Spinal Muscular Atrophy (SMA)
A Brief Summary

You can also read this guide on our website at smauk.org.uk/baww where you can follow all the links we give to further information.

This information tells you more about:

- What is SMA?
- Types of 5q SMA
- What causes 5q SMA?
- SMA: The Numbers
- Drug treatments
- Rarer Forms of SMA

1. What is SMA?

Spinal Muscular Atrophy 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. How severely people are affected, and in what way, varies greatly.

There are different forms of SMA with different genetic causes. The most common form is called **'5q SMA'**.

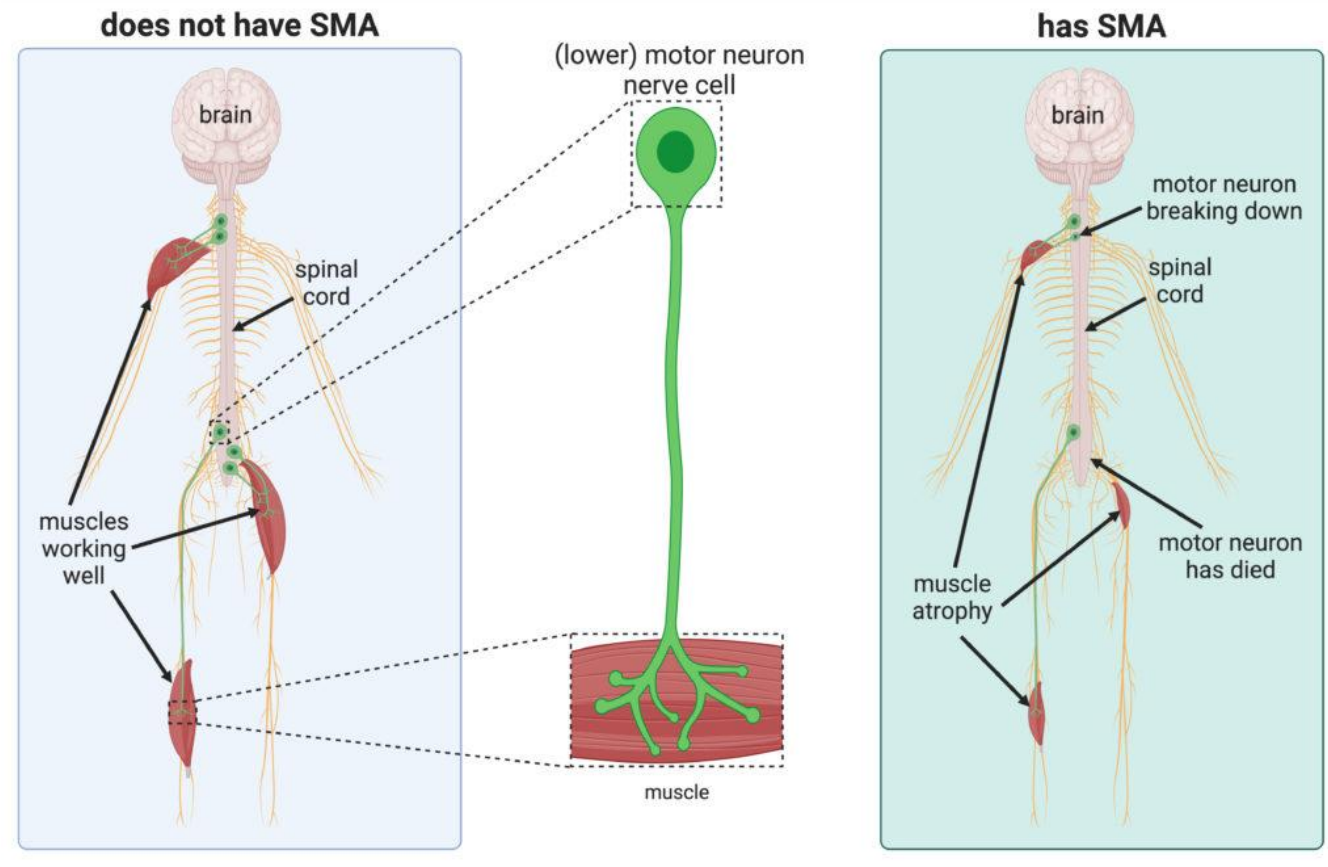
2. What causes 5q SMA?

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 nerves 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.

As a result, the lower motor neurons in the spinal cord deteriorate.

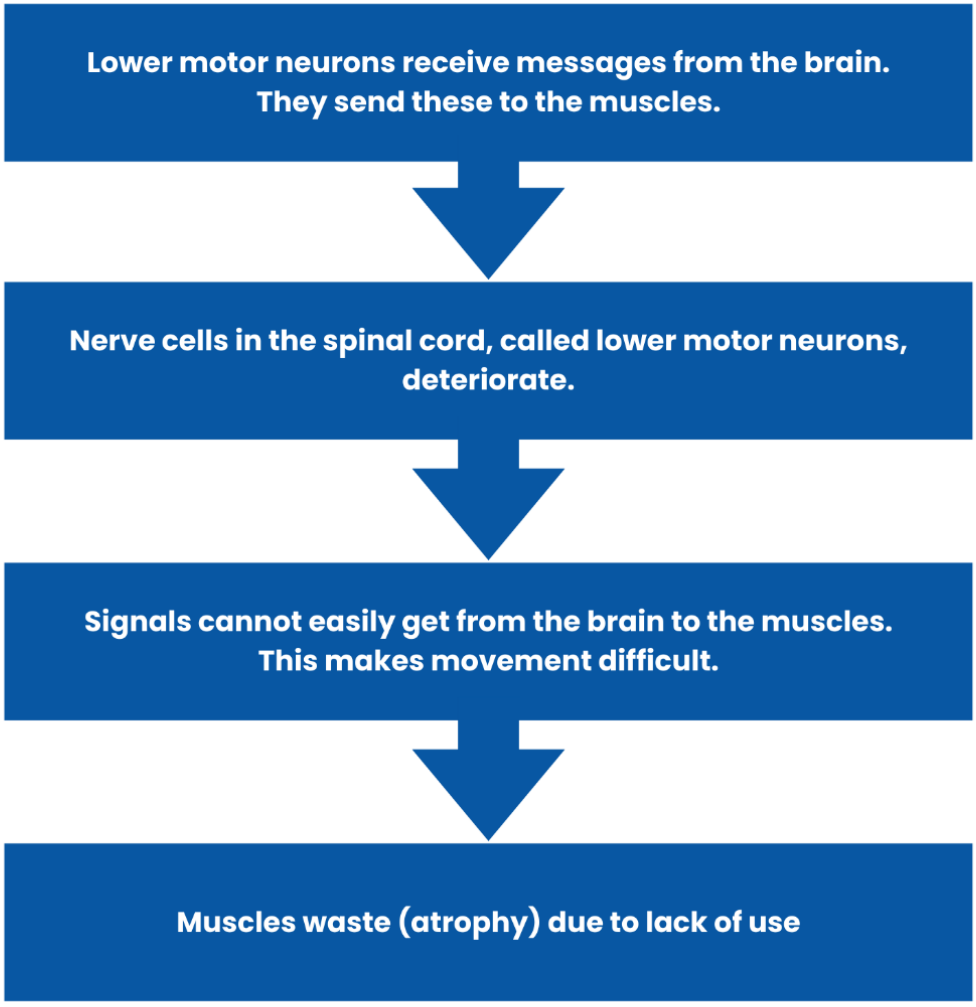


Adapted from Sleigh et al. (2023). Figure adapted with permission from Reference 1 under the CC BY 4.0 licence

This means that signals are not effectively carried from the brain to the muscles. This makes movement difficult.

The muscles then waste due to a lack of use — this is known as muscular atrophy.

In summary:



A second gene also produces SMN protein. This is the *SMN2* gene. It is sometimes referred to as the SMA 'back-up' gene.

Only some of the SMN protein the *SMN2* gene makes works properly. The *SMN2* gene cannot fully make up for the altered *SMN1* genes in people who have 5q SMA.

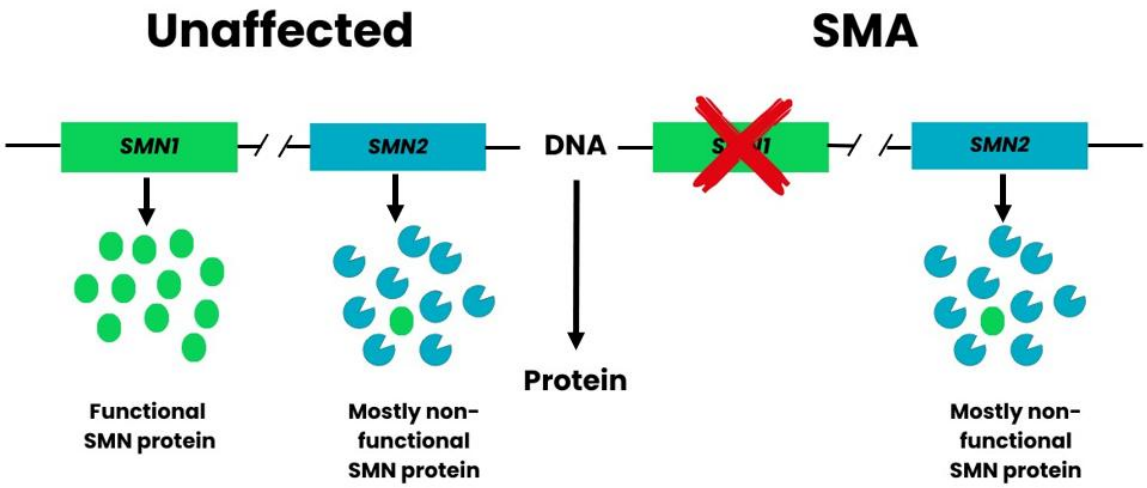


Figure adapted from Burghes, A.H. and Beattie, C.E[3]

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, a person's *SMN2* copy number does not fully predict their SMA Type or severity.

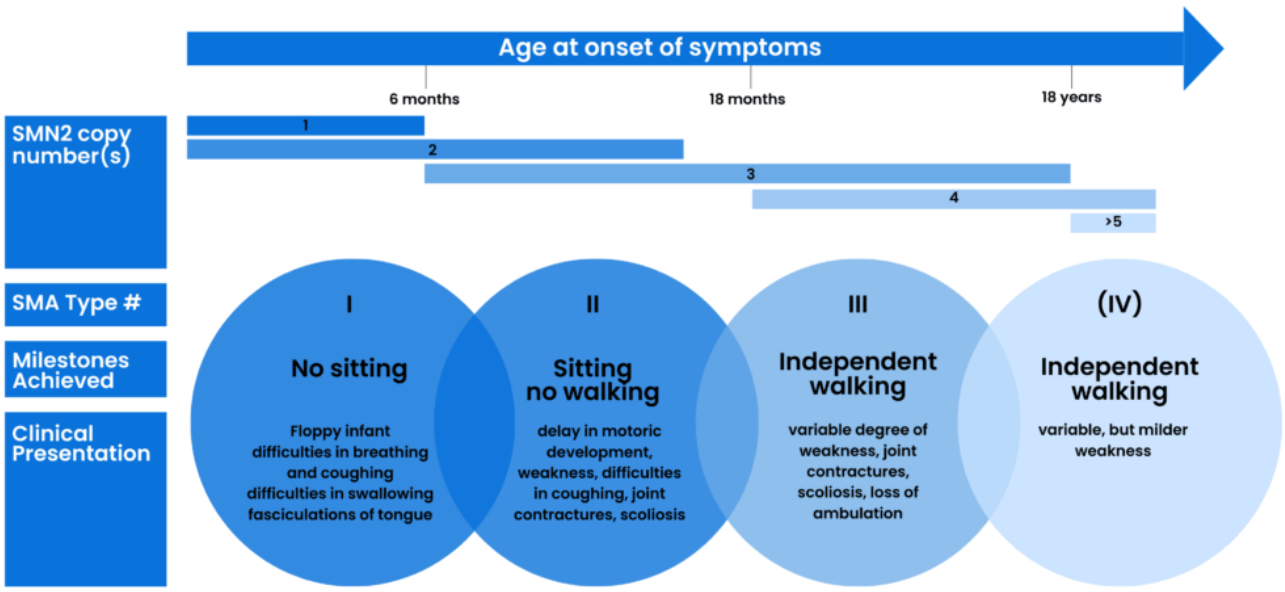
For more information see: What is 5q SMA? >.

3. Types of SMA

Before the new drugs for 5q SMA were developed, clinicians studied the effects of SMA on people. 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. Sometimes a baby is affected before birth; this is called Type 0.

These 'Types' of SMA are based on the age that symptoms begin, and what physical milestones (e.g. sitting, standing, walking) are achieved. How severe and what impact SMA has varies from person to person, both within and between 'Types'. Each child and adult is affected differently.



Clinical classification of SMA subtypes according to onset, milestones achieved, and clinical presentation. Typically associated *SMN2* copy numbers are shown. Figure taken with permission from Reference 2 under the CC BY-NC 4.0 licence .

These clinical classifications are still used by doctors for adults, teenagers and children living with SMA. For many, though, care and treatment are changing the outcome of their SMA.

For more information, see: Symptoms and Effects of 5q SMA >

4. SMA: The Numbers

- **Every month in the UK, 4 babies are born with 5q SMA.**

Recent studies suggest that 5q SMA affects an estimated 1 in 14,000 births. In 2023, this would mean that:

- around 47 babies in the UK were born with 5q SMA.
- approximately 28 of these babies (60%) would have the more severe SMA Type 1.

Between 1st January 2019 and 1st November 2024, 507 children in the UK were registered in the SMA Research and Clinical Hub (REACH) UK database. Of these

- **213 [42%] were diagnosed with SMA type 1**
- **188 [37%] were diagnosed with SMA type 2**
- **106 [21%] were diagnosed with SMA type 3.**

This suggests that approximately 85 babies with a type of 5q-linked SMA are born each year in the UK. This is approximately one baby born every 4 days.

Further UK population research will allow a more accurate measure of SMA incidence in the UK.

- **In 2023 there were up to an estimated 1,365 people living with 5q SMA in the UK.**

As there is no central source of information, exact numbers are unknown. Worldwide between 1 and 2 people in every 100,000 have 5q SMA.

- **Although SMA is a rare condition, an estimated 1 in 40 people carry the altered gene. That's around 1.7 million people in the UK.**

When two 'carriers' of the altered gene have a child, for **each and every pregnancy** there is a one in four chance that the child will have SMA.

For more information see: What is 5q SMA? >

5. Drug Treatments for SMA

There is no cure for SMA and until late 2019 there were no NHS-approved drug treatments specifically for SMA in the UK. There are now three NHS-funded drug treatments:

- **Spinraza™ / nusinersen >**
- **Evrysdi™ / risdiplam >**
- **Zolgensma™ / onasemnogene abeparvovec >**

These treatments are not suitable for everyone who has 5q SMA, but most people who have SMA Type 1, 2 or 3 can receive one of them.

These drugs and better care and management of the condition can change what motor milestones (e.g. the ability to sit, stand and walk) babies and children may be able to achieve, and improve their general health.

The treatments work best if started before there is any muscle weakness, or when this is minimal. It is therefore important for treatment to be started as soon as possible.

This is why clinicians and patient groups are calling for the earliest possible introduction of **Newborn Screening for SMA >** in the UK.

For adults living with SMA, drug treatment that can stabilise the condition later in life may also make a positive difference – for example, helping with fatigue or preventing the loss of the ability to use a finger to control a powerchair or laptop.

6. Rarer Forms of SMA

Much more is known about 5q SMA than the other very rare forms of SMA which have different genetic causes and inheritance patterns. For information about these conditions, including what is known about their symptoms and causes, please see **Rarer Forms of SMA >.**

7. References

1. Sleigh J et al. (2023) Spinal Muscular Atrophy: A Rare but Treatable Disease of the Nervous System. Frontiers for Young Minds 11:1023423 Under the CC BY 4.0 licence >.

2. Schorling D *et al.* (2019) Advances in Treatment of Spinal Muscular Atrophy – New Phenotypes, New Challenges, New Implications for Care > *J Neuromuscul Dis* 7: 1-13, under the CC BY-NC 4.0 licence >.

3. Burghes, A.H. and Beattie, C.E. (2009) 'Spinal muscular atrophy: why do low levels of survival motor neuron protein make motor neurons sick?' *Nature Reviews Neuroscience*, 10, pp. 597-609.

For more detailed information and a full list of references please see: What is 5q SMA? >

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