Smith-Magenis Syndrome Australia

rare voices AUSTRALIA

Navigating Care for Rare Disease Developmental Delay

Practical Tips for Parents and Carers: Through Diagnosis and Accessing Health and Allied Health Services



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About this project

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].

For this project, Smith Magenis Syndrome (SMS) Australia:

- 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.

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

Developmental delay[4] may be temporary or linked with a permanent disability.

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.

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.

This resource **aims to help parents and carers through diagnosis and with access to health and allied health services.**

This resource is split into 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

It includes:

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

For more information about this project, including the lived experience that informed this resource, please download *Navigating Care for Rare Disease Developmental Delay: A Guide for Parents and Carers Through Diagnosis and Accessing Health and Allied Health Services.* You may need support to use this resource.

A summary guide is also available.

Section 1

Diagnosis – Getting a diagnosis and receiving a diagnosis



Getting a Diagnosis: Tips and Questions to Ask

Prepare for appointments by writing down any concerns or questions you have. Suggestions include identifying:

Any specific concerns regarding development. For example, not rolling, not sitting, speech delay, low muscle tone, feeding issues, facial features.

How these concerns came about. For example, other children you know are sitting at 6 months, but your child is not sitting at 8 months.

You may also like to bring a trusted friend or family member to appointments. They can help with backing up your concerns about your child. They can also help with confirming your experience at appointments.

During the appointment:

Share information about the specific rare disease disability you are concerned about. You can ask for testing. Provide research and evidence if you can.

Be upfront about your expectations.

Ask for further investigations or referrals for an identified developmental delay. For example, physio or speech, or somewhere that can assist with feeding issues.

Ask what any further testing is for and why it's being done.

You can also ask for a plan and next steps before leaving the appointment. For example:

1	Will there be another appointment, and when?
2	How will your concerns about your child be monitored?
3	Are there any referrals or services you should be seeking in the meantime?
4	When will testing and further investigation be done?

If you don't feel like you are getting the answers or responses you were hoping for:

1	Ask your doctor if they have concerns about your child's development. Ask why or why not.
2	Ask your doctor when they may be concerned about a developmental milestone you feel your child is not meeting. For example, when would you consider taking further action? What would that action be?
3	Raise your concerns about being dismissed in a calm manner. For example, I don't feel like my concerns are being heard. I would like to explore other options.

If you don't get the outcome you are hoping for, consider seeing the same doctor again. Repeat visits for the same issue are considered a 'red flag'.

The doctor should re-evaluate if any more steps need to be taken on the next visit. Your doctor may also be following advice or guidelines that you're unaware of. That means seeing another doctor may produce the same result.

Record everything



Keep medical records, referrals and results together in a file or safe storage place. Keep development specific assessments or ask for a copy of the results.



Record your concerns and your child's progress to keep a history so you can show health professionals. The Raising Children Network[5] provide information for parents on understanding development delay and milestones.



Visit a maternal health service and ask for a developmental check.



Document your visits with a summary of what was talked about and responses. This means you will have a history of service responses.

Most importantly, trust your gut! If it feels wrong, it probably is. If you aren't satisfied with a provider or feel dismissed, seek a second opinion. You can also look for a new doctor you can trust.

Receiving a Diagnosis: Questions to Ask

Here is a list of questions you might like to ask following diagnosis. What you ask is a personal choice for you as a parent. Doctors may not know the answers to your questions.

1	What might I expect in the next few years?
2	Where can I get more information about this rare condition?
3	How could developmental delays play out over my child's life?
4	What services can help with developmental delay? For example, with behaviours and speech.
5	What can we do at home to support my child?
6	What symptoms or red flags should I watch out for? What should I do when these things come up? This might include co-occurring conditions, such as anxiety, or autism and ADHD.
7	What regular appointments will be necessary? For example, annual hearing tests, eye checks, blood tests.
8	Where can I find supports for myself and my family?
9	If there are 3 things I should do now, what are they? This will give you something tangible, but manageable, to follow up.

Ask about a plan and next steps before leaving the appointment. For example:



Will there be another appointment with the same doctor, and when? If the appointment is more than a month away, ask if you can schedule a follow up for a few weeks' time. This gives you the chance to ask questions after you have had time to process the information.



Who will we be seeing regularly about my child's rare condition? What is the frequency of visits?



How will concerns about my child be monitored and actioned?



Ask if the doctor can write down necessary services and appointments so that you can follow up.



Ask if the doctor can be contacted between appointments, and how. By phone or email?

Section 2

Looking After Yourself and Your Family



Here are some suggestions from parents in our study about how family and friends might be able to support you.

They may offer to:



Care for your child for a short time. This might allow you to spend time together as a couple, or with your other children. It may also give you a chance to have some rest.



Take care of other children so you can go to hospital and/or appointments.



Pre-cook meals or deliver groceries or other essential items. This can help with balancing caring responsibilities with hospital visits, taking your child to appointments and so on.



Help with cleaning, laundry, and/or other household tasks.



Provide a quiet space for you to catch up on life admin, work, or to rest or sleep. Feeding you while you are there is a bonus!



Listen and provide emotional support without judgement or wanting to fix things.



Check in regularly, however it best suits you. For example, you might prefer text messaging to phone calls.



Recognise that sometimes you might not want help. You might just want space with your family, and/or some alone time. You may not want to talk.

Not everyone has this kind of support in their lives. They might not have family or close friends. Their family and close friends might live elsewhere. If this is you, take it easy on yourself and find relief where you can. For example:

- Prioritise what needs to be done. You might not be able to cook a meal every night and that is OK. Laundry might not get put away, and the lawns can wait.
- Outsource household tasks like cleaning and gardening. Check if your NDIS package can pay for these.
- Check if any organisations can provide relief packages. For example, Carer Gateway. These packages are not permanent and require a process. However, they may provide some relief at times.
- Check Core Supports available through the NDIS for in-home supports. An inhome support worker can provide help in the home. They may also be able to take your child out.
- You might enrol your child in daycare. This can give you a break or help with spending time with school age children after school. Check with individual daycare providers to see what's available.
- Look at local services in your area. There may be some disability organisations that provide before or after school care, holiday programs or day programs for children with disability

How to find online groups

Participants in our study suggested joining the online group for the rare and/or undiagnosed disease your child has. The parents we spoke to said this is the best thing you can do. These are usually national or international groups. For example, in our project, some of these groups had 40 or 50 members internationally. Others were more established.

Look for support groups in your local area. For example, the Australian Government offers a group called MyTime for parents and carers. You can also look online or contact Carer's Australia. Check what is available in your state or territory.

These groups may offer informal supports. They might also have information about your child's rare condition and be able to help with finding services and supports.

Section 3

Navigating Health and Allied Health Services



Here are some suggestions for navigating health and allied health services.



Keep paperwork and record visits to show existing and new providers

This was a common suggestion from the parents we spoke to. This could include documentation of the milestones your child is meeting and/or not meeting, reports from health and allied health services, diagnosis letters, recording visits to services, and supporting documentation for applications. For example, carer impact statements for the NDIS.



Provide information and research about your child's rare condition

A good therapist or health worker is interested in your child's rare condition. They will look into how best to support your family based on the evidence.



Prioritise supports

Ask your GP or paediatrician about which supports to prioritise. A referral from a GP or paediatrician won't influence waitlists. However, asking them about what your child needs for their specific case, and how services are going to help, was a suggestion.



Ask allied health providers for their expert opinion on what would best help your child

Reframe questions about goals. Ask about what evidence-based practices providers would recommend for your child, based on their individual assessment.



Co-ordinated supports

The best-case examples from our study involved supports being coordinated. This could be through the Early Childhood Development Program (ECDP), or health centres focused on childhood development delays. Ask your GP, paediatrician or an allied health provider who might be local to you about the services they provide.

How to get on waitlists

1. Ask for advice from parents who have been through similar journeys. For example, through disability support groups.
2. Even though there are waitlists, you can still try to source quality providers that meet the needs of your child.
3. A suggestion given by many participants in our project was to stay on waitlists even when you find someone. That way, if you decide to leave a provider, or if they leave you, you still have options.

Questions you may ask a new or prospective provider:

1. Do you have experience working with rare and/or undiagnosed disease? What type/s?
2. Do you have experience working with children with disabilities? And what is your approach to their care?
3. Do you have experience working with [diagnosis, and/or specific development delay/s]?
4. This is my child's diagnosis, and these are the things they need help with. How can you assist?
5. How do you manage behaviours and engagement?
6. How will you assess and track my child's progress? And how will you measure the effectiveness of interventions?
7. How will you involve us as parents in our child's therapy and support plan? And what will you do or provide to help us implement strategies?
8. How can I communicate my child's delays and limitations without my child present? This ensures your child is not hearing negative things about themselves.

Note: It might be difficult to ask these questions if you're speaking with a receptionist at first. You can ask for someone to call you. Or, you can go on the waitlist and ask the practitioner in the first appointment. Other providers might require an assessment before being able to answer these questions.

Some red flags for 'not so great' health and allied health providers might be:

- Not feeling listened to as a parent who knows your child.
- Dismissive or uninterested in your child's diagnosis. And/or not adapting interventions to suit your child's needs.
- Not researching your child's condition. And/or dismissive of information you give them.
- Not considering your child's life in the context of the family. For example, the realities of how you will implement strategies at home, the impact on siblings, avoiding carer burnout etc.

Always trust your gut. This was the most common advice given by participants in our project!

If something doesn't feel right, change providers. This can be daunting when waitlists are long and you feel lucky to get in anywhere. However, the experience of most participants in our project was that staying with a provider you feel is not helping or isn't working out doesn't usually have many benefits.

Best case scenario

It is a waste of time and stressful for you and your child.

Worst case scenario

It can have unintended consequences such as trauma from a bad experience and/or carer burnout.

Some useful services and links

- Australian Government Department of Social Services (DSS): Provides information about services for carers and for people with a disability and carers.
- Carers Australia: The national peak body representing unpaid carers in Australia.
- Carer Gateway: An Australian government program providing supports and services for carers.
- Little Dreamers: An organisation supporting young people, aged 4-25 years, that provides unpaid care to family or friends affected by disability.
- My Time: A free support network for parents and carers of children under 18 years who require additional support because of disability or other needs.
- National Disability Insurance Scheme: An Australia Government program providing funding to eligible people and carers with permanent and significant disabilities.
- Raising Children Network: An Australian Government funded parenting website providing independent, evidence-based, and expert-reviewed information for parents and carers.

Note: This resource was developed for the Peer Support and Capacity Building grant for the NDIS.

Notes and references

1. Australian Government, Australian Public Service Commission, 'Definition of disability', accessed 23 September 2026, https://www.apsc.gov.au/working-aps/diversity-and-inclusion/disability/definition-disability.
2. 20 x interviews were conducted online, via Microsoft Teams, in 2026. Participants came from Queensland, New South Wales, Victoria, and South Australia - 3 x regional. Participants were parents of children with a diagnosed or undiagnosed rare disease/disability, of which development delay played a part. 9 of the children had Smith Magenis Syndrome, caused by a genetic deletion or mutation, 9 x children had other syndromes caused by genetic deletions and/or duplications/ mutations, 1 x child had a syndrome characterised by multiple physical disabilities, and 1 x did not have a diagnosis. They had a genetic abnormality (one-of-a-kind in the world). Ages of children of the participants were: 4 x 5 years and under, 10 x 6-10 years, 4 x 11-16 years, 2 were adult children - 23 years (diagnosed in 2024 at age 21), and 34 years (diagnosed in 2020 at age 8).
3. 'Developmental Delay Factsheet', accessed 9 August 2026, https://www.schn.health.nsw.gov.au/factsheets/developmental-delay; Early Childhood Intervention & Developmental Delay Therapy | Early Start Australia, n.d., accessed 9 August 2026, https://www.earlystartaustralia.com.au/what-we-help-with/developmental-delay/.
4. Chronic Conditions Manual, 'Developmental Delay or Disability Child', Chronic Conditions Manual, 17 June 2024, https://www.ccm.health.qld.gov.au/management-of-diagnosed-conditions/developmental-delay-or-disability-child.
5. 'Developmental Delay', Raising Children Network, accessed 9 August 2026, https://raisingchildren.net.au/guides/a-z-health-reference/developmental-delay.