Symptoms & Effects of 5q Spinal Muscular Atrophy – Type 4

You can also read this guide on our website at smauk.org.uk/mr4v where you can follow all the links we give to further information.

This is for families, friends and healthcare professionals who want to know more about SMA Type 4.

SMA Type 4 is a form of 5q SMA. Please see our guide: **What is 5q SMA?** for information about causes, diagnosis, inheritance and how many people are affected.

This information sheet tells you more about:

- **The Natural History of SMA**
- **Symptoms and effects of SMA Type 4**
- **Supportive care and management**
- **Related information**

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1. The 'Natural History' 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.

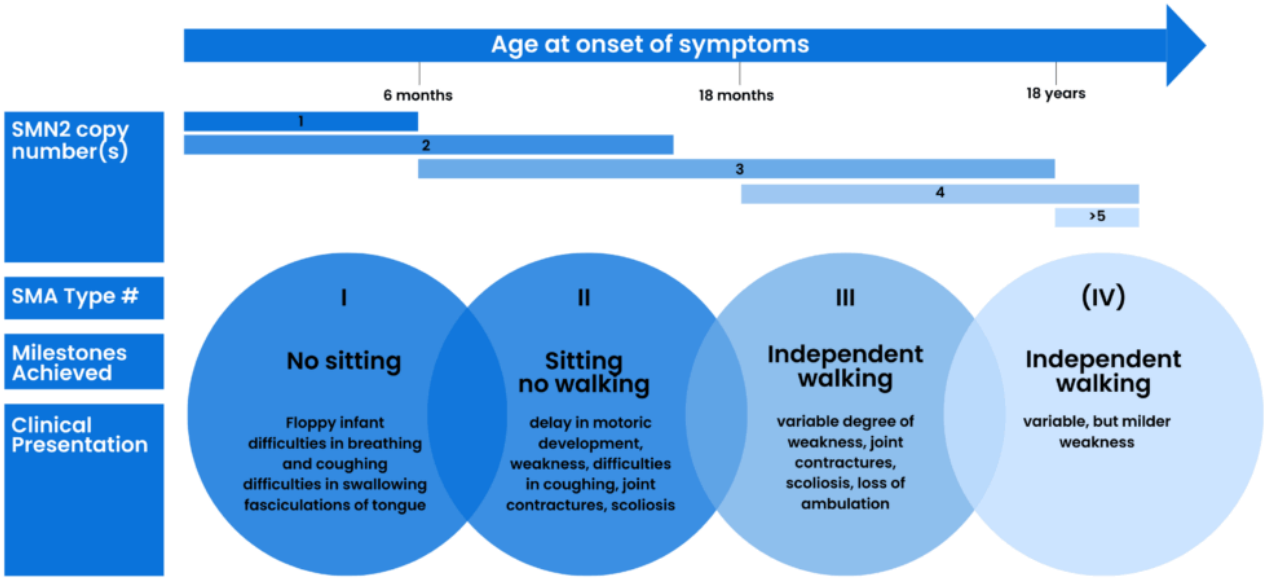
These ‘Types’ of SMA are based on the age that symptoms begin, and what physical milestones (e.g. sitting, standing, walking) are achieved. Clinicians agreed that there could be variation both within and between ‘Types’.

A specialist clinician will examine any child or adult with suspected SMA.

They will also consider:

- the age of the child or adult when the *SMN1* gene deletion test confirmed SMA
- any symptoms of SMA and when these first started
- how many *SMN2* gene copies the test result showed
- any family history of SMA.

They will refer to this summary:



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 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.

2. Symptoms and Effects of 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 usually progresses slowly over time. It causes 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
- does not affect intelligence
- life expectancy is not affected[1].

SMA Type 4 affects the lower motor neurons. This is different from Motor Neurone Disease (MND), also known as Amyotrophic Lateral Sclerosis (ALS) which affects both the upper and lower motor neurons. Though SMA is also classed as a 'motor neuron disease', it is not MND. Whereas MND is almost always life-limiting, SMA Type 4 is not.

3. Supportive Care and Management

Access to treatments for those clinically diagnosed with SMA Type 4 varies between countries. This may depend on the decision made by the regulatory authorities. For example, in some countries access is approved for all who have 5q SMA. It may also depend on how distinct the 'onset of symptoms' age boundary is considered to be between Type 3b and Type 4.

In the UK, access to NHS-funded drug treatment is only possible for those who have a clinical diagnosis of SMA Type 1, 2 or 3 and who meet the eligibility criteria. No drug treatments are currently available in the UK for people who have been given the clinical classification diagnosis SMA Type 4. Discussions leading to this diagnosis should include a careful review of the history of the person's onset of symptoms.

In 2024 / 5, the National Institute of Health and Care Excellence (NICE) is reviewing whether the NHS in England will continue to fund the two drug treatments, nusinersen and risdiplam. SMA UK is advocating for adults diagnosed with SMA Type 4 to have access to drug treatment. You can read more about the review >.

Support and advice can help manage the symptoms and effects of SMA Type 4. Nationally and internationally expert clinicians, researchers and people living with SMA are working together to review and update guidelines for best supportive care and management of symptoms.

- See: **SMA Care UK >**

It is important that you feel able to discuss any questions you have with your clinical team.

4. Related Information

- **Looking After Yourself If You Have Adult Onset SMA >**
- **Living With SMA – information and ideas for daily living >**
- **The Genetics of 5q SMA >**

5. References

1. Lunn MR & Wang CH (2008) Spinal muscular atrophy. *Lancet, 371*: 2120-2133

Version 5

Author: SMA UK Information Production Team

Last updated: June 2025

Next full review due: June 2028

Links last checked: June 2025.

This publication, and its links, provides information. This is meant to support, not replace, clinical and professional care.

Find out more about how we produce our information .

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