with ictal rhythm over the L anterior temporal region. After being missed on initial review, re-review of MRI revealed a left TE (8×5×6 mm) with ipsilateral temporal lobe hypometabolism on 18FDG PET. Patient A remains drug-resistant and is scheduled for epilepsy surgery. Patient B presented with two presumed FBTC's and daily FIC seizures prior. EEG showed mild left posterior asymmetry without interictal discharges. MRI identified a left TE (8×6×6 mm). He commenced levetiracetam monotherapy and remains under evaluation.

Conclusion: TE are a rare but potentially surgically remediable cause of paediatric TLE, often overlooked at initial epilepsy diagnosis. Optimal detection relies on clinical awareness, advanced epilepsy MRI protocols, expert neuroradiology interpretation, and PET which provides valuable diagnostic clues in presumed non-lesional cases.

14:34

The Caregiver Burden of Developmental and Epileptic Encephalopathies: A Scoping Review

Jeremy New [1], *Jeremy Welton* [1,2], *Kate Riney* [3,4], *Hansoo Kim* [1]

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[4] *Faculty of Medicine, University of Queensland, South Brisbane, QLD, Australia*

Background: Developmental and epileptic encephalopathies (DEE) are a group of severe neurological disorders characterised by seizures, cognitive impairment, and developmental delay. The complex symptomology and high care needs place a substantial and multifaceted burden on caregivers.

Methods: A structured literature search was conducted in Embase, supplemented by hand-searching, to identify research focused on caregiver burden in DEE. Inclusion criteria required primary research focused on caregiver burden. Eligible studies were thematically analysed across five key burden domains: emotional, physical, financial, social and relationship, and time and work.

Results: Twenty-six studies met the inclusion criteria, encompassing caregivers of patients with Dravet syndrome, Lennox-Gastaut syndrome, and other rarer DEE. Despite methodological heterogeneity, caregivers consistently reported significant burden in all domains. High levels of emotional stress, anxiety, physical fatigue and sleep deprivation were frequently cited. High direct and indirect costs disrupted social and family lives, and a significant disruption to the caregivers' careers were reported. Variability in study design and inconsistent use of validated instruments limited cross-study comparability.

Conclusions: The burden of care for caregivers of patients with DEE is pervasive but inconsistently measured across the literature. Future research should be aimed at standardisation in caregiver outcome reporting in DEE, and developing a specific outcome measures to assess the holistic impact of caregiving for people with DEE. Including such a tool in clinical trials would enable more accurate evaluation of quality-of-life outcomes for both patients and caregivers to support more comprehensive health technology assessments and policy decisions.

14:46

Effectiveness and safety of an unregistered cannabidiol formulation in a compassionate access scheme for children with uncontrolled epilepsy

Jia Yi Leong [1], Jeremy Freeman [1], Emma Macdonald-Laurs [1,2], Jill Bicknell-Royle [1]

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Background: Cannabidiol (CBD) is an effective adjunctive anti-seizure medication (ASM) for uncontrolled epilepsy in children with Dravet syndrome, Lennox-Gastaut syndrome, and tuberous sclerosis complex (TSC).[1,2,3] Our audit aims to assess effectiveness and safety of an unregistered CBD product in an open-label compassionate access scheme for children with uncontrolled epilepsy.

Methods: Audit of a prospectively collected case series of 115 children treated in an epilepsy clinic. Eligibility criteria: daily seizures or admissions for severe seizures, and failed treatment with >4 other ASMs. Effectiveness was assessed after 3 months of treatment and at last follow up based on seizure diaries. Adverse events were based on parental reports. Primary outcome: proportion of children with >50% seizure reduction after three months. Secondary outcomes: adverse events, proportion who stopped treatment and reasons for cessation.

Results: 62 children (54%) had >50% seizure reduction after 3 months, 8 children (7%) were seizure-free. Adverse events were seen in 75 children (65%). 30 children (26%) stopped treatment after the initial evaluation period due to lack of effectiveness. 5 children (4.3%) stopped treatment due to side effects. Concurrent use of clobazam was associated with risk of somnolence [OR 6.7 (95% CI 2.9 – 15.3), $p < 0.001$]. Concurrent use of sodium valproate was associated with risk of liver enzyme abnormality or worsening. [OR 8.5 (95% CI 3.1 – 23.1), $p < 0.001$].

Conclusions: An unregistered CBD product might be effective and safe adjunctive treatment for children with uncontrolled seizures due to a broad range of epilepsy syndromes and causes.

References:

(1) *Devinsky O, Cross JH, Laux L, et al. Trial of Cannabidiol for Drug-Resistant Seizures in the Dravet Syndrome. N Engl J Med. 2017;376:2011-20.*
(2) *Thiele EA, Bebin EM, Bhathal H, et al. Add-on Cannabidiol Treatment for Drug-Resistant Seizures in Tuberous Sclerosis Complex: A Placebo-Controlled Randomized Clinical Trial. JAMA Neurol. 2021;78:285-92.*
(3) *Thiele EA, Marsh ED, French JA, et al. Cannabidiol in patients with seizures associated with Lennox-Gastaut syndrome (GWPCARE4): a randomised, double-blind, placebo-controlled phase 3 trial. Lancet. 2018;391:1085-96.*

14:58 **Does a treatment strategy for neonatal seizures stratified by seizure burden work prospectively?**

Cynthia Sharpe [1], *Suzanne Davis* [1], *Hannah Jones* [1]

[1] *Paediatric Neurology, Starship Children's Hospital, Auckland, New Zealand*

Background: Retrospective analyses suggest that seizure density of 10 minutes per hour can separate neonates with seizures into two groups. Those with high seizure burden warrant immediate treatment with phenobarbital. Those with lower seizure density can be treated first with non-neurotoxic anti-seizure-medication and will remain responsive to treatment with phenobarbital if needed for several hours. However, seizure density can rapidly escalate and there have not been any prospective studies. Within the NEOLEV3 trial study neurologists calculate seizure density to decide whether to treat with Levetiracetam or phenobarbital. There is ongoing monitoring of seizure density throughout the study.

Methods: The number of patients requiring treatment with phenobarbital in patients stratified to initial treatment with LEV will be reported. Maximum seizure density, total seizure burden, time to treatment with phenobarbital and efficacy of phenobarbital will be reported.

Results: Of 32 consecutive patients with electrographic seizures 27 were stratified to initial treatment with LEV. 8 required 60mg/kg LEV only. 6/27 had increasing seizure density and moved to standard of care treatment with PHB. 13 remained below threshold and received randomized higher dose LEV or PHB. 8/13 breached seizure density of 8 min/hr. Data analysis of additional metrics as described above is underway but not yet complete.

Conclusions: Prospective identification of patients with lower seizure density is difficult, a margin is required. First-line treatment decisions should take into account that patients emerging from burst-suppression HIE will inevitably escalate to high seizure density. Time to treatment can be reduced by advanced preparation of medications.

References:

[1] *Sharpe C, Rennie-Younger CR, Han DY, Davis SL, Nespeca M, Pisani F, Gold JJ, Reiner GE, Wang S, Haas RH. Efficacy of phenobarbital is maintained after exposure to mild-to-moderate seizures in neonates. Epilepsia Open. 2025 Apr 5. doi: 10.1002/epi4.70020. Epub ahead of print. PMID: 40186516.*

DAY 2	THURSDAY 4TH SEPTEMBER	
Time	**Event**	**Speaker**
08:00 - 17:00	**Registration Desk open** Plaza Foyer	
08:30 - 10:30	**Symposium 3: Neuromuscular disorders** Chairs: Melinda Nolan & Hugo Sampaio	
08:30	Translational research in early-onset myopathies: from bedside to bench and back	*Ana Ferreiro* *Sponsored By* Inspired by patients. Driven by science.
09:10	Rhabdomyolysis	*Emily Oates*
09:40	Limb girdle muscular dystrophy	*Ian Woodcock*
10:10	Update in ataxia telangectasia	*Matthew Lynch*
10:30 - 11:00	**Morning Tea & Poster viewing**	
11:00 - 13:00	**Symposium 4: Neuroimmunology** Chairs: Simon Williams & Twinkle Ghia	
11:00	Autoimmune movement disorders	*Hannah Jones*
11:30	Myelin Oligodendrocyte Glycoprotein Antibody -Associated Disease	*Stefan Blum*
12:00	Investigations in neuroinflammation	*Fabienne Brilot-Turville*
12:30	Psychiatry in neuroimmunological disorders	*James Scott*
13:00 - 13:10	**2025 Kate Sinclair Memorial Scholarship award**	
13:10 - 14:00	**Lunch & Poster viewing**	
14:00 - 15:00	**Platform Session 3 – Abstract Presentations** Chairs: Hannah Jones & Sameer Dal	
14:00	Stroke management in paediatric bacterial meningitis – a 10-year retrospective observational study at a single tertiary paediatric centre *(p26)*	*Halla Malik*
14:12	Optimising Neurovascular Surveillance in Loeys-Dietz Syndrome Using Electronic Medical Records: A Paediatric Cohort Audit *(p27)*	*Olivia-Paris Quinn*
14:24	Paediatric-onset multiple sclerosis: A Single Centre Study 2014-2024 *(p28)*	*Rebecca Leung*
14:36	The effectiveness and safety of therapeutic plasmapheresis for idiopathic acute transverse myelitis: A systematic review of the literature *(p29)*	*Elizabeth Cole*
14:48	Positive impact of miglustat treatment in five children with CLN3 Batten disease *(p31)*	*Alexandra Johnson*

15:00 - 15:30	**Afternoon Tea & Poster viewing**	
15:30 - 16:30	**Symposium 5: Ethics Session - Precision Medicine** Chair: Anita Cairns	
	Why N=1 clinical trials are ethical	*Cynthia Sharp*
	Why N=1 clinical trials are unethical	*Michelle Lorentzos*
	Discussion	*Rebecca Doyle*
16:30 - 17:00	**Wisdom of Our Elders** Chair: Matthew Lynch	
	Lessons learnt from fingers burnt	*Robert Smith, Annie Bye & Lakshmi Nagarajan*
19:00 - 22:30	**Meeting Dinner - *The Jetty***	

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1. Australian Government Department of Health and Aged Care. Pharmaceutical Benefits Schedule. www.pbs.gov.au. **2.** Australian Approved Product Information for Koselugo® (selumetinib).

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ALEXION AstraZeneca Rare Disease

PLATFORM ABSTRACTS - SESSION 3:

14:00 **Stroke management in paediatric bacterial meningitis – a 10-year retrospective observational study at a single tertiary paediatric centre**

Halla Malik [1], Rebecca Ward [1], Adriane Sinclair [1]

[1] Queensland Health, Brisbane City, QLD, Australia

Background: Paediatric stroke complicating bacterial meningitis, occurring in 17-43% of cases, is a devastating combination with poorer outcomes including higher rates of long-term disability and mortality. [1,2] There are no best practice guidelines on stroke management in bacterial meningitis, including use of corticosteroids and anti-thrombotic therapy. This study aims to understand the current clinical characteristics and outcomes in this patient cohort.

Methods: Retrospective audit, analysing electronic records of children, aged 4 months to 18 years, admitted to Queensland Children's Hospital over a 10-year period (2015-2025) was conducted. Inclusion criteria included: confirmed bacterial meningitis on cerebrospinal PCR or culture and neuroimaging confirmed stroke.

Preliminary-Results: 13 patients with bacterial meningitis had confirmed stroke (4 females, 9 males), 3 months to 10 years of age, 4/13 of pacific islander heritage. Organisms isolated: Streptococcal pneumoniae (9/13), Neisseria meningitidis (2/13), Escherichia coli (1/13) and Haemophilus influenzae (1/13). Stroke type: ischaemic (11/13; multi-territory 9/11), haemorrhagic (1/13). Stroke management: corticosteroids (4/13), aspirin (2/13), heparin (2/13) and no treatment (8/13). 5/13 were on empiric corticosteroids prior to stroke diagnosis. Stroke outcome: mortality (5/13), morbidity (5/13) and no disability (3/13).

Conclusion: Stroke complicating bacterial meningitis was associated with high mortality (38%) and neurodisability (63%) in survivors. Majority were multi-territory ischaemic strokes (81%) caused by Streptococcus pneumoniae (70%) with age under 4 and pacific islander groups over-represented. Stroke treatment choice and decision not to treat varied, with unclear outcomes of effect. More research is needed to ascertain what factors can help lessen stroke severity, to guide how and when to intervene.

References:

(1) Defeating meningitis by 2030: a global road map. Geneva: World Health Organization; 2021. Licence: CC BY-NC-SA 3.0 IGO.

(2) Dunbar M, Shah H, Shinde S, Vayalumkal J, Vanderkooi OG, Wei XC, Kirton A. Stroke in pediatric bacterial meningitis: population-based epidemiology. Pediatric Neurology. 2018 Dec 1;89:11-8.

14:12

Optimising Neurovascular Surveillance in Loeys-Dietz Syndrome Using Electronic Medical Records: A Paediatric Cohort Audit

Olivia-Paris Quinn [1], *Nicola Fearn* [1]

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

Introduction: Monogenic vasculopathies in childhood, such as Loeys-Dietz syndrome (LDS), carry significant neurovascular risks, including both ischaemic and haemorrhagic stroke. LDS is an autosomal dominant connective tissue disorder associated with cardiovascular, skeletal, and craniofacial abnormalities. Cerebrovascular features may include arterial tortuosity, aneurysms, and dissections. Despite these risks, there is no standardised approach to surveillance or patient education. This project leverages electronic medical record (EMR) capabilities to identify affected children and optimise neurology referrals and care pathways.

Methods: A customised EMR report was developed at the Royal Children's Hospital (RCH) to identify patients under 18 with a diagnosis of LDS. Demographic details, clinical history, neuroimaging status, and involvement of neurology services were reviewed.

Results: Twenty-six children were identified (median age at diagnosis: 7 years, IQR 7–8.75). Of these, 77% had undergone magnetic resonance angiography (MRA) of the head and neck, and 65% had been reviewed by the RCH neurovascular service. Among those imaged, 71% demonstrated arterial tortuosity and one patient had a haemorrhagic stroke. Neurovascular surveillance was initiated in previously unmonitored patients, and neurology referrals were made where absent.

Conclusion: This audit underscores the cerebrovascular risks in LDS and the need for consistent, standardised neuroimaging protocols. Harnessing EMR tools facilitates early identification, enhances workflow, and may serve as a scalable model for other monogenic vasculopathies. Broader adoption of such approaches could inform a national strategy for paediatric neurovascular surveillance in genetic disorders.

14:24

Paediatric-onset multiple sclerosis: A Single Centre Study 2014-2024

Rebecca M Leung [2, 1], Kate Riney [2]

[1] PA Hospital, Woolloongabba, QLD, Australia
[2] Queensland Children's Hospital, South Brisbane, QLD, Australia

Background: Epidemiological data of paediatric-onset multiple sclerosis (POMS) in Australia is limited. Objective: To determine the demographic and clinical characteristics of POMS in Queensland, Australia.

Methods: A retrospective study was conducted to assess the clinical characteristics of multiple sclerosis (MS) patients who were treated at the Queensland Children's Hospital between 2014-2024. POMS was defined as those who had disease onset prior to the age of 18 and fulfilled the International Paediatric MS Study Group (IPMSSG) criteria for MS.

Results: Of the 10 POMS who were assessed, there was an even gender split. The mean age at onset was 11.9 years. Supratentorial clinical manifestations (90 %) were the most frequent presentation at disease onset, followed by visual pathway (50 %) and spinal (20 %) symptoms. At the last follow-up visits, all patients (100 %) remained in a relapsing-remitting phenotype. The annual relapse rate (ARR) was 0.48 throughout the first 2 years while on treatment. At the baseline visit, the 90 % of the cohort were commenced on platform therapies. Breakthrough disease and adverse events (60 %) were the most common indications to escalate or switch to other disease-modifying drugs (DMTs).

Conclusion: Most POMS patients continued to be in a relapsing phenotype in our longitudinal study. Disease breakthrough is common in POMS especially when using platform therapies.

References:

(1) *syndrome (GWPCARE4): a randomised, double-blind, placebo-controlled phase 3 trial. Lancet. 2018;391:1085-96.*

14:36

The effectiveness and safety of therapeutic plasmapheresis for idiopathic acute transverse myelitis: A systematic review of the literature

Elizabeth Cole [1, 2], Ahmed Kaki [1], David Metz [3, 4, 5], Eppie M Yiu [1, 3, 6]

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[3] *Murdoch Children's Research Institute, Melbourne, VIC, Australia*
[4] *Nephrology department, The Royal Children's Hospital, Melbourne, VIC, Australia*
[5] *Monash University, Melbourne, VIC, Australia*
[6] *The University of Melbourne, Melbourne, VIC, Australia*

Background: Acute transverse myelitis (ATM) is a rare neuroinflammatory condition of the spinal cord which can cause life-changing neurologic sequelae.[1,2] Therapeutic plasmapheresis (TPE) is used as second- or third-line therapy after corticosteroid failure.(3) Given the paucity of evidence for its' efficacy and safety in children with ATM, we assess evidence for TPE in this population.

Methods: Systematic literature review (SLR) was conducted using Medline, Embase and PubMed to screen literature from 1946 – April 2024. Titles and abstracts were screened in duplicate by at least two reviewers and included if they reported data on idiopathic ATM TPE recipients < 18 years old. Evidence was graded according to Australian National Health and Medical Research Council (NHRMC) evidence hierarchy. Evidence was assessed using Joanna Briggs Institute (JBI) Critical Appraisal Checklist for case series.

Results: Seven case series describing 56 paediatric idiopathic ATM cases who received TPE were included.[3-10] Female to male ratio was 1.3:1. For children with outcome measures available, there was a positive TPE response in 72%. Early initiation of TPE did not affect outcomes. Adverse effects occurred in 40% of children but only 12% of cycles performed; 80% were mild. Evidence grading was NHMRC level IV.

Conclusion: This SLR was limited to case series with inherent bias risks and data which may not be applicable across all population groups. Pre- and post-TPE comparison outcomes improved in most cases. TPE had a low risk of serious AEs. TPE should be considered as second-line rather than third-line therapy for idiopathic ATM.

References:

[1] *Lopez Chiriboga SF, E. P. Myelitis and Other Autoimmune Myelopathies. CONTINUUM: Lifelong Learning in Neurology. 2021;27(1):62 - 92.*
[2] *Beh SC, Greenberg BM, Frohman T, Frohman EM. Transverse Myelitis. Neurologic Clinics. 2013;31(1):79 - 138.*
[3] *Celik H, Aksoy E, Oztoprak U, Ceylan N, Aksoy A, Yazici MU, et al. Longitudinally extensive transverse myelitis in childhood: Clinical features, treatment approaches, and long-term neurological outcomes. Clinical Neurology and Neurosurgery. 2021;207*

(4) *Akcay N, Barlas UK, Bektas G, Kihtir HS, Sevketoglu E. Therapeutic plasma exchange in pediatric patients with acute demyelinating syndromes of the central nervous system: A single-center experience. Transfusion and Apheresis Science. 2022;61(4).*
(5) *Bigi S, Banwell B, Yeh EA. Outcomes after early administration of plasma exchange in pediatric central nervous system inflammatory demyelination. J Child Neurol. 2015;30(7):874-80.*
(6) *Noland DK, Greenberg BM. Safety and efficacy of plasma exchange in pediatric transverse myelitis. Neurol. 2018;8(4):327-30.*
(7) *Prytula A, Vande Walle J, Verhelst H, Eloot S, Claus S, De Jaeger A, et al. Therapeutic plasma exchange in children with acute autoimmune central nervous system disorders. Int J Artif Organs. 2015;38(9):494-500.*
(8) *Savransky A, Rubstein A, Rios MH, Vergel SL, Velasquez MC, Sierra SP, et al. Prognostic indicators of improvement with therapeutic plasma exchange in pediatric demyelination. Neurology. 2019;93(22):e2065-e73.*
(9) *Shah YD, Eksambe P, Fomani K, Louie J, Shefali K, Kothare S. Feasibility & safety of plasma exchange in paediatric neuro-immunology: A single center experience. Eur J Paediatr Neurol. 2020;27(pp 94-97):94-7.*

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SMA=spinal muscular atrophy. **References: 1.** SPINRAZA® Approved Product Information. **2.** Hagenacker T, *et al. Lancet Neurol.* 2020;19(4):317–25. **3.** Coratti G, *et al. Orphanet J Rare Dis.* 2021;16(1):430. SPINRAZA® and Biogen® are registered trademarks of Biogen MA Inc. ©2025 Biogen. Biogen Australia, Macquarie Park, NSW. ABN 30 095 760 115. Biogen-270318. BIOG1333/EMBC. Date of preparation: August 2025.

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14:48

Positive impact of miglustat treatment in five children with CLN3 Batten disease

Alexandra Johnson [1,2], Alice E Dutton [3,4], Ineka T Whiteman [5,6], Michael M Jones [3,4], Katie E Geering [3], Stephanie Crofts [3], Soheil Afshar [1], John R Grigg [3,4]

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[2] University of New South Wales, Sydney, NSW, Australia
[3] Ophthalmology, Children's Hospital at Westmead, Sydney, NSW, Australia
[4] Save Sight Institute, Sydney, NSW, Australia
[5] Batten's Disease Support and Research Association (Australia, Columbus), Sydney, NSW, Australia
[6] Beyond Batten Disease Foundation, Austin, Texas, USA

Background: CLN3 disease is a childhood dementia syndrome presenting with visual loss, cognitive decline, seizures, motor decline and premature death. At present there is no approved disease-modifying treatment. Miglustat has shown promise of disease stability in CLN3 disease cell and animal models, and a small adult cohort. We hypothesized a further positive effect if commenced early in the disease course.

Methods: Five patients with genetically confirmed classical CLN3 disease were prescribed oral miglustat (weight-based to maximum 600mg daily), commenced between September 2023 and October 2024. Main outcomes were visual acuity, functional assessments (Vineland-3, Unified Batten Disease Rating Score (UBDRS)). Subjective comment regarding vision, behaviour, cognition and motor skills was sought from families, carers and educators.

Results: Patients were aged between 8 years 10 months and 12 years 10 months (median 9.9 years) at treatment commencement, with current treatment length between 6-20 months. Improvements in visual acuity were reported in two patients. Vineland-3 and UBDRS assessments demonstrate stability/improvement for all patients to date. Subjective observations were grouped, and noted improvements from early in treatment (starting at 3 months) for all five children in overall behaviour, improved learning (commensurate with peers for some), and also the functional impact of improved vision in two children.

Conclusions: Five children with early stage CLN3 disease treated with miglustat show meaningful improvements in behaviour, cognition and visual function. In some patients visual acuity also improved. Further studies are needed to assess this effect over the longer term.

DAY 3	FRIDAY, 5TH SEPTEMBER	
Time	**Event**	**Speaker**
07:30 - 17:00	**Registration Desk open** Plaza Foyer	
08:00 - 10:30	**Symposium 6: Updates in Neurology** Chairs: Lakshmi Nagarajan & Harry Singh	
	Session Sponsored By PTC THERAPEUTICS	
08:00	Update in migraine	*Sophie Calvert*
08:25	Update in seizure classification	*Melinda Nolan*
08:50	Update in Anti-Seizure Medication	*Sameer Dal*
09:15	Update in epilepsy surgery	*Ellen West*
09:40	Update in concussion	*Karen Barlow*
10:05	Update on stroke	*Adriane Sinclair*
10:30 - 11:00	**Morning Tea & Poster viewing**	
11:00 - 12:20	**Symposium 7: Neonatal neurology and neuropathology** **Chris Burke memorial session** Chair: Stephen Malone	
11:00	Vale Chris Burke	*Barry Appleton*
11:05	Neonatal epilepsy	*Lakshmi Nagarajan*
11:30	Hypoxic ischaemic encephalopathy	*Paul Colditz*
11:55	Neuropathology	*Abel Devadass / Krishnan Iyengar*
12:20 - 12:30	**Awards & Close** Anita Cairns & Manoj Menezes	
12:30 - 13:30	**Lunch**	

POSTERS

#3259 **A Pathogenic Variant in The Micu1 Gene: Siblings with Myopathy with Extrapyramidal Signs and a Literature Review**

Nezir Özgün [1], Uğur Gümüş [2]

[1] Mardin Artuklu University Medicine Faculty, Artuklu, MARDIN, Turkey
[2] Center for Genetic Diseases Evaluation, Dr. Ersin Aslan Education and Research Hospital, Gaziantep, Turkey

Introduction: Myopathy with Extrapyramidal Signs (MPXPS) develops due to homozygous pathogenic variants in the Mitochondrial Calcium Uptake 1 (MICU1) gene. The disease is characterized by early-onset proximal myopathy, learning disability and progressive extrapyramidal movement disorder. In this article, the clinical and genetic findings of two siblings are examined in the light of the literature, in order to make MPXPS both rare and to increase the evidence about genotype and phenotype.

Method: Since the patient was not diagnosed despite detailed examinations, whole exome sequencing was performed.

Result: A 6-year-old male patient and his sister presented to our clinic with complaints of weakness and frequent falls while walking. They creatine kinase and liver function tests were high. No etiology could be detected in the muscular dystrophy gene panel, metabolic tests and other tests performed. WES was done. The analysis revealed a homozygous, nonsense alteration [NM_001195518 c.553C>T p.(Arg185*)] in the *MICU1* gene.

Conclusion: MPXPS should be considered as a possible diagnosis, especially in pediatric patients who have myopathies with multisystemic involvement, whose symptoms appear after an infection or whose clinical conditions deteriorate.

#3727 **Rare occurrence of congenital neuroblastoma and tuberous sclerosis**

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Despite known correlation between neurocutaneous disorders and malignancy, Tuberous Sclerosis and neuroblastoma is quite rare with only one previous case in the literature.[1,2] This case follows patient A.F., a male diagnosed antenatally with congenital right adrenal neuroblastoma, later found to have TSC (tuberous sclerosis) following incidental brain MRI findings. Initial investigations revealed a right adrenal mass and elevated urinary catecholamines, with MRI confirming a suprarenal lesion. At ten months, surveillance imaging identified a brain lesion, not consistent with metastasis. Following surgical resection of the neuroblastoma, repeat MRI brain imaging showed findings consistent with TSC. Targeted genetic testing performed revealed a low-level mosaic nonsense variant in the TSC2 gene. This case highlights extremely rare but significant association of TSC with neuroblastoma.

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#3729 **The effect of dexmedetomidine on the neonatal EEG**

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A term neonate was diagnosed with hypoxic ischemic encephalopathy (HIE), started on therapeutic hypothermia and sedated with dexmedetomidine. Dexmedetomidine usually causes only very subtle changes in the EEG (Mason, O'Mahony, Zurakowski, & Libenson, 2009). In this case profound suppression of the EEG was seen that normalised after the cessation of dexmedetomidine. This has important implications for assessments regarding prognosis in similar cases.

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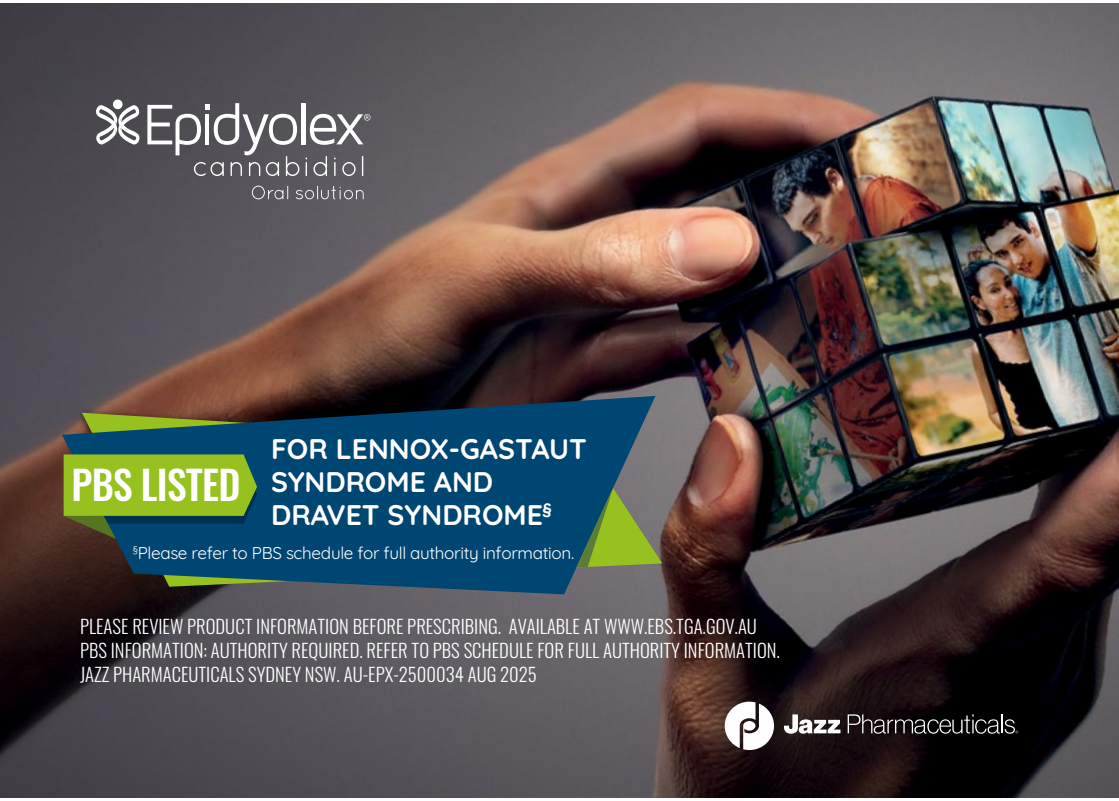

#3736 **Practice of paediatric neurologists in managing neurological manifestations of Sturge Weber Syndrome in a paediatric tertiary centre**

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Background: Sturge-Weber Syndrome (SWS) is a rare neurocutaneous disorder characterized by facial port-wine stains and leptomeningeal vascular malformations, leading to neurological manifestations including seizures, stroke-like episodes, headaches, and neurodevelopmental challenges. Despite existing guidelines, variability in SWS management remains prevalent due to limited high-quality evidence.

Objective: To evaluate clinical practices in managing neurological manifestations of SWS at a tertiary paediatric centre compared to published guidelines.

Methods: This is a mixed-method, retrospective cohort study conducted at The Children's Hospital at Westmead and a cross-sectional survey of paediatric neurologists using a customised Likert-scale questionnaire. Clinical data and survey responses were analysed descriptively and compared against published guidelines.

Results: The cohort included 13 paediatric SWS patients, and 8 paediatric neurologists completed the survey. Drug-resistant epilepsy (DRE) affected 61.5% of patients. No patients were trialled on ketogenic diet or vagus nerve stimulation. One patient underwent hemispherectomy with 10 years seizure freedom. No patients received pre-symptomatic anti-seizure medications. Stroke-like episodes were observed in 70% of patients, none of which had acute neuroimaging changes but 1 had corresponding EEG seizures. Pre-symptomatic aspirin was started in 61.5% of patients. Aspirin did not reduce the frequency of stroke-like episodes. Survey results showed general alignment with current recommendations but variability in investigations, pre-symptomatic treatment, and referral practices.

Conclusion: This study highlights variability in SWS management, underutilisation of advanced epilepsy therapies, overutilisation of acute investigations, limited observed long-term benefit of aspirin, and gaps in neurodevelopmental and mental health support. More robust evidence is needed to standardise care for SWS patients.

#3742 **Trauma- associated Acute encephalopathy with biphasic seizures and late diffusion restriction: insights from two cases**

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Background: Acute encephalopathy with biphasic seizures and late diffusion restriction (AESD) is characterised by acute encephalopathy, biphasic seizures, and subcortical diffusion restriction on subacute imaging.1 While aetiology is not well understood, susceptibility to AESD has both genetic and environmental contributions. It is often preceded by viral infection. Rarely cases of post-traumatic AESD have been reported.[2, 3] Here we report two post-traumatic cases.

Methods: Retrospective records of two cases of post-traumatic AESD presenting to our department were reviewed.

Results: Patient A presented at 8-months of age following a fall from a car seat onto his head with subsequent encephalopathy and seizures. CT head demonstrated subdural haemorrhages. Despite initial improvement, he had ongoing encephalopathy. An MRI brain showed no new findings. A second phase of seizures developed and repeat MRI brain revealed late restricted diffusion. He received supportive care and made a complete recovery.

Patient B presented at 9-months of age with focal seizures and encephalopathy. Initial MRI brain was normal. Seizures recurred. A repeat MRI found extensive diffusion restriction, favouring a diagnosis of AESD. Treatment with IVIg and IV methylprednisolone was initiated. Infectious work-up was negative. The encephalopathy resolved, however, spasticity and developmental delay emerged. A presentation 3-months later with seizures and signs of non-accidental injury raised the possibility of the initial presentation being in the context of trauma.

Conclusions: These two cases highlight that whilst AESD is often thought of as triggered by infection, aetiology is not understood, and trauma should be considered as an alternative cause.

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#3750 **A 13-year-old girl with sixteen syndrome and intractable vomiting**

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Background: Neuromyelitis Optica Spectrum of Disorders (NMOSD) are rare, relapsing neuroinflammatory disorders with predilection for the optic nerves and spinal cord.[1,2] NMOSD is associated with several clinical syndromes and AQP4 autoantibodies, although diagnostic criteria may be met with positive antibodies against myelin oligodendrocyte glycoprotein (MOG), or seronegative status.[1] 3-5% of cases occur in children; these have higher frequency of diverse presentations reported in the literature.[2] This is the first case report of NMOSD causing Sixteen Syndrome in a child.

Methods: We describe the clinical course of a Polynesian 13-year-old girl, presenting to hospital with non-opticospinal NMOSD.

Results: This adolescent presented with intractable vomiting causing 10 kg weight loss. Comprehensive work up was unrevealing including bloods for metabolic conditions, upper GI contrast study, abdomen/pelvis computed tomography, and gastroscopy. Brain MRI performed to assess vomiting from central nervous system causes was normal. Eight weeks later she developed complete bilateral horizontal conjugate gaze palsies with facial diplegia. Repeat Brain MRI showed FLAIR hyperintensity involving paramedian pontine tegmentum and area postrema structures, and bilateral facial and abducens nerve enhancement. CSF showed mild lymphocytic pleocytosis, with absent oligoclonal bands. The phenotype suggested NMOSD causing area postrema syndrome (APS) and sixteen syndrome;[3-6] she commenced intravenous methylprednisolone and plasmapheresis. Rituximab was commenced to prevent relapses. Symptoms and signs resolved. Paired AQP4 antibodies returned positive after immunotherapy initiation.

Conclusion: This case highlights the importance of considering non-opticospinal NMOSD in individuals with complex gaze palsies, particularly in association with APS, and to commence immunotherapy without waiting for autoantibody results.

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#3752 **Presentation and management of primary headaches in children in a paediatric neurology clinic in a metropolitan hospital in Western Australia**

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[2] St John of God Midland Public Hospital, Midland, WA, Australia

Background: Recurrent headaches in children represent a significant health concern that requires specialised care, ideally managed through dedicated paediatric neurology clinics in tertiary children's hospitals for optimal outcomes. We are presenting our audit of primary headache patients at a secondary level, metropolitan hospital seen at an outreach paediatric neurology clinic, evaluating the management against tertiary standards.

Methods: A retrospective review analysed de-identified data from children seen over five years at this clinic in our hospital. Parameters including demographics, headache type, comorbidities, and management strategies were compared with similar studies from tertiary centres.

Results: Among the 52 children audited (29 female, 23 male; age range 1-16 years), of whom 17.3% were preschool age in and 32.8% teenagers. Migraine was most common type (73%), with 67% reporting a family history. Mental health and behavioural comorbidities were prevalent in 48.1% (25% anxiety, 15% mood dysregulation, 7.5% ADHD). Sleep disturbances were frequent (40% increased sleep latency, 19.2% difficulty maintaining sleep). Management included acute pain medications (including triptans in 25%). Prophylaxis was started in 57.7%, of whom 1/3 needed trial trail of second line agent. All patients also had non-pharmacological interventions including lifestyle modifications.

Conclusions: This clinic's demographics and management align with those of tertiary centres, demonstrating the efficacy of such specialised care outreach models. Outreach models are imperative to Australia's healthcare system, mitigating inequities, improving chronic disease management, and ensuring sustainable, patient-centred care. Scaling these initiatives—supported by policy and technology—will be critical to meeting Australian SDG 3 (health equity) goals by 2030.

#3754

The Bidirectional Relationship between Headache and Affective Disorders in Children: a single centre, paediatric neurology clinic experience

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Background: Headache disorders, particularly migraines and tension-type headaches, are prevalent in children and adolescents and show a strong bidirectional association with affective disorders such as anxiety and depression. Recurrent headaches can contribute to emotional and behavioural difficulties, while preexisting mood disorders may increase the frequency and severity of headaches.

Methods: A retrospective review analysed de-identified data from children seen over five years at this clinic in our hospital. Parameters including demographics, headache type, comorbidities, and management strategies were audited, including subgroup analysis within the affective disorders cohort.

Results: Among the 52 children audited (29 female, 23 male; age range 1-16 years), of whom 17.3% were preschool age in and 32.8% teenagers. Migraine was most common type (73%), with 67% reporting a family history. Mental health and behavioural comorbidities were prevalent in 48.1% (25% anxiety, 15% emotional dysregulation, 7.5% ADHD). Sleep disturbances were frequent (40% increased sleep latency, 19.2% difficulty maintaining sleep). Developmental issues and learning difficulties were reported in 25%, and 21.2% had experienced parental separation. Treatment involved not only acute pain management and prophylactic therapy, but also non-drug strategies including lifestyle changes, mindfulness, psychological support, and counselling.

Conclusions: The findings support the complex interaction between chronic headaches and emotional health in children. Integrated, multidisciplinary care—including pharmacologic and psychological interventions—is essential. Routine screening for mood and anxiety symptoms in paediatric headache patients (and vice versa) is crucial to early intervention, improved symptom management, and better long-term outcomes.

#3757 **Evaluating the clinical impact of EEGs in Complex Febrile Seizures in children: A single-centre experience**

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Background: Electroencephalograms (EEGs) are frequently requested in children with complex febrile seizures (CFS), despite growing evidence questioning their routine utility. This audit aimed to evaluate EEG usage and its influence on clinical management in children with CFS.

Methods: A retrospective audit was conducted on 24 patients reviewed in a neurology clinic at a metropolitan, secondary level hospital with a diagnosis of CFS. Data collected included age-at-onset, seizure characteristics, developmental history, EEG use, interventions and outcomes, and assessed against guideline-based criteria.

Results: Twenty-four children were included in the audit (age of seizure onset 4 months to 4 years). EEGs were performed in all but one case. Abnormalities were noted in 30% which did not change the clinical management.

Pharmacological interventions were used in 79%, including buccal Midazolam for seizure rescue (54%), Clobazam for febrile illness (37.5%), and Leviteracetam in selected cases (12%); none of these children had an abnormal EEG. A family history of febrile seizures or epilepsy was documented in 42%, and 33% had developmental or behavioural concerns.

50% had no further seizures and were subsequently discharged, one of whom represented with Rolandic epilepsy and had an abnormal EEG on initial presentation. The rest continue to be annually reviewed. Most families reported infrequent use or discontinuation of prophylactic medications such as Clobazam.

Conclusions: EEGs were frequently performed outside guideline indications and rarely influenced management decisions. These findings support more selective EEG use in CFS, in line with evidence-based criteria, to improve efficiency and reduce unnecessary investigations.

#3758 **A stratified analysis of efficacy and safety of fenfluramine in patients with Dravet Syndrome**

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Background: Dravet syndrome (DS) is a rare, drug-resistant, developmental and epileptic encephalopathy characterized by frequent seizures and motor, behavioral, and cognitive impairments. Fenfluramine was evaluated in three pivotal phase 3 randomized controlled studies (NCT02682927, NCT02826863, NCT02926898) and is approved for the treatment of seizures associated with DS (US, EU, UK, Japan, among others) in patients ≥2 years old. The studies were pooled and stratified by baseline characteristics not originally reported.

Methods: Patients (2-18y) with DS were treated with placebo or fenfluramine (0.2 mg/kg/d or 0.7 mg/kg/d without stiripentol [0.2FFA, 0.7FFA], 0.4 mg/kg/d with stiripentol [0.4FFA]). Data were stratified by fenfluramine initiation age (<4y, ≥4y), severity (1-3, 4-6, and 7+ failed antiseizure medications [ASMs]), and genotype (*SCN1A*+/-). Baseline characteristics, safety, median percentage change in monthly convulsive seizure frequency (MCSF), median longest interval of convulsive seizure-free days (SFDs), and Clinical Global Impression—Improvement (CGI-I) scores (caregiver/investigator) were assessed.

Results: Among 348 patients (0.2FFA, n=85; 0.4FFA, n=43; 0.7FFA, n=88; placebo, n=132), 83.7%-89.4% were ≥4y; 70.6%-81.4% had genotype *SCN1A*+. Failed ASM rates were generally high (4-6: 31.8%-48.8%; 7+:34.8%-57.6% [no 0.4FFA-treated patients]). Treatment-emergent adverse events rates were similar across stratified groups. Fenfluramine treatment was associated with greater decrease in MCSF, increase in longest interval of SFDs, and higher CGI-I scores versus placebo, regardless of stratification.

Conclusions: Fenfluramine is associated with improved outcomes relative to placebo, regardless of age, severity, or genotype. Analyses of stratified groups in larger populations may provide a better understanding of the benefits seen with various DS subpopulations and synergies with concomitant medications.

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	EDUCATION DAY - FRIDAY, 5TH SEPTEMBER For general paediatricians and paediatric neurology trainees Separate registration required to attend	
Time	**Event**	**Speaker**
13:30 - 15:30	**Session 1** Chair: Matthew Lynch	
13:30	Basics of genomic testing	*Chirag Patel*
14:00	Introduction to metabolic genetics and acute metabolic encephalopathy	*Michelle Lipke*
14:30	Metabolic approach to seizures/epilepsy, focussing on treatable causes	*Matthew Lynch*
15:00	An approach to ED presentation with Acute weakness	*Christina Miteff*
15:30 - 16:00	**Afternoon Tea**	
16:00 - 17:00	**Session 2** Chair: Alex Johnson	
16:00	An approach to ED presentation with Gait difficulty	*Kenny Yong*
16:30	An approach to ED presentation with altered level of consciousness	*Chris Troedson*
	EDUCATION DAY - SATURDAY, 6TH SEPTEMBER For general paediatricians and paediatric neurology trainees Separate registration required to attend	
08:30 - 10:30	**Session 3** Chair: Sophie Calvert	
8:30	Learning, brain plasticity and cellular mechanisms of learning	*Michael Duhig*
9:00	Paediatric Speech and Language Disorders – Terminology, Assessment and Diagnosis	*Ciara Durnin*
9:30	Intellectual Disability and Specific Learning Disorders	*Erica Harding-Smith*
10:00 - 10:30	**Morning Tea**	
10:30 - 12:30	**Session 4** Chair: Alex Johnson	
10:30	Research & grant applications	*Emily Oates*
11:00	Wellbeing & looking after yourself	*Simone Ardern-Holmes*
11:30	Neurology Career Advice	*Sophie Calvert, Steve Malone, Erik Andersen*
12:00	Panel Q+A	
12:30	**Close**	

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