Spinal muscular atrophy

FOR FAMILIES RECEIVING A POSITIVE NEWBORN SCREENING RESULT



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Acknowledgements

This work was produced by the Child Neurology team at UNSW Sydney, in collaboration with a multidisciplinary group of healthcare professionals and parents of children with spinal muscular atrophy (SMA) from across Australia and New Zealand.

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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 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 SMA 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., with many children living full and wonderful lives with these treatments.

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 resource for you.

What these resources cover

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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 realised 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."

-Reggies 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 heel prick. This was sent to a lab where all the conditions on the newborn screening panel were tested. SMA has been on the national newborn screening 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 effective choices can be made for your baby quickly.

AFTER THE SCREENING RESULT

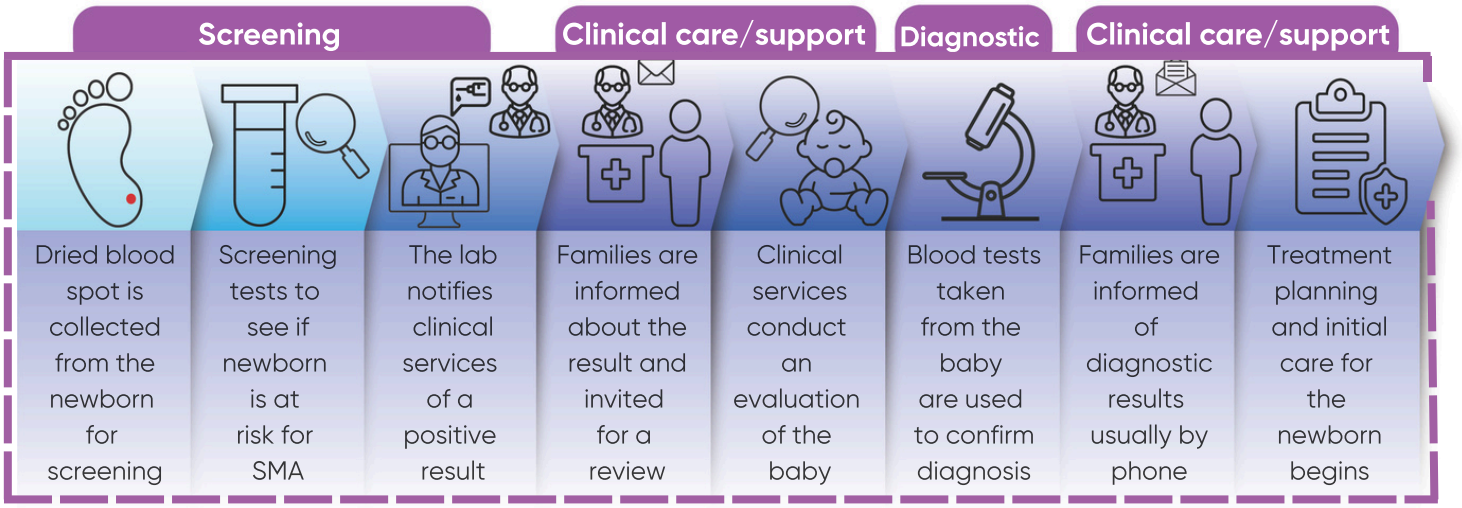
When a positive screen is found, healthcare workers will contact parents. These workers are most often part of a specialist team caring for kids with SMA.

This is a very reliable test. But they still 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

Below is a broad overview of the major steps involving healthcare services, between the moment your child had a blood spot collected after birth, through to the moments after a diagnosis has been confirmed.



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

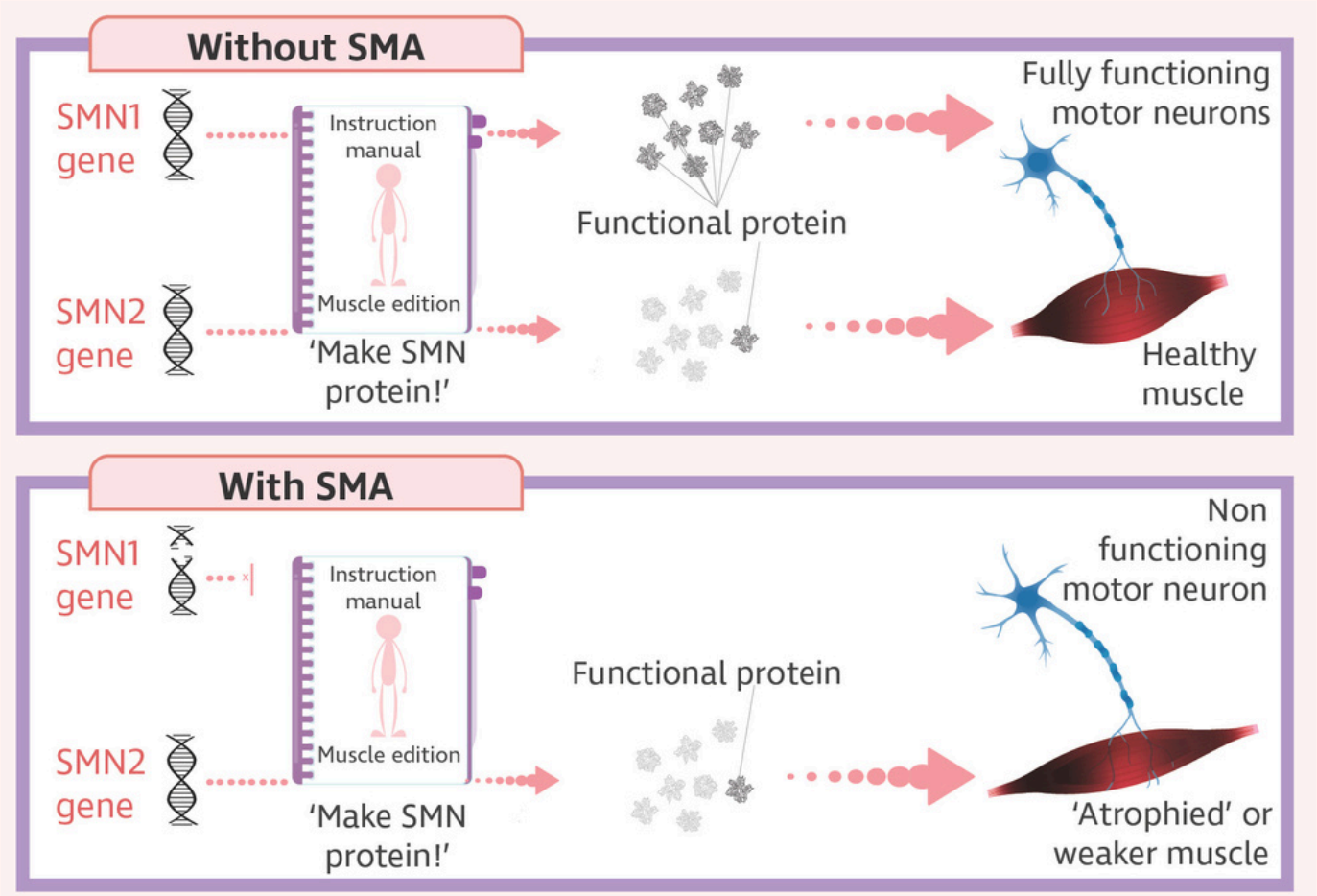
DIAGNOSTIC TESTING & UNDERSTANDING SMA GENETICS AND INHERITANCE.

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. 95% of SMA cases are due to part of the gene being missing, or deleted. This is what newborn screening identifies. 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

Your healthcare team will talk to you about genes and inheritance when they have diagnostic test results for your baby. Sometimes parents ask questions about SMA and their family sooner. We've collected some of the more common questions.

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

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.

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

Your health care team will speak with you about any other children you have. There is a chance that they could be carriers of SMA, which may have implications for the future 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.

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

There may be implications for children you may want to have in the future and grandchildren as well. A genetic counsellor will help you consider these implications and will be able to discuss the risk of having another child with SMA.

FROM SCREENING TO DIAGNOSIS

QUESTIONS FROM PARENTS



Who else should be tested in my family?

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 your sibling(s) and your partner's sibling(s), which can then indicate whether their children also need to be tested.

What does autosomal recessive mean?

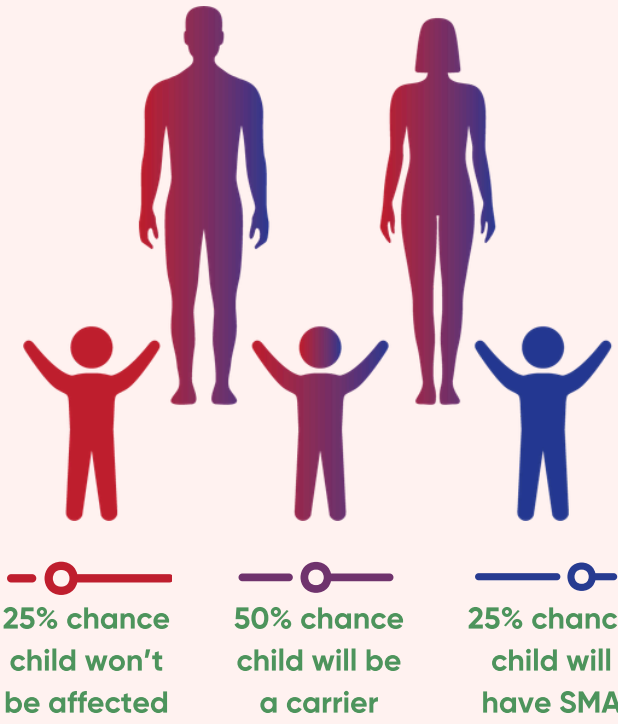
Autosomal recessive is a term used by geneticists to refer to the way in which an inherited disease is passed down within a family. When two parents are unaffected healthy carriers of a genetic condition, they have a 1 in 4 chance of having an affected child. SMA is inherited in this way.

What are the other 5% of cases due to?

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.

How is SMA inherited?

Both parents are carriers



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 New Zealand. Your team will help guide you on which to give your child based on the suitability of each option for your child, eligibility rules, and your choice. There is a set of eligibility rules for government subsidisation, with minimal cost for families (for almost everyone).

No evidence says one is better or worse. They all work best when given early and have their own risks and benefits.

While *SMN1* (Survival Motor Neuron 1) is the missing gene that causes SMA, a backup gene produces small amounts of functional SMN protein. The number of *SMN2* genes (or copies) influences SMA severity. Counting the number of *SMN2* genes is part of diagnostic testing. For people with SMA and 3 or fewer copies of the *SMN2* gene, treatments can be started before there are any symptoms of SMA. 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. You can find a table comparing the treatments on the following page.

Q | Will they cure my baby?

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.

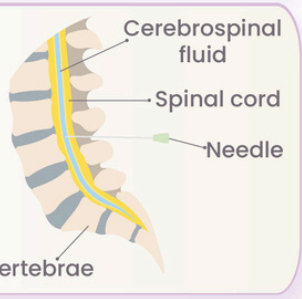
Q | How do they work?

These treatments all work by increasing the level of SMN (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.

MEDICATIONS & OUTCOMES

COMPARING TREATMENTS

Available treatments for SMA

	Evrysdi (Risdiplam)	Spinraza (Nusinersen)	Zolgensma (onasemnogene abeparvovec)
How it works	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.
How it's given			
How often its given	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.
Who can have it	Children and adults with SMA. (1)	Children and adults with SMA (all ages and types). (2)	Children with SMA (age and weight limits vary between countries). (3)
Side effects / risks	Fever, diarrhea, rash, mouth ulcers, joint pains, urinary tract infections. (4)	Upper and lower respiratory tract infections, constipation, pyrexia, urinary tract infection; thrombocytopenia, coagulation abnormalities. (5)	Vomiting, fever, elevated transaminases & tropinin I, acute liver injury/failure, thrombocytopenia, thrombotic microangiopathy. (6)
Other considerations	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/evrysdi
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/Evrysdi_CMI
4. https://www.evrysdi.com/safety-and-side-effects/evrysdi-side-effects.html
5. 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
6. 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 AND OUTCOMES



QUESTIONS FROM PARENTS

Which ones work best?

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.

Do I have to pay for treatment?

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

Can all babies access treatment?

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 ≤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 ≤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 whether that applies to your child by asking your healthcare team. The most common reason to limited access is due to a child having at least 4 copies of *SMN2*. You can find more information about this on page 13.

Are there more tests needed before treatment?

Your healthcare team will talk to you about tests 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.

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"

FOUR COPIES OF SURVIVAL MOTOR NEURON 2

WHY IS FOUR COPIES DIFFERENT?

4 *SMN2* copies generally means a milder form of SMA, with symptoms often developing later. But there is variability and it can't give us exact predictions for each child.

INTERNATIONAL CONSENSUS FOR TREATING FOUR COPIES?



Because of this variability, there is currently no international consistency about when to treat presymptomatic infants with 4 *SMN2* copies. Some recommend/reimburse treatment straight after diagnosis, while others recommend careful monitoring and starting treatment once disease activity is seen. These differences reflect knowledge gaps about the optimal timing of treatment in this group, rather than disagreement about the value of treatment itself.

AUS/NZ RECOMMENDATIONS?

The approach in Australia and New Zealand is to monitor infants with 4 *SMN2* copies identified through newborn screening. Your child will be reviewed regularly by a specialist neuromuscular team experienced in recognising the earliest signs of SMA. This can include assessments of motor development, physical examination, and other investigations as required. This allows us to find the very earliest signs of muscle weakness or changes in motor development so that treatment can be started promptly.

For a detailed breakdown of the international evidence, and the Australian recommendations.



www.unsw.to/nbs-sma

TAKE HOME MESSAGE

While it is natural to feel uncertain following this diagnosis, it is reassuring that many children with 4 *SMN2* copies remain well for a prolonged period. Your neuromuscular team will work closely with you, provide ongoing support, and ensure management decisions are guided by the best available evidence and your child's individual progress.

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.

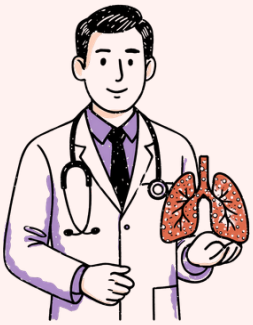
YOUR HEALTHCARE TEAM



Physiotherapist

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

Respiratory physician



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 broadly speaking helps with speech, swallowing, and breath control.

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

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

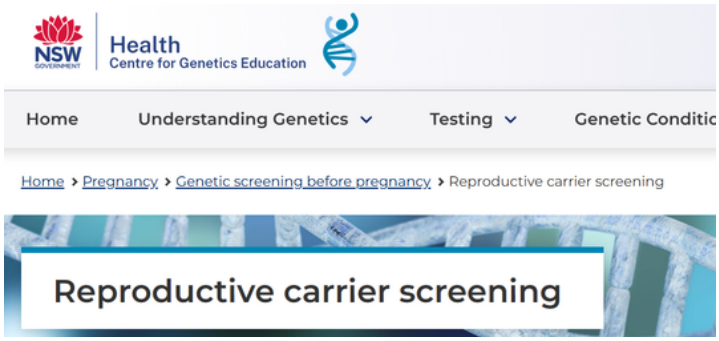
- Avoid sending links from random internet searches, especially if they are frightening or out of date.
- Avoid saying "I know how you feel" unless you have been through something very similar – instead try, "I can't imagine how hard this is, but I'm here for you."
- Avoid making promises such as "everything 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.

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 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 have a low or high risk of having a baby with SMA. This involves testing both parents. For couples with a high risk of having a baby with SMA, this information can help them make reproductive decisions based on their preferences.

Carrier screening is complementary to newborn screening, but is not a replacement.

SUPPORT & INFORMATION

As a parent or caregiver, how can you take care of yourself?

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.



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 for a newborn can be especially difficult. Parents may find this news challenging to cope with. There are organisations that provide support to postpartum parents. Some of these are listed here:

- https://www.cope.org.au/new-parents/getting-help-for-new-parents
- https://panda.org.au/get-support/support-new-parents
- https://forwhenhelpline.org.au/

Between juggling the typical challenges of a newborn and the unexpected challenges of spinal muscular atrophy, it can be easy to forget to enjoy the precious moments with your baby.
Try and be present as a family when the opportunity allows.

SUPPORT & INFORMATION

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

Above are some additional support and information services you may wish to approach. There is no right or wrong way to go about this. Some parents prefer to reach out immediately after receiving a diagnosis, some find them more useful years later, and some may prefer to rely on themselves or their friends and family. Facebook groups are often another source that parents bring up when asked what has been useful for them.

DEFINITIONS

Autosomal recessive
a pattern of genetic inheritance, where both parents pass on a particular gene variant that causes a medical condition. In SMA, this is the *SMN1* gene.

Carriers
someone who carries one working and one non-working copy of a gene. The person is healthy but can pass on the non-working gene to their children.

Carrier screening
a test parents can get to see if the have a low or high risk of their child having a specific condition.

Confirmatory test (diagnostic test)
a test to confirm a diagnosis following a positive newborn screening result, or clinical diagnosis.

Copy number / SMN1 / SMN2
in SMA, copy number refers to the number of SMN (Survival Motor Neuron) genes a person has, specifically the *SMN1* gene (used to tell us if a person has SMA) or *SMN2* gene (used to tell us how severe SMA may be).

- Two copies of *SMN1* means a person does not have SMA.
- One copy of *SMN1* means a person is a carrier for SMA but does not have symptoms.
- Zero copies of *SMN1* (often called a homozygous deletion of *SMN1*) means a person has SMA.

The copy number of *SMN2* varies and is used to predict severity. The fewer copies, the more severe the SMA may be.
The newborn screening result will tell us about the *SMN1* copy number, while the diagnostic tests will tell us about the *SMN2* copy number.

Disease modifying therapies
this means the treatment can change what the disease will do at a biological level, rather than treat what the disease has done.

DNA (Deoxyribonucleic Acid)
contains instructions (like a manual) that informs how your body is built and operated. This determines everything from the colour of your eyes to how your muscles work.

Dried blood spot
a specimen collected for lab testing, using an approved medical device (specified filter paper) and air-dried for transport or storage.

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

Genetic counselling
professional who helps people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.

Newborn screening program
a simple test that identifies babies at risk of becoming seriously ill from a rare condition. Screening aims to improve the health of these babies by allowing early intervention.

Motor neuron
nerve cells in the brain and spinal cord that determine muscle activity. In SMA, the loss of motor neurons means there is less and less of a signal between the brain, spine, and muscles, which leads to muscle weakness and shrinking (or 'atrophy').

Neuromuscular / Neurological
the term 'neuromuscular' concerns the interaction of nervous systems and muscles. 'Neurological' can be thought of as being a bit broader, and covers anything related to the nervous system.

Protein
if genes are the blueprints, proteins are the bricks and builders. They are crucial to the instructions being carried out.

Screening-diagnostic-clinical care
screening concerns the initial test your baby has shortly after birth. Diagnostics are specialised tests to confirm a positive screening result. Clinical care is post-diagnosis, when healthcare specialists can engage with a patient and consider the best management pathways.

The glossary of terms is partly derived from The Clinical and Laboratory Standards Institute (CLSI) Harmonized Terminology Database (updated 2023).