CONDITION GUIDE

# Spinal Muscular Atrophy (SMA)

Spinal Muscular Atrophy (SMA) is a neuromuscular condition that can affect infants, children and adults. It affects nerve cells called motor neurons that are found in the spinal cord and send messages from your brain to your muscles. In SMA the motor neurons do not work properly, meaning your muscles don't receive the messages from your brain. This causes muscles to gradually become weak and waste away over time (atrophy). It is a genetic condition, meaning it is inherited through genes from your parents, and affects both males and females.

# Processing a new diagnosis

A diagnosis of SMA can be a huge shock for yourself, parents, siblings, extended family members and friends. It is normal to feel an overwhelming mix of grief, confusion, anxiety, loneliness and helplessness as your life has been forever changed. Everyone will have different ways of getting through this time but it's important to know that help and support are available for you, your child and family. Hear advice on processing your diagnosis from a community member with shared experience.

The video is available at https://youtu.be/A9XM71knDVw.

## Getting help processing a new diagnosis

There are many ways you can seek support as you process a new diagnosis:

- A psychologist is a university-qualified health professional who can help you talk about your thoughts and feelings to understand and cope with the challenges you and your family are facing. Visit our page on Psychology for more information.
- A counsellor is a trained professional who can help you talk about and work through problems. Visit our page on Counselling for more information.
- A social worker can provide information and support to people experiencing a range of issues including family problems, anxiety, depression, crisis and trauma. Visit our page on Social Work for more information.
- A genetic counsellor can help you understand how SMA is inherited and whether there are any implications for other members of your family. Your specialist will be able to advise whether a genetic counsellor is required. Visit our page on Genetic Counselling for more information.
- Your state or territory neuromuscular organisation can provide support, advice and information about living with SMA. Visit our page on our state and territory neuromuscular organisations for more information.

- Your GP can talk to you about a mental health plan and how you can use this to help you access the support and services you need. Visit our page on Wellbeing for more information.
- Other people and families living with SMA have also experienced the emotional rollercoaster that comes with a diagnosis of SMA and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with SMA in The Loop Community on our Forum.

# Understanding SMA and How it's Diagnosed

## About SMA

Spinal Muscular Atrophy (SMA) is a neuromuscular condition that can affect infants, children and adults. It affects nerve cells called motor neurons that are found in the spinal cord and send messages from your brain to your muscles. In SMA the motor neurons do not work properly, meaning your muscles don't receive the messages from your brain. This causes muscles to gradually become weak and waste away over time (atrophy). It is a genetic condition, meaning it is inherited through genes from your parents, and affects both males and females.

SMA affects muscles throughout the body, including muscles that control movement of your arms and legs, the muscles for feeding and swallowing, and the muscles involved in breathing and coughing. In recent years there have been promising treatments developing for SMA.

## How to talk to your child or others about a new diagnosis

Having a conversation with a loved one about a diagnosis of SMA, is not easy, in fact it will probably be one of the hardest talks you will have. But, like all difficult conversations it is important and necessary.

There are many reasons why people may not want to have this conversation, mostly centred on wanting to protect their loved ones and themselves.

But avoiding the topic is not helpful. People may have noticed differences and changes that have occurred and/or heard the condition being discussed.

So it is crucial that you have a role in your loved one learning about their/your diagnosis. This will enable you to support them as they process the information and to be on hand to answer any questions they may have. If you are the person with SMA it will provide you with support and someone to talk to. Together, you can learn about the journey ahead.

It will also allow you to:

- Provide answers to questions in an age appropriate way

- Correct any misinformation they have heard or read
- Show them that you have faith in their ability to handle difficult conversations

There is no right time to start this conversation. Research suggests the earlier you talk to your loved ones about your/their condition, the more natural the conversation will become. The important part is to pick a time and commit to it.

Some important do's and don'ts:

- Do be positive but also realistic.
- Do reassure them that no one has done anything wrong and this is not their fault.
- Do tell them they/you will do many wonderful things in their/your life – they/you may just do them differently.
- Do tell the truth. Answer all their questions.
- Do consider your language. Try to avoid negative or emotive words, such as 'suffering from a condition'.
- Do use daily living examples that they can relate to.
- Don't avoid answering questions and don't shut the conversation down.

If you are having difficulty starting the conversation, just ask a couple of questions such as 'How are you feeling today?' or 'Do you know why you/they are having difficulty with your/their muscles?'

Remember it's okay to be upset during the conversation but try to avoid breaking down as this will only cause greater distress. And if speaking with a child, remember children are resilient and they generally handle information of this nature far better than adults do.

## What causes SMA?

The most common form of SMA is caused by a change (also called a mutation) in a gene (the parts of a cell that tell the cell what to do). The gene involved in SMA is called survival motor neuron 1 (SMN1 gene). This gene usually produces a protein called survival of motor neuron (SMN). This protein is needed to maintain healthy functional motor neurons (nerve cells in the spinal cord that send signals to muscles throughout the body and control their movement). The genetic mutation in SMA means the entire gene is missing or some of the code of the gene is changed, making it inactive. If enough SMN protein is not made, motor neurons start to break down and can't send messages to the muscles to make them move. Over time, the muscles become weaker and smaller due to inactivity.

The reason that some people are affected much more severely by the SMN1 gene change than others is mainly due to the presence of another gene called SMN2.

Both SMN1 and SMN2 are located on chromosome 5. SMN2 also produces a small amount of SMN protein and can help compensate for the faulty SMN1 gene. People can have multiple copies of this SMN2 gene. Having more than

three copies of the SMN2 genes can result in enough SMN protein being produced, reducing the severity of the disease. As a general rule:

- People with the most severe type of SMA (type 1) have one or two copies of SMN2
- Most people with SMA type 2 have three copies of SMN2
- People with least severe types of SMA (types 3 and 4) have four or more copies of SMN2.

There are other forms of SMA that are caused by mutations in in different genes. These other types of SMA vary greatly in their symptoms and severity.

## What are the types of SMA?

There are several types of SMA, each with differences in age of onset, symptoms, and how quickly it progresses:

**SMA linked to the SMN1 gene**

There are 4 types of SMA that all result from mutations to the SMN1 gene, causing a deficiency of the SMN protein:

- Type 1 SMA – This is the most severe form of SMA, with symptoms present at birth or before the age of 6 months. Babies with this type of SMA never learn to sit independently and often have difficulty swallowing and sucking.
- Type 2 SMA – Symptoms of this type of SMA present between 6 and 18 months, before the child can stand or walk. These infants typically gain the ability to sit but not to stand.
- Type 3 SMA – Symptoms appear after the age of 18 months into the teenage years. Most children with this type of SMA can walk but may develop problems with balance and walking as they get older.
- Type 4 SMA – This type of SMA causes symptoms in adulthood adults, usually after the age of 30.

**SMA not linked to the SMN1 gene**

Other forms of SMA caused by genes other than SMN1 include:

- SMA with respiratory distress (SMARD) - In this severe form of SMA, infants have respiratory distress (breathing difficulties) in addition to muscle weakness.
- Distal SMA - This form of SMA affects the hand and feet muscles, with varying symptoms and severity depending on the gene involved.

## How is SMA diagnosed?

If SMA is suspected a doctor will perform a physical examination and gather information about symptoms. A blood test (genetic testing) will check for the presence or absence of the SMN1 gene to help diagnosis. The person will need

to be assessed by a Neurologist (a doctor specialising in muscle conditions). If genetic testing confirms the diagnosis then treatment options can be discussed. Further information about genetic testing can be found on the **Healthdirect website**.

# The genetics of SMA

Everybody has two copies of the SMN1 gene – one inherited from each parent. People with SMA most commonly have a change or mutation in both copies of the SMN1 gene that they have inherited. This is what is called an 'autosomal recessive' inheritance. The parents of a person with SMA each carry one copy of the faulty SMN1 gene and are known as ‘carriers’. They do not show signs and symptoms of the condition. Often a family has no idea that some members are carriers until a child is born with SMA. To have a child affected by SMA, both parents must pass on a faulty SMN1 gene to their child. If both parents are carriers, the likelihood of a child inheriting the condition is 25 per cent, or one in four pregnancies. In Australia about one in every 35 people is a carrier of the faulty gene that causes SMA.

It is possible to have a test to see if you are a carrier of SMA. If you are a carrier you won’t have any signs or symptoms of SMA. However if your partner is also a carrier there is a 1 in 4 chance that you could have a child with SMA.

In rare cases, SMA can be inherited from a faulty gene from just one parent.

## Genetic counselling

If you or your child have been diagnosed with SMA, or if it runs in your family, it may be helpful to speak to a genetic counsellor. Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand SMA and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.If SMA runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

Visit our page on Genetic Counselling for more information.

# Long-term outlook

Due to medical and scientific advances, quality of life is improving for people with SMA and life expectancy is increasing.

Living with SMA usually means life, and the future, looks different from what you had planned. However many children and adults with SMA can lead fulfilling, rewarding lives just like everyone else. School, friendships, sports and recreation, arts, university, work, having a family and travel are all possible for many people with SMA although some may need significant medical support.

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# Understanding and Planning for Changes

Learning about SMA and surrounding yourself with the right healthcare providers, services and support can help you feel more in control of what lies ahead. Although every person's journey with SMA will be unique, the information below will help you understand this journey and how to prepare and plan for changes in the future.

### What are the main symptoms of SMA?

In infants with Type 1 SMA, symptoms are generalised muscle weakness, a weak cry and trouble breathing, swallowing and sucking.

Symptoms of Type 2 SMA include muscle weakness in arms, legs and lower torso, weak respiratory (breathing) muscles and spinal curvature (scoliosis). This curvature often requires bracing and surgery.

Children with Type 3 SMA show weakness in the leg, hip, shoulder, arm and breathing (respiratory) muscles.

People with Type 4 SMA have symptoms including mild muscle weakness, tremor and twitching.

# Community Advice

Hear from a community member who has walked the path before you.

The video is available at https://youtu.be/_6oJ9Mc0UO8.

# Life Stuff

To find out more about living life with a neuromuscular condition and to access stories and peer-advice from the community, visit our Living Life section.

### Where to find more information about living with SMA

**Spinal Muscular Atrophy Australia Inc** An organisation whose mission is to “Improve care for all Australians living with SMA, so that the community have access to all possible research, diagnosis and treatment options.”

**SMA fact sheet** From Spinal Muscular Atrophy Australia Inc, 2017. This fact sheet contains facts about SMA.

**SMA Type 1 fact sheet** From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

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**What is Spinal Muscular Atrophy A guide for parents and families** From Muscular Dystrophy Association, United States, February 2019. This guide includes what are the signs and symptoms of SMA?, what should I know about SMA?, how is SMA treated? and a glossary of terms.

**A Guide to the 2017 International Standards of Care for SMA**The guide was developed by Spinal Muscular Atrophy UK, Cure SMA, SMA Europe, Muscular Dystrophy UK and TREAT-NMD Alliance in 2019 and outlines the 2017 International Standards of Care Recommendations for SMA.

**Standards of care recommendations for SMA**

Below are links to the more detailed standards of care recommendations for SMA on which A Guide to the 2017 International Standards of Care for SMA is based. Although these are written in medical terms they may be useful for discussions with specialists, doctors, nurses and allied health professionals

**Part 1: Diagnosis and Management of SMA; Recommendations for diagnosis; rehabilitation; orthopaedic and nutritional care.**

**Part 2: Diagnosis and Management of SMA; Pulmonary and acute care; medication; supplements and immunisations; other organ systems; and ethics.**

**International organisations**

**Cure SMA**Cure SMA leads the way to a world without spinal muscular atrophy, the number one genetic cause of death for infants. We fund and direct comprehensive research that drives breakthroughs in treatment and care, and we provide families the support they need for today.

**Spinal Muscular Atrophy UK**If you or someone in your family has SMA, and you live in the UK, you have come to the right place for support and information about the condition and the latest developments with new treatments.

**SMA Europe**SMA Europe is an umbrella organisation, founded in 2006, which includes Spinal Muscular Atrophy (SMA) patient and research organisations from across Europe.

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# Support for SMA

## Treatments for SMA

A medication called spinraza (nusinersen) is the first commercial treatment for SMA worldwide. In Australia it was approved by the Pharmaceutical Benefits Advisory Committee (PBAC) on 1 May 2018 for those under 19 years of age with Type 1, 2 and 3a SMA. It works by increasing the production of SMN2 protein to increase motor function. The treatment is provided via an injection into your spine (lumbar puncture) with 4 loading doses and then a maintenance

dose every 4 months. SMA Australia is currently campaigning to allow access to this treatment for people over 19 years of age.

SMA Australia is also involved with other treatments that are being researched and developed around the world.

Zolgensma is ‘gene therapy’ which involves one injection at birth to replace the faulty gene with one that works correctly. Australia is part of current trials for this revolutionary treatment and it is currently under review with the Food and Drug Administration (FDA) in America. SMA Australia is monitoring the outcome of the FDA decision, to enable an application to the regulatory bodies here to proceed when appropriate.

Risdiplam is a medication that you swallow. It is taken daily to increase more functional SMN protein to better support motor neurons and muscle function. The current clinical trials in America are working with different groups of people affected by SMA, looking at a range of type, ages and severity of SMA. Current clinical trials within Australia are focusing on newborn babies.

Please contact SMA Australia if you have any questions about these treatments. You can also register with the SMA Patient Register here to be kept up to date with developments about trials and treatments.

## International Standards of Care

The 2017 International Standards of Care for SMA contain recommendations for diagnosis and management of SMA. The guide to these standards contains information in relation to:

- Genetics and diagnosis
- Physiotherapy and occupational therapy
- Orthopaedic management
- Nutrition, growth and bone health
- Breathing
- Other organs and systems
- Medications
- Emergency care
- Anaesthetics
- Administration of new treatments for sma and
- Ethics and choices.

Although this list might seem long and daunting, it can be useful to review the guide to the Standards and have discussions with your specialists, doctors, nurses and other health professionals for further clarification if you have questions about managing SMA.

### SMA Registry

The Australian Neuromuscular Disease Registry (ANMDR) aims to establish a database of all patients, children and adults in Australia diagnosed with neuromuscular disease. By creating this database, they hope to form a network of patients, clinicians, researchers, and industry to further research into neuromuscular diseases, ease the process in finding and selecting participants for clinical trials, and create an additional support for patients and their families. Joining the Registry is entirely voluntary and Australian families affected by SMA are encouraged to register or contact the Registry for more information via anmdr@mcri.edu.au

## Other helpful support services

The video is available at https://youtu.be/BDaCNsd9R_s.

- Your state or territory neuromuscular organisation can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing. Visit our page on our state or territory neuromuscular organisations for more information.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn more about what services are available and how to access them here. Visit our pages on Counselling, Social Work and Wellbeing for more information.
- Living with SMA may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on Disability Support Workers for more information.
- Other people and families living with SMA have also experienced the emotional rollercoaster that comes with a diagnosis of SMA and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with SMA in The Loop Community on our Forum.

## Medical Alert Card

Muscular Dystrophy Queensland has developed a Medical Alert Card for SMA Type 2. A Medical Alert Card can help you communicate your care needs in a medical emergency situation. If you would like a Medical Alert Card for SMA Type 2 to keep in your wallet or purse, please call (07) 3243 9700 or complete the form on the Muscular Dystrophy Queensland website.

# How to have better conversations when communicating your needs

To learn how to have better conversations when communicating your needs, visit the following pages:

Employers

Educators

Living Life: Education

# Support for SMA

## Treatments for SMA

A medication called spinraza (nusinersen) is the first commercial treatment for SMA worldwide. In Australia it was approved by the Pharmaceutical Benefits Advisory Committee (PBAC) on 1 May 2018 for those under 19 years of age with Type 1, 2 and 3a SMA. It works by increasing the production of SMN2 protein to increase motor function. The treatment is provided via an injection into your spine (lumbar puncture) with 4 loading doses and then a maintenance dose every 4 months. SMA Australia is currently campaigning to allow access to this treatment for people over 19 years of age.

SMA Australia is also involved with other treatments that are being researched and developed around the world.

Zolgensma is ‘gene therapy’ which involves one injection at birth to replace the faulty gene with one that works correctly. Australia is part of current trials for this revolutionary treatment and it is currently under review with the Food and Drug Administration (FDA) in America. SMA Australia is monitoring the outcome of the FDA decision, to enable an application to the regulatory bodies here to proceed when appropriate.

Risdiplam is a medication that you swallow. It is taken daily to increase more functional SMN protein to better support motor neurons and muscle function. The current clinical trials in America are working with different groups of people affected by SMA, looking at a range of type, ages and severity of SMA. Current clinical trials within Australia are focusing on newborn babies.

Please contact SMA Australia if you have any questions about these treatments. You can also register with the SMA Patient Register here to be kept up to date with developments about trials and treatments.

## International Standards of Care

The 2017 International Standards of Care for SMA contain recommendations for diagnosis and management of SMA. The guide to these standards contains information in relation to:

- Genetics and diagnosis
- Physiotherapy and occupational therapy
- Orthopaedic management
- Nutrition, growth and bone health
- Breathing
- Other organs and systems
- Medications
- Emergency care
- Anaesthetics
- Administration of new treatments for sma and
- Ethics and choices.

Although this list might seem long and daunting, it can be useful to review the guide to the Standards and have discussions with your specialists, doctors, nurses and other health professionals for further clarification if you have questions about managing SMA.

### SMA Registry

The Australian Neuromuscular Disease Registry (ANMDR) aims to establish a database of all patients, children and adults in Australia diagnosed with neuromuscular disease. By creating this database, they hope to form a network of patients, clinicians, researchers, and industry to further research into neuromuscular diseases, ease the process in finding and selecting participants for clinical trials, and create an additional support for patients and their families. Joining the Registry is entirely voluntary and Australian families affected by SMA are encouraged to register or contact the Registry for more information via anmdr@mcri.edu.au

## Other helpful support services

The video is available at https://youtu.be/BDaCNsd9R_s.

- Your state or territory neuromuscular organisation can provide information and advice on what support they can offer such as local support groups, camps, programs, services in the local area, access to cough assist machines, advocacy or assistance in times of crisis. They can also provide an ear to listen if you need someone to talk to or guide you to get the assistance you are needing. Visit our page on our state or territory neuromuscular organisations for more information.
- Seeing a psychologist, counsellor or social worker can be incredibly helpful if you, your child or other members of the family are having a tough time or struggling with negative thoughts and feelings. Learn

more about what services are available and how to access them here. Visit our pages on Counselling, Social Work and Wellbeing for more information.

- Living with SMA may mean that you or your child may need some assistance for everyday activities. Find out more about how a disability support worker could help and how to engage this type of support. Visit our page on Disability Support Workers for more information.
- Other people and families living with SMA have also experienced the emotional rollercoaster that comes with a diagnosis of SMA and are able to understand exactly how you're feeling. You are not alone. Connect with other people and families living with SMA in The Loop Community on our Forum.

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# About SMA

**SMA fact sheet** From Spinal Muscular Atrophy Australia Inc, 2017. This fact sheet contains facts about SMA.

**SMA Type 1 fact sheet** From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

**SMA Type 2, Type 3 and Type 4 fact sheet** From Muscular Dystrophy New South Wales, 2017. This fact sheet includes considerations for future planning.

**What is Spinal Muscular Atrophy A guide for parents and families** From Muscular Dystrophy Association, United States, February 2019. This guide includes what are the signs and symptoms of SMA?, what should I know

about SMA?, how is SMA treated? and a glossary of terms.

# Care Recommendations

**2017 International Standards of Care for SMA** Contains recommendations for diagnosis and management of SMA.

**A Guide to the 2017 International Standards of Care for SMA** The guide was developed by Spinal Muscular Atrophy UK, Cure SMA, SMA Europe, Muscular Dystrophy UK and TREAT-NMD Alliance in 2019 and outlines the 2017 International Standards of Care Recommendations for SMA.

Below are links to the more detailed standards of care recommendations for SMA on which A Guide to the 2017 International Standards of Care for SMA is based. Although these are written in medical terms they may be useful for discussions with specialists, doctors, nurses and allied health professionals

**Part 1: Diagnosis and Management of SMA; Recommendations for diagnosis; rehabilitation; orthopaedic and nutritional care.**

**Part 2: Diagnosis and Management of SMA; Pulmonary and acute care; medication; supplements and immunisations; other organ systems; and ethics**

# Support Organisation

**Spinal Muscular Atrophy Australia Inc** An organisation whose mission is to “Improve care for all Australians living with SMA, so that the community have access to all possible research, diagnosis and treatment options.”

# International Organisations

**Cure SMA** Cure SMA leads the way to a world without spinal muscular atrophy, the number one genetic cause of death for infants. We fund and direct comprehensive research that drives breakthroughs in treatment and care, and we provide families the support they need for today.

**Spinal Muscular Atrophy UK** If you or someone in your family has SMA, and you live in the UK, you have come to the right place for support and information about the condition and the latest developments with new treatments.

**SMA Europe** SMA Europe is an umbrella organisation, founded in 2006, which includes Spinal Muscular Atrophy (SMA) patient and research organisations from across Europe.

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**State & Territory Neuromuscular Organisations**These State and Territory organisations across Australia work to empower, connect and support people with neuromuscular conditions, including SMA. Although many retain the name Muscular Dystrophy in their title, these organisations welcome families with Spinal Muscular Atrophy. Reach out to your nearest organisation for information, and the local supports and services available.

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