A Guide to:

Symptoms, diagnosis & effects of 5q Spinal Muscular Atrophy

Many people – families, friends and professionals who haven’t come across SMA before – will want to know more about its symptoms, how it’s diagnosed and what impact it may have. This guide explores these questions for the most common form of SMA, known as 5q SMA due to its genetic cause. 5q SMA includes the different ‘types’ or clinical classifications – Types 1, 2, 3 and 4.

For more information about the causes and genetics of 5q SMA please see:
- ‘What is Spinal Muscular Atrophy’.
- ‘The Genetics of 5q SMA’

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

Includes

SMA Type 1
SMA Type 2
SMA Type 3
SMA Type 4

Muscular Dystrophy UK thanks Spinal Muscular Atrophy Support UK (SMA Support UK), a certified member of The Information Standard, who have produced this information sheet. Muscular Dystrophy UK works closely with SMA Support UK and endorses the information provided here.
It’s important as you read this guide to remember that:

➢ Each child and adult is affected differently

➢ Although SMA is clinically classified into different ‘Types’ which reflect the severity of its impact, it is considered a spectrum

➢ For children and adults, the severity of the condition varies from person to person, both within and between ‘Types’

➢ Although there is currently no cure for SMA, this doesn’t mean that nothing can be done. There are a range of options aimed at managing symptoms, reducing complications of muscle weakness and maintaining the best quality of life. These are all covered in the International Standards of Care for SMA\(^1,2\).

➢ There are also promising drug treatments emerging

How is 5q SMA diagnosed?

Any child or adult with suspected SMA will be physically examined. This may be by their GP, paediatrician, neurologist or neurological specialist who will ask about their medical history and concerns. A GP may have met few children or adults with SMA so may make an immediate referral to a specialist neuromuscular centre. Once SMA is suspected, a blood sample for DNA testing will be arranged. The blood sample is tested for a deletion mutation in the *Survival Motor Neuron 1 (SMN1)* gene on chromosome 5. It is also now recommended\(^1\) that the number of *SMN2* copies is also assessed as this can be a helpful indicator of what effects the condition will have. Clinical trials of new treatments often have entry criteria that specify the number of *SMN2* copies someone must have to be eligible to take part. For more information on this, see: ‘What is Spinal Muscular Atrophy’.

The *SMN1* deletion test result is usually available within 2 – 4 weeks. Other tests may take longer.

If there’s any uncertainty about the diagnosis, further tests such as an electromyogram (EMG) which records the electrical activity of muscles may be discussed, but this isn’t usually needed to confirm 5q SMA.

Parents / carers will often have been concerned about symptoms of weakness in their child. Symptoms may have been noticed by doctors or the health visitor or community nurse.

Adults who are later diagnosed with SMA Type 4 may have had concerns about their muscle weakness or fatigue.
Overview of 5q SMA clinical classification of ‘Types’

There is a wide spectrum of how severely children, young people and adults are affected, both within and between ‘Types’. Broadly, though, they are as follows:

<table>
<thead>
<tr>
<th>SMA Type</th>
<th>Usual age of symptoms</th>
<th>Impact of muscle weakness on sitting / walking</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type 1</td>
<td>Younger than 6 months</td>
<td>Unable to sit or roll independently</td>
</tr>
<tr>
<td>Type 2</td>
<td>6 – 18 months</td>
<td>Able to sit but not walk independently</td>
</tr>
<tr>
<td>Type 3a</td>
<td>18 months – 3 years</td>
<td>Able to walk, though may lose this ability over time</td>
</tr>
<tr>
<td>Type 3b</td>
<td>3 years – 18 years</td>
<td>Able to walk, though may lose this ability over time. Difficulties usually appear later than for children who develop symptoms earlier.</td>
</tr>
<tr>
<td>Type 4</td>
<td>Over 18 years</td>
<td>Mild walking difficulties</td>
</tr>
</tbody>
</table>

Adapted from Tillmann et al. 2018²
SMA Type 1

The symptoms and effects of SMA Type 1 usually begin from birth or within the first few weeks or months of life. Generally, the earlier the onset of symptoms, the more severe the condition.

Each child is affected differently, but in general, babies with early onset SMA are:

- bright, alert and responsive; their intelligence isn’t affected
- able to smile and frown as their facial muscles aren’t severely affected
- often described as ‘floppy’ babies due to their low muscle tone (hypotonia) and severe muscle weakness.
- unable to support or lift their head due to their weak neck muscles
- unable to sit unsupported and have difficulty rolling over
- able to move their hands and fingers but have difficulty lifting their arms and legs

They have:

- breathing muscle weakness, which can cause a weak cry and difficulties with breathing and coughing
- an increased chance of chest infections, which can be life-threatening
- difficulty swallowing their saliva and other secretions, which may make them sound chesty or make them cough
- difficulties feeding and gaining weight
- an increased risk of fluids or food passing into their lungs (aspiration), which can cause choking and, sometimes, chest infections or pneumonia

It’s not possible to predict life expectancy accurately but for most children, without intervention for breathing difficulties, this has previously been estimated as less than two years. Evidence suggests that since the International Standards of Care for SMA introduced more proactive managements in 2007, children have been living longer.
SMA Type 2

The symptoms and effects of SMA Type 2 usually begin between 6 and 18 months of age. Generally, the earlier the onset of symptoms, the more severe the condition.

Each child is affected differently, but in general, children with SMA Type 2 are usually bright and engaging. However, due to SMA, they are likely to experience:

- muscle weakness on both sides of their body
- muscle weakness closest to the centre of their body as these muscles are more severely affected than muscles furthest away
- difficulties moving their arms, but their hands and fingers less so
- difficulties lifting their legs
- legs that are weaker than their arms

As they get older, their intellectual and sexual development isn’t affected but SMA usually causes them:

- muscle weakness that may make it difficult for them to keep up with their daily activities. For example, if they have been able to crawl or roll, they may lose this ability
- a tendency to become weaker after infections and at times of major growth spurts such as puberty
- weak breathing muscles, making it difficult for them to cough effectively and more vulnerable to chest infections
- muscles supporting the spinal column that are weak meaning that most children will develop a sideways curvature of their spine (scoliosis)
- reduced ability to move so that some joints may become tight (contractures) and further restrict their range of movement

Children and adults will need help with daily tasks like washing, dressing and undressing. Though their bladder and bowel control isn’t usually affected, they will need help transferring from their wheelchair to the toilet.

SMA Type 2 can weaken chewing and swallowing muscles. For some children, their tongue and shoulder muscles may twitch and they may have a slight tremor in their hands.

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

Please see our guide for parents and carers:

‘Looking after your child with SMA who has had a recent diagnosis of SMA Type 2’

Our website tells you more about what stage emerging treatments have reached:

www.smasupportuk.org.uk/drug-treatments-whats-happening-now
SMA Type 3

The symptoms and effects of SMA Type 3a usually begin between 18 months and 3 years.

The symptoms and effects of SMA Type 3b usually begin after 3 years, but before adulthood.

Each child is affected differently, but in general, children with SMA Type 3 are bright and engaging. However, their SMA causes:

- muscle weakness on both sides of their body
- muscle weakness closest to the centre of their body as these muscles are more severely affected than muscles furthest away
- legs that are weaker than arms

As they get older, their intellectual and sexual development isn’t affected, but their SMA usually causes them to have:

- difficulties with standing and walking. This usually happens later for children with SMA Type 3b than for children who develop the first symptoms at an earlier age
- difficulties keeping up with daily activities. For example, if they have been able to walk or climb stairs, they may lose this ability. Some children may fall more easily because of their muscle weakness. If they’re sitting on the floor they may need help to get up
- muscles supporting the spinal column that are weakened. This means that some children develop a sideways curvature of their spine (scoliosis)
- a reduced ability to move due to some joints becoming tight (contractures), restricting their range of movement.
- a tendency to become weaker after infections and at times of major growth such as puberty.

Some children, young people and adults will need help with daily tasks like washing, dressing and undressing. Though their bladder and bowel control isn’t affected, some may need help getting to and sitting on the toilet.

Most people with SMA Type 3 don’t have breathing problems and their life expectancy isn’t affected. Most can live long, fulfilling lives.

Please see our guide for parents and carers:

‘Looking after your child with SMA who has had a recent diagnosis of SMA Type 3’

Our website tells you more about what stage emerging treatments have reached:

www.smasupportuk.org.uk/drug-treatments-whats-happening-now
SMA Type 4

The symptoms and effects of SMA Type 4 begin in adulthood.

Each person is affected differently, but in general, symptoms can include:

- tired, aching muscles
- a feeling of heaviness
- numbness
- cramp
- a slight shaking of the fingers and hands
- fatigue

SMA Type 4 progresses steadily and slowly over time causing increased muscle weakness with age. This may impact on daily living activities such as walking, dressing and bathing.

SMA Type 4:

- rarely affects swallowing or breathing
- doesn’t affect intelligence, and life expectancy is normal.

It’s important not to confuse SMA Type 4, which affects the lower motor neurons, with Motor Neurone Disease (MND) - also known as Amyotrophic Lateral Sclerosis (ALS). MND affects both the upper and lower motor neurons. Though SMA is also classed as a motor neuron disease, it isn’t MND. Whereas MND is almost always life-threatening, SMA Type 4 isn’t.

Please see our guide:

‘Looking after yourself: adults who have had a recent diagnosis of SMA’

Our website tells you more about what stage emerging treatments have reached:

www.smasupportuk.org.uk/drug-treatments-whats-happening-now
Resources and support

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

SMA Support UK

Phone: 01789 267 520
Email: supportservices@smasupportuk.org.uk
Website: www.smasupportuk.org.uk

We provide free information and support to families in the UK affected by SMA. Our outreach workers are able to visit you at home. They offer personalised support and information and are available to answer questions. They can discuss with you the support you and your family can access. Please note, we don’t give medical advice.

Our Route Maps for SMA have other information about day to day life with SMA and signpost to possible sources of support and advice. At the moment they are organised according to Type of SMA. You can find these at: www.routemapforsma.org.uk

We are currently re-organising this information so that it’s more accessible. It will be located on a new part of our website which we will call ‘Living with SMA’. As soon as it’s ready we’ll let people know via our monthly e-news.

Muscular Dystrophy UK

Phone: 0800 652 6352 from 8.30am to 6pm, Monday to Friday
Email: info@musculardystrophyuk.org
Website: www.musculardystrophyuk.org

MDUK provide information and emotional support, advocacy services and grants towards specialist equipment for people affected by a range of neuromuscular conditions, including SMA. If they can’t help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

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References


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.smasureportuk.org.uk/our-writers-and-reviewers-panel or requested from supportservices@smasureportuk.org.uk

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