



Looking after your child who has SMA Type 2

Information for parents and carers of children who have had a recent diagnosis



Includes

Treatment

Healthcare teams who support you

Posture, Movement & Mobility

Breathing and Eating

Transport

Support and resources



Patient information awards
Highly commended



What's in this guide?

This guide tells you more about the practical side of caring for your child who has SMA Type 2. It combines information about the healthcare your child may need, along with tips and suggestions that have worked for other families. The guide is designed to be used alongside medical advice which should always come from your child's medical team.

One of the first questions many families ask their medical teams is: "What treatment might be possible for my child?". Since late 2016, the gradual worldwide introduction of drug treatments for some children, young people and adults who have SMA Type 2 has seen positive outcomes for many. Not every treatment is suitable or possible at the moment; your child's medical team will talk this through with you.

In the UK, treatment is possible for many children who have SMA Type 2:

- **Nusinersen (Spinraza™)** is funded by the NHS. You can read about it in our information sheet: **Nusinersen (Spinraza™) treatment for those diagnosed with SMA Type 2 or 3.**
- **Risdiplam (Evrysdi™)** is not yet funded by the NHS but may currently be prescribed in certain, very limited circumstances.

Another treatment you may have heard about: **onasemnogene aberparvovec (Zolgensma™)** is funded by the NHS only for eligible groups of children who have SMA Type 1.

Your child's medical team will have the latest information about treatments, and you will be able to discuss possible options with them. Whether or not your child receives drug treatment, your medical team will talk with you about the International Standards of Care for SMA (SoC)^{1,2} which are all about best care for children who have SMA.

The impact of SMA Type 2 varies greatly between individuals and may change over time.

You can find out more about nusinersen and other drug treatments at:

www.smauk.org.uk/drug-treatments-screening-whats-happening-now

Read about risdiplam here:

www.smauk.org.uk/risdiplam

More information about the Standards of Care is at:

www.smauk.org.uk/international-standards-of-care-for-sma

or contact us for a copy.

The most common form of Spinal Muscular Atrophy (SMA) is also known as 5q SMA due to its genetic cause. 5q SMA includes SMA Type 2. For more information about this please see:

- **What is Spinal Muscular Atrophy?**
- **Symptoms, Diagnosis & Effects of 5q Spinal Muscular Atrophy**
- **The Genetics of 5q Spinal Muscular Atrophy**

You can find these at: www.smauk.org.uk/about-sma

Who will be in my child's medical team to support us?

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 specialists in:

- neuromuscular conditions
- hospital or community paediatrics
- physiotherapy
- occupational therapy
- breathing (respiratory) care
- orthopaedics
- dietetics
- speech and language therapy
- general practice, community health care.

The aim of everyone involved in your child's care is that your child will stay healthy and enjoy a good quality of life. Different members of the team will meet you regularly, both to measure any change in your child's health and to offer advice and interventions at the right time. As well as looking at your child's medical needs, when they talk with you about options for care of your child, they will also want to be aware of your family's social, cultural and spiritual needs.

You should be given time to ask questions at every appointment with your child's team so that you can jointly decide what support and treatment is best for your child.

What vaccinations should my child have?

The SoC recommend that anyone with SMA should have the same vaccinations as given to any child in the UK. The only extra vaccinations are the pneumococcal vaccine (over the age of two this should be the vaccine with wider coverage known as '23 valent') and annual flu vaccination.

You can find out more about how these people can help in our information sheet, '**Who's Who of Professionals**'.

One of the team should act as your keyworker. Their role is to help co-ordinate services for your family.

You may want to try to avoid direct contact with others who have coughs and colds but remember that it's impossible for you to prevent your child catching all possible viruses.

What about my child's posture and mobility?

Children with SMA who can sit but who have difficulties standing need specialist support for their posture and mobility. They'll need their physiotherapist (physio) to design their own personal exercise routine which may include exercises to:

- help maintain their range of motion
- reduce any discomfort
- stretch any tight muscles
- prevent joints becoming tight

Some of your child's joints may become tight (contractures) because of your child's reduced ability to move them. Regular gentle stretching can help to reduce any pain. If your child does have any pain, it's important to talk to their doctor and physio.

Regular moderate exercise will help with your child's fitness and energy levels. Activities like swimming and horse riding can be adapted to suit your child and are a fun way to exercise.

Your occupational therapist (OT) will advise you what sort of seating will give your child the best, most comfortable support so that it's easier for them to play with toys, eat independently and join in at home and at school.

Your physio or OT will provide equipment to support your child's standing and positioning. Some children find it helpful to have splints (sometimes called orthoses) for support. These would be made specifically for your child by an orthotist who would explain how to use them and how they can help.

Warm water helps with buoyancy so your child may enjoy doing these exercises in the bath, or in a swimming or hydrotherapy pool.

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 many things including breathing, blood circulation, bladder, bowels, bones and joints.

Children with SMA who are able to sit and are as young as 18 months can often manage to steer a 'Wizzybug'. This 'first wheelchair' with its fun practical design, means they can explore and join in with other children and members of the family. Later they may use a lightweight manual wheelchair or a small, powered wheelchair which will make a big difference to joining in at nursery or school and when going out. Your child's physio and OT will be able to advise you what will work best for your family.

To make sleeping more comfortable, your OT may suggest your child has a sleep system to support their back, arms and legs. They can also give you advice about other adaptations and equipment that will help with your child's everyday home and school activities, such as writing, playing, washing, dressing and eating.

As your child gets older it's important that your team keeps an eye on what impact their SMA and muscle weakness is having on how straight they can sit. Most children with SMA Type 2 develop a sideways 'S'-shaped curvature of their spine (scoliosis). Your team will take into account your child's age and how much their spine is curving and talk to you about how it can best be managed. Although this won't stop scoliosis progressing, initially your child may be given a spinal brace which helps with support but if it's contributing to breathing difficulties, preventing comfortable sitting or the spine is curving beyond a certain point, surgery is likely to be suggested.

You can read Elsie's story about getting her Wizzybug at:

www.smauk.org.uk/elsie-and-her-wizzybug

What can be done to help my child's breathing?

When we breathe in, muscles called **inspiratory muscles** act as bellows to expand our lungs, enabling oxygen to be pulled in. The most important inspiratory muscle is the diaphragm. SMA causes the inspiratory muscles to be weakened, resulting in a reduction in lung volume.

Breathing out the waste gas (carbon dioxide) from the lungs is known as expiration. This doesn't need particularly strong muscles, but coughing does.

If children do have problems, it's often due to having a weak cough and only being able to take in smaller breaths. Every child is affected differently, but the main problems caused by weak breathing muscles are that:

- It makes it difficult to cough and therefore clear mucous (secretions) from lungs.
- Lungs can't get rid of enough of the waste gas produced by breathing - which includes carbon dioxide. This is known as 'hypoventilation'.
- It may make it difficult to take in enough oxygen while asleep.

Regular respiratory checks

Though breathing problems occur less often than for children who are unable to sit, the SoC still recommend that children should have a physical examination at least once every six months. At this appointment, children should have their breathing checked. If they can understand how, they will use a machine called a spirometer and the strength of their cough will also be measured.

If your child has poor sleep quality, headaches or daytime sleepiness, these may be symptoms of night-time breathing difficulties in which case you would expect them to have an overnight sleep study. This may be at home with a small clip on a finger recording information, or in hospital with small sensors attached to your child's face, head, arm and chest, and monitoring them overnight with a blood test.

Weak breathing muscles are common for children, young people and adults with SMA Type 2. This can result in breathing problems which are a leading cause of health problems.

All these tests help the doctors and you decide on the best options for managing your child's day-to-day breathing.

Possible options for managing breathing

If your child has a weak cough, it's difficult for them to clear mucous and other secretions from their lungs and it makes them more vulnerable to chest infections. There are a number of options to help manage this which you can discuss with your child's respiratory specialist and medical team. Not all of them will be appropriate for your child.

Options that may be discussed include:

- **Chest physiotherapy** to keep your child comfortable and help clear secretions from their chest. You'll be advised how often is best for your child and may be trained to do it yourself.
- **Cough assist** is the name often given to a mechanical insufflator – exsufflator machine. This helps to clear secretions from your child's lungs. Your medical team will discuss if it would be helpful for your child and, if so, give you training so that you feel confident about using it. For older children it may be possible for your child to be shown how to manage to use this themselves.
- **A suction machine** to help remove your child's excess secretions. If this is needed, your physio or another member of the team would talk to you about why and when you should use it and give you training to feel confident about it.
- **Non-invasive ventilation (NIV)**, a machine with a mask providing gentle pressure to breathe against, helps to keep the lungs inflated longer. This can help your child get rid of carbon dioxide and take in more oxygen making breathing easier. This is individually fitted for your child by a respiratory specialist. When your child's breathing triggers the ventilator, it delivers a supported breath in. As they start to breathe out, the machine cycles into exhalation, allowing them to breathe out normally. There are a number of different products available; BiPAP is one example.

Even if your child's breathing is usually fine, they may struggle to deal with a chest infection compared to children without SMA. One of these options may be needed to support them whilst unwell.

You may wish to watch our 2021 video about Cough Assist for Younger Children:

www.livingwithsma.org.uk/children/breathing

If your child needs any equipment, you'll be given training so that you feel confident to use it.

You may wish to watch our 2021 video about Non-Invasive Ventilation for Younger Children:

www.livingwithsma.org.uk/children/breathing

Information from sleep studies may lead to a change in the NIV settings so that they better suit your child's needs.

- **Medication** used to breakdown secretions (mucolytics) are **not** recommended for long-term use.
- **Antibiotics** aren't recommended by the SoC as a way of trying to prevent chest infections (prophylactic use), but the medical team will have a 'low warning' threshold for when they should be started.

Support from Professionals

If your child has breathing problems, your medical team will include a respiratory specialist. Your team understands the possible impact of SMA and will get to know how it's affecting your child, so will be able to discuss the different appropriate options with you. You can also talk to them about any worries you have about managing your child's breathing and what to do in an emergency.

Air Quality

All children can be more susceptible to the effects of poor air quality, for example cigarette smoke. Children who are exposed to second-hand smoke are more likely to contract a serious respiratory infection that requires hospitalisation. Children with SMA who have respiratory challenges are especially vulnerable.

Advice on how to quit smoking is available from the Smokefree website:

www.nhs.uk/smokefree

What advice is there on eating, drinking and diet?

Due to their muscle weakness, your child may have difficulties with chewing food and find eating tiring. If they can't swallow safely, there'll be a risk they might inhale (aspirate) food which can cause choking and chest infections.

Eating difficulties can mean children don't get enough food and become underweight. However, some children can become overweight because their muscle weakness makes it difficult to exercise. Extra weight can increase the stress on muscles, bones and joints, making physical activity and breathing even more difficult.

Regular Checks

You may find that mealtimes are taking longer, or your child is having difficulties chewing and swallowing food. You'll be able to discuss any concerns like this with your medical team at your child's regular clinic checks. To help them gain a clearer picture, you'll be asked if your child has been bringing back up what they've swallowed (gastroesophageal reflux), had difficulties passing their bowel motions (constipation), or been sick (vomiting).

If your child has choking or coughing episodes when feeding or eating this may be a sign they are inhaling their food (aspirating) and should be investigated. It's not always obvious if someone is aspirating as it can be silent. If your child has frequent colds or respiratory infections this may be a sign of silent aspiration.

Guidance for Diet

A dietician should work out what diet your child needs. Their diet will be adjusted depending on if they are underweight or overweight so that they are getting the right calories, protein, fat and carbohydrates. It'll also take into account the need for the right amount of fluids and nutrients – especially calcium and vitamin D which are needed for bone health. Their glucose levels will be checked so that their diet can be corrected if the level is too high (hyperglycaemia), or too low (hypoglycaemia).

Team members who can give you advice and support on safe eating and diet include: your consultant, dietitian, speech and language therapist, OT, physio.

The SoC recommend that all children who are able to sit have their diet checked soon after diagnosis. For younger children this should then be every 3 – 6 months, then annually once older.

The test recommended to assess swallowing is a Video Fluoroscopic Swallow Study which uses a type of x-ray and swallowing of a special drink.

Due to weakness of the muscles of the bowels, it's common for children to become constipated. This can lead to discomfort and a fullness in the stomach that reduces the movement of the diaphragm which, in turn, restricts breathing and coughing. A fibre-rich diet may be recommended along with extra fluids. Your child may also be given medications to help.

It's very important generally that your child has plenty of fluids especially if they're unwell. You'll be advised about correct amounts and how often. Your medical team should be keen to restart food as soon as it is safe to do so during an illness.

Making Eating Easier

If your child has had difficulties, your team will talk to you about the best positions for eating and drinking. They may also suggest foods to avoid because they're more difficult to chew and swallow.

Children who struggle to feed themselves can dislike not being able to be in control and having to depend on others. Long mealtimes due to eating difficulties can also put pressure on other family members and activities. Your physio and OT will be able to suggest positioning and seating options, and orthotic devices that will help your child eat more independently.

Currently, professional opinion is divided about whether the Amino Acid diet, which is based on an elemental formula, is beneficial. Professionals do agree that the type of diet and how it's given should be based on each child's individual reactions and should be regularly reviewed.

Pureed food or a semi-solid diet can help with chewing difficulties and can reduce the length of mealtimes. Thicker liquids such as milkshakes might help avoid aspiration.

Families have found some of this equipment helpful:

- Valved straws which reduce the effort of drinking by keeping the liquid at the top of the straw
- Elbow supports
- The Neater mobile arm supports or eating / feeding aid which enables people to use their own movements to feed themselves: www.neater.co.uk
Other companies are available.

If Eating Gets More Difficult

If your child is no longer swallowing safely and / or not gaining enough weight, your team may suggest additional ways for them to take in enough food safely. You should be given time to discuss and ask questions about the reasons for any suggestions so that you understand the possible benefits and risks for your child.

Additional short-term options may include feeding through a:

- **Nasogastric (NG) tube** - a thin flexible feeding tube passed through the nose into the stomach
- **Nasojejunal (NJ) tube** - through the nose into the middle part of the small intestine (the jejunum)

A longer-term option could be a:

- **Gastrostomy (PEG) tube** - placed in the stomach via a surgical procedure and also called a PEG - percutaneous endoscopic gastrostomy. Children who have a PEG tube to help them gain weight are usually encouraged to eat some food by mouth.

Whichever option is decided on, you'll be provided with training and support so that you can feed your child safely at home.

Looking After Teeth

Because of their muscle weakness, it may be difficult for your child to open their mouth wide. This can cause problems with eating and with teeth cleaning and dental care. Regular dental check-ups are important, and your team can suggest ways to manage difficulties and help prevent complications.

How can I be prepared for an emergency?

Your child's team should work with you to develop an emergency health plan (EHP). This records the treatment you wish your child to receive if there is an emergency or if their health deteriorates. Although they can be difficult, these discussions are important and should be had before a child is unwell. The team will discuss possible respiratory problems and the different ways that these can be managed.

The plan can be reviewed, and you can change your mind about the contents of the EHP at any time.

With your permission, this plan can be shared with professionals supporting your child, including ambulance services, so that everyone is aware of your wishes. You should have your own copy of your child's EHP so that you can give it to hospital services if you're away from your home area.

It's important that you know how to access medical care in case your child has a respiratory emergency. Your medical team can arrange open access to your local children's ward.

You can upload a copy of your child's latest EHP to your mobile using our app. Find out more: <https://ehpapp.com>

What about 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 visit the website: www.gov.uk

There are also a number of charities that may assist you with the cost of general household goods, specialist equipment and holidays or days out. Please contact SMA UK for more information or see the Living With SMA area of our website (more details at the end of the booklet).

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

What about travel by car?

Your child may be able to use a standard car seat, but this may be too upright for them. If they're uncomfortable in their car seat, you can ask your physio or OT for their opinion of any suitable alternatives that provide recline options and enough support. A small neck support and additional head support may also be useful.

The **Blue Badge scheme** is run by local authorities. This entitles people with severe mobility problems to parking concessions. Although the minimum age for Blue Badge applications is 3 years, in certain circumstances it's possible to apply for children under 3 years old with medical conditions which require them to be accompanied by bulky equipment, or if they need to remain near their vehicle in order to access treatment. To find out more about these special circumstances and how to apply for a Blue Badge, visit the GOV.UK site: www.gov.uk/blue-badge-scheme-information-council

If you find you need a **wheelchair accessible vehicle (WAV)**, this can be leased through the Motability Scheme if a child is over 3 years old and receives the enhanced (higher) rate mobility component of Disability Living Allowance. Currently this option is not available to children under the age of 3, so some parents purchase their own vehicle.

This is such a lot to take in and manage, what other help can I get?

Though you may feel some relief that you now have a diagnosis, you may well also be feeling shock, disbelief, confusion, anger and sadness. You may find it difficult to take everything or anything in.

Everyone's different, but it's important that you and your family have access to emotional support and plenty of time to talk and ask questions. This can be with members of your child's medical team, your local GP, health visitor, social worker, psychologist or a counsellor. Family and friends, spiritual leaders, support groups, or online communities can also be helpful to talk to.

Even though you'll be focused on your child, try and look after yourself too. Things like remembering to keep up-to-date with your own health checks and getting the 'flu' jab are important.

SMA UK can also help with information on car seats and possible sources of funding.

If your child can't use a car seat for medical reasons, you can contact your GP for a certificate which will explain why – this is a '**certificate of exemption from compulsory seatbelt wearing**'. You'll need to carry this document at all times in your car. For more information, please see: www.gov.uk/seat-belts-law

The 24 hour-a-day responsibility of caring for a child with potentially complex needs can be physically, emotionally and psychologically exhausting.

In some areas, local children's hospice services may be able to support you. They may offer short breaks at your home or at the hospice. Some local authorities also have residential short breaks provision for children with disabilities. Ask your GP, community nurse, health visitor or social worker for more information.

Over time, so that your child can participate fully in activities, you may need further information and support on mobility, education, equipment and sources of funding. You can find out more by talking to your child's healthcare team, SMA UK's Support Services Team, and the other people and organisations listed at the end of this booklet.

Perhaps ask friends and family to help out with practical things such as shopping and cleaning which can help save you some time and energy.

Support and Resources

The International Standards of Care for Spinal Muscular Atrophy (2017) can be read / downloaded from here: www.smauk.org.uk/international-standards-of-care-for-sma

SMA UK

Phone: 01789 267 520

Email: supportservices@smauk.org.uk

Website: www.smauk.org.uk

We provide a free **Support & Outreach Service** for families by email, phone, text and outreach home-visiting. Our experienced team offer personalised support and information and are available to answer questions and talk things through. Though we don't give medical advice, we can discuss with you the support you and your family can access.

You can upload a copy of your child's latest **emergency health plan** using our **EHP app** which follows the NHS guidelines for data privacy. It's free to download on Android and iOS. You can find out more at: <https://ehpapp.com>

If you're wondering about an aspect of life with SMA, we hope the **Living With SMA** area of our website will be a helpful starting point, giving you useful information and ideas. It builds on knowledge and advice from the SMA Community and SMA UK's Support Services Team, and covers a whole host of topics, including: health & wellbeing, equipment, homes, education, work, transport, leisure, holidays, financial, and emotional & social support: www.livingwithsma.org.uk

Any family affected by SMA, living in the UK, can request a free copy of the following books:

- **Smasheroo** - written by the mother of a child who has SMA, with illustrations by Mary Hall, this uplifting story highlights that everyone is different and everyone is special in their own way.
- **SMA Type 2 and Me** - an illustrated book published in 2016 for children aged 8 – 12 years, but it could also be read to a younger child. As of October 2019, it remains factually correct and may still be useful. We will not be updating this book and instead are building new resources in the Living With SMA area of our website: www.livingwithsma.org.uk/children/resources-for-parents-health-wellbeing

You'll find more information **about nusinersen** and what is happening in the UK here: www.smauk.org.uk/nusinersen Our information sheet '**Nusinersen (Spinraza™) treatment for those diagnosed with SMA Type 2 or 3**' can be read / downloaded from here: www.smauk.org.uk/treatment-information-leaflets

This website section tells you **about other research developments**: www.smauk.org.uk/drug-treatments-screening-whats-happening-now You may also find our information sheet '**How Clinical Trials Work**' useful: www.smauk.org.uk/clinical-trials

Muscular Dystrophy UK

Phone: 0800 652 6352

Website: www.muscular dystrophyuk.org

MDUK provide information, support, advocacy services and grants towards specialist equipment for people affected by a range of neuromuscular conditions. They also have condition specific 'alert cards' which can be used to provide medical professionals with information. A link to their alert card for SMA Type 2 can be found at: www.muscular dystrophyuk.org/about-muscle-wasting-conditions/information-factsheets/conditions/alert-cards-and-care-plans/alert-cards/

Contact for Families with Disabled Children

Phone: 0808 808 3555

Website: www.contact.org.uk

Contact provide information and support to families who have a child with a disability, including information on benefits and grants.

Children's hospices

Phone: 'Together for Short Lives' on 0808 8088 100

Website: www.togetherforshortlives.org.uk

Located throughout the UK, these 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 and more information is available on their website.

Online communities

These have been set up by people in the SMA Community. If you're interested in any of the support groups offered, please contact the group directly. You can find details of some of them here: www.smauk.org.uk/online-sma-communities



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This information booklet is one of our 'Looking After...' series, which was highly commended in the 2019 British Medical Association Patient Information Awards.

Patient information awards

Highly commended

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We are grateful to the writers and reviewers who assist us in our information production. A list of who this includes may be requested from office@smauk.org.uk

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If you have any feedback about this information, please do let us know at:

office@smauk.org.uk

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