5Q SPINAL MUSCULAR ATROPHY: INFORMATION FOR SCHOOLS

Who this is for

This summary is for schools where there is already a child who has Spinal Muscular Atrophy (SMA,) and schools where parents are thinking of applying for a place. It aims to give a brief overview of this rare health condition.

What is SMA?

Spinal Muscular Atrophy (SMA) is a rare, neuromuscular condition. It causes progressive muscle wasting (atrophy) and weakness. It may affect crawling and walking ability, arm, hand, head and neck movement, breathing and swallowing. The most common form is **5q SMA**.

Types of 5q SMA

Before drug treatments (also known as 'disease-modifying' treatments) became available for 5q SMA, clinicians and researchers gathered information about what impact someone with SMA might expect the condition to have on them.

This is called the **'natural history'** of the condition.

This led to 5q SMA being divided into four main types of SMA: Types 1, 2, 3, and 4.

These 'Types' of SMA were based on the age that symptoms began, and what physical milestone (e.g. sitting, standing, walking) could be achieved. It was agreed that there could be variation both within and between 'Types.'

SMA Type	Age symptoms usually begin	Motor milestones
Type 1	0-6 months	Unable to sit or roll independently
Type 2	7-18 months	Able to sit but not walk independently
Type 3	18 months – 18 years	Able to walk though may lose this ability over time
Type 4	18 years +	Mild walking difficulties

This classification system is still used for adults, teenagers and children living with SMA in the UK even though for many, care and treatment is changing the outcome of their SMA.

Does 5q SMA just affect motor milestones?

Depending on the severity of the condition, SMA may affect a child's:

- Arm and hand movement
- Ability to eat and drink independently and safely
- Breathing ability – making them more vulnerable to respiratory infections
- Ability to communicate in a way that can be easily understood
- Bones and joints

SMA doesn't usually affect a child's intelligence or thinking.

The child who has SMA who is (or will be) at your school, along with their parent(s) / guardian(s), and any supporting health, education and social care professionals, are the best people to tell you about the impact of their SMA. They will also be able to tell you what support and equipment the child needs so that they can be fully included in every learning, social and playing opportunity.

What causes 5q SMA?

Most people have two genes called the ***Survival Motor Neuron 1 (SMN1)*** gene. This produces Survival Motor Neuron (SMN) protein which keeps lower motor neuron nerve cells healthy. These are essential for activating muscles used for crawling and walking, the movement of arms, hands, head and neck, as well as breathing and swallowing.

Most people have two healthy copies of the *SMN1* gene. People who have 5q SMA have two 'altered' copies of the SMN1 gene. This means they are unable to produce enough SMN protein to have healthy lower motor neurons.

A second gene also produces SMN protein. This is the ***Survival Motor Neuron 2 (SMN2)*** gene. It is sometimes referred to as the SMA 'back-up' gene. Although this gene makes some functional SMN protein, it cannot fully make up for the altered *SMN1* genes in people who have 5q SMA.

People can have between 0 – 8 copies of the *SMN2* gene (*SMN2* copy numbers). Having more *SMN2* copies is generally associated with less severe SMA symptoms. However, the *SMN2* copy number alone cannot accurately predict the Type of 5q SMA or the severity of its impact.

Is there a cure or treatment?

There is no cure for SMA, but there are now three NHS-funded drug treatments. These are changing what motor milestones babies and children may be able to achieve and improving their general health. These drugs must be given as early as possible to maximise the potential benefits.

Children who are already in school will almost all be receiving one of these ongoing treatments:

- **nusinersen(Spinraza™)**
- **risdiplam (Evrysdi™)**

In July 2021, a 'one-off' gene therapy became available in the UK for children who had been diagnosed with SMA Type 1:

- **onasemnogene abeparvovec (Zolgensma™)**

For some children who had previously been receiving one of the ongoing treatments, this was stopped and instead they received the gene therapy.

All these treatments continue to work as a child grows and develops. What the outcomes are, or will be, will vary for each individual child. It will depend on factors such as how early treatment was started and how severe the impact of a child's SMA was at the time. The appropriate, ongoing, individualised health management and care is also critical.

How rare is it for a child to have SMA?

Every month in the UK, 4 babies are born with 5q SMA.
5q SMA affects an estimated 1 in 14,000 births. In 2022, this would have been around 48 babies in the UK who were born with 5q SMA, of which some 60% (29 babies) would have the more severe SMA Type 1.

In mid-2022 there were up to an estimated 1,350 people living with 5q SMA in the UK.
Between 1 and 2 people in every 100,000worldwide have a Type of SMA. In mid-2022 this would have meant there were up to an estimated 1,350 people living with SMA in England, Scotland, Wales and Northern Ireland. As there is no central source of information, exact numbers are unknown.

Although SMA is a rare condition, an estimated 1 in 40 people carry the altered gene.
That's around 1.69 million people in the UK.
When two carriers have a child, for each and every pregnancy there is a one in four chance that the child will have SMA.

Further Information and Support

You can find further information on the SMA UK website:

- About SMA: smauk.org.uk/support-information/about-sma
- About life with SMA: smauk.org.uk/living-with-sma

Our Support Team may be contacted by any family affected by SMA. We are also happy to be contacted by schools seeking support: smauk.org.uk/support-team

You will find details of other organisations offering support and information in the 'Getting the Right Support' part of our Schools Pack.

Trusted Information Creator
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The information provided in this guide, on our website, and through links to other websites, is designed to complement not be a substitute for clinical and professional care and advice.

For more detail about how we produce our information, please see: smauk.org.uk/support-information/our-information-service

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

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