Spinal Muscular Atrophy Type 3

This information sheet briefly explains the cause, effects and management of Spinal Muscular Atrophy (SMA) Type 3. It includes sources of further information and support. It is for the families of children diagnosed with SMA Type 3. It may also be useful for healthcare and other professionals.

SMA Type 3 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 from our website at: www.smasupportuk.org.uk/glossaries

Further information on SMA Type 3 and sources of support are available from SMA Support UK’s route map for SMA Type 3: 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) 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. 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:

![Diagram](attachment:image.png)

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**

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; 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

What is SMA Type 3?

SMA Type 3 is sometimes called Kugelberg-Welander disease. With SMA Type 3a: symptoms usually begin between 18 months and 3 years of age. Children can stand and walk, although this will become more difficult and they will need more support with this over time. With SMA Type 3b, symptoms usually begin after 3 years. Difficulties with standing and walking usually occur later than for children who have earlier symptoms. Depending upon the individual impact of their condition, children and adults with SMA Type 3 may be described as ‘sitters’ or ‘walkers’. They don’t usually have breathing problems, their life expectancy is not affected and most can live long, fulfilling lives.

How is SMA Type 3 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 test result is usually available within 2 – 4 weeks.

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

What are the effects of SMA Type 3?

This section describes the effects of SMA Type 3 in general terms. However, it’s important to remember that each child with SMA Type 3 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). Generally, children with SMA Type 3 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 walk or climb stairs, they may lose this ability as they get older. Some children may fall more easily because of their muscle weakness. If they are sitting on the floor they may need help to get up. Children are generally able to stand and walk until late childhood and sometimes into adolescence. About 50% of children with SMA Type 3 lose the ability to walk independently by the age of 14, although some people are still able to walk in adulthood.

In SMA Type 3, the muscles supporting the spinal column are weakened. This means that some 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.

Muscle weakness in children with SMA Type 3 mainly affects their limb movement. SMA doesn’t affect a child’s sexual or intellectual development or bladder and bowel control. Some children will though need help with, for example, getting to and sitting on the toilet, washing, dressing and undressing.

Children with SMA Type 3 may become weaker after infections and at times of major growth spurts such as puberty.

What healthcare and support is needed for SMA Type 3?

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 3. There are two sections for SMA Type 3 – one headed ‘sitters’ and one ‘walkers’ - whichever is most appropriate for guiding best practice and management.

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 3 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**

Generally, children with SMA Type 3 do not have difficulties with their breathing, but their breathing strength and cough effectiveness should be checked regularly.

You can read an overview of what good respiratory management involves in the SoC. As each child with SMA Type 3 is affected differently, it’s important to discuss any queries you have with your child’s medical team.

- **Nutrition**

Children with SMA Type 3 rarely have difficulty with their chewing and swallowing but your child’s medical team will provide you with advice and support if necessary.

A healthy diet is important for everyone. If your child has reduced mobility, they may become overweight. 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\(^1\). Additionally, children with SMA Type 3 can become constipated which may cause discomfort.

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.

- **Posture, movement and mobility**

SMA Type 3 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 contractures
Your child might 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 any pain that contractures can cause. If your child does have any pain, do talk to their doctor and physiotherapist.

Although your child will lose muscle strength over time, it’s important that they maintain activities like standing and walking for as long as possible. Walking can help delay the development of scoliosis and standing is good for breathing, blood circulation, bladder, bowels, bones and joints. Regular moderate exercise will also help your child maintain their fitness and stamina and activities such as swimming and horse riding can be adapted to match their physical ability.

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, walking and positioning. Some children with SMA Type 3 find that it’s helpful to have splints (sometimes called orthoses) to support their standing and walking. 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.

Specialist equipment will enable your child to take part in home and school activities. If your child finds walking increasingly difficult, or they are having falls, a walking frame or a manual wheelchair may help. If your child’s weakness increases, a powered (electric) wheelchair may help their independence. Your child’s physiotherapist and occupational therapist will be able to advise you about walking aids and wheelchairs.

As already mentioned, children with SMA Type 3 may 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. This is more likely if your child is no longer able to walk. 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.

Occupational therapists can 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.

What other help is available?

A diagnosis of SMA Type 3 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 General Practitioner (G.P.), health visitor, social worker, psychologist or a counsellor.

So that your child can participate fully in activities at home, school and in their community, you will need information, advice and support on mobility, education, equipment and sources of funding. You can find out more by talking to your child’s healthcare team, SMA Support UK’s Support Services Team, and the other people and agencies listed in this leaflet. SMA Support UK’s route map for SMA Type 3 is also a useful online resource available at: www.routemapforsma.org.uk

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 3 or families caring for a child with SMA Type 3 who are part of our Shared Experiences Network. Information about these services is available on our website: www.smasureportuk.org.uk/how-we-can-support-you or please phone us on 01789 267 520 or email: supportservices@smasureportuk.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 check website: www.musculardystrophyuk.org/get-the-right-care-and-support/people-and-places-to-help-you/care-advisors/

- **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 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.contact.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
**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 3 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 help you to understand how SMA is passed on and what the chances are of other people in your family being affected. Genetic counselling also provides you with the opportunity to discuss your choices for any future pregnancies. You will be able to go back to your genetic counsellor later if you have more questions.

As your child and any siblings grow up, they can also ask for genetic counselling, particularly if they are considering having children.

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](http://www.smasupportuk.org.uk/the-genetics-of-sma)


**Is there a treatment or cure for SMA Type 3?**

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 Sq 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 our 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](http://www.smasupportuk.org.uk/about-sma)

Any UK family with a child with SMA Type 3 may request a free copy of each of the following publications:

- **SMA Type 3 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](mailto: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](http://www.smasupportuk.org.uk/merchandise)

**Muscular Dystrophy UK**

Provides 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 at [http://www.musculardystrophyuk.org/](http://www.musculardystrophyuk.org/) and a link to the alert card for SMA Type 3 is at [http://www.musculardystrophyuk.org/app/uploads/2016/11/SMA-alert-card-Type-3-v2.pdf](http://www.musculardystrophyuk.org/app/uploads/2016/11/SMA-alert-card-Type-3-v2.pdf)

**The UK SMA Patient Registry leaflet**

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](http://www.treat-nmd.org.uk/registry)

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If you have any feedback about this information, please do let us know at [supportservices@smasupportuk.org.uk](mailto:supportservices@smasupportuk.org.uk)
We are grateful to the writers and reviewers who assist us in our information production. A list of who this includes may be viewed on our website: [www.smasupportuk.org.uk/our-writers-and-reviewers-panel](http://www.smasupportuk.org.uk/our-writers-and-reviewers-panel) or requested from supportservices@smasupportuk.org.uk

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References


