childhood dementia Symposium 2023

PROGRAM

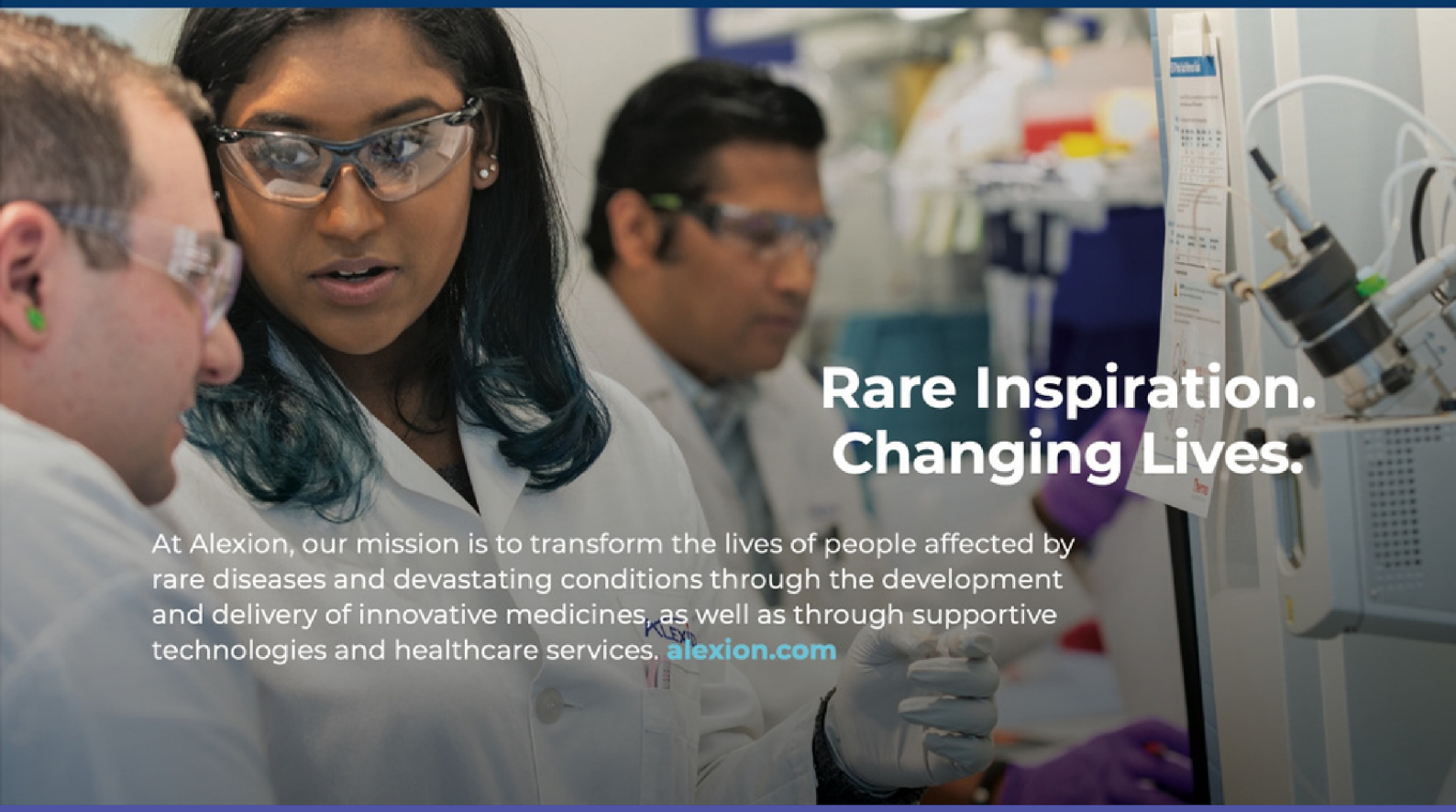
#childhooddementia2023

childhood dementia INITIATIVE

Thank you to our Silver Childhood Dementia Symposium Sponsor, Alexion.

LEADING IN RARE DISEASES FOR 30 YEARS

ALEXION
AstraZeneca Rare Disease



You can support better research and improvements for children with dementia across health and policy. Donate today, or join us on an upcoming life-changing trekking adventure to raise funds and awareness for childhood dementia.

Learn more: 3 Peaks Challenge

Make a donation

Welcome

Welcome to the 2023 Childhood Dementia Symposium and thank you for being a part of the unprecedented growth in childhood dementia research.

In the last year, an estimated 50,000 babies were born with a condition that causes childhood dementia. Sadly, around 75% are not expected to reach adulthood.

The progress we are all driving today gives hope. Hope that more and more of these children will be able to live long lives. And hope their families will see them thriving and experiencing a quality of life out of reach for children today.

The inaugural Childhood Dementia Symposium was held just one year ago. Together, we've made great progress since then:

- The Childhood Dementia Research Alliance has surpassed 120 members and continues to grow.
- Groundbreaking proposals were submitted to the MRFF in response to the world's first dedicated government research funding for childhood dementia in the world.
- We launched the Childhood Dementia Knowledgebase to provide deep data – an example of the shared infrastructure that can propel global research.

Alongside these strides is growing awareness and interest in the new, innovative research that is enabled by viewing childhood dementia conditions as a collective. I can't wait to see all that emerges from the 2023 Symposium.

Today is possible thanks to our generous sponsors: Alexion, Sanofi, Biomarin and Business Events Sydney. We thank them and acknowledge their significant support. Thank you also to our esteemed Scientific and Medical Advisory Committee, the speakers and session chairs. We couldn't do this without you.

If you haven't yet joined the Research Alliance, viewed the Knowledgebase, or read our latest report, State of Childhood Dementia, I encourage you to do so using the QR codes below. Information sessions on childhood dementia are being held in most states too.

Thank you for being a part of change for children with dementia and their families. Your contribution will no doubt be felt for many years and generations to come.

Dr Kris Elvidge
Head of Research
Childhood Dementia Initiative

Childhood Dementia Research Alliance

Childhood Dementia Knowledgebase

Report: State of Childhood Dementia

Information sessions

#childhooddementia2023

childhood dementia INITIATIVE

Noah

"Noah is from the idyllic island of Bali. He was fostered at 11 hours old and then adopted by Geoff and I, his Australian family, while we were living and working in Bali. He was a happy and healthy baby and met all his milestones. He spent his days going to the beach, riding horses, playing with friends and his beloved giant groodle dog, Nyame. His world was full of joy and laughter.

Noah absolutely loved performing. He just loved dancing, singing and was front and centre at kinder and school concerts, loving the audience and proud of remembering his steps. As a family, we travelled throughout Bali and Indonesia. Noah loved it. He loved adventures and going on boats and planes. He loved food and lived for spicy dishes and 'nasi' rice.

Noah was always smiling and never cried – not ever – nor did he ever throw tantrums. We used to say he is calm and almost zen because he is Balinese, and many share these gentle and kind traits.

In hindsight, these characteristics may have been a symptom. There were other things too. Like an ever so slightly slurred speech (almost an American accent) and the occasional facial tick when he was tired. Any concerns I had, as a first-time mother, were always dismissed by others as being overprotective or living in a bilingual home (English and Indonesian).

Noah started having noticeable problems jumping and keeping up with his peers from about 3 1/2 to 4 years old. He loved horse riding, but when he got on the horse, it made him so happy, that he passed out. We later found out he had cataplexy and narcolepsy brought on by joy and happiness. Noah then started having problems breathing and developed pneumonia too many times to count. We travelled all over Indonesia searching for answers but with no luck. The adoption process took almost 5 years, and while we loved our life in Bali, we could not travel to Australia for tests or a diagnosis. The government does not grant passports for children undertaking international adoption for fear that families will simply leave the country and not return.

Noah and I finally arrived in Australia in 2019 and, after 5 months of tests, he was diagnosed with Niemann-Pick Type C (NPC). NPC is a rare degenerative genetic disorder that is fatal. The body slowly malfunctions to the point where it can no longer go on. At this time, Noah was very unsteady on his feet. He could no longer run without falling over. He lost words and began answering questions with either single words or gestures. He also coughed and choked when eating. Up to 6 hours a day was spent slowly feeding, one small mouthful at a time. Around this time was when we first heard the words 'childhood dementia'. As so many of his symptoms were unknown to me, the word 'dementia' stood out. It highlighted the severity of his disease.

The diagnosis was a most devastating and heart-breaking experience. It was a complete shock and very difficult to understand that our son would never grow up or grow old. I can't put into words the pain. As Noah was neurologically symptomatic, he was given only a few short years to live.

Noah's NPC is very aggressive and has taken so much from him in the last few years. He can no longer walk, talk or even sit up. This is because he has ataxia and muscular hypotonia. He is fed via a tube in his tummy because he cannot swallow any of the food he loves so much (dysphagia). He has loads of scary seizures (epilepsy, cataplexy) and can't express to anyone how he feels anymore. We try to interpret his slight gestures and what they mean and act accordingly. I talk to him about activities and which he may prefer. Sometimes he can kick his legs if it is something he wants to do or give me a sideways glance, meaning, "no way, mum!"

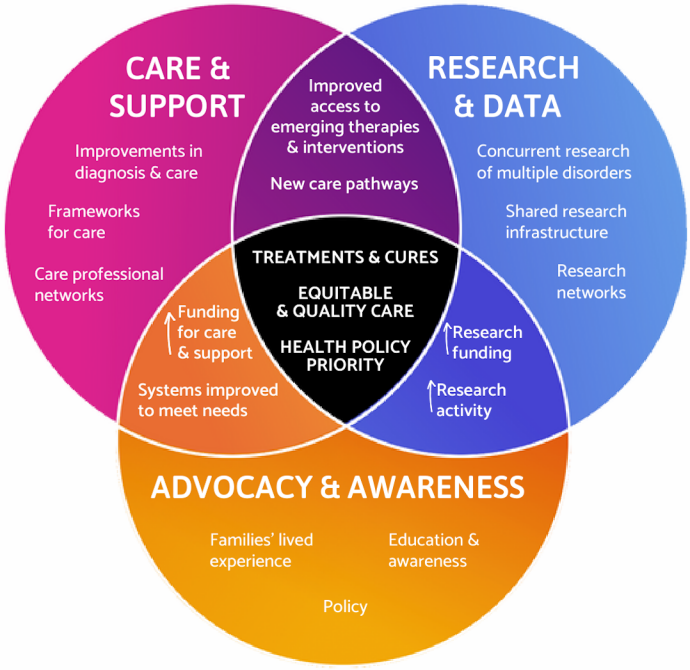
His future is very uncertain, and every day, he loses a little more control of his body and brain. He now requires 24-hour support to help him with movement, self-care, and live his best life. He no longer recognises people and places but is most happy doing the things he used to love as a 2 or 3-year-old. He loves music, especially anything Zumba, movies, and people from his past and shows his recognition."
- Jane, Noah's mum

We are very sad to share that Noah died in 2022. He was 8.

It's time for change

Childhood Dementia Initiative is driving world first action for every child with dementia. We are finding better ways to research and treat childhood dementia. Through bold, innovative approaches and systemic change we are improving outcomes for children with dementia across the world. Our Framework for Systems Change (pictured here) guides our work to achieve:

- Treatments and cures for childhood dementia
- Equitable and quality care for children and families
- Recognition of childhood dementia as a health policy priority



#childhooddementia2023

childhood dementia INITIATIVE

Program

0830 Registration

Session 1 Introductory session
Session chair: Tiffany Boughtwood, Managing Director, Australian Genomics and Director, Childhood Dementia Initiative

0900 Welcome

0910 Why we are here: the family perspective
Insights from parents Louise Jessop and Peta Murchison

0940 Understanding concerns and common challenges for children with dementia
Jason Djafar, UNSW

1000 The childhood dementia burden study
Dr Kristina Elvidge, Childhood Dementia Initiative/Dr Nicholas Smith, Women's and Children's Hospital Adelaide

1020 The Childhood Dementia Initiative
Megan Maack, Founder and Director of Childhood Dementia Initiative

1035 Session questions and discussion

1045 MORNING TEA

Session 2a Childhood dementia disorders: the commonalities, the opportunities & what we can learn from other fields. Session chair: Professor Kim Hemsley, Flinders University
SPONSORED BY ALEXION

1105 Introduction

1110 Clinical data of blarcamesine in Alzheimer's disease, Parkinson's dementia, and Rett syndrome - relevance to childhood dementia
Christopher U Missling, PhD, Anavex Life Sciences, USA

1150 Repurposing drugs to treat childhood dementia
Associate Professor Anthony White, QIMR

1210 Neuroinflammation in childhood dementia
Professor Sarah Spencer, RMIT

1230 Session questions

1240 **LUNCH**

Session 2b Childhood dementia disorders: the commonalities, the opportunities and what we can learn from other fields (continued)
Session chair: Professor John Christodoulou

1320 **Introduction**

1325 **Leukodystrophies: opportunities for improved treatments**
Professor Richard Leventer, Murdoch Children's Research Institute

1345 **Why are non-mammalian models of childhood dementia worth the effort?**
Associate Professor Michael Lardelli, The University of Adelaide

1405 **Brief outline of newly funded MRFF projects (if embargo lifted)**

1435 **Session questions**

Session 3 Networking session

1445 **Facilitated networking - small group discussions & afternoon tea**

Session 4 Clinical and clinical trial aspects of childhood dementia
Session Chair: Professor Michelle Farrar

1545 **Introduction**

1550 **Call for consensus on defining and measuring developmental regression: Next steps**
Dr Kirsten Furley, Monash Childrens Hospital

1610 **AAV Gene Therapy: the ultimate brain delivery system**
Dr Andrea Perez-Iturralde and Dr Florencia Haase, Children's Medical Research Institute

1630 **Prognostic biomarkers for childhood dementia**
Dr Prashant Bharadwaj, Edith Cowan University

1650 **Session questions**

1700 **Closing remarks**

1710 **REFRESHMENTS**

Speakers

Dr Prashant Bharadwaj has over 10 years of research experience in autophagy, protein aggregation and neurodegeneration. His work has identified new autophagy-lysosomal genes involved in Alzheimer's Disease (AD). He has developed cell models for high-throughput screening for AD and identified a neuroprotective function for the drug latrepirdine, now in Phase 3 clinical trials. Notably, his findings have guided the discovery of novel pro-autophagic/anti-neurodegeneration compounds to improve neurogenesis.

Jason Djafar is a 5th Year UNSW medical student with a keen interest in neurology and aspires to be a clinician-researcher. He achieved First Class Honours for his project which gathered the key phenotypes and target symptoms for childhood dementia disorders, under supervision of Professor Michelle Farrar and Dr. Alexandra Johnson. He works with Michelle's team as a research assistant whilst balancing his studies and co-curricular roles at UNSW.

Dr Kristina Elvidge has worked with rare disease patient organisations since 2008, defining research strategies, facilitating collaborations and communicating research to all stakeholders. She is Head of Research at the Childhood Dementia Initiative and has previously worked with Sanfilippo Children's Foundation and several muscular dystrophy organisations in Australia and the UK. Kristina completed a PhD in molecular biology at University of Western Australia and a postdoc at University of Oxford.

Dr Kirsten Furley is a developmental paediatrician and researcher studying developmental regression through a PhD at Monash University. She helped develop a novel, research-embedded clinical service at Monash Children's Hospital. She is passionate about improving care for children and families experiencing skills loss by collaborating with them and community partners. Kirsten is chief investigator on a Medical Research Future Fund grant to establish a unique multi-specialist, collaborative and accessible developmental regression clinic.

Dr Florencia Haase was born in Buenos Aires, Argentina. She moved to Sydney in 2013 and earned her Bachelor's Degree in Science (Immunology & Biology) from the University of Sydney. She then completed her Ph.D. in Medicine (Molecular neurobiology) from the University of Sydney in 2022. She joined the Translational Vectorology Research Unit at Children's Medical Research Institute to study and design AAV-based gene therapies for rare neurological disorders.

Louise Jessop is a health and disability advocate and mum to 3 boys. Her eldest, Dylan, was diagnosed with Sanfilippo Syndrome. Louise considers Dylan her greatest teacher – on life, unconditional love and the need for societal change in all things disability and rare childhood disease. Louise completed a Masters of Social Work in 2017 and is today a Specialist Support Coordinator.

Assoc. Prof. Michael Lardelli obtained a B.Sc. (Hons) degree from the University of Sydney before Ph.D. research on Drosophila developmental genetics in the UK and a postdoc in Sweden identifying Notch genes. At the University of Adelaide his research focuses on Alzheimer's disease genetics, particularly PRESENILIN genes. His laboratory uses genome editing to introduce neurodegeneration-related mutations into zebrafish followed by transcriptome and metabolomics analyses to understand how these mutations cause disease.

Prof. Richard Leventer is a consultant paediatric neurologist at the Royal Children's Hospital (RCH) in Melbourne and Group Leader of Neuroscience Research at the Murdoch Children's Research Institute (MCRI). He is a Professor in the University of Melbourne Department of Paediatrics, Director of the RCH/MCRI Brain Malformation Program and Clinic and neurologist at RCH neurogenetics clinic. His main clinical and research interests are disorders of early brain development and the use of imaging and genomic technologies to understand their causes.

Megan Maack founded Childhood ementia Initiative in 2020 to drive accelerated action on childhood dementia. She is the organisations' CEO and a Director. Prior to establishing Childhood Dementia Initiative, Megan was the founder and leader of the Sanfilippo Children's Foundation. She holds an MBA and has 15 years' international experience as a Change Manager, Management Consultant and Project Manager. Megan's work is inspired by her children, Isla and Jude, who both have childhood dementia caused by Sanfilippo syndrome.

Dr Christopher U. Missling is President and CEO of Anavex Life Sciences Corp. He has over 20 years of healthcare industry experience. His work is dedicated to finding potential cures for neurodegenerative and neurodevelopmental diseases by advancing potential treatments through clinical trials. Dr. Missling has an MS and PhD from the University of Munich in Chemistry and an MBA from Northwestern University Kellogg School of Management and WHU Otto Beisheim School of Management.

Peta Murchison founded Bounce4Batten in 2014, after her daughter Mia was diagnosed with Batten Disease. She shares her lived experience to advocate for positive change for rare disease, inclusive education and dying at home. Peta received a standing ovation at the Sydney Opera House for her TEDx talk "Finding hope in hopelessness".

Dr. Andrea Perez-Iturralde was born in Pamplona, where she obtained her bachelor's degree in Biochemistry from University of Navarra. She completed a master's degree in Drug Research, Development & Innovation and earned her PhD in Applied Medicine and Biomedicine. During these years, Andrea studied in detail the transduction mechanism of Adeno-associated viral (AAV) vectors. She joined the Translational Vectorology Research Unit at CMRI to develop different variants of AAV vectors for therapeutic purposes.

Nicholas Smith is a Neurologist at the Women's and Children's Hospital, South Australia. He obtained his PhD from the University of Cambridge investigating the neuropathology of Gaucher Disease. Returning to Australia, his research focuses upon the pre-clinical and translational development of therapeutic technologies for childhood neurodegeneration. A strong advocate for improving childhood dementia clinical care, he is the PI for multiple international gene-transfer clinical trials and serves as an advisor and board member for patient advocacy groups including the Childhood Dementia Initiative.

Prof. Sarah Spencer is Head of the Neuroendocrinology of the Obese Brain Research roup, and the Leader of the Healthy Foundations Research Program at RMIT University. Since obtaining her PhD in Physiology from University of Queensland, she works as a neuroendocrinology researcher focusing on understanding how early life events that precipitate brain inflammation influence lifelong health and cognitive outcomes. She has expertise in developmental, stress, and neuroimmune physiology.

Assoc. Prof. Anthony White obtained a PhD in neuroscience from Murdoch University and then investigated Alzheimer's and prion diseases as a postdoc at University of Melbourne andImperial College of Medicine, UK. In 2004 he established a research group at University of Melbourne investigating the role of biometals in neurodegeneration and development of metal-based treatments. He is currently developing new patient derived cell models of neurodegeneration to improve translational outcomes for neurotherapeutics.

Notes

Thank you

The Childhood Dementia Symposium was made possible by the following generous sponsors

ALEXION®
AstraZeneca Rare Disease

sanofi

BIOMARIN®

#childhooddementia2023

childhood dementia INITIATIVE

childhooddementia.org

hello@childhooddementia.org

childhood dementia INITIATIVE