



Childhood Dementia
National Action Plan
Funding Request

July 2021

Introduction

Right now, 2,300 Australian children are suffering and dying slowly with dementia, without access to care or therapies - and no one knows about it. 75% of these children will die before they turn 18.

In 2020 [Childhood Dementia Initiative](#) commissioned a Burden of Disease study undertaken by THEMA consulting that outlines the truly unacceptable impact of childhood dementia in Australia. We urge you to read through this as it highlights the urgency for us to act as a collective in support of these children. <https://www.childhooddementia.org/burdenstudy>

The critical issue that has led to an underinvestment in research and inadequate support of children with dementia, has been the singular focus on the individual genetic causes of the dementia, rather than the commonality of presentation: i.e. dementia. This has resulted in a cost of \$389 million annually to the Australian economy, and a cost to the families who love these children that is immeasurable.

To change the national discourse for children with dementia we must develop a coordinated national framework; an evidence based national action plan (Action Plan) that drives systemic change to help children with dementia live fulfilling and long lives through access to therapies, alongside excellent medical and social care.

Globally, there has never been a movement to address childhood dementia disorders under one umbrella. Through national leadership this endeavour will drastically impact the lives of thousands of Australian children suffering, along with their families, while setting a framework that is a leader in the global response.

Federal Funding Commitment

This submission outlines activities that require the investment of \$1,467,000 in order to deliver the Action Plan which will provide a clear, actionable, and effective national strategic framework for childhood dementia. We have articulated in further detail the deliverables for each project below, however in summary these costs are:

	Total
Childhood dementia evidence base	\$504,000
National Childhood Dementia Consumer Network	\$260,000
Define Therapeutic Research Priorities	\$665,000
Develop, publish and launch National Action Plan	\$38,000
Total 2021/22	\$1,467,000

Childhood Dementia: Devastating, overlooked and under-researched

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 the disorders that result in childhood dementia have treatments.

The disorders that lead to childhood dementia include Batten disease, Sanfilippo syndrome, Niemann-Pick disease, Tay-Sachs disease, metachromatic leukodystrophy, Rett syndrome and some mitochondrial disorders.

The alarming statistics¹

- 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)².
- Every year more than **90 young Australians die**, having lived their short lives suffering from childhood dementia. This is a similar number to children who die from paediatric cancer aged 0-14.³
- The years of life lost due to early deaths is **1096 each year**.
- The total economic cost of childhood dementia in Australia is **\$389 million annually**. The cost to the families who love these children is immeasurable.

Globally the impact of childhood dementia⁴ 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.

¹ 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>

² 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](https://www.mndaust.asn.au/Influencing-policy/Economic-analysis-of-MND-(1)/Economic-analysis-of-MND-in-Australia.aspx)

³ 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>.

⁴ Childhood Dementia Initiative Report (2020). Childhood Dementia: the case for urgent action. <https://www.childhooddementia.org/whitepaper>.

A lack of research

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. The majority of childhood dementia disorders remain under researched and poorly understood. Additionally, there is no research underway that considers them holistically with commonality of mechanism, 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. Additionally, these single gene disorders share common pathophysiological mechanisms with aging dementia and provide an effective disease model to study, therefore research will inform the broader group of dementia disorders.

A lack of adequate care

Families with children suffering from childhood dementia report that care and support is inadequate, poorly coordinated and inconsistently delivered. The complexity of diagnosis, care requirements and a lack of awareness means that children with dementia experience unique challenges that are not well served by 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.

In order to advocate for and enable the systemic changes necessary to support 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.

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.

Why The Childhood Dementia Initiative

Transforming the approach to childhood dementia

The Childhood Dementia Initiative is driving awareness, research, advocacy and systemic change to urgently and drastically impact the way children, families and society at large continue to be affected by childhood dementia.

Launched in Australia, the Initiative is leading the collective consideration of all childhood dementia disorders, a world-first. 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 benefit⁵, accelerating the drug development pipeline for the individually rare childhood dementia disorders,

⁵ Brooks PJ, Tagle DA and Groft S (2014) 'Expanding rare disease drug trials based on shared molecular etiology'. *Nat biotechnol*;32(6):515-8

and also delivering significant outcomes for the management and care of all children who suffer dementia.

The Initiative's approach is to drive systemic change while focussing on efficacy in expanding currently available services. We are already realising results such as Dementia Support Australia and Dementia Australia agreeing to extend services to children with dementia this year, access to support these children have never had before, just one example and the start of what can, and needs to be done for these children.

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

Leadership

The Childhood Dementia Initiative's Board, supported by a Strategic Advisory Group and a Scientific Medical Advisory Committee, 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, 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. Megan's depth of experience with childhood dementia also stems from her lived experience through the complex and often siloed systems, as both her children have the childhood dementia disorder, Sanfilippo.
- 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 \$100 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 2020⁷.

⁶ Rare Voices Australia (2020, Feb). [National Strategic Action Plan for Rare Diseases](#). Australian Government Department of Health.

⁷ Childhood Dementia Initiative Report (2020). Childhood Dementia: the case for urgent action. <https://www.childhooddementia.org/whitepaper>.

National Action Plan for Childhood Dementia

The Childhood Dementia Initiative seeks to deliver the first nationally coordinated effort to address childhood dementia in Australia, indeed the first national framework that addresses childhood dementia in the world. The Initiative recognises that a robust and clear National Action Plan will drive the alignment of activities across the multitude of vital stakeholders within the medical, social care and research communities in order to:

- **Deliver therapeutic solutions** for childhood Dementia.
- **Improve the quality of life** for children living with dementia and their families.

The three projects outlined below will deliver evidence that will guide the articulation of a clear, viable and effective response to childhood dementia. The Action Plan will represent the views of families living with childhood dementia, and the care and research sectors, to deliver a collaborative and evidence based approach. The following are important outcomes of each project that will be critical to the formulation of a meaningful Action Plan:

1. Documented lived experience of families of children with dementia through their voices.
2. Evidence of the health and social care systems' current management of childhood dementia and the unmet needs of children with dementia and their families within it.
3. Buy in and commitment by critical stakeholders, including consumers, patient groups, health and social care providers, and researchers, to proactively participate in actioning the National Action Plan, due to their involvement in its development.
4. The landscape of current global research within this space is understood, and the research priorities clearly identified.
5. The identification of opportunities for greater efficacy and collaboration within research to accelerate therapeutic solutions.

Project 1: 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 Psychosocial 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 parents' physical and psychological well being. 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 counterparts⁸. In order to improve the lives of those

⁸ 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.

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, gaps and recommendations for improvements. 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: \$504,000

Project 2: National Childhood Dementia Consumer Network

While the burden of disease study in 2020⁹ 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 psychosocial 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: \$260,000

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 3: Define Therapeutic Research Priorities

Childhood Dementia Initiative will bring great minds together to identify the key research priorities to deliver urgently needed new treatments to children suffering from childhood dementia. With guidance from our esteemed Scientific and Medical Advisory Committee (Including industry representation) and input from the National Childhood Dementia Consumer Network we will:

- 1) Define therapeutic development focus areas and gather leading experts in each area. We anticipate focus areas will include topics such as diagnosis and screening, data collection and sharing, therapeutic development and clinical trials.
- 2) Convene the first ever symposium dedicated to childhood dementia. Facilitated workshops for each focus area will define research gaps and opportunities and establish new collaborations. We estimate that 200 researchers will attend this 2 day symposium.
- 3) Devise a research strategy that will enable funders within Australia and abroad to invest in research that takes advantage of economies of scale and scope to accelerate the development of much needed therapies

A strategic coordinated approach to investment in childhood dementia research, as will be defined in the National Action Plan, will deliver:

- Progress towards earlier diagnosis through screening programs to enable access to earlier treatment and optimal standards of care.
- Systems to collect and share patient data and biosamples to support research (including clinical trials).
- Therapy platforms (e.g. gene therapy and high throughput drug screening).
- New multidisciplinary collaborations to discover new treatments, in particular cross indication therapies.
- A network of clinical trial centres ready and able to conduct investigator led trials, attracting trials from abroad.

Implementation cost: \$665,000

Thank you

We are deeply grateful for the opportunity to represent the needs of the forgotten children, those with dementia, and their families. Our goals to accelerate therapeutic development and provide access to excellent medical and social care, is an aspiration for every Australian, regardless of their child's medical diagnosis. As a collective, we can and should do better for these children so they are no longer left behind.

Thank you for your consideration of this endeavour.

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