

Navigating care for rare disease developmental delay

A Guide for Parents and Carers Through Diagnosis and Accessing Health and Allied Health Services

Smith-Magenis Syndrome Australia

rare voices AUSTRALIA

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Introduction

At Smith Magenis Syndrome (SMS) Australia, we are parents of children with a rare disease that is also considered a developmental disability. Developmental delay, when combined with a rare or undiagnosed disease, can be misunderstood.

About 2 million Australians live with a rare disease that impacts their daily life over a long time. For many people, these impacts meet the Australian Government's definition of disability[1].

As parents, we experienced challenges navigating services and systems. It seems we are not the only ones.

For this project we:

- Spoke with 20[2] parents. Their children live with a rare or undiagnosed disease. They also experience developmental delays.
- Facilitated a project steering group with 9 parents of children with rare or undiagnosed disease developmental delay. We went to this group at different stages of the project.

This guide was hard to write.

Many challenges identified through interviews relate to problems in practice and systems. Parents were often doing the right things and asking the right questions.

There are problems in health and allied health practice. The National Disability Insurance Scheme (NDIS) and systems need to be improved. For example, the public health and school systems. Parents also need help with finding and using these services and systems.

Understanding Developmental Delay

Childhood developmental delay[3] occurs when a child does not reach the following milestones within the usual age range:

- Physical
- Cognitive
- Communication
- Social
- Emotional

The first 5 years are critical.

Developmental delay[4] may be temporary or linked with a ***permanent*** disability. Some children experience delays related to illness, life events or disruptions in caregiving.

Early intervention can support them to catch up. Other children may have permanent developmental disabilities. ***Global developmental delay*** means a child is developing slower than expected across 2 or more areas. It is often used as a temporary diagnosis.

Children with a rare disease often present with developmental delays that are ***subtle, fluctuating and unusual.***

Many children with rare disease disability will receive a global developmental delay or idiopathic autism diagnosis. This explains their symptoms and how it impacts them while they wait for more testing and support.

In rare disease disability, developmental delay varies greatly but can include:

- Challenges with speaking
- Challenges in understanding language
- Differences in muscle tone
- Changes in the skeleton and bones
- Differences in facial features

It is important to acknowledge the significant and often lifelong impacts of rare disease disability. It's also important to recognise the impact on the daily lives of children and families.

In Australia, children's development is monitored by maternal and child health services, general practitioners (GP) and early childhood services. If needed, children may be referred to allied health professionals, a paediatrician, Medicare-funded services or the NDIS, if eligible.

Australian policy has increasingly prioritised the early identification and support of children with developmental delay and disability.

About this guide

The aim[5] of the following sections is to support parents and carers of children with rare or undiagnosed disease in seeking support to access health services and systems for their child.

The guide has 3 sections:

Section 1: Diagnosis – Getting a Diagnosis and Receiving a Diagnosis

Section 2: Supporting Yourself and Your Family

Section 3: Navigating Health and Allied Health Services

More detailed information, including the lived experience that informed this resource, is available in the full guide. You may need support to use this resource.

Use the guide with the **Practical Tips for Parents and Carers companion resource**, which includes:

- Helpful tips and suggestions
- Suggested questions you might like to ask
- Useful services and links



Section 1

Diagnosis

Getting a diagnosis and receiving a diagnosis

Early diagnosis helps

Diagnostic tools like genetic testing are becoming more available. In our project, people diagnosed earlier generally reported better experiences accessing supports. There were still challenges.

Getting a diagnosis

Parents told us that their experience with diagnosis and healthcare came down to who they were seeing. They also said a good paediatrician or general practitioner (GP) came down to luck.

Some people said they had a wonderful paediatrician. They picked up on developmental delays early and monitored their child closely.

Getting a diagnosis and accessing health services were both easier if a child had physical health problems.

Delayed diagnosis when developmental delays were present was a ***significant issue.*** Many participants in our study said their concerns were dismissed. The 'anxious mum' was a common theme.

> "At six weeks old, I picked it. I remember saying to my GP [has she] got a syndrome? And he laughed at me, and then she got formally diagnosed with a blood test—ahh [at] about 2 [years old], I think…"

In our study, developmental delays alone did not generally appear to be a significant risk factor or red flag for health professionals[6].

Parents said the doctors and paediatricians they were seeing didn't consider or explain developmental delays. For example, being late to sit on their own or walking. Saying the child 'would get there in their own time' was common.

Two participants who were migrants ended up getting a diagnosis overseas. This was after raising multiple concerns with doctors in Australia.

> "We told [concerns about development delays] to [our] GP, but she said 'she will take time'…"

Sometimes children were diagnosed with global developmental delay before diagnosis. However, no one explained:

- What this meant
- What services should be accessed
- If there was a plan to track progress
- If more tests were needed

> "I'm still yet to have a medical professional tell me about the syndrome [at 2 years of age, diagnosed at birth] or what is a global delay? What is a developmental delay? What does this mean? Where to go from here? Everything that we are doing is because of self-research…
>
> "I actually remember the last paediatrician appointment… we went in and they did the weight and the measurements and all of those sorts of things. I said to them, look, she's been steady weight now for the last... 6 months or whatever it is. She's not moving anywhere. And they said, oh yeah, but she's at a healthy weight, so that's fine… and then they said to me, oh, but what do you want from us now? What do you need from us?
>
> "And I was like, well, I don't actually know. I feel like they should have been saying, alright, well, she's 1 1/2, so we should be doing this and she's not reaching these markers. So, this is where you need to go from here. Here's some resources or, you know, these are the therapies that you should talk to NDIS about including in a plan next. But there's nothing like that."

Health professionals come from different backgrounds, training and expertise. Their concerns or approach may not match your expectations or needs. Feeling dismissed may be the result of health professionals lacking knowledge or time.

Medical Gaslighting[7] is when a healthcare professional doesn't listen or take someone's health concerns seriously. It impacts several groups more, including people with poorly understood conditions like rare disease disability.

Receiving a diagnosis

Receiving a rare disease diagnosis is not easy. It can be life changing, devastating and scary. You are finding out your child won't have the life you thought they would. You also don't know about the future or how to help them.

There can be very little support for parents during this time. Most parents said their **experience of receiving a diagnosis** looked something like being given a pamphlet or printed information about the condition. They were often told not to Google.

In most cases, there was limited follow-up. Additionally, there was not much of an explanation of the condition or development delay. There was often no explanation or referral to allied health services.

Parents were told to join the NDIS. They were then left to do this on their own.

Referrals and supports after a diagnosis were generally only done where there were physical health problems.

> "… because no one knew about [rare disease]… And I remember when she was little, the only reason why I guess we got supports was because she was hearing impaired."

Parents often said no information was provided about how developmental delays relate to diagnosis. Often, no information was given about what a child's condition or progression may look like.

It's important to understand this situation is complex. Many parents shared that their doctors said they didn't necessarily know how developmental delay would play out over a child's lifetime. They said every child with rare disease disability is different.

> "[Doctor] was quite honest with us and said, 'I have no idea basically how this might affect her'. At that point, there were only 40 registered cases of her deletion worldwide. So, it was very rare… But basically, he just said, 'we'll just keep an eye on her, get early intervention and just tackle things as they come up'."

Some parents said they didn't necessarily want to know how developmental delays would impact their child. They said they'd prefer to wait and see in line with the doctor's advice.

Although, others said looking back, they would have liked to ask more questions about what was to come. This could have helped them prepare.

> "… like, how does this play out sort of later? Because I feel like that kind of gives parents like, almost like the initial stuff. Like she did tell us a lot of this you won't see until she's about two. So, we enjoyed, like, the bliss. And I think that was helpful.
>
> "I wish I'd asked more questions about, like, how does this look long term for them in their life? Or to adults? Because I think that would have given me personally a better understanding… like definitely [about] the behaviours. I wish I'd known a little bit more.
>
> "Not that it would have changed anything, but I think I had a very different view of what that was going to look like…"

Others said they'd asked all the questions they could think of. They didn't think anything would change by asking more questions.

Adapting to life with rare disease disability can be challenging and takes time. It can help to look at the broader picture beyond diagnosis and developmental milestones. For example:

- How a child participates in daily activities
- What they can and cannot do independently
- What supports are needed
- How rare disease disability affects learning, communication, behaviour, safety, emotional wellbeing, and family life more broadly



Section 2

Supporting yourself and your family

Grief

You will likely experience grief after your child is diagnosed. It is OK to grieve. Many participants in our study expressed guilt.



> "… when [child] was first admitted into hospital, I was like, I just don't know why I'm struggling with it all. And [counsellor] was like, you're grieving. And I was like, what do you mean? Like, [child's] not dead. She's still alive. And she's like, no, no, you've just had a baby and you're coming up to Christmas, and you had in your head this picture of what life was going to be like. And it's been totally turned upside down.
>
> "She's like, you can grieve the picture in your head of what life would be like. And that really stuck with me because ever since then I've been like, you do have doubts of like, oh, this is what I imagined would happen. Even if you think you've, you know, with developments like, OK, we're going to reach this milestone, you might then regress and so then you grieve like it is. There's so much grieving that's not to do with death, but it's the same process."

Grieving after a diagnosis doesn't make you a bad parent. It doesn't mean that you see your child as a 'burden'. But the reality is, caring for a child with rare disease disability changes your life in many ways. It is OK to recognise the life changing nature of diagnosis and to:

- Grieve the life you thought your child would have
- Grieve the family you imagined
- Feel fear over what is going to come next
- Feel fear about 'not knowing' how life is going to look for your child, for you, or your family

There are some well documented stages of grief[8] you might go through:

1 Denial

Initial shock or refusal to accept the diagnosis. This acts as a temporary buffer to help process overwhelming information.

2 Guilt

Thinking there were things with the pregnancy or the birth that were your fault. You may feel guilt over how this will affect your other children.

3 Anger

Feelings of frustration, helplessness, or resentment. These may be directed at oneself, others, or the situation.

4 Bargaining

Attempts to negotiate or make sense of the diagnosis. This often involves 'what if' or 'if only' thoughts.

5 Depression

Deep sadness, withdrawal, or emotional heaviness. This can come in waves and vary in intensity.

6 Acceptance

Acknowledgment of the reality of the loss. This does not mean being 'OK' with it but learning to live with it.

None of these experiences or stages happen in order. People experience them in different ways, and at different times.

Some participants in our project described the grief going 'in waves'. They said they'd go through phases of acceptance and then grief might come again as children missed certain milestones. For example, going to Special School instead of mainstream school. Other people close to you might also go through these things, and at different times to you.

The parents we spoke to said that another part of grief looked like navigating changes to life and how other people reacted or dealt with their child's rare condition.

Some participants talked about how they were the first in their family or immediate circle to have a child with rare disease disability. This made it harder to understand and navigate.

Sometimes friends and acquaintances could misunderstand the diagnosis or developmental delays. They said things like 'they will get there' or pointed out that they also struggle with tantrums with their children. Sometimes these words might be well-meaning with the intent of comfort. But it was also draining for some to talk with parents of children without rare disease disability about milestones or achievements.

Being 'pitied' for having a child with rare disease disability was also draining. Some people said they withdrew or closed off from some parts of their life, or people in it, at times.

> "I realised that I was becoming a little bit bitter about other people. [They] just don't understand. And you find this disconnect. People start to isolate. Either you talk to them and you're open and then suddenly they can't relate, and so they feel bad and they stop talking to you about normal stuff. They think they can't complain. Or you start to become a bit resentful of other people who just have no idea. And there becomes this disconnect."

But parents in our study also spoke about working through grief and finding a 'new normal'. The same participant said:

> "But I didn't want to spend my life focused on disability. In just the negative and the weight of disability… It is the weight and the worry I will always have for that child is there. She's in this world, it's here. But sometimes to diffuse that and have your focus on other things. It's like going to work when you've had bad news… You want, you need a breather."

> "… But there's times where it can be such a hard day and the funny things that come out of her mouth or the things she does and you're just, there's not a day I don't laugh. And I'm like that is really important. And it also keeps you connected with the rest of the world… It's great, I love connecting with people in disability, but it's also nice to still feel like you can connect with the greater world and that you're not like, it's us and them, I don't even understand that, and you don't get me."

A project steering group member talked about a comparison that offered comfort for them – 'Welcome to Holland' by Emily Perl Kingsley. This describes planning for a trip to Italy, but you end up in Holland. They likened this to the experience of raising a child with rare disease disability.

When planning a trip to Italy, you might buy guidebooks and make plans specific to Italy. You might anticipate wonderful experiences you have heard from others. But instead, the plane lands in Holland. Holland is a different place to Italy.

It's not what you had prepared for so there will be challenges. But Holland still offers unique and beautiful experiences which you can come to learn and enjoy.

Just know that grieving is normal. It is OK and you are not the only one, even though it can feel very lonely at this time.

Looking after each other

It is important to look after yourself and your family. You can be pulled in many different directions at this time. For example, appointments and getting help from services and systems.

Most of the responsibility for caring and putting early support in place rests with parents and carers (especially mothers). Remember that for systems and services, this is a job. For you, this is your life. And caring for a child with rare disease disability is lifelong.

Carer Burnout[9] is real. You need to look after yourself. You don't need to have all the answers. You don't need to do it all by yourself, although it can seem that way.

For example, some parents in our study found that they had to change work arrangements. This helped with managing the needs of their child. Many had other children and talked about the impact on the entire family, including siblings.

For you

Counselling is an option for ***yourself individually***, as a couple, or ***as a family***. Ask your GP about a referral and a mental health care plan.

For siblings

Little Dreamers is an organisation dedicated to supporting ***young carers ranging from 4-25 years old.*** It provides counselling and group work, holiday programs, tutoring programs and so forth. Check eligibility for where you live as eligibility and programs differ by state/region.

In our study, family and friends were generally discussed as supportive. But some people didn't have family. Others had family living overseas or far away.

Some parents in our study also said their families were grieving too. For example, grandparents. Parents were also relied on for information and support, which was draining.

The following parent talked to some of these struggles. They suggested a model called the Ring Theory[10] for parent support.

> "I didn't realise why I was struggling with other people telling me how they felt about our situation… if you're at the centre of it, you can't be taking on board how everyone else feels about your situation. It's too much emotion. You vent outwards and then it's not that your mother or your sister can't feel sorry for you, but they can't put that on you. They have to go further out, and they have to vent to friends and family.
>
> "Because otherwise, living in a world where your child is at the centre of this as a parent, it's really heavy to constantly have people telling you how they feel about your child's condition. So, you've sort of got to teach people that it's OK to feel that way. Just don't tell me about it. Go and tell other people and other friends. Let me vent to you. And that also was helpful for me in sort of navigating all these things as that model of coping."

It's important to centre yourself and your immediate family and 'grieve out'. Identify what support you need from people close to you. Sometimes friends and family don't know what you are going through, or what they can offer.

Support groups for parents of children with disabilities will likely be your best friend.

> "… talk to other parents [of children with disabilities]… At the special schools, you get more information from parents than you get from most other people because even though all our kids have got something different, there is usually somebody [that's] tackled what you've needed to tackle."



Section 3

Navigating health and allied health services

The choice between private and public health comes down to finances and personal choice. In our project, private health wasn't always the answer. However, private healthcare did have benefits regarding paediatricians and more timely access to specialist services.

Co-morbid physical conditions alongside developmental delays appeared to be a more straightforward path through the health system.

For developmental delays, like delayed speech, walking and motor skills, allied health services generally provided the **best interventions.** For example, speech therapists and occupational therapists (OT).

In most cases, participants said allied health services were not explained to them. They weren't told who to access and what could help with development delays and rare disease disability.

Referrals were only usually made in cases where there were physical co-morbidities. It was up to parents to find allied health supports.

> "... It has probably [been] a lot of Googling, and trial and error with different therapists and staff as well to find... That would have been helpful to, you know, having doctors or even the paediatrician or people that we were coming in contact with saying, you know, like, here… this is like the priority area. Here is a good OT or you know."

Parents said **waitlists were incredibly long.** Sometimes 1-2 years for speech therapy. Waitlists and a lack of access to services meant less choice of providers.

This also added to worries about children not accessing services that could help with developmental delays and their rare disease disability.

Not every provider offers the services needed. Many said they found good providers but that the provider left the service or moved. There was never a handover or referral. The parent had to find a new provider themselves.

After being on a waitlist, some parents said they eventually found that a service or practitioner didn't have the skills to work with their child's needs.

> "I asked before… that we are using [a] AAC [Augmentative and Alternative Communication] device. How familiar are you with the AAC device? And [therapist said] oh, no, not a big deal. Yeah, I can do it. Now I came to know [therapist] is using [a] AAC device [for the] first time… because I can't find anyone now onwards, right? And it will be another wait time."

Parents also said that developmental delays were viewed by services in terms of 'goals'. For example, aligning with goals in a NDIS plan.

Goals were often described as generic and unrealistic. They were not changed for a child with rare disease disability. This kind of goal setting could be frustrating or distressing for parents. They were aware their child may not achieve the goal.

Some parents expressed that goals were deficit based. They didn't recognise the strengths of their child. They also didn't recognise that being different and having rare disease disability is OK.

Asking parents what their goals were for their child was also described as unhelpful. Parents want realistic goals and expert opinions.

> "Well, I think it has to be a really collaborative approach, right? Like you as a parent can only do so much, but the therapist has to like put the hard work in to actually try and reach that goal, right?... And it's hard for parents like, oh my god, how much more do you want us to like make a decision on?"

Participants got the impression from services that developmental delays were not lifelong and could be caught up on. Generally, this wasn't the case where there was a diagnosed rare disease disability. Or children might catch up with some developmental delays, but others remain and become stronger over time. For example, a child might catch up on their speech delay, but intellectual disability is lifelong.

In many cases, children would improve in skills but then lose other skills and/or regress.

> "… you feel like you've mastered one skill, or [child's] mastered the skill, and then you move on to the next skill and then she seems to forget the previous skill.
>
> "... I've never felt so ignored and powerless… you're almost scared to tell the NDIS any progress that's been made with development delay because are they going to then cut funding? And then the next year [they're] going to go backwards and they're going to regress."

Given our study was on rare disease developmental delay, most parents talked about the frustrations of providers not knowing about their child's condition. They said it was tiring to go through the same assessments and repeat information. They then eventually came to find that the allied health provider does not know about the rare disease. Sometimes information provided was dismissed with standard 'one-size-fits-all' interventions applied.

Parents said using too many standard services at once meant too many appointments. In the end, they also found that long awaited services didn't help in the way they had imagined.

> "I feel like it was very overwhelming in terms of like, these are all the things that [child] she could struggle with later down the track, and these are the things she is struggling with. So, we'll just cover all bases and just, and we probably overloaded her and ourselves in terms of getting every single therapy.
>
> "It's like your trial and error. It's like, OK, well, now I've wasted 12 months of all this money in therapy, but I figured out… no, that's not the good one to go to. Like, oh, this person needs to specialise in this, or this is what we're looking for. But it's purely the parent is educating themselves on all these rare things."

Looking for more practical tips? Download the Practical Tips for Parents and Carers companion resource.



Notes and references

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5. NB It is not the aim of this guide to provide health advice on development delay, rare disease or disability. If you have concerns about your child's development and/or health, please see a relevant health provider.
6. We acknowledge that this is a small sample, and that the intent of this project was not to conduct health and/or clinical research. However, this finding is considered significant across 20 interviews, and we would suggest warrants further investigation via research and advocacy.
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