Spinal Muscular Atrophy Type 2

This information sheet briefly explains the cause, effects and management of Spinal Muscular Atrophy (SMA) Type 2. It includes sources of further information and support. It is for the families of children diagnosed with SMA Type 2. It may also be useful for healthcare and other professionals.

SMA Type 2 is a complex condition; there is a lot of information to take in and every child with SMA is different. You may come across medical terms you haven’t heard of before now. We do our best to explain them in this information sheet, but if anything isn’t clear, you can always ask your child’s medical team to go over any of this information with you. (You may find it helpful to have a copy of our ‘Glossary of terms’ which you can download via the website at www.smasupportuk.org.uk/glossaries)

Further information on SMA Type 2 and sources of support are available from SMA Support UK’s route map for SMA Type 2: www.routemapforsma.org.uk

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a rare, genetically inherited neuromuscular condition. It causes progressive muscle weakness and loss of movement due to muscle wasting (atrophy). This may affect crawling and walking ability, arm, hand, head and neck movement, breathing and swallowing. There are different forms of SMA and a wide spectrum of how severely children, young people and adults are affected.

The more common forms of SMA are often described as ‘Types’. Symptoms of SMA Types 1, 2 and 3 begin at different ages in childhood. SMA Type 4 is an adult onset form. The impact of the condition, however, varies greatly within and between each type. Sometimes, to try to describe this, doctors will add ‘a’, ‘b’ or ‘c’ to the type of SMA or, as well as being diagnosed with a type of SMA, a child may also be described as ‘strong’ or ‘weak’ or having ‘early onset’ or ‘later onset’. A child / young person’s ability to sit, stand or walk and how their breathing ability is affected by their SMA are all also very important – especially when it comes to managing the condition.
What causes SMA?

• The *SMN1* gene

The main types of SMA affect the nerve cells called lower motor neurons which run from the spinal cord to the muscles. These lower motor neurons carry electrical signals from the brain to move the muscles used for crawling and walking. These signals also control movement of arms, hands, head and neck as well as breathing and swallowing. For these lower motor neurons to be healthy, our *Survival Motor Neuron 1* genes (*SMN1* genes\(^2\)) must produce enough Survival Motor Neuron (SMN) protein.

Most people have two copies of the *SMN1* gene. People with the main types of SMA have two faulty copies of the *SMN1* gene, which means they are unable to produce enough SMN protein to have healthy lower motor neurons\(^3\). This causes their lower motor neurons in the spinal cord to deteriorate. This restricts the delivery of signals from the brain to their muscles, making movement difficult. Their muscles then waste due to lack of use; this is known as muscular atrophy. In summary:

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\begin{align*}
\text{Brain sends signals along the spinal cord via lower motor neuron nerve cells} \\
\begin{array}{c}
\text{*SMN1* genes don't produce enough SMN protein to keep these cells healthy in people who have SMA} \\
\text{Lower motor neurons in the spinal cord deteriorate} \\
\text{Signals can't efficiently get through to the muscles making movement difficult} \\
\text{Muscles waste (atrophy) due to lack of use}
\end{array}
\end{align*}
\]

The *SMN1* gene is on the fifth chromosome in the region labelled ‘q’. This is why the main types of SMA are often referred to as ‘5q SMA’.

• The *SMN2* gene\(^2\)

A second gene also has a role in producing SMN protein. This is the *Survival Motor Neuron 2* gene (*SMN2*), sometimes referred to as the SMA “back-up gene”.


However, most of the SMN protein produced by SMN2 lacks a key building block that is usually produced by SMN1. This means that SMN2 cannot fully make up for the faulty SMN1 gene.

The number of SMN2 genes can vary greatly from person to person, from 0 – 8 copies. The severity of SMA has been linked to how much SMN protein a person makes\(^6\); individuals with more SMN2 copies typically have a less severe form of SMA than those with fewer copies.

For more information on the inheritance of SMA please see ‘The Genetics of Spinal Muscular Atrophy’: [www.smasupportuk.org.uk/the-genetics-of-sma](http://www.smasupportuk.org.uk/the-genetics-of-sma)

### What is SMA Type 2?

SMA Type 2 is sometimes called intermediate SMA or chronic infantile SMA. The symptoms of muscle weakness and floppiness (low tone / hypotonia) usually begin between 7 and 18 months of age.

Each child with SMA Type 2 is different. Some children will sit independently whilst others will require some support. Usually children with SMA Type 2 will need supportive aids for standing and a wheelchair to get around.

Though this is a serious neuromuscular condition that may shorten life expectancy\(^7\), improvements in care standards mean that the majority of people can live long, fulfilling lives.

### How is SMA Type 2 diagnosed?

The doctor will need to physically examine your child and find out about their medical history. They will then take a blood sample for DNA testing. The blood sample is tested for a deletion mutation in the *Survival Motor Neuron 1* (SMN1) gene on chromosome 5. The result of this test is usually available within 2 – 4 weeks.

If there is any uncertainty about the diagnosis further muscle tests such as an electromyogram (EMG) may be discussed with you, but this is not usually needed to confirm SMA.

### What are the effects of SMA Type 2?

This section describes the effects of SMA Type 2 in general terms. However, it’s important to remember that each child with SMA Type 2 is affected differently and the severity of the condition varies from child to child.

Children’s muscle weakness is usually the same on both sides of their body (symmetrical). The muscles closest to the centre of their body (proximal muscles) are usually more severely affected than the muscles furthest from the centre of their body (distal muscles). This can make it difficult for children with SMA Type 2 to lift their arms and legs but they will still be
able to use their hands and fingers. Generally, children with SMA Type 2 find that their legs are weaker than their arms.

As your child grows, their muscle weakness may make it difficult for them to keep up with their daily activities. For example, if your child has been able to crawl or roll, they may lose this ability as they get older. They may become weaker after infections and at times of major growth spurts such as puberty.

Because a child with SMA Type 2 has weak breathing (respiratory) muscles, it can be difficult for them to cough effectively. This can make them more vulnerable to chest (respiratory) infections.

In SMA Type 2, the muscles supporting the spinal column are weakened. This means that most children develop a sideways curvature of their spine (scoliosis). Also, because the condition reduces children’s ability to move, some joints may become tight (contractures) which restricts their range of movement.

SMA Type 2 can weaken children’s chewing and swallowing muscles (bulbar muscles). Some children find that their tongue or shoulder muscles twitch (fasciculation) and they may have a slight tremor in their hands. SMA doesn’t affect a child’s sexual or intellectual development or bladder and bowel control. Children will though need help with, for example, transferring from their wheelchair to the toilet, washing, dressing and undressing.

What healthcare and support is needed for SMA Type 2?

The 2017 Standards of Care for Spinal Muscular Atrophy (SoC) provide a summary of guidelines for health professionals covering the three more common forms of SMA, which includes Type 2. The sections in the SoC headed ‘sitters’ covers best practice and management SMA Type 2.

Your child should receive care from a multidisciplinary healthcare team. The number of people in this team can feel a bit overwhelming, but they all have an important role to play. You may have contact with a hospital or community consultant paediatrician as well as specialists in:

- neuromuscular conditions,
- respiratory medicine,
- orthopaedics,
- physiotherapy,
- occupational therapy,
- speech and language therapy,
- dietetics,

You can find out more about how these people help in our information sheet ‘Who’s Who of Professionals’: www.smasupportuk.org.uk/whos-who-of-professionals. If possible one of the team should be your keyworker who will help co-ordinate services for your family.
Children with SMA Type 2 should be seen by their medical team regularly to measure any change in their health and to offer advice and interventions at the right time. The aim is to enable your child to remain healthy and enjoy a good quality of life.

At every appointment with your child’s medical team you should be given time to ask questions and then jointly decide what support is best for your child.

- **Breathing**

Children with SMA Type 2 often have weak breathing (respiratory) muscles – that’s both the ones for breathing in (inspiratory muscles) and the ones for breathing out (expiratory muscles). This can cause them breathing problems, such as ‘under breathing’ (hypoventilation) which is when the breaths they take are too small and their breathing rate may also be slower than normal. Because of this, their oxygen levels may drop and also waste gas (carbon dioxide) that they are not getting rid of, may build up. Typically, this happens overnight and is called ‘nocturnal hypoventilation’. Signs of nocturnal hypoventilation include frequent turning at night, morning headaches, fatigue during the day and poor concentration.

If your child has signs of nocturnal hypoventilation, they may need a sleep study. This usually involves attaching a small sensor to their ear or arm and monitoring them overnight. Sometimes, they may need to have a more detailed study which involves a few more sensors attached to their face, head, arm and chest.

If the study shows your child is under breathing at night, the recommended treatment is non-invasive ventilation (NIV), for example Bilevel NIV. This consists of a small breathing machine attached to a mask that is worn by your child when they are sleeping. When they are asleep, NIV provides your child with a deeper breath than they can make themselves and it can also increase the number of breaths they can make when they are asleep. This then corrects oxygen and carbon dioxide levels overnight and improves any daytime sleepiness and lack of concentration.

Children’s muscle weakness can also lead them to have an increased risk of chest (respiratory) infections. Any sudden (acute) chest infection causes their muscle weakness to temporarily become worse, so NIV is sometimes used to manage this. NIV can also help to rest the breathing muscles and make your child less short of breath when they are unwell.

Some children may need help with their coughing. Your respiratory physiotherapist will assess each child individually to suggest the best way to manage this. They may suggest using a ‘mechanical insufflator-exsufflator’(MI-E) device, sometimes known as a ‘cough machine’. This helps children clear secretions from their lungs.

Chest physiotherapy is an important part of management and will be tailored to your child’s needs. Your respiratory physiotherapist will train you and any carers on the techniques they advise for your child.

You can read an overview of what good respiratory management involves in the SoC\textsuperscript{11} and this article ‘Airway clearance techniques in neuromuscular disorders’\textsuperscript{12}. As each child with
SMA Type 2 is affected differently it’s important to discuss any queries you have with your child’s medical team.

- **Eating, drinking and nutrition**

Children with SMA Type 2 sometimes have difficulty with their chewing, swallowing and nutrition.

A healthy diet is important for everyone. Your child may have problems putting on weight if eating becomes tiring for them or if they have illnesses or infections. Equally, your child may become overweight due to their reduced mobility. If this happens the extra weight can increase the stress on muscles, bones and joints, making physical activity even more difficult. Limited mobility can also mean that some individuals with SMA have a lower bone density than others. Vitamin D and calcium supplementation may help improve bone strength. Additionally, children with SMA Type 2 can become constipated which may cause discomfort and make breathing more difficult.

A dietitian will be able to advise on a healthy diet that will suit your child. You can ask them or your child’s medical team how to manage any constipation and for guidance on any supplements. They, and your child’s health visitor, consultant and speech and language therapist may also provide you with other advice and support on eating and nutrition. Occupational therapists and physiotherapists may also advise on positioning, seating or arm supports to help your child to eat independently.

If your child has difficulty swallowing there is a risk that they may inhale liquids or food into their lungs (aspirate) which can cause chest infections. If this is happening, or your child is not putting on weight, your child’s medical team may suggest alternative ways for them to take in food. This may involve your child’s food going through a tube directly into their stomach (gastrostomy (G) tube).

Muscle weakness may make it difficult for your child to open their mouth widely. This can cause problems with not only eating, but also teeth cleaning and dental care. Regular dental check-ups and getting help with these sorts of problems early may help to prevent complications such as getting food into the lungs.

- **Posture, movement and mobility**

SMA Type 2 will mean that your child will have difficulties with their posture, movement and mobility. They will need their own exercise routines designed by their physiotherapist to help with this. Routines may include exercises to:

- help maintain their range of motion
- reduce any discomfort
- stretch any tight muscles
- prevent joints becoming tight
Your child may enjoy doing these exercises in the bath, or a swimming or hydrotherapy pool as the warm water aids buoyancy.

Regular gentle stretching of tight joints can help to reduce the pain that contractures can cause. If your child does have any pain, do talk to their doctor and physiotherapist.

Regular moderate exercise will also help your child to maintain their fitness and stamina. Activities such as swimming and horse riding can be adapted to match their physical ability.

Although your child will lose muscle strength over time, it’s important that they maintain activities like supported standing for as long as possible. Standing is good for breathing, blood circulation, bladder, bowels, bones and joints.

As your child’s physical abilities change, an occupational therapist will advise what sort of seating will give them the best, most comfortable support. This will make it easier for them to play with toys, eat independently and join in at home and at school.

A physiotherapist will assess your child and provide appropriate equipment to support their standing and positioning. Some children with SMA Type 2 find that it’s helpful to have splints (sometimes called orthoses) for support. These include ankle foot orthoses (AFOs) and knee, ankle, foot orthoses (KAFOs). These will be made specifically for your child by an orthotist who will explain how they will help.

Your child’s physiotherapist and occupational therapist will be able to advise you about powered wheelchairs. This should be at around the time that your child would have been learning to walk. This equipment will mean that they will be able to explore and join in much more easily which will help with their physical, emotional, social and educational development.

As already mentioned, most children with SMA Type 2 develop a sideways curvature of their spine (scoliosis). It’s important that the medical team monitors your child regularly so that any increase in curvature is noticed early. The degree of the curvature and your child’s age will be factors in deciding how to manage this. Initially this may be with a spinal brace or jacket but surgery to correct scoliosis may be recommended if the scoliosis is contributing to breathing difficulties, preventing comfortable sitting or has progressed beyond a certain point.

Your occupational therapist may advise you about a sleep system to support your child’s back, arms and legs and make sleeping more comfortable. They can also give you advice about other adaptations and equipment that will help with your child’s everyday activities both at home and school, such as writing, playing, washing, dressing, cooking and eating. With appropriate encouragement, adaptations and support, your child will be able to lead a fulfilling life.
- Emergency health plans

Your child’s medical team may work with you to develop an emergency health plan. This records the treatment you wish your child to receive if there is an emergency or if their health deteriorates. You should have your own copy so that you can give it to hospital services if you are away from your home area. The plan can be reviewed and you can change your mind about its contents at any time.

What other help is available?

A diagnosis of SMA Type 2 with all its complexity can have an enormous impact on families. It’s important for you and your child to have emotional support and plenty of time to talk and ask questions. This can be with members of your child’s medical team, your local G.P., health visitor, social worker, psychologist or a counsellor.

As your child gets older, their needs will change. So that your child can participate fully in activities at home, school and in their community, you will want information, advice and support on mobility, education, equipment and sources of funding. You may also want advice when it comes to adolescence, for example, how to manage periods for girls and sex education in general. Your child’s healthcare team, SMA Support UK’s Support Services Team, and the other people and agencies listed in this leaflet are all helpful sources of information. SMA Support UK’s route map for SMA Type 2 is also a useful an online resource to explore at: www.routemapforsma.org.uk (e.g. see the section daily living/adulthood and SMA)

SMA Support UK’s Outreach Workers can visit you at home and discuss the health, social, educational, financial and care support that you and your child may be entitled to. As well as providing emotional support, SMA Support UK can also put you in touch with individuals with SMA Type 2 or families caring for a child with SMA Type 2 who are part of our Shared Experiences Network. Information about these services is available on our website: www.smasupportuk.org.uk/how-we-can-support-you or please phone us on 01789 267 520 or email: supportservices@smasupportuk.org.uk

Muscular Dystrophy UK also provides information, support and advocacy services, including grants towards specialist equipment, for people affected by a range of neuromuscular conditions. Their website is: www.musculardystrophyuk.org or you can phone them on 0800 652 6352 or e-mail: info@musculardystrophyuk.org

Regional care advisors and sometimes neuromuscular nurse specialists are attached to NHS neuromuscular clinics in various regions of the UK. They provide support and information to children and adults with muscle diseases and their families. They link up with other professionals and services so that people receive the local health and social support they need. Regional care advisors’ contact details are available on Muscular Dystrophy UK’s website: www.musculardystrophyuk.org/get-the-right-care-and-support/people-and-places-to-help-you/care-advisors/
Children’s hospices throughout the UK also offer a wide range of services and support to children and families; some also offer short breaks. Details of hospice services are available from ‘Together for Short Lives’ on 0808 8088 100 and more information is available on their website: [www.togetherforshortlives.org.uk](http://www.togetherforshortlives.org.uk)

### Financial Support

Families living in the UK may be eligible for a number of financial benefits to help towards the cost of providing the extra care their child may need. This does depend on your individual circumstances.

For further information about financial benefits visit the Gov.UK website [www.gov.uk](http://www.gov.uk) and look at the sections ‘Benefits’ and ‘Carers and Disability Benefits’.

**Contact** provide information and support to families who have a child with a disability. This includes information on benefits and grants. They can be contacted on 0808 808 3555 or through their website: [www.contactafamily.org.uk](http://www.contactafamily.org.uk)

**Disability Rights UK** publishes free factsheets on a range of benefits and the ‘Disability Rights Handbook’ annually. For further information visit: [www.disabilityrightsuk.org](http://www.disabilityrightsuk.org)

**Turn2Us** is a charity which helps people access money available to them through welfare benefits, grants and other help. They can be contacted on 0808 802 2000 or through their website: [www.turn2us.org.uk](http://www.turn2us.org.uk)

Your health visitor, neuromuscular care advisor, family support worker, social worker or outreach worker may be able to help you with applications for financial benefits.

There are also a number of charities that may assist you with the cost of general household goods, specialist equipment and holidays / days out. Please contact SMA Support UK for more information or see the Daily Living section of the SMA Type 2 route map: [www.routemapforsma.org.uk](http://www.routemapforsma.org.uk)

### Genetic Counselling

As a parent with a child with SMA you should be offered a referral for genetic counselling. You can also request a referral from your General Practitioner (G.P.).

Genetic counselling is with a healthcare professional who has expert training in genetics. They will aim to explain results from genetic testing in an easily understandable way, and answer any questions you might have about the genetic aspects of the diagnosis. Common issues to discuss in genetic counselling might include implications or options for a future pregnancy, and whether there is a need to discuss the diagnosis with other family members, who might wish to seek genetic counselling and testing themselves.
For more information on the genetics of SMA, the chances of having a child with SMA and the tests that can be carried out, please see our leaflet ‘The Genetics of Spinal Muscular Atrophy’: www.smasupportuk.org.uk/the-genetics-of-sma

For more information on ‘Future Options in Pregnancy’ please see: www.smasupportuk.org.uk/future-options-in-pregnancy

Is there a treatment or cure for SMA Type 2?

Although there is currently no cure for SMA, the range of options aimed at managing symptoms and reducing complications of muscle weakness all help maintain the best quality of life.

- **Nusinersen/ Spinraza™**

The first (and currently, the only) potentially available drug treatment for SMA is called nusinersen. Essentially, the drug is designed to modify the product of the *SMN2* gene to produce more functional SMN protein.

In collaboration with researchers, nusinersen was developed by Ionis Pharmaceuticals and Biogen Idec, which have run clinical trials with infants and children affected by SMA Types 1, 2 or 3. There have not yet been any clinical trials of nusinersen with anyone with SMA Type 4. On June 1st, 2017, the European Commission approved nusinersen for marketing under its brand name Spinraza™ as a treatment for those with 5q SMA. This is a broad term, that includes SMA Types 1, 2, 3 and 4.

Currently in the UK, nusinersen is only available if the medical team and family agree that an infant with SMA Type 1 is eligible and may potentially benefit from the treatment. This is possible in Scotland via the NHS and in the rest of the UK via what is called an Expanded Access Programme (EAP).

To find out more about how nusinersen works and the clinical trial results, please explore this section of our website: http://www.smasupportuk.org.uk/nusinersen Here, you will also find out how the UK drug approval system works, what stage nusinersen has reached, and the collaborative efforts being made to widen access to the treatment.

Research and further developments

The UK SMA Patient Registry is a database of genetic and clinical information about people affected by SMA. As new treatments for SMA are being developed, they need to be tested in clinical trials. Researchers wanting to find people interested in joining a clinical trial contact the Patient Registry which then contacts the people who have registered to let them know about the potential opportunity. If this is of interest to you, you can sign up with the Patient Registry.
The Registry also helps specialists gain more knowledge about the condition and the number of people affected by SMA. This information helps to develop and improve worldwide standards of care for people with SMA. You can find out more by looking at their website: www.treat-nmd.org.uk/registry e-mailing: registry@treat-nmd.org.uk or phoning: 0191 241 8640.

SMA Support UK’s website also notifies the SMA community about latest developments with other drug treatments, the science behind them and what clinical trials and other research is going on: www.smasupportuk.org.uk/research We alert people to updates via our social media and monthly E-news. You can sign up for mailings at www.smasupportuk.org.uk/sign-up-for-mailings

Further Resources

Standards of Care for Spinal Muscular Atrophy (2017)

You can read about and download the 2017 internationally agreed Standards of Care from here: http://www.smasupportuk.org.uk/international-standards-of-care-for-sma

SMA Support UK information sheets and books for children

Information sheets and other resources may be downloaded from the SMA Support UK website: www.smasupportuk.org.uk/about-sma

Any family with a child with SMA Type 2 may request a free copy of each of the following publications:

- **SMA Type 2 and Me** – an illustrated book written for children
- **Smasheroo** – an illustrated book for young children affected by SMA Type 2 or SMA Type 3

For hard copies of information sheets or free copies of these books, contact supportservices@smasupportuk.org.uk or phone 01789 267520

Further copies of the children’s books may be ordered from the shop on SMA Support UK’s website: www.smasupportuk.org.uk/merchandise

Muscular Dystrophy UK

Provide information, support, advocacy services and grants towards specialist equipment for people affected by a range of neuromuscular conditions. They also have specific ‘alert cards’ which can be used to provide medical professionals with information about your child’s condition. You can find more about MDUK: www.musculardystrophyuk.org/
The link to the alert card for SMA Type 2 is:

The UK SMA Patient Registry
This leaflet describes the work of the Registry and how to sign up. A hard copy may be requested from SMA Support UK. It can also be downloaded from:
www.treat-nmd.org.uk/registry

If you have any feedback about this information, please do let us know at
supportservices@smasupportuk.org.uk

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References


