Exploring the issues series

Childhood Dementia: Family experiences of health systems in New South Wales

March 2024

A Childhood Dementia Initiative report

childhood dementia INITIATIVE

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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.

Childhood Dementia Initiative considers the voices of families as central to improving awareness and understanding of childhood dementia and to creating change. We thank and acknowledge the parents and families who contributed to this important resource.

Thank you to NSW Health for requesting this report, for listening to families, and committing to address the needs of families impacted by childhood dementia.

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Executive summary

Dementia is usually only associated with the elderly. Tragically, hundreds of thousands of children across the world suffer from dementia. It's caused by more than 100 life-limiting neurodegenerative disorders, many of which are not yet understood. A baby is born every 2 days in Australia with a childhood dementia disorder from which they will die, most likely before their 18th birthday[1]. Children with dementia live with chronic, increasingly severe symptoms as well as progressive intellectual and physical disabilities, until they die prematurely. Childhood dementia causes 91 deaths in Australia every year. This is a similar number to deaths from childhood cancer for patients aged 0-14 years[2].

Yet most health professionals are unaware of childhood dementia. Correspondingly, finding care and support is extremely difficult for families. Additionally, research is scant and there are no cures or treatments.

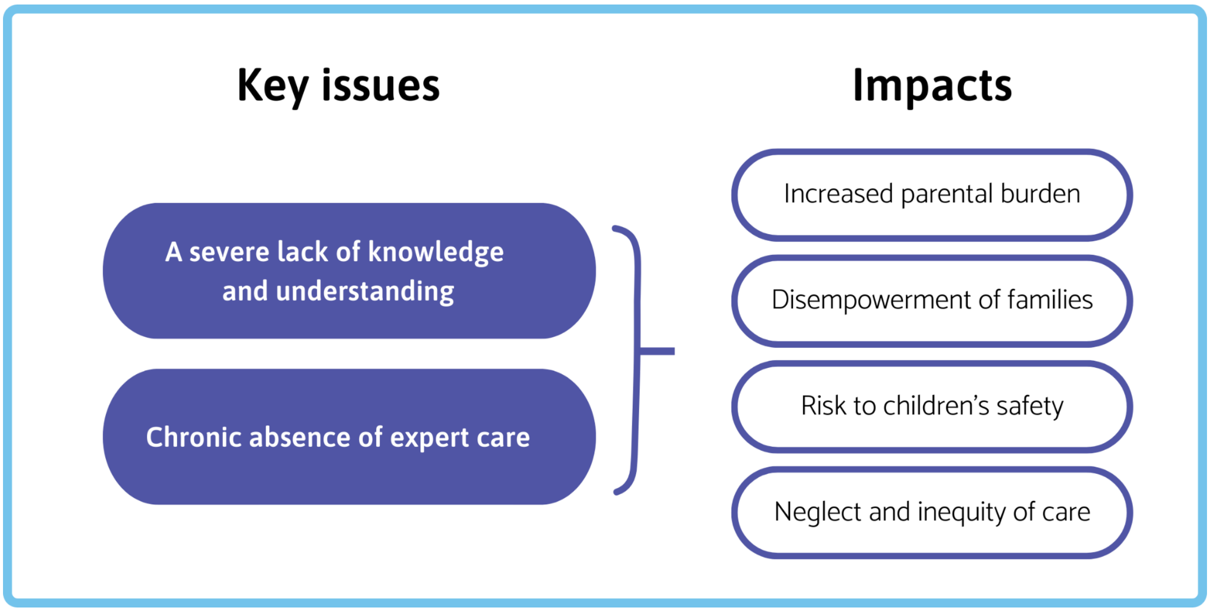
Childhood Dementia Initiative (CDI) was formed in 2020 to transform this situation. Listening to families is at the heart of CDI's approach. In late 2023, CDI conducted a series of roundtable discussions and interviews and a short follow up survey with parents and caregivers about their experiences with health systems in New South Wales (NSW).

Key Findings

From this research, 2 key issues were identified: **a severe lack of knowledge and understanding**, and **chronic absence of expert care**. These key issues have resulted in the following impacts for families:

- **increased parental burden**
- **disempowerment of families**
- **risk to children's safety**
- **neglect and inequity of care**

This report is intended to be used as input into further consultation and exploration by NSW Health and other bodies to improve systems of care in NSW and ultimately deliver greater outcomes for children with dementia and their families.



Key issues

A severe lack of knowledge and understanding

Chronic absence of expert care

Impacts

- Increased parental burden
- Disempowerment of families
- Risk to children's safety
- Neglect and inequity of care

Background

This report explores family experiences of the health system for children with dementia in New South Wales, Australia. Through their voices, Childhood Dementia Initiative seeks to communicate how families interact with the health systems in NSW, and to provide a basis for future improvements.

Childhood dementia is uniquely devastating and severely under-recognised. The condition is unknown to most people, including health professionals. As such, there are tremendous unmet needs in treatment, research and psychosocial support.

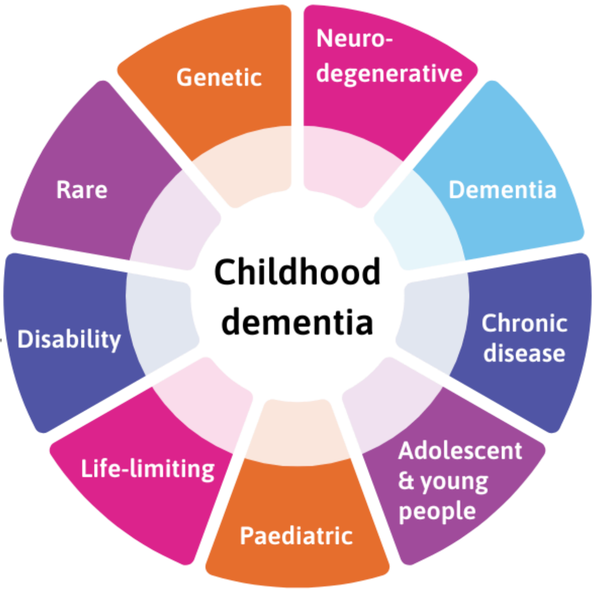
A baby is born every 2 days in Australia with a childhood dementia disorder from which they will die. 50% of these children will die before their 10th birthday and 70% of them before they reach adulthood[1]. Nearly 100 will die every year. This is about the same number of children who will die from childhood cancer (ages 0-14)[2].

Childhood Dementia Initiative commends NSW Health for requesting this report and its commitment to addressing the needs of families enduring this disease. NSW Health is undertaking further qualitative research in this area with a broader stakeholder group. Once complete, these results, in combination with this report, will form a comprehensive foundation for action.

About childhood dementia

Childhood dementia is caused by 100+ neurodegenerative genetic disorders. These conditions have recently been grouped to define this unique and under-recognised cohort. All childhood dementia disorders are life-limiting and there are no cures.

The specific attributes of childhood dementia and the lack of a coordinated approach means the children with dementia face a uniquely devastating situation in the Australian health system.



- There are no cures for childhood dementia and it is life-limiting for all affected children.
- Children experience chronic, increasingly severe symptoms and their intellectual and physical disabilities progress until they die prematurely. Children suffer from confusion, distress, unhappiness, and pain. The complex childhood dementia disorders can also cause seizures, loss of vision and hearing, and problems with bones, joints and cardiovascular, respiratory, or digestive systems.
- Due to the lack of awareness of the disease, disability, health and education systems don't cater for the unique needs of children with dementia. This results in heightened carer responsibilities for families. The psychosocial challenges they face are broad, encompassing physical, economic, social, emotional and psychological implications[4].
- Children with dementia are often excluded from essential services due to their dementia symptoms.
- The challenges facing children with dementia are not currently addressed by any national dementia policies globally.
- There is, and has always been, a complete inequity and underinvestment in research. This has resulted in no notable improvement in survivorship for children with dementia[5].
- Childhood dementia disorders are individually rare. Families report struggling to get diagnoses[3 4], and can subsequently go on to have more than one child without knowing about their genetic risk.

The challenges described by families in this report accumulate and are exponential when there are multiple children with dementia in a family. Given the genetic nature of childhood dementia, this is not uncommon.

Education for health professionals about childhood dementia is urgently required, as is a commitment to clinical trials and psychosocial support. Until then, children will continue to die and families will continue to experience the ordeals outlined in this report.

Childhood Dementia Initiative

Childhood Dementia Initiative is driving world-first action for every child with dementia, bringing all the genetic conditions that cause dementia in childhood together under a single umbrella.

Childhood Dementia Initiative works to ensure that Australian children and their families can access the treatments and support they need. This will change the lives of thousands of Australians now as well as the lives of future generations. Childhood Dementia Initiative's work is informed by the *Framework for Childhood Dementia Systems Change* and underpinned by the key principles of evidence, people and co-design. The framework aligns with the National Strategic Action Plan for Rare Diseases which recognises the need for research into rare diseases to be collaborative, person-centred and systematically address gaps.

Framework for Childhood Dementia Systems Change

Established in 2020, Childhood Dementia Initiative drives vital change by:

- Building evidence that is validated by families, experts and empirical data.
- Building networks. We bring cross-disciplinary experts together to enable change. This includes families, researchers, health professionals and service providers, and policymakers.
- Enabling partnerships, collaborations and co-design. This ensures sustainable, effective solutions are implemented.
- Advocating for enablers. This includes investment, policy and practice changes, as well as greater awareness. Shifts in funding, policy and awareness are already having an impact.

Methodology

This report summarises key insights that emerged during a series of roundtable discussions and interviews facilitated by Childhood Dementia Initiative at the request of NSW Health in late 2023. Twelve parents and primary caregivers of children with dementia participated in this consultation. For more information on the methodology, see Appendix 1.

Key Issues

Two key issues emerged from the discussions.

Severe lack of knowledge and understanding across health systems

The lack of awareness and knowledge of childhood dementia means that:

- Parents find it extremely difficult to find and use relevant, reliable information.
- Almost every health professional they encounter is unfamiliar with the disease and how it should be managed.

Chronic absence of expert care and services

At a systemic level, where there are services, there are no care pathways or integration of services. Additionally, parents report 'cobbling together' care teams across disciplines, geographies and the public/private systems.

These issues result in the following impacts on families:

- **Increased parental burden -** In the absence of expert teams and defined care pathways, parents act as researchers, care coordinators, advocates and project managers, further increasing their stress. This burden is exacerbated for parents with multiple children with dementia.
- **Disempowerment of families -** Parents report that the knowledge they have of their child's condition is frequently dismissed and/or disrespected by health professionals, that they feel disconnected from and unsupported by health professionals, and information is not made readily available to them.
- **Neglect and inequity of care** - The level of care a child receives is dependent on their family's geographic and socioeconomic circumstances, and whether they are treated in a paediatric, specialist adolescent or an adult setting or access care in the private and/or public system. The gaps in care as well as neglect and isolation reported by families are concerning. When compared to other chronic paediatric conditions, they demonstrate significant inequity.
- **Risk to children's safety -** When their child is being cared for by a health professional unfamiliar with childhood dementia or their child's specific condition, parents worry about risks to the child's safety if they are not physically present and available to manage their care.

Each of these impacts are accumulative and inter-related. They are not isolated impacts and all participants in this study reported experiences in each impact area.

Impacts on families

There is a severe lack of knowledge and understanding across health systems and a chronic absence of expert care. Families who care for a child (or children) with dementia reported the following resulting impacts:.

Increased parental burden

In the absence of expert teams and defined care pathways, parents act as care coordinators, advocates and project managers, further increasing stress and burden.

This study has found that parents are leading the investigation into their child's dementia condition, searching for answers with multiple healthcare professionals in different healthcare settings, and over prolonged timelines. **More than 50% of families consulted reported attending over 15 appointments with different healthcare practitioners** before receiving an accurate diagnosis and 30% reported the diagnosis process took more than 5 years.

Parents reported being left to coordinate their children's healthcare at all stages of their conditions. This included needing to source clinicians and care, managing communication between different specialties of care, managing medication and performing daily care. They also carry a significant mental load, feeling they are solely responsible for getting their children and family all they require. The burden of care falls predominantly on one parent.

"You're exhausted all the time, and you're concerned about your child, because you're there in an emergency, in a crisis. And you have to relate everything from the beginning about your child's complex, progressive condition. And it's very, very stressful."

"Every time we went to Liverpool hospital, I would say, 'You should have all this information on the computer.' And they're like, no, they have to start again, and they've got to start from fresh. And I found it quite strange that they couldn't just log in. And so, what I would do is, if we were rushing in an emergency, I would email the neurologist, and he would get the neurology team if he wasn't on to come down and see me and take over."

"I'm holding all of the information and it's the history and the clinical information and the medications all in one place for the system to access. It's in my head and on my Google Drive. And we participate in a clinical trial outside of the state so there is a big chunk of her history in another state."

"I used to carry a folder around everywhere with all the meds that she was on. And then the diary of what we've given, so we could show the doctors and nurses because there's just way too much to remember off by heart. I'd have a spreadsheet and we'd always have it and they just take it and photocopy it, and so, it was easier for them."

"I was the one gathering the research and articles and bringing it to the appointments. This is what the overseas doctors said, and the local doctors were willing to work with that. We still occasionally contact those German doctors, and they've been really supportive, and they don't charge us if we need to consult with them."

Parents need to be relentless advocates for their children to receive the care required. They felt that they carried the sole responsibility of coordinating care for their child within a complex web of clinical disciplines.

"We were in such desperate need of answers to try to understand what was going on. It was, 'Let's just do everything, do whatever we can.' So we think back now and say that if we hadn't done that test, where would we be, you know, 2 years later?"

> "I am absolutely terrified because there's nobody who's going to do what I do for my kids and navigate them through this broken system and get them what they need and be looking out for them."

In addition to coordinating the care for their children, responsibility for seeking clinical trials for children's dementia conditions and communicating this information with healthcare professionals falls to parents. Participating in clinical trials is the only chance for families to access treatment that can alter their child's prognosis, as there are currently no curative treatments for childhood dementia conditions. In some instances, families reported needing to travel over state and territory borders to access clinical trials; this travel resulted in significant impacts on the family unit.

> "Because we pushed for it, we had testing done in order to understand the mutations that the kids had. That information was really important because I was at the same time investigating international clinical trials and mutations were a factor. This doctor actively discouraged me from pursuing international trials, but I had to, they were our only hope. The results of these tests sat on the doctor's desk for 6 weeks before they were shared with us."

> "Our interaction then with Westmead, we felt was lacklustre. We just went, were weighed, measured, answered questions and discussed things. There were not any trials or research or things available, we had to go and find the information and bring it back to the doctor, and then suggest the trials to her to then say yes, or no, that kind of thing, which was a bit again, underwhelming. I would have thought we could have been presented with the information of 'this is what's out there', or that sort of stuff."

There is an absence of care coordination, despite children being diagnosed with conditions that are genetic, progressive and life-limiting. Families whose children live long enough to transition from paediatric to adult services reported additional challenges.

“He was honest and said, ‘Look, I don’t have time to learn a lot about this disease. But I’m happy for you to consult with others. And I’m happy to listen to what you tell me.’ We sort of thought, well, that’s the best we’re going to get now.”

“I’ve got an ambulance care plan now for my daughter and I’ve got an enduring guardianship now. But I could only get that once she was 18. But between, I think there’s a lot of gaps there, transitioning from child to adult, in that kind of thing.”

“Having a small team would be able to control the number of directions we as a family were being pulled.”

“We’ve never had any multidisciplinary teams. It’s very, ‘Here’s the hospital system and we are dealing with everyone else’. My wife and I are definitely leading our daughter’s care.”

“Paediatric wards in adult hospitals do not have the expertise to look after a dementia child.”

There is insufficient collaboration and communication between clinicians, and families experience additional challenges within health system hierarchies. Some parents shared that they became the decision-makers for their children’s care because of the health system’s siloed nature.

“All the doctors couldn’t agree and we were literally the meat in the sandwich.”

> "For diagnosis, there was no coordination, no sharing, just tests were being done and other doctors didn't even know about them."

> "I have to ask the neurologists to communicate with each other and I provided their email addresses so they can send each other their letters. I ask, 'Can you please send it?' Because otherwise it's word of mouth from me."

Parents called for care coordination implemented by multidisciplinary teams and better coordination between intersecting systems, including the NDIS.

> "A quicker process and communication with the NDIS and them respecting the reports (from) NSW Health would be a big improvement."

> "One good clinical lead, preferably a neurologist and good working relationships between all practitioners including myself would have produced the biggest positive impact for us all. If for some reason, issues between clinicians arose, I as a parent would have somewhere to go to resolve it diplomatically. That would be a great improvement, doctors could save face, but my child's health would remain utmost important""

Disempowerment of families

Parents report that the knowledge they have of their children's conditions is frequently dismissed and/or disrespected by health professionals, that they are disconnected from and unsupported by health professionals, and information is not made readily available to them.

Parents shared examples of good practice, and nearly all parents had an example of individual health professionals who had provided excellent care for their children. However, these individuals were almost always stumbled upon by chance. Overall, parents reported a number of challenges in their long-term experience of the health system.

Specific instances of poor, and at times offensive, communication with individual health professionals were shared. These experiences increased stress and left parents feeling disrespected and disempowered.

"I've had awful times where doctors would say to me while seeking diagnosis, 'What are you worried about? She doesn't look funny.'"

"I haven't until very recently had any interaction with health professionals that has made me feel heard, respected, like my child's an equal human or my concerns listened to. It has been 10 years of battling."

Parents need to work in partnership with health professionals. **Despite shouldering the burden of care and becoming disease experts themselves, parents reported being unable to ask questions or challenge decisions.** They felt unable to make choices around preferred treating clinicians and that they had to 'keep doctors happy' in order to get the care their children needed.

"I remember thinking, 'Why do we not have a neurologist? This is a brain disease.' And so I asked our clinician to refer us to a neurologist. And they said no, because they were the lead clinician. So I actually privately referred to a neurologist."

"Feeling I had to keep the doctors happy, to not make any waves or rock the boat so that my daughter would receive the best care and support and keep things smooth. I avoided conflict at all times and went out of my way to be nice. In the past, if I voiced concerns or was upset, I felt I was labelled 'difficult' or just another 'neurotic mum', and it made the relationship much harder. I felt like I had to defer to them and not be as direct as I would like to be."

> “I wanted to make it smooth sailing, I didn’t want any conflict. There was often a mismatched agenda in what we were discussing at appointments, and so I would kind of just leave going with what was the point? None of my concerns or issues were addressed in that meeting. And it was almost too draining.”

> “I was assigned a lead specialist, with whom, immediately, we clashed. I will take some of the responsibility for that as a newly diagnosed parent who is probably quite angry, but they never listened to me and they were dismissive of my child. I said, ‘I do not have faith in this clinician, and the care that we are receiving as a family. I would like to be moved to a different specialist within the team’. They said to me, ‘I’m sorry, we do not switch patients to different specialists. If you want a new lead specialist, you have to go to a different hospital. And the only way you can go to a different hospital is to move states because as long as you’re in the New South Wales health system, you come to Westmead’.”

Moreover, as well as describing challenges of feeling unsupported and misunderstood themselves, **parents reported a lack of continuity and connection between health professionals and their children.**

> “I think practitioners, especially specialists, underestimate their role in these children’s lives. These kids are visiting the health care professionals so frequently and it is a real battle if they have no connection with those professionals.”

> “I don’t expect our doctor to be across every single symptom and detail of every rare disease they treat, but I do expect they will listen to me when I bring them information I have found. We parents often become experts in their child’s disease, and this needs to be recognised, not dismissed. This has not been my experience.”

> “Doctors with more open minds about other types of treatment would be a great improvement.”

Receiving a diagnosis is a life-changing event for both children with dementia and their families. **Seeking and receiving a diagnosis is confusing and fraught, and often exacerbated by delays and inadequate information and communication.**

> "One of the greatest challenges was the time it took to get a diagnosis, and all the implications that this had on our family and our daughter such as potential for any treatment or earlier interventions, and also just the stress of having five different possible diagnoses at one time."

> "When we first saw the hospital team, they said you're going to have global developmental delay, a possible mental disability, she's never going to walk and talk or communicate - it was heartbreaking. And then they did all these tests. They couldn't find the sample and it ended up being a whole year to get the sample back, which was quite frustrating. When it did come back, they said, 'We want to see you at the hospital.' I went up alone and got the diagnosis. I burst into tears."

> "They explained the biological cause of the condition. They'd used the word 'progressive', but I didn't understand what that meant. They said, 'The name of the condition is Sanfilippo syndrome, and we've made an appointment for you to see a specialist in two weeks' time at Westmead. And don't Google it.' They hadn't told me that it was terminal."

> "We did lots of irrelevant tests. And our form of that disease can be diagnosed with a blood test, one simple blood test. It took a long time to get to the neurologist to listen to us. By the time we got our diagnosis, we sat down, he said, 'Look, I think you already know the answer. You're right'."

Sufficient information about their children's diagnosis, dementia condition and prognosis is not provided to families.

"I don't know what we should be accessing in terms of allied health, in terms of other specialists and what we need to be monitoring. You kind of work out from other parents what you need to offer your child."

"The thing that I reflect on often is of all of those people in the room, why did nobody flag actually, 'You've not understood what we've said when we said progressive'? They certainly did not say terminal. We left that room not knowing that she was going to die. They gave us a piece of paper which was photocopied from the UK which was generic, about 15 years old. So I Googled it. And I found a high school creative writing exercise that I read that told me that my daughter was dying, and quite possibly my son."

Neglect and inequity of care

Family insights indicate that the level of care a child receives is dependent on their family's geographic and socioeconomic circumstances, and whether they are treated in a paediatric, specialist adolescent or an adult setting, and whether they access care in the private and/or public system. The gaps in care as well as neglect and isolation reported by families are concerning. When compared to other chronic paediatric conditions, they demonstrate significant inequity.

Adequate psychological support is not provided to either parents or children at critical events, such as receiving a diagnosis.

There was a notable lack of counselling and psychological support offered to families during the diagnosis process. **All participants in this study reported there was no counsellor or psychologist present when their children's diagnosis was delivered. None of the participants reported being offered counselling or psychological support during any follow-up care.**

"I wasn't offered any support. I wasn't even given Lifeline's number."

"For me, I have anticipatory grief. I'm not a bereaved parent yet. But right now, the number of times she's had close calls, do you know what I mean? Or the behaviour is out of control. I just need someone to talk to."

"I struggled finding a psychologist for my daughter. First, she had a psychologist in the children's hospital but then they got busy, and they were supposed to be finding the one when she went to the adult hospital, it never happened, they couldn't find one or something."

"So after 2 years talking to my doctor, I have finally admitted that I needed some help with antidepressants. And really, that's about it. There's been no talk of any counselling, mental health psychology, anything for us as parents."

"My child talks about death a lot. I haven't told her what's going to happen. But she's seen other kids die and she's putting two and two together. She's seeing that she's having seizures, she's losing mobility and she's asking questions. There's just not that support there for her."

"I find it astounding that both my children were diagnosed with a terminal condition and I have never been offered any form of counselling."

Experiences of palliative care and end-of-life care are varied and inconsistent. Some children are ineligible for provision of palliative care due to their dementia condition and there are gaps in the services available.

"The nurses and the paediatrician who did a home visit on the Monday or over the weekend, both indicated that their hands were tied, or told me their hands were tied. The dose of medication, morphine or something, that had been prescribed by this head physician was conservative and very low. And it was no wonder that our child was still in pain, but their hands were tied. There was really no compassion and it felt really heavy handed, it felt like the 11th hour. I'd had no rapport with this physician, I'd never had any interaction besides 2 very brief appointments over a 6-year period. This was despite us having discussions around our end-of-life plan with our paediatrician, we had Hammond Care involved, and we had Bear Cottage involved."

> “We did approach someone about supporting us as parents and our older kids and they said I should find Bear Cottage but we’re not at, you know, the really palliative stage. But when I spoke to them, they said that we’re not eligible.”

> “The head palliative care doctor came in heavy-handed and made decisions we disagreed with, we felt there was a complete disregard for our family’s wishes and respect, and that she knew better and made it very stressful. This was someone we never had a formal consultation with over a 6-year period and was then taking the lead in the last 5 days of our daughter’s life.”

> “I want to love palliative care and I want to get something from them. But right now, they’re just another thing to do. They haven’t helped in any way, unfortunately. I remember saying to them, can you not help me with care coordination or anything? ‘No, no, your paediatrician will need to help you.’”

> “They said, ‘We’re happy for you to come and look around the facility, but there’s no way we’re taking her because her needs are too high.’ So ironically, the child who’s requiring one-to-one care, whose family probably needs the respite most, is the child that they can’t actually look after. So we left, our terminally ill child got rejected from Bear Cottage.”

Parents reported inequitable access to care across different healthcare settings. Access to suitable care depends on their age (whether they are eligible for paediatric, young adult or adult services) and the specific services available in their geographic location. **A major factor in whether they can access care is their parent’s ability to educate health professionals, coordinate care and advocate for their child’s needs.**

> “If I didn’t have a history working in health, I wouldn’t have known how to navigate the system.”

"Three or 4 times, the head nurse said she's in the wrong ward. She would say, 'We can't take this kind of a patient, because she would get up and she would shout as well, that scares kids.' So, it was sort of like she didn't fit. Where does she fit?"

"I noticed in the children's hospital, they would have someone one-on-one when children were at high risk, but I've noticed they don't do that in an adult hospital. Anytime we've gone, I've had to stay in an adult hospital and you don't have a parent bed, like in a children's hospital. I would share the single bed with my child and sleep in there with them overnight. I knew the nurses didn't have time or the resources because it is two nurses to 30 patients in a ward."

"It's good that there is AYA palliative care in Manly now, but that's from age 15 and we're 13. So, we don't fit in the Bear Cottage system, and we don't fit in the AYA palliative care [hospice], we're just kind of in between."

"Now we're going to see a private gynaecologist. There's no gynaecologist that we've been referred to in the public system to work with the neurologist to get my daughter's IUD replaced. So, I've organised all that myself. We've had to do that privately. I want to be clear that having access to a gynaecologist who will work with a neurologist is critical to my daughter's care."

Families are not always able to access care that is required in their local areas, this includes but is not limited to, regional and rural areas. 25% of participants reported crossing state/territory borders in order to access care and support for their child, including clinical trials.

"At our local regional hospital, in terms of services, I get a little bit worried because he's got a port and things like that, and they get a little bit worried about touching him. So, if we have a problem, 9 times out of 10, I will ring Queensland to find out what to do and then they ring the local hospital."

"Less travelling and easier access for rural patients would have the biggest impact for our family."

"We live quite close to the Queensland border. In Queensland, we get exactly the kind of coordinated care for their disease. So, we see a neurologist and the metabolic team are up there, and they coordinate all our care. And then if we have emergencies, like last year, we get flown straight to Queensland. It makes sense to have that as we're only 3 hours from Brisbane. The only reason we go to Sydney is to literally access medication. We can't be sponsored through Queensland."

Follow-up care is inconsistent. This includes access to cascade genetic testing for siblings, referral to post-diagnostic care, palliative and end-of-life care, and access to clinical trials.

"I had no paediatrician, I had nobody who could prescribe me the medication that my kids needed. They couldn't allocate a paediatrician to me who had experience with palliative children or neurodegenerative conditions. There was a paediatrician who was open to learning, but I had already trained 2 others previously in my kids' condition, I couldn't face a third."

"I was left for 2 years without a psychiatrist for my daughter. We couldn't have the review for the behaviour or medications review and that was very stressful, because we're having a lot of behavioural issues. And then they said to me, well we don't have anyone that's qualified because she has to be 18. And she was 17. So, there was no one to look after a 17-year-old in an adult hospital because they didn't have the qualifications. So where does she sit then? And you've wasted this much time when I could have found a psychologist myself out of there."

“We recently were referred to the spine clinic, where the doctor had never heard of the disease before. And we needed a referral to a dietitian, and we went through the hospital system trying to get a dietitian, because the NDIS believed that that was a health responsibility, but we couldn’t get one.”

“We had to actively seek the diagnosis of our second child. When we received the diagnosis of our first child our paediatrician had indicated that the metabolic team or the genetic counsellors would contact us. We hadn’t heard from anyone. When we had our first appointment with a metabolic team we said, ‘Well, there is a sibling involved, and we need to get a sibling tested.’”

Risk to children’s safety

When their child is being cared for by a health professional unfamiliar with childhood dementia or without deep knowledge on their child’s specific condition, **parents worry about risks to their child’s safety especially if they can not be physically present and available to manage their care.**

“My daughter was upset with me, and they wouldn’t let me in the ambulance. And if there’s no one else around, what do I do? Let her go by herself, having seizures? The paramedics don’t understand what’s going on.”

“If I wasn’t there, in hospital, in the emergency, in the ambulance, my child probably wouldn’t be alive to be quite honest.”

“Our child cannot talk or communicate, and we don’t want to put her with people that she doesn’t trust or know.”

“I am terrified about what happens if I’m not around to look after my kids.”

Parents are responsible for complex medication management, including communication of dose changes and care team compliance. They felt they were not provided with adequate information or support on this.

"If I go to the neurologist, they might say to me, 'Increase the Risperdal to twice a day'. I didn't feel right that I was verbally giving that authority [to the broader care team], I thought it should have been from them."

"Medication management was 100% my responsibility. Sometimes there wouldn't even be a script, it would be an email from the neurologist that would be like increase this, decrease this. There'd be a general script at the pharmacy, but I would then be communicating exactly what that was. They often get it wrong. So then you get your Webster-paks and check everything."

"The nurse had to intervene and give an injection to calm her down and she said, 'You need two people doing this, you shouldn't be doing this. You shouldn't be having to manage this kind of a situation.' But of course a parent is going to put your hands on and it's your child. But at the same time, I don't want to be. I don't want to be holding my child and having her resent me at her end of life or at any point. I shouldn't be the bad guy. It's like it's a clinical situation. It should be a clinical person."

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Appendix 1 - Methodology

Overview

The study was undertaken in two steps: the first an online interview or face-to-face roundtable session, this was followed up with an online survey to all participants.

Study Participants

All study participants were recruited from the Childhood Dementia Initiative Family Advocates Program. They were parents of a child or children with a rare genetic condition that causes childhood dementia. Representation included:

- Different diagnoses of conditions that cause childhood dementia.
- Residing in different locations across New South Wales and the Australian Capital Territory in Australia, including metro, regional and rural settings.
- Bereaved families
- Families with more than one child with childhood dementia
- Primary carers (mothers and fathers)
- 12 participants (11 families represented)

Questions and topics from roundtable sessions and interviews

Question areas

- What are some of the challenges you and your child experience with health systems in NSW?
- What are some of the things that have worked well for you and your child in health systems in NSW?
- What improvement would make the biggest impact for you and your child?

Topics explored

- seeking diagnosis
- receiving a diagnosis
- post-diagnostic support and services
- palliative care
- intersecting systems including disability, education, housing, social services.
- transitions in care

Follow up survey questions

1. Please indicate the different health care practitioners you spoke to when seeking a diagnosis for your child/children?
    - General practitioner
    - Paediatrician
    - Speech therapist
    - Occupational therapist
    - Neurologist
    - Genetic counsellor
    - Geneticist
    - Social worker
    - Psychologist
    - Psychiatrist
    - Early childhood nurse
    - Other (please specify)

2. How many appointments with health care professionals did you attend in seeking a diagnosis?

    - 1 - 2 appointments
    - 3 - 5 appointments
    - 5 - 10 appointments
    - 1 - 15 appointments
    - greater than 15 appointments
    - Other (please specify)

3. In what year did you receive your child/children's diagnosis?

4. How old was your child/children when they were diagnosed?

5. Please indicate the different health care practitioners who were present when you received the diagnosis for your child/children:
    - General practitioner
    - Paediatrician
    - Speech therapist
    - Occupational therapist
    - Neurologist
    - Genetic counsellor
    - Geneticist

Social worker
Psychologist
Psychiatrist
Early childhood nurse
Palliative care specialist
Counsellor
Other (please specify)

6. How long did it take for you to receive a diagnosis for your child/ren?

1 - 3 months
3 - 6 months
6 - 12 months
1 - 2 years
2 - 3 years
3 - 5 years
more than five years

7. Please indicate the different health care practitioners you speak to since your child/children's diagnosis:

General practitioner
Paediatrician
Speech therapist
Occupational therapist
Neurologist
Metabolic
Genetic counsellor
Geneticist
Social worker
Psychologist
Psychiatrist
Early childhood nurse
Palliative care specialist
Counsellor
Ophthalmology

Other (please specify)
None of the above

8. Which specialty has your child/children been referred to as their clinical lead?

9. Do you cross state/territory borders to access care and support for your child/children, including clinical trials? If yes, please specify which states and territories you travel to to access care and support, including clinical trials.

10. What are some of the greatest challenges you and your child experience with health systems in NSW?

11. What are the most significant things that have worked well for you and your child in health systems in NSW?

12. What improvement would make the biggest impact for you and your child?

13. Is there anything else you would like to add?

14. Please describe your connection to childhood dementia
    - mother of child/children with dementia
    - father of child/children with dementia
    - carer/guardian of child/children with dementia
    - Other (please specify)
    - none of the above

15. Are you a bereaved family member?

16. How would you describe where you live?
    - Metropolitan
    - Regional
    - Rural
    - Remote Other (please specify)