Australian Childhood Dementia Research Funding Report 2024

A Childhood Dementia Initiative report

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Acknowledgments

In the spirit of reconciliation, Childhood Dementia Initiative acknowledges the Traditional Custodians of country throughout Australia and their connections to land, sea and community. We pay our respect to their elders past and present and extend that respect to all Aboriginal and Torres Strait Islander peoples today.

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Background

A recent study[1] showed that childhood dementia is caused by more than 145 rare genetic disorders which affect 1 in every 2900 births. Modelling in this paper estimated that in Australia:

- two babies are born every week who will develop childhood dementia
- childhood dementia is so severe, that half of these children will die before they reach the age of 10 years
- someone dies from childhood dementia every 4 days.

Not only are the lives of children with dementia short, they are extremely difficult.[2,3] As a result of the progressive cognitive decline, children lose communication skills and experience changes in eating, motor function, sleep, and behaviour resulting in complex medical issues and needs. Parents watch their child(ren) suffer increasing levels of confusion, distress, unhappiness, and pain. Childhood dementia is also associated with significant carer stress, anxiety, and challenges in care. Psychosocial challenges are numerous and encompass physical, economic, social, emotional and psychological implications.[2,4]

Treatments and cures are needed to both improve length and quality of life for children with dementia and their families. However there has been a long-standing disparity in allocation of funding to childhood dementia research. As a result, there is a lack of childhood dementia clinical trial options and few new treatments gaining regulatory approval globally.[5]

The level of childhood dementia research funding was compared to childhood cancer, another severe group of paediatric diseases which cause a similar number of deaths each year in Australia[1]. It is worth noting the comparative prevalence of cancer and dementia in children aged 0-14 (Figure 1). In Australia approximately 1.4 times more children are undergoing treatment for cancer at any one time than the number living with dementia, and this was taken into account in our analysis.

Thanks to intensive medical research in recent decades, death rates from cancer almost halved between 1997 and 2017 in children aged 0–14[6] in Australia and in high-income countries, more than 80% of children with cancer are cured (Figure 2).[7] In contrast, childhood dementia has had no notable overall improvement in survival. We endeavour to learn from the progress in childhood cancer and achieve similarly impactful improvements in length and quality of life for children with dementia.

Figure 1: Children 0-14 years in Australia living with childhood dementia and childhood cancer.

Figure 2: Children 0-14 years in Australia who will survive dementia and childhood cancer.

Until now, each of the 145 genetic conditions that cause childhood dementia have been considered and viewed individually, with little awareness, research or support. Childhood Dementia Initiative was founded in 2020 to bring the conditions that cause childhood dementia together and challenge this siloed approach in order to enable sustainable global health solutions for children with dementia.

Collectively addressing childhood dementia is a world-first approach that is providing opportunities for greater scale, impact and acceleration of therapy development. It is unlocking opportunities to work across multiple childhood dementia disorders at once, develop platforms for therapy development and put in place research infrastructure such as biobanks, data collections and collaboration

opportunities. Importantly, the awareness raised of these disorders is attracting researchers from other fields including adult dementia to work in this area due to the untapped opportunity to make progress for not only childhood dementia but other neurodegenerative diseases.

Australian funding analysis

Childhood dementia research funding overall

We analysed the funding allocated to childhood dementia through the Federal Government's National Health and Medical Research Council (NHMRC) and Medical Research Future Fund (MRFF) from 2017 to 2023.

In total, 35 projects have been funded by the NHMRC and MRFF into conditions that cause childhood dementia totalling $23.4 million (Table 1 and Supplementary Tables 1 and 2). This includes research into individual childhood dementia conditions, for example Sanfilippo syndrome and Rett syndrome and four projects researching multiple childhood dementia conditions concurrently. It was challenging to decide which projects to include because many were not explicitly researching dementia in childhood, but we believe we have reached a reasonable and conservative estimate based on expert advice. Where a project is broader than just childhood dementia, a proportion was calculated and the rationale for this calculation can be found in the Appendix.

The analysis revealed:

- **Childhood cancer received 4.6 times more funding than childhood dementia per patient** (Figure 3).
- On average $22 million dollars has been invested in childhood cancer per year over the past 7 years compared to $3.3 million per year for childhood dementia.

Table 1: NHMRC and MRFF funding from 2017 to 2023

	Total	Estimated patient population (Australia)	$ per patient	Ratio
Childhood dementia	$23,392,611	1394*	$16,781	1
Childhood cancer (0-19)	$153,748,380	1984^	$77,494	4.6

*Elvidge et al., 2023, defined as those diagnosed with a childhood dementia disorder before the age of 18.
^Childhood Cancer prevalence was calculated assuming an average treatment duration of 2 years. Prevalence equals the number diagnosed in 2020 and 2021 aged 0-19 (2,253) minus the number of deaths in those years (271). Australian Institute of Health and Welfare 2022. Cancer data in Australia. Canberra: Accessed: July 2022; https://www.aihw.gov.au/reports/cancer/cancer-data-in-australia

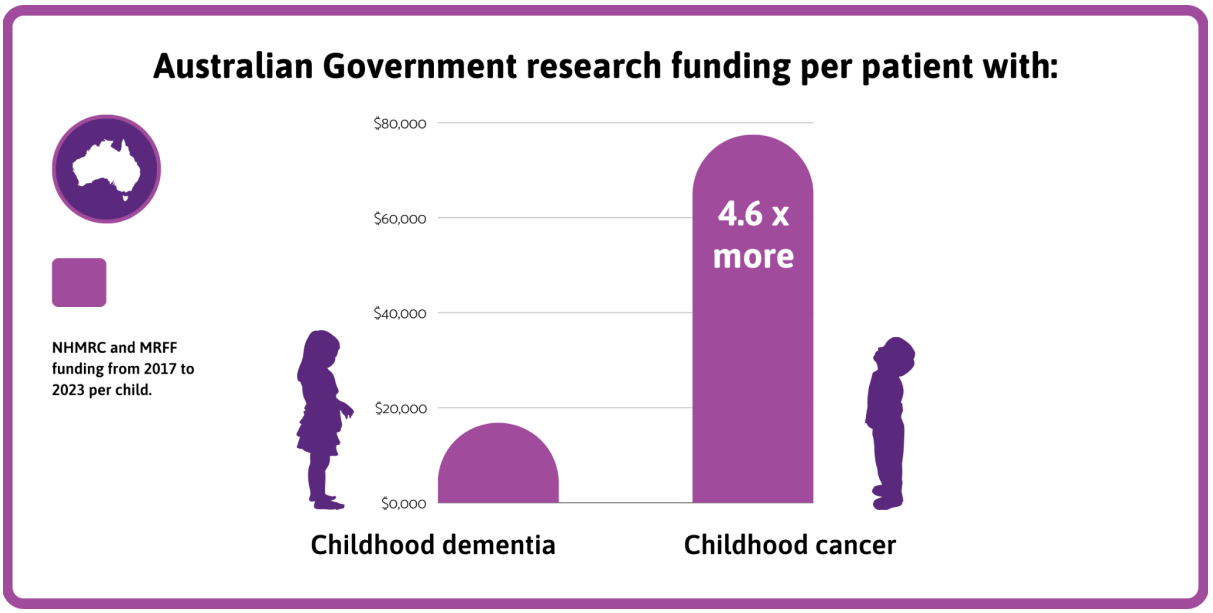

Figure 3: Australian Government research funding from 2017 to 2023

Research funding subgroup analysis

We noted in our analysis that 20 of the 35 projects (50% of research funding) for childhood dementia was for mitochondrial diseases. Mitochondrial disease accounts for approximately 9% of the childhood dementia patient population[1].

In 2023 there was a $15 million MRFF grant awarded that was necessary to implement mitochondrial donation in Australia, after the law was changed in 2022 to allow the use of this technology. Mitochondrial disease can affect many systems and organs in the body and symptoms can start at any age. It is estimated that approximately 2% of people diagnosed with mitochondrial disease have childhood dementia[1]. Mitochondrial donation is welcomed and will help some families affected by childhood dementia caused by mitochondrial disease avoid passing it down to future generations. This is important because IVF techniques available to couples at high risk of many other types of childhood dementia, are not applicable to mitochondrial disease caused by changes to mitochondrial DNA. Like all childhood dementia disorders, mitochondrial disease currently has no treatments, the biology is complex, and is difficult to diagnose, therefore, investment into mitochondrial disease research must continue. However, this funding skewed the funding analysis. To get a sense of the disparity in the remaining 91% of the childhood dementia population, an additional calculation was made excluding mitochondrial disease.

Research into subtypes of childhood dementia that affect 91% of the childhood dementia patient population is relatively neglected. Excluding mitochondrial disease from the patient population estimate and research funding amount, it was revealed that there was only $9,256 of research funding per patient and this is **8 fold less than childhood cancer.**

Discussion

Research funding in Australia is lacking and does not align to need

This is made particularly apparent when comparing research funding per patient for childhood cancer and childhood dementia. **Over the period 2017 to 2023, childhood dementia received 4.6 times less funding than childhood cancer per patient**.

This is despite great unmet need for medical research. Death rates for children with cancer have almost halved, steadily and dramatically declining so much that, between 2008 and 2017, the 5-year survival after a cancer diagnosis for children was 87%.[6] By contrast, **childhood dementia is terminal for all children and has had no notable overall improvement in survival in recent decades** underscoring the great unmet need for investment in medical research.

It is anticipated that this lack of research funding for childhood dementia is replicated around the world. This is due to the lack of awareness of this group of conditions which have traditionally been seen as individually rare conditions, rather than grouped together based on their similar clinical presentation. This historic and ongoing lack of research funding is contributing to a global lack of clinical trials and subsequent extremely limited treatment options for children with dementia.

However, recent analysis indicates that research inequity is particularly severe in Australia. A 2024 analysis of clinical trials globally showed that, per patient, there were 24-fold fewer clinical trials recruiting children with dementia than children with cancer in December 2023. In Australia the disparity was even greater with a 43 fold difference in clinical trials for children with dementia than children with cancer. **Of 54 clinical trials recruiting patients globally, only 2 of these trials were listed as recruiting in Australia, and no new trials started in Australia in 2023.**[5]

Opportunities to transform treatment of childhood dementia

In a world first, in 2022, the Australian Government announced a dedicated research funding call for childhood dementia. $2.7 million was allocated to 5 childhood dementia research projects in 2023 through the Medical Research Future Fund. This enabled the first projects that are studying multiple childhood dementia disorders concurrently. This is expected to give unique new insights into childhood dementia, demonstrate the economies of scale that can be achieved and accelerate the development of therapies. This is a step in the right direction, but this was a one off opportunity.

Clinical trial results released by companies[8–12] and published in peer reviewed journals,[13–16] have demonstrated positive results, especially in children treated early in their disease course. So, it is not that the development of therapies for this group of diseases is too difficult, or not possible. There is now an opportunity to transform treatment of childhood dementia, but **to capitalise on these advances, large scale, coordinated and collaborative research funding is needed.**

In conclusion, advances in genomics and development of therapeutics in recent decades have enabled effective treatments to be within reach for childhood dementia, however **increased research funding for childhood dementia is needed to address the historic inequity in attention to childhood dementias, and to progress and deliver effective treatments to patients.**

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Appendix

Supplementary Table 1: Childhood dementia related projects funded by the MRFF 2017- 2023

MRFF Initiative	Grant Opportunity	Organisation	Project Name	Chief Investigator A	Contract Start Date	Total Funding	Proportion	$ allocated	Type of CD
Clinical Trials Activity	2019 Rare Cancers, Rare Diseases and Unmet Need - General	The University of Queensland	Ataxia-telangiectasia: treating mitochondrial dysfunction with a novel form of anaplerosis	Professor David Coman	1/6/2020	$2,459,666.00	100%	$2,459,666	ataxia telangiectasia
Clinician Researchers	2017 Next Generation Clinical Researchers	University of Sydney	Improving diagnosis, treatment and prevention of mitochondrial disease	Professor Carolyn Sue	1/1/2019	$257,388.25	2%	$5,663	mitochondrial disease and other neurological diseases due to impaired mitochondrial function(1)
Emerging Priorities and Consumer Driven Research	2018 Accelerated Research - Sanfilippo Syndrome	Sanfilippo Children's Foundation (NSW)	Development of a personalised medicine approach for Australian children with Sanfilippo Syndrome (MPS III) utilising patient specific neuronal cell models	Not applicable	18/02/2019	$2,000,000.00	100%	$2,000,000	Sanfilippo syndrome
Emerging Priorities and Consumer Driven Research	2019 Accelerated Research - Leukodystrophy Flagship	Murdoch Children's Research Institute	Massimo's Mission	Not applicable	1/4/2019	$3,000,000.00	100%	$3,000,000	leukodystrophy
Emerging Priorities and Consumer Driven Research	2021 Chronic Neurological Conditions	Monash University	Early, novel and accessible intervention for children with developmental regression	Professor Katrina Williams	1/4/2022	$1,995,974.54	10%	$199,597	childhood dementia + autism spectrum disorder (2)
Emerging Priorities and Consumer Driven Research	2022 Effective Treatments and Therapies	University of New South Wales	Improving health outcomes by identifying biomarkers to delineate common mechanistic pathways and to monitor therapeutic effect of clinical trials in childhood dementia	Associate Professor Michelle Farrar	1/1/2023	$595,955.60	100%	$595,956	childhood dementia - all
Emerging Priorities and	2022 Effective Treatments and	University of Sydney	RTTomics: Towards developing new treatments and therapies for Rett	Associate Professor	1/1/2023	$595,972.93	100%	$595,973	Rett syndrome

Consumer Driven Research	Therapies		syndrome individuals using cortical brain organoids	Wendy Gold					
Emerging Priorities and Consumer Driven Research	2022 Effective Treatments and Therapies	University of Tasmania	A new substrate reduction strategy to treat childhood dementias: Glucosylceramide synthase-targeting antisense oligonucleotides	Associate Professor Anthony Cook	1/1/2023	$599,977.30	100%	$599,977	childhood dementia - multiple
Emerging Priorities and Consumer Driven Research	2022 Effective Treatments and Therapies	The University of Adelaide	Developing Nanoparticle Mediated Gene Transfer for Childhood Dementia	Doctor Nicholas Smith	1/1/2023	$302,148.00	100%	$302,148	Sanfilippo syndrome
Emerging Priorities and Consumer Driven Research	2022 Effective Treatments and Therapies	University of Melbourne	Developing an mRNA-based gene therapy strategy for Niemann-Pick Disease Type C1: a blueprint to treat childhood dementia	Doctor Ya Hui Hung	1/1/2023	$599,650.36	100%	$599,650	Niemann pick type C
Emerging Priorities and Consumer Driven Research	2022 Mitochondrial Donation Pilot Program	Monash University	Introducing Mitochondrial Donation into Australia: The mitoHOPE (Healthy Outcomes Pilot and Evaluation) Program	Professor John Carroll	1/6/2023	$15,000,000.00	50%	$7,500,000	Mitochondrial disease (6)
Genomics Health Futures Mission	2019 Projects	Monash University	Preventing mitochondrial disease using genomics	Not applicable	30/06/2020	$499,417.00	2%	$10,987	adult and childhood onset mitochondrial disease (1)
Genomics Health Futures Mission	2020 Genomics Health Futures Mission	Murdoch Children's Research Institute	Mitochondrial Diagnostic Network for Genomics and Omics	Professor David Thorburn	1/6/2021	$2,999,999.66	2%	$66,000	adult and childhood onset mitochondrial disease (1)
Stem Cell Therapies Mission	2022 Stem Cell Therapies	Flinders University	Pre-clinical iPSC-neuron screen of repurposed drugs for children with a form of dementia	Associate Professor Cedric Bardy	1/2/2023	$738,228.02	100%	$738,228	Sanfilippo syndrome
TOTAL								**$18,673,845**	

[14 grants]

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Supplementary Table 2: Childhood dementia related projects funded by the NHMRC 2017- 2023

APP ID	Date Announced	CIA Name	Grant Type	Grant Title	Admin Institution	Total	Proportion applicable to CD	$ allocated to CD	Type of CD
1140851	11/10/2017	Dr David Stroud	Career Development Fellowships	Systems approaches to understanding mitochondrial function and dysfunction in disease	Monash University	$431,000.00	2%	$9,482	adult and childhood onset mitochondrial disease (1)
1140906	6/12/2017	Dr David Stroud	Project Grants	Systems approaches to understanding the assembly of mitochondrial machines	Monash University	$600,005.00	2%	$13,200	adult and childhood onset mitochondrial disease (1)
1154352	13/8/2018	A/Pr Daniel Hatters	Research Fellowships	Proteostasis mechanics of neurodegenerative diseases	University of Melbourne	$649,175	5%	$32,459	Huntington's disease (4)
1155244	13/8/2018	Prof David Thorburn	Research Fellowships	Minimising the impact of mitochondrial disease by discovery and translation	Murdoch Childrens Research Institute	$860,385	70%	$602,270	primarily childhood onset mitochondrial disease (3)
1164459	12/12/2018	Prof Michael Ryan	Project Grants	Dissecting the functions of accessory subunits in mitochondrial complex I	Monash University	$722,284	2%	$15,890	adult and childhood onset mitochondrial disease (1)
1164479	12/12/2018	Prof David Thorburn	Project Grants	Deciphering the pathogenetics of rare diseases by multi-omic approaches: disorders of mitochondrial energy generation as an exemplar	Murdoch Childrens Research Institute	$1,041,548	70%	$729,084	primarily childhood onset mitochondrial disease (3)
1165217	12/12/2018	Prof Michael Ryan	Project Grants	Defining molecular pathways for COX2 maturation in mitochondrial Complex IV	Monash University	$595,788	2%	$13,107	adult and childhood onset mitochondrial disease (1)
1179029	6/10/2020	Prof Carolyn Sue	Partnership Projects	Delivering precision diagnosis to patients with mitochondrial disease: Using digital technologies to enhance the delivery pathway to provide an accurate genetic diagnosis for patients with mitochondrial disease	University of Sydney	$1,273,553.50	2%	$28,018	adult and childhood onset mitochondrial disease (1)
1184166	12/7/2019	A/Pr Daniel Hatters	Ideas Grants	The cascade of consequences in Huntington Disease from mutant Httex1 synthesis and aggregation	University of Melbourne	$747,700.00	6%	$44,862	Huntington's disease (4)
2000723	15/12/2020	Prof Justin St. John	Ideas Grants	UNDERSTANDING THE BENEFITS AND LIMITATIONS OF METAPHASE II SPINDLE TRANSFER	University of Adelaide	$1,629,373	70%	$1,140,561	primarily childhood onset mitochondrial disease (3)

2001112	15/12/2020	Prof John Carroll	Ideas Grants	Mitigating the risks of mitochondrial donation	Monash University	$1,063,748	70%	$744,624	primarily childhood onset mitochondrial disease (3)
2001536	15/12/2020	Dr Wendy Gold	Ideas Grants	Developing exon replacement gene therapy to cure Rett syndrome: an innovative model for neurodevelopmental disorders	University of Sydney	$475,105	100%	$475,105	Rett syndrome
2009732	9/14/2021	Dr David Stroud	Investigator Grants	Developing a multi-omics platform for the diagnosis of mitochondrial disease	University of Melbourne	$1,570,120.00	2%	$34,543	adult and childhood onset mitochondrial disease (1)
2010149	9/14/2021	Dr Luke Formosa	Investigator Grants	Understanding complex I assembly for better diagnosis and future treatment	Monash University	$650,740.00	2%	$14,316	adult and childhood onset mitochondrial disease (1)
2010332	11/4/2021	Prof Aleksandra Filipovska	Ideas Grants	Programmable correction of mitochondrial DNA mutations	University of Western Australia	$760,442.50	2%	$16,730	adult and childhood onset mitochondrial disease (1)
2010939	11/4/2021	Prof Michael Ryan	Ideas Grants	Molecular mechanisms underlying the pathogenesis of complex I dysfunction and mitochondrial disease	Monash University	$1,370,808.00	2%	$30,158	adult and childhood onset mitochondrial disease (1)
2019993	14/12/2022	Dr Julia Pagan	Ideas Grants	Tuning mitophagy in mitochondrial diseases	University of Queensland	$684,080.00	2%	$15,050	adult and childhood onset mitochondrial disease (1)
2021085	14/12/2022	Prof David Thorburn	Ideas Grants	Modelling of mitochondrial disease in specific cell lineages to understand pathomechanisms and develop effective targeted therapies	Murdoch Childrens Research Institute	$1,360,059.40	70%	$952,042	primarily childhood onset mitochondrial disease (3)
2022156	17/11/2022	Lottie Morison	Postgraduate Scholarships	Improving outcomes for children with complex communication needs	Murdoch Childrens Research Institute	$99,112.50	50%	$49,556	Batten Disease (5)
2026191	15/12/2023	Dr Ian Harding	Investigator Grants	Hereditary Cerebellar Ataxias: Next-Generation Biomarker Discovery on a Global Scale	Monash University	$1,586,190.00	2%	$31,714	Hereditary cerebellar ataxias (7)
2026315	15/12/2023	Prof Aleksandra Filipovska	Investigator Grants	Tackling mitochondrial dysfunction: understanding and treating metabolic diseases	University of Western Australia	$2,697,165.00	2%	$59,338	adult and childhood onset mitochondrial disease (1)
TOTAL								**$4,766,351**	

[21 grants]

Notes for supplementary tables 1 and 2

(1) Mitochondrial disease affects 1 in 4300 people.[1] With an Australian population of 25.69 million, this means 5974 people in Australia. The prevalence of childhood dementia caused by mitochondrial disease is estimated to be 129 in Australia (incidence of 7 per 100,000 births, life expectancy of 6.1 and birth rate of 300,000 per year in Australia).[2] Since childhood dementia constitutes 2.2% of the mitochondrial disease population, this proportion was applied.
(2) Clinicians estimate that children with dementia would constitute approximately 10% of the patients at the developmental regression clinic
(3) Approximately 70% of patients with childhood onset mitochondrial disease have childhood dementia.[2]
(4) 5% of Huntington's disease cases have the juvenile form of the disease.[3]
(5) This project also includes Kleefstra Syndrome which is typically not a childhood dementia disorder so 50% of the amount was allocated
(6) It is estimated that approximately half of the funding relates to preventing childhood dementia and related conditions
(7) Some types of hereditary cerebellar ataxias are known to cause childhood dementia such as SCA7 and SCA17. SCA7 represents 2% of all SCAs[4] and SCA17 incidence is unknown. Estimate 2% allocation to childhood dementia.

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