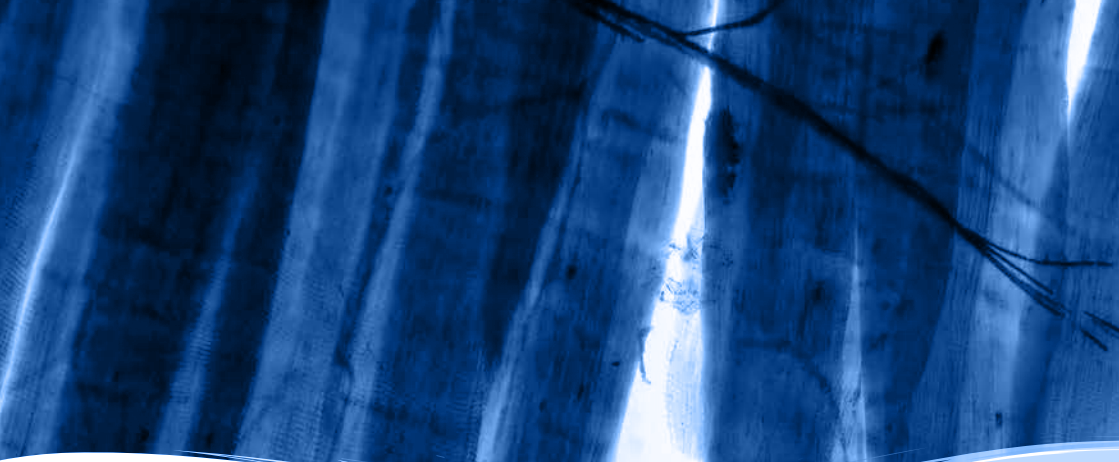
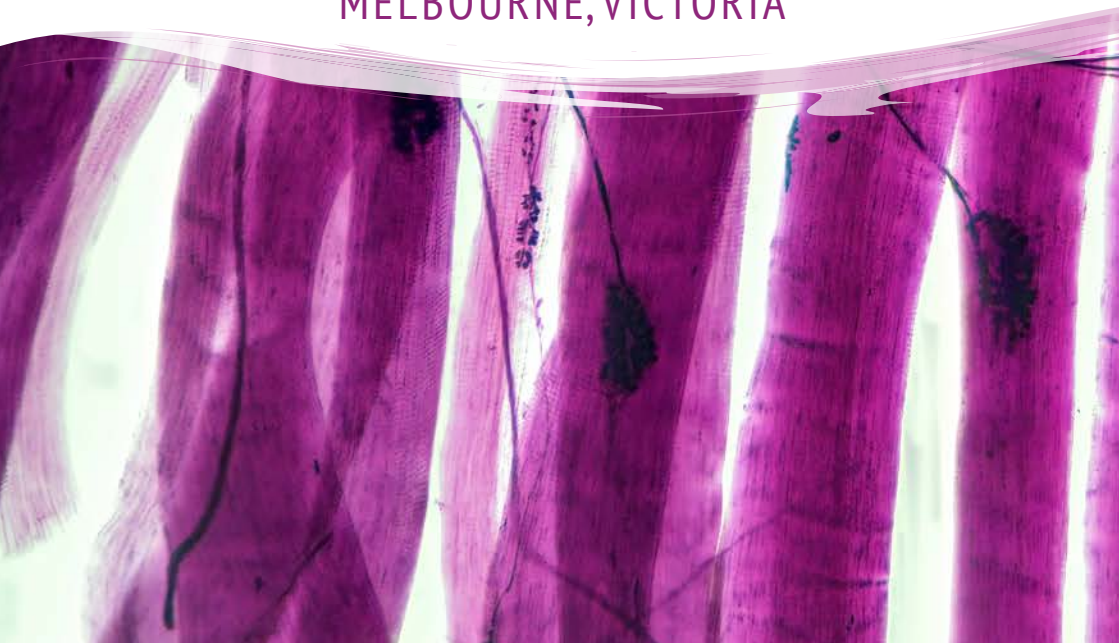


Australasian Neuromuscular Network 2024

27TH - 29TH MAY 2024

SHERATON MELBOURNE HOTEL

MELBOURNE, VICTORIA



AUSTRALASIAN NEUROMUSCULAR NETWORK (ANN) ANNUAL CONGRESS 2024

CHANGING LANDSCAPES IN NEUROMUSCULAR DISEASE: DIAGNOSIS, CARE, TREATMENT & MANAGEMENT

SHERATON HOTEL BALLROOM
27 LITTLE COLLINS STREET
MELBOURNE, AUSTRALIA

(IN-PERSON EVENT)

Cover photograph: Histology microscope image of motor unit synapse of muscle fibers (100x) © DrWD40 - stock.adobe.com

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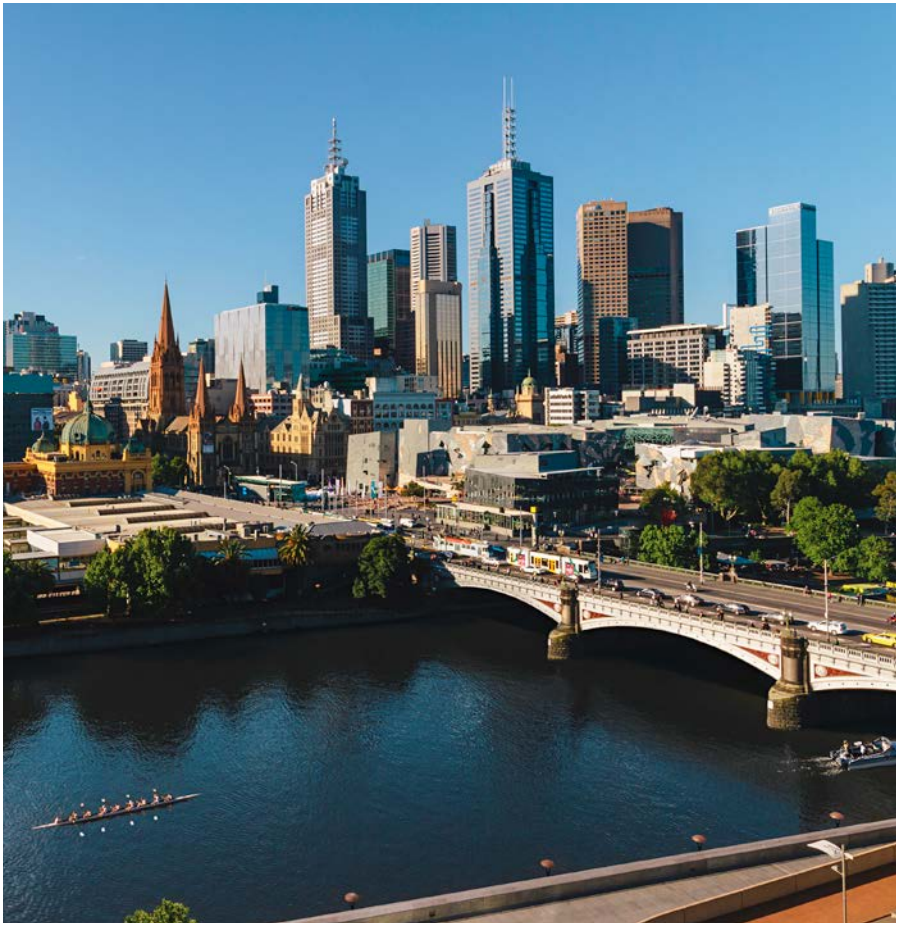

Melbourne City Skyline © Roberto Seba

WELCOME

On behalf of the organising committee, we welcome you to the ANN Congress 2024

This year, we are pleased to welcome our guest international speaker Dr Chris Weihl, Professor of Neurology and Head of the Neuromuscular Section at Washington University School of Medicine in St. Louis, Missouri (USA). Dr Weihl's lab aims to understand the molecular mechanisms of protein inclusion formation, disaggregation, and clearance in myodegenerative and neurodegenerative diseases, such as inclusion body myopathy and limb-girdle muscular dystrophy.

We are grateful to our invited local speakers, from all over Australia and New Zealand, who have agreed to present at the congress. We look forward to learning from their wealth of expertise and experience. We extend our gratitude to each person who submitted abstracts for this year's Congress. The calibre of abstracts submitted this year were of a high standard and showcased the depth and breadth of knowledge in our community. Thank you to the independent abstract review committee for their evaluations.

The committee are also greatly indebted to World Muscle Society, who have generously sponsored awards this year for our Early Career Researchers (ECRs) Rapid-fire presentations and awards for best poster and best oral presentations.

Each year, we reflect on the original mission for the Australian Neuromuscular Network which was to be a *coordinated and collaborative voice* by *establishing a cohesive, integrated neuromuscular network which enables people to work together across Australia and New Zealand, for the well-being of patients*. And each year, we see this at work with ANN Congress that brings together an increasing number of clinicians and researchers from across Australia and New Zealand to achieve our goal of *excellence in diagnostic methods and clinical management*. We would like to thank you all for being part of that and welcome you to this year's Congress.

Robin Forbes *Chair* | Anita Cairns | Zoe Davidson | Chantal Coles | Vanessa Crossman

The local organising committee

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Conrad "Chris" Weihl

Conrad "Chris" Weihl, MD, PhD, is a Professor of Neurology, Head of the Neuromuscular Section and Director of the Muscular Dystrophy Association Clinic at Washington University School of Medicine in St. Louis, Missouri, USA. He received his undergraduate degree from the University of Illinois at Urbana/Champaign and his graduate (PhD) and medical (MD) degrees from the University of Chicago, where he trained in Neuroscience in the laboratory of Dr. Raymond Roos where he studied the role of presenilin-1 in familial Alzheimer's disease. Dr. Weihl completed his residency in Neurology at Washington University and then pursued a Neuromuscular Fellowship at Washington University with Dr. Alan Pestronk. Upon seeing a patient with dementia and inclusion body myopathy in clinic with Dr. Pestronk, he was sparked by the connections between neurodegenerative diseases and muscle diseases. This led him to pursue a postdoctoral fellowship with Dr. Phyllis Hanson at Washington University in the Department of Cell Biology where he generated the first mouse model of hereditary inclusion body myopathy associated with mutations in VCP. Dr. Weihl began his faculty appointment at Washington University in the Department of Neurology in 2007 and was promoted to full professor in 2018. He was appointed Chief of the Neuromuscular Division in 2022. His research program focuses on both genetic and acquired myopathies with an emphasis on how dysfunction in protein quality controls lead to muscle degeneration. More specifically, his work delineated the molecular mechanism of VCP associated inclusion body myopathy and identified the genetic cause of LGMDD1. He has received a number of honors including the Derek Denny-Brown Young Neurological Scholar Award from the American Neurological Association. He is currently a member of the WMS meeting planning committee and the ANA scientific program advisory committee. More recently, he is an active member ClinGen Neuromuscular Working Group and Chair of the ClinGen LGMD gene and variant curation expert panels. Dr. Weihl has a strong commitment to the training of future neuromuscular clinicians and myologists. He was awarded a K24 mid-career investigator award to mentor residents, fellows, junior faculty and early stage scientists in patient oriented research. Clinically, Dr. Weihl sees patients in a multidisciplinary muscular dystrophy association clinic and recently established a clinic devoted to the care of patients with IBM.

The benefits of joining the WMS

The World Muscle Society is a dynamic community that aims to promote, disseminate, and share all aspects of neuromuscular physiology and diseases, from basic science to patient care. It encompasses a broad range of scientists and healthcare professionals who share our mission and values as well as a common interest in neuromuscular disorders.

Members benefit from:

- Interaction with a valued community dedicated to muscle research by attending the annual Congress and using the website resources
- Reduced WMS Congress registration fees
- Regular, informative newsletters via email
- Monthly copies of the official WMS monthly journal, Neuromuscular Disorders
- Access to the Neuromuscular Disorders online archive through the WMS website

Our active membership pursues a number of advances for fellow members through a number of dedicated committees. This includes a Myology Developments Across the World Committee, which seeks to extend our reach to those areas where myology is still an emerging science. We also have a Guidelines Committee, dedicated to optimise the implementation of best care practices for neuromuscular disorders worldwide. Join us today and help advance our work.

Supporting the meeting

The WMS mission is to be a global, multidisciplinary community committed to advancing the science of neuromuscular disorders and the care for people living with them.

As a demonstration of our commitment to develop the field of neuromuscular disorders through clinical research and patient care, the WMS supports projects, such as conferences including the Australasian Neuromuscular Network 2024 Congress. We want to break down the barriers to access for our community and ensure that scientists around the world can network, learn from one another, share science and publish their work.

Along with hosting our own annual Congress, facilitating a global network of people working in the field of neuromuscular disorders helps us to bring about change more quickly for the people who are living with life-limiting diseases and future generations of patients. We wish you all a very successful Congress.

2024 WMS Congress website: wms2024.com

WMS World Muscle Society

#WMS2024

	MONDAY 27TH MAY 2024
08:00 - 09:00	**REGISTRATION**
09:00 - 10:35	**SESSION 1 - Welcome & Opening** Chair: Ian Woodcock
09:00	Welcome on behalf of organising committee - *Robin Forbes*
09:05	ANN Inc - *Ian Woodcock (President)*
09:15	**KEYNOTE ADDRESS** **Clinical Trial Readiness in LGMD: identifying patients, biomarkers and treatments - *Chris Weihl***
10:15	FSHD Update - *Ian Woodcock*
10:35 - 11:00	**MORNING TEA**
11:00 - 12:45	**SESSION 2 - Care in adults with neuromuscular conditions** Chair: Gayatri Jain
11:00	Challenges in respiratory care of the neuromuscular patient - *Mark Howard*
11:30	Cardiac care in neuromuscular disease - *David Prior*
12:00	Obstetric considerations for the neuromuscular patient - *Renuka Sekar*
12:30	Question panel
12:45 - 13:00	**LUNCH**
13:00 - 14:15 NOVARTIS	**LUNCH SESSION - New paradigm of care in the paediatric SMA patient** Supported by Novartis *(Due to Medicines Australia rules, members from parent advocacy and support groups are not able to attend this symposium)*
13:00	Welcome and update - *Novartis*
13:05	Car seats & safety assessments - *Ashlee Cruz*
13:20	Complexities of genetic counselling in new treatment era - *Pauline McGrath*
13:30	Disease modifying therapies in SMA: Implications for nutritional management from the current evidence base - *Katie O'Brien*
13:40	SMA Emergency care plans - *Anita Cairns*
13:50	Question panel
14:15 - 15:45	**SESSION 3 - Collaborations and future directions** Chair: Miriam Rodrigues
14:15	JPH1 - *Anita Cairns & Mridul Johari*

14:30	RFC1 - *Richard Roxburgh, Caroline Scriba & Luciana Pelosi*
14:45	FGF14-SCA27B - *Catherine Ashton & Caroline Scriba*
15:15	Diagnostic gene discovery for genetically unresolved pre-adult-onset Australian neuromuscular patients - *Emily Oates*
15:35	Question panel
15:45 - 16:10	**AFTERNOON TEA & POSTER SESSION 1** (delegates with odd numbered posters)
16:10 - 16:45	**SESSION 4 - Early Career Researchers Rapid Fire Presentations** Supported by Awards from World Muscle Society (WMS) Chairs: Kate Carroll & Kristy Rose
16:10	Body Fat is associated with disability severity in children with Charcot-Marie-Tooth disease - *Gabrielle Donlevy*
16:15	Pre-clinical Evaluation of Long-term Dimethyl Fumarate Treatment in Dystrophic Mice: An Interim Report - *Cara A. Timpani*
16:20	Feasibility and accuracy of home polysomnography in children with neuromuscular disorders - *Adelaide Withers*
16:25	Preparing for clinical trials in childhood-onset FSHD – the MOVE Peds protocol - *Katy de Valle*
16:30	Question panel
16:45 - 17:45	**ANN Inc AGM** Sheraton Ballroom
18:00 - 19:00	**ANN 2024 Mixer drinks and canapes**

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One dose, continuous possibilities*[1–8]

*All patients in the long-term follow-up studies maintained and/or gained motor milestones, with no regression up to 7.5 years (LT-001; n=10) or 4.3 years (LT-002; n=61) of follow up.[2–8]

PBS listed for presymptomatic SMA patients with 1 to 3 copies of *SMN2* or patients with Type 1 SMA under 9 months of age[9]

3,700 babies treated worldwide as of April 2024[10]

PBS: Pharmaceutical Benefits Scheme; SMA: spinal muscular atrophy; *SMN2*: survival of motor neuron 2 gene. **References: 1.** Zolgensma Approved Product Information. **2.** Mendell JR *et al. N Engl J Med* 2017;377(18):1713–22. **3.** Day JW *et al. Lancet Neurol* 2021;20(4):284–93. **4.** Mercuri E *et al. Lancet Neurol* 2021;20(10):832–41. **5.** Strauss KA *et al. Nat Med* 2022;28(7);1381–9. **6.** Strauss KA *et al. Nat Med* 2022;28(7);1390–7. **7.** Mendell JR *et al.* Long-Term Follow-Up of Onasemnogene Abeparvovec Gene Therapy in Symptomatic Patients with Spinal Muscular Atrophy Type 1. Poster presented at: 2023 MDA Clinical and Scientific Conference. March 19–22, 2023. **8.** Connolly AM *et al.* Intravenous and Intrathecal Onasemnogene Abeparvovec Gene Therapy in Symptomatic and Presymptomatic Spinal Muscular Atrophy: Long-Term Follow-Up Study. Poster presented at: 2023 MDA Clinical and Scientific Conference. March 19–22, 2023. **9.** Pharmaceutical Benefits Scheme. Available at www.pbs.gov.au/pbs/search?term=zolgensma (accessed April 2024). **10.** Novartis. Data on File.

PBS Information: Section 100 Public Hospital Authority Required for the presymptomatic treatment of SMA patients with 1 to 3 copies of the SMN2 gene or patients with Type 1 SMA under 9 months of age. Refer to PBS Schedule for full authority information.

▼ This medicinal product is subject to additional monitoring in Australia. This will allow quick identification of new safety information. Healthcare professionals are asked to report any suspected adverse events at www.tga.gov.au/reporting-problems.

WARNING: HEPATOTOXICITY • Acute serious liver injury, acute liver failure and elevated aminotransferases can occur with ZOLGENSMA. Cases of acute liver failure with fatal outcomes have been reported. • Patients with pre-existing hepatic impairment may be at higher risk. • Prior to infusion, assess liver function of all patients by clinical examination and laboratory testing (e.g., hepatic aminotransferases [aspartate aminotransferase (AST) and alanine aminotransferase (ALT)], total bilirubin, prothrombin time, albumin, partial thromboplastin (PTT) and international normalised ratio (INR)). Administer systemic corticosteroid to all patients before and after ZOLGENSMA infusion. Continue to monitor liver function for at least 3 months after infusion and at other times as clinically indicated.

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NOVARTIS

MONDAY 27TH MAY
NOTES

	TUESDAY 28TH MAY 2024
08:30 - 09:00	**REGISTRATION**
09:00 - 10:45	**SESSION 5 - Oral Research Presentations** Chair: Katrina Morris
09:00	Treatment with nusinersen in adults with Spinal Muscular Atrophy: clinical results from the SMA cerebrospinal fluid biobank - *Lauren Sanders*
09:10	A 50 year retrospective cohort study describing survival in Duchenne muscular dystrophy - *Zoe Davidson*
09:20	Creating an immune cell atlas of the peripheral blood for Facioscapulohumeral muscular dystrophy (FSHD) - *Chantal Coles*
09:30	A new genetic cause for a lethal brain disorder and what it teaches us about the biology of neurodevelopment - *Michaela Yuen*
09:40	Efficient Single-AAV Delivery of CRISPR Therapy for Duchenne Muscular Dystrophy with Exon-50 Deletion - *Fatwa Adikusuma*
09:50	Question panel
10:00	From Neuromuscular Registry to Pūnaha Io - the New Zealand NeuroGenetic Registry & Biobank - *Miriam Rodrigues*
10:10	National Muscle Disease Bio-databank (NMDB) - *Emily Galea*
10:20	ACTN3 genotype influences androgen response in skeletal muscle - *Jane Seto*
10:30	Putting one foot in front of the other – an update on the Ambulate NMD project - *Kate Carroll*
10:40	Question panel
10:45 - 11:15	**MORNING TEA & POSTER SESSION 2** (delegates with even numbered posters)
11:15 - 13:00	**SESSION 6 - Gene therapy treatment and clinical trials in Australia** Chair: Kristi Jones
11:15	Adult clinical trial landscape - *Christina Liang*
11:45	Gene therapy treatment and trials in Australia - *Michelle Lorentzos*
12:15	Pharmacy perspective - *Donna Legge*
12:45	Question panel
13:00 - 13:45	**LUNCH** **Including AHNA Lunch Meeting** Chair: Zoe Davidson & Katy de Valle

13:45 - 15:15	**SESSION 7 - Research presentations** **Early Career Researchers - Rapid Fire Presentations** Supported by Awards from World Muscle Society (WMS) Chairs: Kate Carroll & Kristy Rose
13:45	Bionano optical genomic mapping for FSHD1 molecular diagnosis - *Harmony Clayton*
13:50	Respiratory outcomes of nusinersen and risdiplam treatment in children with Spinal Muscular Atrophy - *Archana Chacko*
13:55	Comprehensive collation and analysis of variants in the skeletal muscle α-actin (ACTA1) gene - *Joshua Clayton*
14:00	Implementation of Physical Activity and Exercise for Children and Young People Living with Neuromuscular Disease: A Churchill Fellowship - *Rachel Kennedy*
14:05	ClinicalOutcomeMeasures.org: Implementing a web-based training and quality assurance program for standardising the evaluation of CMT trial endpoints - *Kayla Cornett*
14:10	Evaluation of the Queensland Paediatric Neuromuscular transition model - *Rebecca Leung*
14:15	Question panel
	Oral Research presentations Chair: Dr Jane Seto
14:25	Dissecting the genetic basis of, mechanisms in, and therapies for muscle diseases using zebrafish - *Avnika Ruparelia*
14:35	Oxidised albumin levels in plasma and skeletal muscle, as a biomarker of muscle necrosis and inflammation, to measure treatment efficacy in DMD - *Miranda D Grounds*
14:45	PYROXD1 myopathy update: from phenotypic expansion to therapy development - *Frances Evesson*
14:55	Developing pre-clinical models of childhood onset FSHD - *Peter Houweling*
15:05	Question panel
15:15 - 15:45	**AFTERNOON TEA & POSTER SESSION 3** (general viewing)
15:45 - 17:00	**SESSION 8 - Newborn screening for neuromuscular conditions 2024 and beyond** Chair: Danya Vears, Murdoch Children's Research Institute
15:45 - 17:00	Interactive panel and audience participation
19:00	**ANN 2024 Congress Dinner** *(Henry and the Fox, 525 Little Collins St)*

TUESDAY 28TH MAY
NOTES

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revvity

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With 2022 revenue of more than $3 billion and over 11,000 employees, Revvity serves customers across pharmaceutical and biotech, diagnostic labs, academia and governments. It is part of the S&P 500 index and has customers in more than 190 countries.

	WEDNESDAY 29TH MAY 2024
08:00 - 08:45	**BREAKFAST SYMPOSIUM - SMA Transition in 2024** Supported by Biogen *(Breakfast is provided to all delegates, but due to Medicines Australia rules, members from parent advocacy and support groups are not able to attend this symposium)* Chair: Lauren Saunders
08:00	Introduction - *Andrew Corbett, Biogen*
	Interactive audience participation
09:00 - 10:35	**SESSION 9 - New Horizons in Neuromuscular Research** Chair: Eppie Yiu
09:00	Establishing a global certification standard for evaluating CMT and related neuropathies - *Josh Burns*
09:30	Questions
09:45	Australian Neuromuscular Disease Registry Update - *Robin Forbes*
09:50	**KEYNOTE ADDRESS** **Aggregate and Vacuolar myopathies: common and distinct pathomechanisms - *Chris Weihl***
10:35 - 11:05	**MORNING TEA**
11:05 - 12:15	**SESSION 10 - Oral Research Presentations** Chair: Gina O'Grady
11:05	Respiratory health, sleep dysfunction and mental health in children and adolescents with a neuromuscular disorder: a descriptive qualitative study - *Adelaide Withers*
11:15	Proposal for an Asia-Pacific guideline for management of Spinal Muscular Atrophy in adults - *Lauren Sanders*
11:25	Staying on their feet – a study of walking in Duchenne muscular dystrophy - *Kate Carroll*
11:35	Baseline continuous glucose monitoring in children diagnosed with spinal muscular atrophy in infancy on disease-modifying therapies - *Sophie Manoy*
11:45	Stress exacerbates glucose intolerance in the mdx mouse model of Duchenne Muscular Dystrophy - *Angus Lindsay*
11:55	Differential respiratory function response in paediatric spinal muscular atrophy types 2 and 3 treated with nusinersen over 3 years - *Archana Chacko*
12:05	Question panel
12:15 - 12:30	**AWARDS & CLOSE**

ANN 2024
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SEE YOU AT THE SUNSHINE COAST FOR THE 2025 ANN CONGRESS 26TH - 28TH MARCH 2025