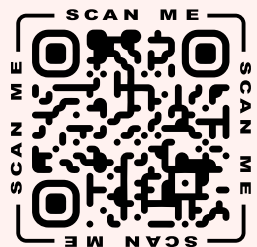


# Spinal muscular atrophy

EDUCATIONAL  
RESOURCES FOR  
PARENTS AND FAMILIES



SCAN FOR ELECTRONIC  
VERSION & LINKS



# A LETTER TO PARENTS



You are here because you were told your child (or a child of someone close to you) has a newborn positive screening result for spinal muscular atrophy (often called SMA).

These resources will help you understand SMA, the next steps, and how to talk with your healthcare team to make the best choices for your child. Please know there is hope and you are not alone.

You may have never heard of spinal muscular atrophy before. This news can be shocking and frightening. And it can be confusing since your baby looks well and the next steps are uncertain. We know this is a difficult time and the emotions you are going through are normal given such an unexpected situation. While SMA is a serious disease if left untreated, treatments are available.

Some information online may not be up to date and relevant to you and your child. In order to make sure the information here is relevant for you, Australian and New Zealand families who have been through this, alongside experts, have made this website for you.

## What these resources cover

The newborn screening test	3
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# HEAR FROM REGGIE'S MUM



“When our son Reggie was born, he looked just like any other baby—perfect and deeply loved from the moment we met him. Not long after his birth, we learned that he had Spinal Muscular Atrophy (SMA). Because of early screening, Reggie was able to begin treatment straight away. Hearing those words was overwhelming, and like any parents we worried about what the future might hold. But very quickly we realized that Reggie is so much more than a diagnosis.



Now seven months old, Reggie surprises us every day. He is strong, determined, and works so hard during his physio sessions. Watching him learn new movements and reach little milestones—like getting up on all fours and starting to move himself into sitting—fills us with so much pride. Every small step feels like a big celebration in our house.

Reggie also has two big brothers who absolutely adore him. Our home is busy, loud, and full of life. They cheer him on, make him laugh, and remind us every day that he is simply their little brother first.

SMA is part of our story now, but it doesn't define who Reggie is. He is curious, happy, and determined, and we are so excited to watch him keep growing and discovering what he can do.

Sharing our story is important to us because awareness and early support can make such a difference for families like ours. Most of all, we hope that when people read about Reggie, they see what we see every day—a beautiful little boy with so much ahead of him.”

-Reggie's mum

# I've received a screen positive result for my baby...what does this mean?

Your baby has received a screen positive result for spinal muscular atrophy (SMA). Parents will often ask what was found and what this means.

## THE NEWBORN SCREENING TEST

After your child's birth, a midwife asked if it was ok to do a newborn screen test for your baby. This tests for rare but serious conditions that can be helped through treatment if found early.

Your baby had a small amount of blood collected from a heelprick. This was sent to a lab where all the conditions on the newborn screening panel were tested. SMA has been on the national NBS panel in Australia since 2022, and New Zealand in 2024. Your baby's result means they most likely have SMA. This is a rare condition that most people often do not know much about.

It is a serious condition of the nerves and muscles involved in muscle strength and movement. But, living with SMA is not the same today as it was even a few years ago.

Treatments now exist, and are most helpful when started early. This is why SMA is part of newborn screening. So the most helpful choices can be made for your baby quickly.



## AFTER THE SCREENING RESULT

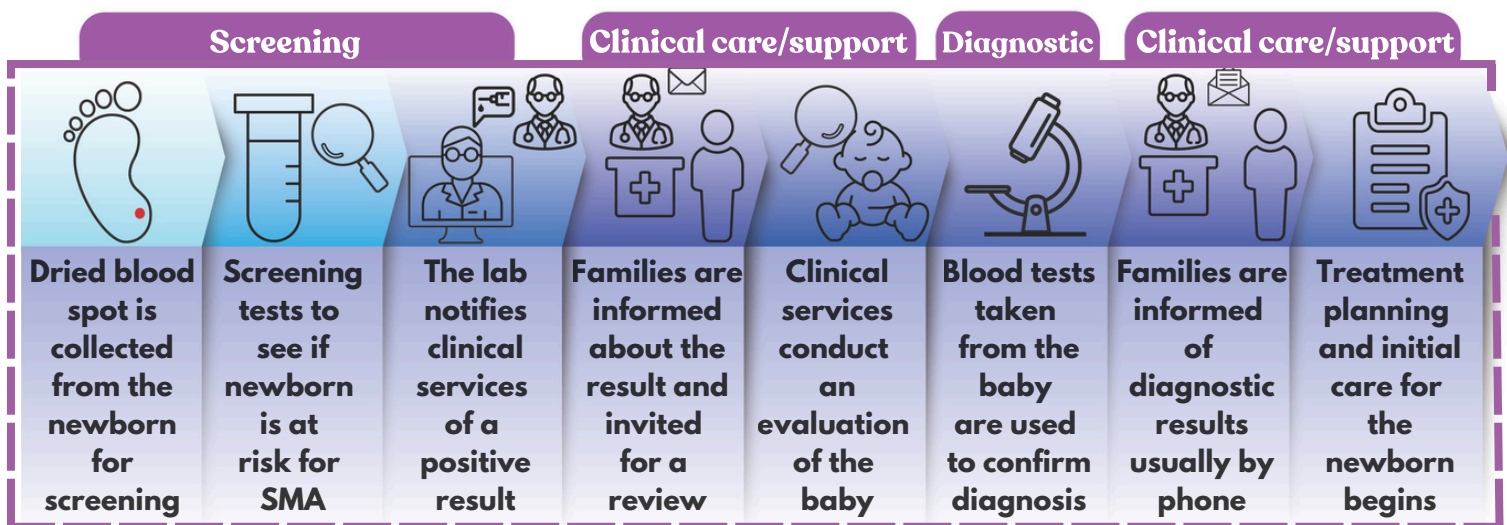
When a positive screen is found, healthcare workers will contact parents. These workers are most often part of a specialist team caring for kids with SMA.

This is a very reliable test. But they still have to double check with a second blood test. This test is often called a confirmatory test or diagnostic test. You will be quickly directed to a centre where this occurs, and where your healthcare team can examine your baby and provide information, care and support.

# I've received a screen positive result for my baby...what does this mean?

## FROM SCREENING TO TREATMENT

This is a broad overview of the major milestones between the moment your child had a blood spot collected after birth, through to the clinical care and support you and your family can expect to receive.



## A LITTLE MORE ABOUT SMA

- Other words to describe spinal muscular atrophy (SMA) include 'neuromuscular disorder', or 'motor neuron disease'.
- It affects the nerves in the spinal cord that control movement. In SMA, muscles shrink or atrophy, giving the name spinal muscular atrophy. This is most commonly evident during childhood.
- Symptoms can vary, and every child with SMA is different. More testing can give more information about how SMA may impact your child. Without treatment, symptoms can get worse and affect voluntary movement, breathing and swallowing. Therefore, newborn screening positive results are really important to allow treatment as early as possible.
- Some information online and via social media may not be up to date and may not apply to you and your child.

# + FROM SCREENING TO DIAGNOSIS

## WHAT TO EXPECT

After a positive screen, you will be contacted by a healthcare professional who specialises in caring for children with spinal muscular atrophy (SMA). You are offered an appointment at a specialist centre with a paediatric neurologist. For families in rural and remote parts of Australia and New Zealand this may be with your local healthcare professional via video chat.

We know it can be hard to act quickly, but it is important. If needed, you can bring a support person to help you.

### At this appointment member/s of your healthcare team will:

- Examine your baby



- Talk to you about SMA



- Organise diagnostic tests



*These include tests to also understand how SMA may impact your baby*

Treatment options can begin to be discussed at this appointment. Sometimes blood tests are also taken to guide the best treatment options.

### At the end of this appointment your healthcare professional team will:

- Give you a contact number
- Arrange your next appointment



It is OK to call while you are waiting for results, if you want to ask questions or have concerns. While waiting for results, if your baby is eligible, please get your baby's Medicare number (for those in Australia). This will help you access treatment once you have a diagnosis.

<https://www.servicesaustralia.gov.au/enrolling-your-baby-medicare?context=60092>



# FROM SCREENING TO DIAGNOSIS

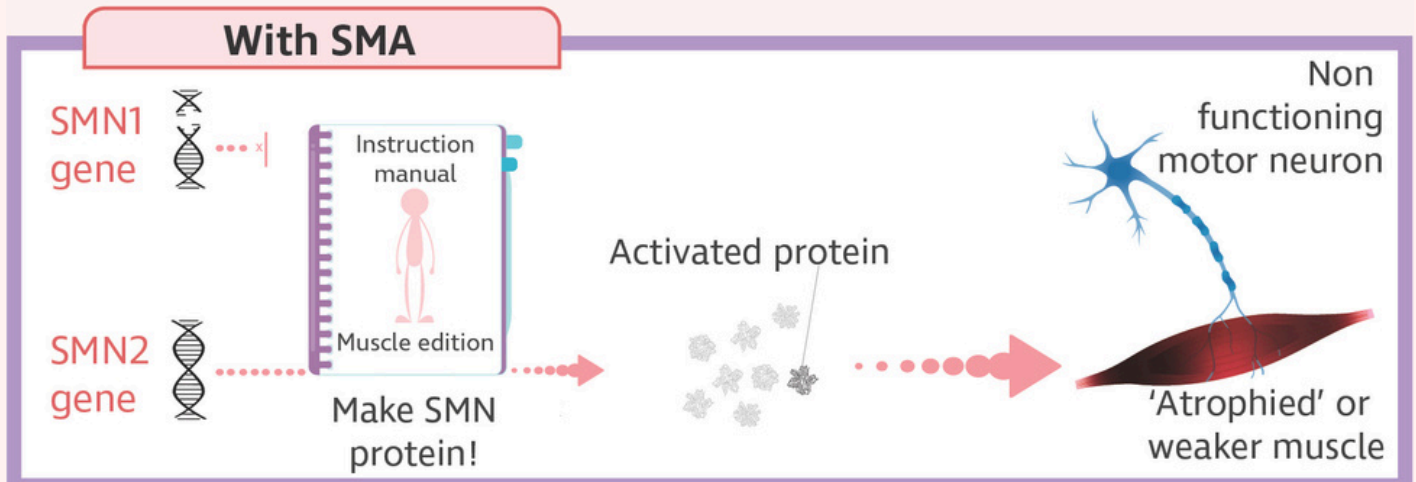
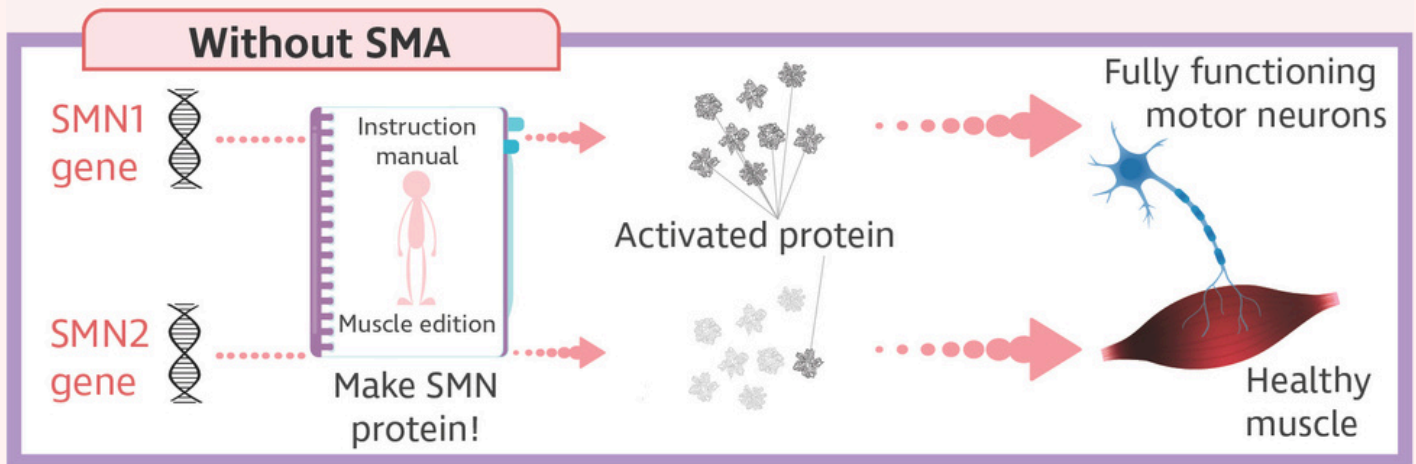
## DIAGNOSTIC TESTING & UNDERSTANDING SMA GENETICS AND INHERITANCE.

Spinal muscular atrophy (SMA) is an inherited genetic condition. Everyone has a gene called the survival motor neuron 1 gene (SMN1). SMN1 is important because it makes a protein called survival motor neuron (SMN).

Genes = Found in every cell and act like the blueprint for our body. They contain instructions for things like what colour our eyes are.

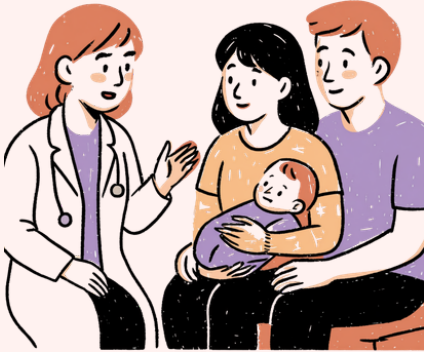
Proteins = If genes are the blueprints, proteins are the bricks and builders. They are crucial to the instructions being carried out.

If we don't make enough of this protein, we lose motor neurons, until they can no longer send messages to muscles to move. Most people have two working copies of the SMN1 gene. SMA occurs when both are not working. In 95% of people with SMA this is because part of the gene is missing, or deleted. This is what newborn screening helps identify. A second blood test is done to confirm the deletion in the SMN1 gene and the diagnosis of SMA.



# + FROM SCREENING TO DIAGNOSIS

## QUESTIONS FROM PARENTS



The health care team will talk about genes and inheritance when they have diagnostic test results for your baby. Sometimes parents ask questions about SMA and their family sooner.

### **Q Why does no one else in the family have SMA if it is genetic?**

**A** Parents of a child with SMA are often “carriers”. This means they each have one fully working survival motor neuron 1 gene, and no symptoms. About 1 in 50 people are carriers of SMA, but often people don’t know it. A DNA test is the only way to find out. If both parents are carriers, there is a 1 in 4 chance that each of their kids can have SMA. A genetic counsellor will help organise your carrier testing after your baby’s genetic tests are available and when you are ready.

### **Q What does this mean if I have older children? Could they also have SMA?**

**A** Your health care team will speak with you about any other children you have. There is a small chance that they could be healthy carriers of SMA, which may have implications when planning a family of their own. There is a smaller chance that a sibling might be affected – your doctor will discuss whether any other children need review by a neurologist or genetic testing.

*As a reminder, you have no control over the genetic information you pass on. No one causes gene variants, and they cannot be prevented. You did not cause this to happen.*

### **Q What are the implications (if any) for my future kids? And my kids kids?**

**A** A genetic counsellor will help you consider these implications and will be able to discuss the risk of having another affected child. There may be implications for your other kids and their kids. A genetic counsellor will help you work out who else in your family might need to be tested.



# FROM SCREENING TO DIAGNOSIS

## QUESTIONS FROM PARENTS

**Q**

**Who else should be tested in my family?**

**A**

Once the results of your “carrier testing” are available, a genetic counsellor will meet with you to work out who else in your family might need testing. This might include siblings of yourself and your partner.

**Q**

**What does autosomal recessive mean?**

**A**

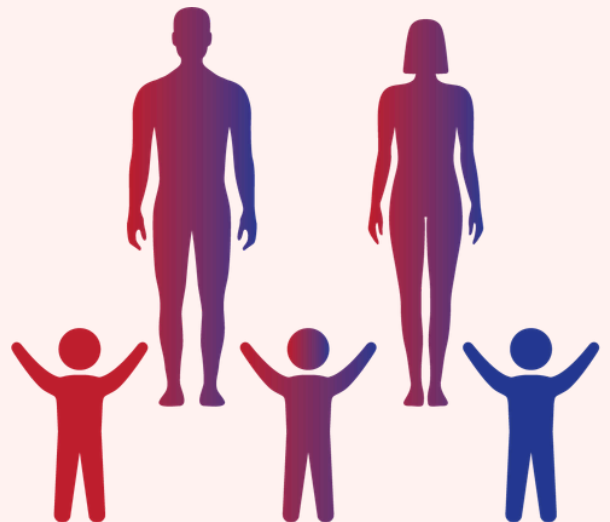
Autosomal recessive is a term used by geneticists to refer to the way in which an inherited disease is passed down within a family. When two parents are unaffected healthy carriers of a genetic condition, they have a 1 in 4 chance of having an affected child. SMA is inherited in this way.

**Q**

**How is SMA inherited?**

**A**

**Both parents are carriers**



—○—  
25% chance  
child won't  
be affected

—○—  
50% chance  
child will be  
a carrier

—○—  
25% chance  
child will  
have SMA

**Q**

**What are the other 5% of cases due to?**

**A**

5% of individuals with SMA have a different genetic change in the survival motor neuron 1 gene. It interferes with the function of the SMN1 gene, but is not detected by the technology used in the newborn screening test.



If you have questions about the specifics of your carrier status your team/ genetics counsellor can provide more information

# + MEDICATIONS & OUTCOMES

There are currently 3 treatments available in Australia and 2 in NZ. Your team will help guide you on which is best for your child. No evidence says one is better or worse. They all work best when given early, have their own risks and benefits, and all are subsidized by the government at minimal cost for families (for almost everyone). The treatments are called:

Spinraza (nusinersen)

Zolgensma (onasmenogene aberparvoec-xioi)

Evrysdi (risdiplam)

Here, we use the names of the treatments most often used in Australia and New Zealand. These treatments differ by the way they work, the way they are given, and how often they are given.

## QUESTIONS FROM PARENTS

**Q How do they work?**

**A** These treatments all work by increasing the level of SMN, or survival motor neuron protein (low levels of this protein cause SMA). All three treatments target the motor neurons in the spinal cord. Some treatments also reach other cells around the body. The long terms benefits and risks of this are unknown.

**Q Which ones work best?**

**A** All three treatments have been shown to work and there is no comparison that shows one is better than the other. Since we know all three treatments work best if given early, your healthcare team will work hard to prevent delays to treatment.


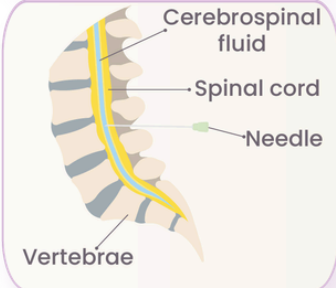

**Q Will they cure my baby?**

**A** None of the treatments are cures. These treatments are known as 'disease modifying therapies'. This means they can be lifesaving and allow progress or stability in movement, breathing and feeding. Outcomes vary and your healthcare team will be able to talk to you.

# + MEDICATIONS & OUTCOMES

## COMPARING TREATMENTS

### Available treatments for SMA

	Evrydsi (Risdiplam)	Nusinersen (Spinraza)	Zolgensma (onasemnogene abeparvovec)
<b>How it works</b>	Increases the amount of SMN made by the SMN2 gene. Risdiplam is a small molecule drug.	Nusinersen is a short strand of genetic material that binds to SMN2 gene and increases the amount of SMN protein that it makes.	Gene replacement therapy. An SMN gene in a virus shell (called AAV-9) is given to the baby. This virus shell does not cause illness.
<b>How it's given</b>			
<b>How often its given</b>	Once a day, every day.	Loading phase: doses on days 1, 15, 29, 64 Maintenance: every 4 months.	One time dose. Can't be repeated as your baby makes antibodies against the treatment, meaning it won't be effective again.
<b>Who can have it</b>	Children and adults with SMA. <b>1</b>	Children and adults with SMA (all ages and types). <b>2</b>	Children with SMA (age and weight limits vary between countries). <b>3</b>
<b>Side effects / risks</b>	Fever, diarrhea, rash, mouth ulcers, joint pains, urinary tract infections. <b>4</b>	Upper and lower respiratory tract infections, constipation, pyrexia, urinary tract infection; thrombocytopenia, coagulation abnormalities. <b>5</b>	Vomiting, fever, elevated transaminases & tropinin I, acute liver injury/failure, thrombocytopenia, thrombotic microangiopathy. <b>6</b>
<b>Other considerations</b>	The liquid form must be stored in the fridge.	Treatment usually occurs in a hospital where staff have done many lumbar punctures on babies before.	Prior to drug: test blood for antibodies (can take time). Corticosteroids and blood tests for 2 months after to manage immune reactions.

SMN = Survival Motor Neuron. SMA = Spinal Muscular Atrophy. AAV=Adeno Associated Vector.

1. <https://www.tga.gov.au/resources/auspmd/evrydsi>

2. <https://www.tga.gov.au/sites/default/files/auspar-nusinersen-heptadecasodium-180813.pdf>

3. <https://www.tga.gov.au/resources/auspmd/zolgensma>

4. [https://medsinfo.com.au/consumer-information/document/Evrydsi\\_CMI](https://medsinfo.com.au/consumer-information/document/Evrydsi_CMI)

4. <https://www.evrydsi.com/safety-and-side-effects/evrydsi-side-effects.html>

5. [https://www.spinraza.com/en\\_us/home/why-spinraza/safety-profile.html](https://www.spinraza.com/en_us/home/why-spinraza/safety-profile.html)

5. [https://medsinfo.com.au/consumer-information/document/Spinraza\\_CMI?drug\\_id=13787&documenttype=cmi](https://medsinfo.com.au/consumer-information/document/Spinraza_CMI?drug_id=13787&documenttype=cmi)

6. [https://medsinfo.com.au/consumer-information/document/Zolgensma\\_CMI?drug\\_id=18167&documenttype=cmi](https://medsinfo.com.au/consumer-information/document/Zolgensma_CMI?drug_id=18167&documenttype=cmi)

6. <https://www.zolgensma.com/safety>

\*\* 4 / 5 / 6 adapted from: Balaji, L., Farrar, M. A., D'Silva, A. M., & Kariyawasam, D. S. (2023).

<https://doi.org/10.1080/14737175.2023.221854>

# + MEDICATIONS & OUTCOMES

## QUESTIONS FROM PARENTS

**Q Do I have to pay for treatment**

**A** In Australia, treatments are covered by the Pharmaceutical Benefits Scheme (PBS). In New Zealand, treatments are covered by the PHARMAC. This means that if your baby is eligible, you will not need to pay.

**Q Can all babies access treatment**

**A** As a general rule, access to treatments in Australia is currently for:

- Newborns and children who do not have signs and symptoms of spinal muscular atrophy (SMA) and have  $\leq 3$  copies of the SMN2 (survival motor neuron 2) gene.
- All newborns and children with signs and symptoms of SMA.

Access to treatments in New Zealand is currently for:

- Newborns and children who do not have signs and symptoms of SMA and have  $\leq 3$  copies of the SMN2 gene.
- All newborns and children with signs and symptoms of SMA, provided they have onset of symptoms less than 4 years of age.

There are some limits in access to the treatments. You can find out more about those by clicking 'here', or by asking your healthcare team.

**Q Are there more tests needed before treatment**

**A** Your healthcare team will talk to you about tests that they may want to do before they treat your baby. These can include blood tests to make sure their blood count, kidney and liver tests are normal. They may want to make sure the kidneys are healthy by taking a urine sample for testing. More tests might be needed based on which treatment you choose.

**Q My baby has 4 SMN2 copies. Can they be treated**

**A** .....

# + MEDICATIONS & OUTCOMES

## OUTCOMES

Your healthcare team will talk with you about expectations for your baby. For children with 2 copies of SMN2 that are NOT treated, muscle weakness and floppiness appear before the age of 6 months. Children lose strength and do not learn to sit or walk and only 10% live until the age of 2 years;

Babies treated quickly and with no symptoms can have development that is similar to children without SMA or slight motor delay. Follow up for about 8 years shows that children may have differences in walking for long distances, running and climbing stairs

### How can I help my baby have the best outcomes?

We know that children who are treated quickly have better outcomes, and your healthcare team will work quickly so that your baby can be treated without delay. We also know that babies who have the care of a team of people (multidisciplinary team) both in their local communities and in specialist teams have better outcomes. We want babies to thrive and that means they need the care of a whole team around them



“We wanted to have treatment, take on all of the information and **get as many opinions as possible**, but at the same time we knew that we **had to move quickly**. Talk to your healthcare team so that you can understand what is going on and all of your options. In the end you have to do what feels right and is the best decision for your child. We made the best call with the information we had available and that is what you can do”

# + YOUR HEALTHCARE TEAM

## WHO IS INVOLVED IN CARING FOR YOUR CHILD

While every child with spinal muscular atrophy (SMA) is different, there are a number of healthcare professionals that may be involved at different times. Your healthcare team will refer your child to the right professional when they are needed. This care, treatment and support is based on your child's age, abilities and individual needs to help them manage daily activities. Below are more details on health professionals you might speak with such as **General practitioner, Geneticist/Genetics counsellor, Neurologist (Paediatric), Nutritionist/Dietician, Occupational therapist, Paediatrician, Respiratory physician, Speech therapist, Physiotherapist, Social worker, and Nurse specialist.**

## WHY DO I NEED SO MANY PEOPLE

We know that babies who have the care of a team of people (multidisciplinary team) both in their local communities and in specialist teams have better outcomes. We want babies to thrive and that means they need the care of a whole team around them. Because our muscles impact so many different parts of our body, we need the specialists for all those different parts to give your child the best chance at a healthy future.

## MEET THE TEAM



### Nurse specialist

They are an essential part of your team who manages care with the wider team and will co-ordinate treatments, provide support and education to help simplify your hospital visits.

### General practitioner

Your GP is a very important and will monitor your child's health and treat usual childhood illnesses (e.g. fever, ear infections) and give immunisations. They will also look after your health and wellbeing. They communicate with your specialists who know more about SMA.



# + YOUR HEALTHCARE TEAM

## Geneticist / Genetic counsellor

They help you understand genetics and inheritance, assist in reproductive decision making that is right for you and can help share information about SMA with other family members who want to know if they are carriers.



## Neurologist (Paediatric)

A paediatric neurologist is a doctor who specialises in children's medicine, and then even more specifically they specialise in neurological conditions and the brain. This is particularly important in SMA because your child's nerves are impacted by the condition. They will have a role in diagnosis, treatment, and ongoing management.

## Nutritionist / Dietician

Nutritionists/dieticians help your child get the right nutrition to maintain a healthy weight, muscle mass and overall health.



## Occupational therapist

An occupational therapist helps your child to perform daily activities without help from others. They may use special devices and equipment.

## Paediatrician

This person specialises in children's medicine. They will likely meet with you and your child when you come to a hospital for consultations. They can also help working out what specialists you will need and when.





# YOUR

# HEALTHCARE TEAM

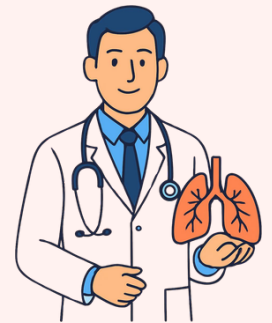


## Physiotherapist

Physical therapy helps to improve and maintain your child's muscle strength, flexibility and range of motion through exercise.

## Geneticist / Genetic counsellor

They assess breathing and coughing abilities. They help with keeping the airways clear, performing breathing exercises, and supporting breathing.



## Social and emotional support

Helps to support the mental health of you and your child. This can include counselling, support groups, psychologists and more.

## Speech therapist

A speech therapist helps with speech and swallowing.



# + FOR FAMILY & FRIENDS



This information is for you to share with family and friends to help explain spinal muscular atrophy (SMA). Parents of children with SMA have provided suggestions for your family and friends on how they can support you through these early moments of diagnosis.

## WHAT SHOULD I KNOW? WHAT SHOULD I DO?

Someone you care about has just been told that their baby has, or is very likely to have, SMA. This news is usually unexpected and can be frightening. SMA is a genetic condition that affects the nerves that help muscles move. Many babies who screen positive for SMA look completely well at first. In Australia and New Zealand, newborn screening and new treatments mean that SMA can often be treated before symptoms appear.

## WHAT THIS DOES AND DOES NOT MEAN

- SMA is serious, but there are now treatments that were not available in the past.
- Information you find online may be old, or may describe children who did not receive early treatment.
- No one caused SMA by anything they did or did not do during pregnancy or birth. It is nobody's fault.

## HOW YOU CAN BE SUPPORTIVE

### Things to Try

- Listen first. Parents may need to talk, or they may need quiet. Follow their lead.



- Ask what they need: help with childcare for siblings, meals, transport to appointments, or company.

- Respect their choices about treatment and about who they tell, and when.

# + FOR FAMILY & FRIENDS



## Things to Avoid

- Avoid sending links from random internet searches, especially if they are frightening or out of date.
- Avoid saying “I know how you feel” unless you have been through something very similar – instead try, “I can’t imagine how hard this is, but I’m here for you.”
- Avoid making promises such as “everything will be fine” or, on the other extreme, focusing only on worst-case scenarios.
- Try not to ask for detailed explanations when parents are exhausted; you can offer to read the information they share with you instead.

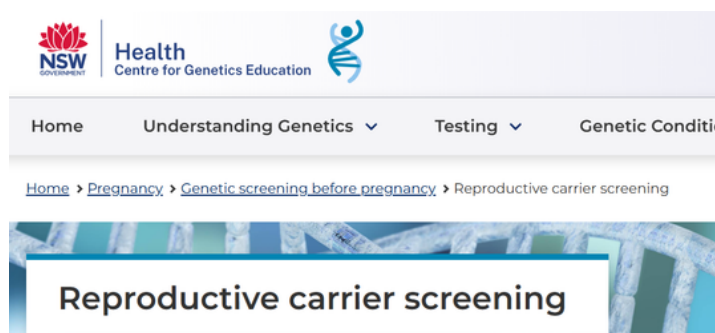
Knowing they are not alone helps parents. Being present, willing to listen and offering practical help can make a real difference.

## GENETIC CARRIER SCREENING FOR FAMILY

SMA is usually inherited. Parents of a child with SMA are often “carriers”, which means they each carry one copy of the SMA gene change without having symptoms themselves. Relatives may also be carriers without knowing it. In Australia, carrier screening for conditions like SMA is increasingly available. If you are a close relative and are thinking about having children in the future, you may wish to talk to your GP or a genetic counsellor about whether carrier screening is right for you.

For a more detailed explanation of reproductive carrier screening, please follow the below link to the centre for genetics education, hosted by the NSW government.

<https://www.genetics.edu.au/SitePages/Reproductive-carrier-screening.aspx>



# + FOR FAMILY & FRIENDS



## A FREQUENTLY ASKED QUESTION FROM PARENTS IS “WHY NOW, WE HAD ALL THE TESTS DONE DURING PREGNANCY?”

During pregnancy a number of tests are carried out routinely, e.g. nuchal translucency, NIPT (non invasive prenatal test). These test for some important inherited conditions, like Down Syndrome or other chromosomal differences, but not all genes (e.g. SMA).

Reproductive carrier screening is available (before or in early pregnancy) to find out if you are at risk of having a baby with SMA. This involves testing both parents and based on results, their risk can be high or low. For couples with a high risk of having a baby with SMA, this information can help them make reproductive decisions based on their preferences.



Not everyone has carrier screening for SMA and rarely, people with a low risk can be carriers. These are additional reasons why it is important to screen newborns for SMA.




# + SUPPORT & INFORMATION

## Some take home messages

As a parent or caregiver, how can I take care of myself?

The most important thing to remember is that you are not alone.

- Reach out to your current support system, such as your family and friends.
- While spinal muscular atrophy is 'rare', there is a community (locally and globally) that can connect you with resources and support.
- Try to take care of yourself. At times it can be easy to overlook (or hard to manage) but your sleep, fitness, and diet, can help fuel the physical and emotional energy you need to take care of your baby. This is of course, easier said than done even at the best of times.
- Prioritize your mental health. Supports are also available for you and it is OK to ask for help. Reach out to a professional counsellor who is trained to help.
- Receiving a SMA diagnosis in a newborn can be especially difficult. Parents may find this news difficult to cope with. There are organizations that provide support to postpartum parents:

	<a href="https://lifeline.org.au">lifeline.org.au</a>	Crisis support hotline (AUS)
	<a href="https://curesma.org">curesma.org</a>	Advocacy group for people living with neuromuscular conditions (U.S)
	<a href="https://mdnsw.org.au">mdnsw.org.au</a>	Advocacy group for people living with muscular dystrophy (AUS)
	<a href="https://smaaustralia.org.au">smaaustralia.org.au</a>	Advocacy group for people living with spinal muscular atrophy (AUS)
	<a href="https://smauk.org.uk">smauk.org.uk</a>	Advocacy group for people living with spinal muscular atrophy (U.K)
	<a href="https://australianmdregistry.org.au">australianmdregistry.org.au</a>	Registry for people with a neuromuscular disease (AUS)
	<a href="https://beyondblue.org.au">beyondblue.org.au</a>	Organisation supporting mental health (AUS)
	<a href="https://rarevoices.org.au">rarevoices.org.au</a>	National peak body for Australians living with a rare disease
	<a href="https://servicesaustralia.gov.au">servicesaustralia.gov.au</a>	Australian government website for accessing medicare/PBS services