childhood dementia INITIATIVE

2021-22 Federal Pre-Budget Submission

January 2021

Introduction

About childhood dementia

Dementia is usually only associated with the elderly. Sadly, children can also suffer from dementia. Today, there are an estimated 700,000 children and young people living with dementia worldwide. Their short lives are shaped by progressive brain damage, social isolation, pain and suffering. Most will not live to adulthood, some will die in their infant years.

Childhood dementia is more common than you might think. It is estimated that 1 in 2,800 children are born with a childhood dementia disorder, that is more common than well-known disorders like cystic fibrosis (1 in 2,874 live births)[1].

Childhood dementia is caused by more than 70 genetic conditions with shared presentation and mechanisms such as Batten disease, Sanfilippo syndrome, Niemann-Pick disease, Tay-Sachs disease, metachromatic leukodystrophy, Rett syndrome and some mitochondrial disorders. Childhood dementia disorders are neurodegenerative, progressive, severe, and devastating. They are complex disorders with high care needs, which result in poor quality of life for patients and impact entire families. Alarmingly, less than 5% of these 70 childhood dementia disorders have treatments.

The alarming statistics[2]

- Childhood dementia is caused by more than **70 individual genetic conditions**.
- **Fewer than 5%** of the conditions causing childhood dementia have a treatment.
- **One in 2,800** children born will develop childhood dementia.
- Collectively, the life expectancy for childhood dementia is estimated to be **28 years**. Many die in early childhood, even infancy.

For Australians the impact is:

- Each year, **129 babies are born** with a condition that will lead to childhood dementia. That is one born every 3 days.
- An estimated **2,273 Australians currently suffer from** childhood dementia. This prevalence is similar to that for motor neurone disease (2094 Australians in 2015)[3].
- Every year more than **90 young Australians die**, having lived their short lives suffering from childhood dementia. This is comparable to the number of deaths from cancer in children[4].
- The total economic cost of childhood dementia in Australia is **$389 million annually**. The cost to the families who love these children is immeasurable.

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[1] Massie RJ, Olsen M, Glazner J, Robertson CF and Francis I (2000) 'Newborn screening for cystic fibrosis in Victoria: 10 years' experience (1989–1998)'. *Med J Aust*; 172: 584-587.

[2] Tilden D, Valeri M and Ellis M (2020) 'Childhood dementia in Australia: quantifying the burden on patients, carers, the healthcare system and our society'. Report for Childhood Dementia Initiative. *THEMA Consulting Pty Ltd*. https://www.childhooddementia.org/burdenstudy

[3] Deloitte Access Economics Report (2015) 'Economic analysis of motor neurone disease in Australia' Report for Motor Neurone Disease Australia, Canberra. https://www.mndaust.asn.au/Influencing-policy/Economic-analysis-of-MND-(1)/Economic-analysis-of-MND-in-Australia.aspx

[4] Australian Institute of Health and Welfare (2020) 'Australia's children'. Cat. no. CWS 69. Canberra: AIHW. https://www.aihw.gov.au/reports/children-youth/australias-children/contents/health/cancer-incidence-and-survival.

Globally the impact of childhood dementia[5] is:

- Each year, 50,000 babies are born with a condition that will lead to childhood dementia.
- An estimated 700,000 individuals currently live with childhood dementia.
- Annually, 48,300 children and young people die prematurely.

Childhood Dementia: Overlooked and under-researched

Childhood dementia has received little recognition, and little research investment from major funding bodies. To illustrate this point, in the past 3 years the NHMRC has granted 38 times more research dollars to motor neurone disease ($17.9 million versus $475,000) and 29 times more research dollars to cystic fibrosis ($13.8 million). This is despite the similar prevalence of childhood dementia and motor neuron disease and the similar incidence of childhood dementia and cystic fibrosis.

Research on childhood dementia is not just limited in resourcing, it is limited in scope too. Sadly the majority of childhood dementia disorders attract little to no research. Additionally, there is no research underway that considers them holistically with commonality of presentation and impact. For many of these rare disorders, holistic research of this kind is the only feasible way progress to treatments will be made.

In addition, children living with dementia and their caregivers experience unique challenges that are not well known nor well responded to within Australian health and social care systems. Quality of life is poor for these children, health and social care support is limited and multidisciplinary care is not coordinated.

All families impacted by childhood dementia deserve to receive an accurate and timely diagnosis, to have treatments that are available and accessible, and to benefit from improvements to their quality and length of life. In short, patients with childhood dementia deserve to live the best life possible. The lack of research, awareness and multidisciplinary support and care means children and families have little hope of this.

Childhood dementia is an unacceptable problem; it is time to transform the way we approach it. Children are dying, we need to act; fast.

Childhood Dementia Initiative

Transforming the approach to childhood dementia

The Childhood Dementia Initiative drives research and advocates to urgently disrupt the impact of the collective group of childhood dementia disorders on children, families and society at large.

[5] Childhood Dementia Initiative Report (2020). Childhood Dementia: the case for urgent action. https://www.childhooddementia.org/whitepaper.

It is timely that the Childhood Dementia Initiative was launched in 2020. *The National Strategic Action Plan for Rare Diseases*[6] was endorsed by the Australian Federal Government in February 2020. The Action Plan recognises the need to ensure that research into rare diseases is collaborative, person-centred and systematically addresses gaps.

Founded in Australia, the Initiative has a global strategy to drive systemic change across research, clinical care and support. The Initiative is leading the collective consideration of all childhood dementia disorders. This is key to enabling transformative, and to date unrealised, economies of scale and scope through the utilisation of common infrastructure and resources. Rare disease experts agree that cross indication approaches will lead to enhanced efficiencies and greater patient benefit[7], accelerating the drug development pipeline for the individually rare childhood dementia disorders, but also delivering significant outcomes for the management and care of all children who suffer dementia.

The Childhood Dementia Initiative's leadership and Board have extensive experience in rare disease and childhood dementia research and first-hand experience and insights into the roadblocks and opportunities to make real progress for children with dementia:

- The Initiative is led by Megan Donnell, the Founding Director of the Sanfilippo Children's Foundation which, under her leadership transformed research into the little-known childhood dementia disorder, Sanfilippo syndrome. Megan pioneered new global partnerships, departed from traditional research funding models and secured funding for 24 distinct research projects, including a world-first gene therapy clinical trial. The Foundation today drives the world's first Sanfilippo research roadmap.
- The Initiative's Board of Directors includes Sean Murray, CEO and Founding Director of the Mito Foundation, a position he has held for the past 12 years. The Mito Foundation supports the Australian Mitochondrial disease community while driving research into mitochondrial disease, some subtypes of which are childhood dementia disorders.
- Tiffany Boughtwood, Managing Director of Australian Genomics, brings expert knowledge of the medical research sector to the Board of Directors. She manages the coordination, delivery and process evaluation of the $80 million Australian Genomics program and has more than 25 years' experience in molecular biology and research management.

Progress on childhood dementia cannot be left only to individual patient groups. These organisations are severely under-resourced, and no matter how skilled and committed the researchers they engage, the progress they make will be limited without building collaborations across disorders. The Initiative has been established and tasked to meet this need. We look forward to working with the federal government to establish initiatives that will finally give relief and hope to those suffering childhood dementia.

The Initiative's approach and recommendations were outlined in a case for action published by the Childhood Dementia Initiative in November 2020[8].

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[6] Rare Voices Australia (2020, Feb). National Strategic Action Plan for Rare Diseases. Australian Government Department of Health.https://rva.blob.core.windows.net/assets/uploads/files/NationalStrategicAPRD.pdf

[7] Brooks PJ, Tagle DA and Groft S (2014) 'Expanding rare disease drug trials based on shared molecular etiology'. *Nat Biotechnol*;32(6):515-8

[8] Childhood Dementia Initiative Report (2020). Childhood Dementia: the case for urgent action. https://www.childhooddementia.org/whitepaper.

Federal Funding Commitment

The projects outlined in this pre-budget submission are designed to facilitate the systemic change, collaborations and research required to:

1. **Find therapeutic solutions** for childhood dementia.
2. **Improve the quality of life** for children living with dementia and their families.

The breakdown of funding sought for 2021/2022 is as follows:

Priority 1	Find therapeutic solutions	$1,325,000
Priority 2	Improve quality of life	$890,000
	Total 2021/22	**$2,2155,000**

Priority 1: Find therapeutic solutions

Project 1.1: National Childhood Dementia Research Network

There is currently little to no active research for the majority of the childhood dementia disorders and none that considers them holistically with commonality of presentation and impact. In order to accelerate progress towards therapies, a multi-disciplinary, collaborative effort is required.

The National Childhood Dementia Research Network will bring together leading researchers from around the country to work collaboratively on the development of therapeutic solutions for children suffering from dementia with the following key objectives:

- Establish, build and nurture collaborative research networks.
- Enable research that concurrently investigates multiple childhood dementia disorders.
- Support and promote the collaborative utilisation of technology, data and infrastructure for research.
- Develop platform technologies for advanced therapeutics.

This network will be managed by the Childhood Dementia Initiative and will include multidisciplinary representatives from leading research institutes, universities and hospitals nationally. Connections and support established for this network include experts from:

- Sydney Children's Hospital Network
- Australian Genomics/Murdoch Children's Research Institute
- University of Queensland
- Flinders University
- SAHMRI
- University of Adelaide

This network will comprise experts in fields such as gene therapy, genomics, high throughput drug screening and repurposing, small molecule drug design, biochemistry and regenerative medicine and will bring together the necessary skills for a holistic approach to solving childhood dementia. The network will also include researchers already working on individual childhood dementia disorders, clinician researchers and consumer and industry representatives.

Implementation cost: $515,000 per annum for 3 years.

Project 1.2: Global Childhood Dementia Solution Summit

Convening the first ever summit dedicated to childhood dementia will bring leading minds together to share knowledge and novel ideas, and forge new collaborations. The National Childhood Dementia Research Network will be the starting point for invitations to this summit, their collaborators and other international experts will also be invited to attend. We estimate that 200 researchers will attend this 2 day summit.

Facilitated workshops at the summit will define childhood dementia research priorities and devise a strategy to accelerate the development of much needed therapeutics.

Implementation cost: $225,000.

Project 1.3: Build a drug screening platform for childhood dementia

This project will develop pipelines for the repurposing of existing drugs to deliver fast relief to children with childhood dementia. Pioneering medical breakthroughs will be used to create neuronal cell models to serve as a cellular avatar of a person's brain. Using induced pluripotent stem cell (iPSC) technology, skin cells from an individual affected by childhood dementia will be engineered into neuronal cells and their health assessed with novel tools. These models will be used to test novel therapies and screen existing drug libraries to identify those that could be repurposed.

The project team has been working together since mid-2019 to create neuronal cell models and conduct a preliminary drug screen for one type of childhood dementia – Sanfilippo syndrome (MPS III). The team has demonstrated their ability to collaborate effectively and deliver project milestones.

This project aims to leverage the established infrastructure and collaborative team to identify individualised therapies for children with other types of childhood dementia, including but not limited to Batten disease and mitochondrial disease. This will expand the pipeline to benefit more types of childhood dementia and further develop and refine this pipeline for even wider application to all types of childhood dementia. This will achieve economies of scale by taking advantage of the already purchased equipment, streamlined protocols and established team.

The Project Delivery Team includes key leaders from the following institutions:

- Women's and Children's Hospital, Adelaide.
- Childhood Dementia Research Group, Flinders University, South Australia.
- Laboratory for Human Neurophysiology and Genetics, South Australian Health and Medical Research Institute (SAHMRI).
- Centre for Nanoscale BioPhotonics, University of Adelaide.

An experienced Project Manager employed by the Childhood Dementia Initiative will oversee and coordinate the activities of this project. Overall guidance and support for this project will be provided by an experienced international steering committee with expert representation for each critical project area: Clinical management; high-throughput drug screening; clinical translation.

Implementation cost: $585,000 for year 1 and $475,000 for year 2.

Priority 2: Improve quality of life

Families with children suffering from childhood dementia report that care and support is inadequate, poorly coordinated and inconsistently delivered. In order to advocate for and enable the systemic changes necessary to ensure consistent, equitable and accessible quality care and support, a thorough understanding of the needs and experiences of families impacted by childhood dementia is required. Additionally, a clear understanding of the current care provision and management across Australia is needed.

Achieving this will involve 2 key steps:

1. Establishing a National Childhood Dementia Consumer Network
2. Undertaking foundational research to evidence the:
   - Experience and unmet needs of those impacted by childhood dementia.
   - Current capacity of the health system caring for these children.

Project 2.1: National Childhood Dementia Consumer Network

In 2020, the Childhood Dementia Initiative commissioned a burden of disease study[9] to quantify the prevalence, incidence and economic cost of childhood dementia in Australia. While the study revealed some significant impacts of childhood dementia, there has been little to no research into the lived experience of those impacted by childhood dementia.

Childhood Dementia Initiative is committed to involving consumers in all aspects of our work including health care policy, research, education and advocacy programs to improve the outcomes and experiences of people affected by childhood dementia. Engaging consumers will add a depth of knowledge that complements the organisational strategy with the reality and practicality of the consumer experience. It is well accepted that consumer engagement results in better health and research outcomes and a more trusted health system.

The National Childhood Dementia Consumer Network will bring together parents and primary caregivers from around the country to:

- Provide input on, and participate in, the research, health care policy, education and advocacy programs led by the Childhood Dementia Initiative.
- Inform foundational research projects that will aim to evidence the experiences and unmet needs of consumers.
- Bring the voice of families impacted by childhood dementia to external consultations. This could include, for example, consulting with Palliative Care Australia and PapCANZ on issues facing the childhood dementia community, to contribute to their work on a National Action Plan for Paediatric Palliative Care.

This network will be managed by the Childhood Dementia Initiative with access to external psychological support for consumers as required.

Implementation cost: $245,000 for year 1 and $175,000 for years 2 and 3.

[9] Tilden D, Valeri M and Ellis M (2020) 'Childhood dementia in Australia: quantifying the burden on patients, carers, the healthcare system and our society'. Report for Childhood Dementia Initiative. *THEMA Consulting Pty Ltd*. https://www.childhooddementia.org/burdenstudy

Project 2.2: Childhood dementia evidence base

The development of a robust evidence base will provide the foundation for Childhood Dementia Initiative to formulate a National Childhood Dementia Action Plan so that all Australian jurisdictions and stakeholders working in dementia care for children and young adults align to, prioritise and work towards common goals and objectives.

There are 2 key components to this project:

Evidence the Experiences and Unmet Needs of Consumers

The complexity and severity of paediatric rare genetic conditions pose substantial challenges to families. Delayed diagnosis, lifelong caring, limited capacity for independent living, lack of treatment options and large health service needs have severe impacts, termed as 'spillover effects', on parent's physical and psychosocial wellbeing. Studies have shown that parents with a child with a rare genetic condition have a significantly reduced quality of life in comparison to their non-impacted counterparts[10]. In order to improve the lives of those impacted by childhood dementia we must first establish the evidence of impact on and unmet needs of families.

Evidence the Health System management of childhood dementia

Understanding how the health system in each jurisdiction is currently organised for the management of children with dementia will enable identification of best practice and gaps. Identifying the strengths and needs of health professionals working in this space will guide future education advocacy programs.

The scope of this project will include the major paediatric hospitals, palliative care models and referral pathways to supportive services nationwide, including regional and remote care.

The outcomes of these two research project components combined will clearly identify the needs and priorities of families and the health sector and enable the development of best practice models of clinical care to improve outcomes for both the diagnosed child, their siblings and caregivers.

Implementation cost: $645,000

Contact

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[10] Wu Y, Al-Janabi H, Mallett A, Quinlan C, Scheffer IE, Howell KB, Christodoulou J, Leventer RJ, Lockhart PJ, Stark Z, Boughtwood T, Goranitis I. Parental health spillover effects of paediatric rare genetic conditions. Qual Life Res. 2020 Sep;29(9):2445-2454.