Symptoms & Effects of 5q Spinal Muscular Atrophy – Type 2

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 the symptoms and effects of SMA Type 2, 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**
- **The 'Natural History' of SMA Type 1: Symptoms and Effects**
- **SMA Type 2 Today**
- **Changing Outcomes**
- **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.

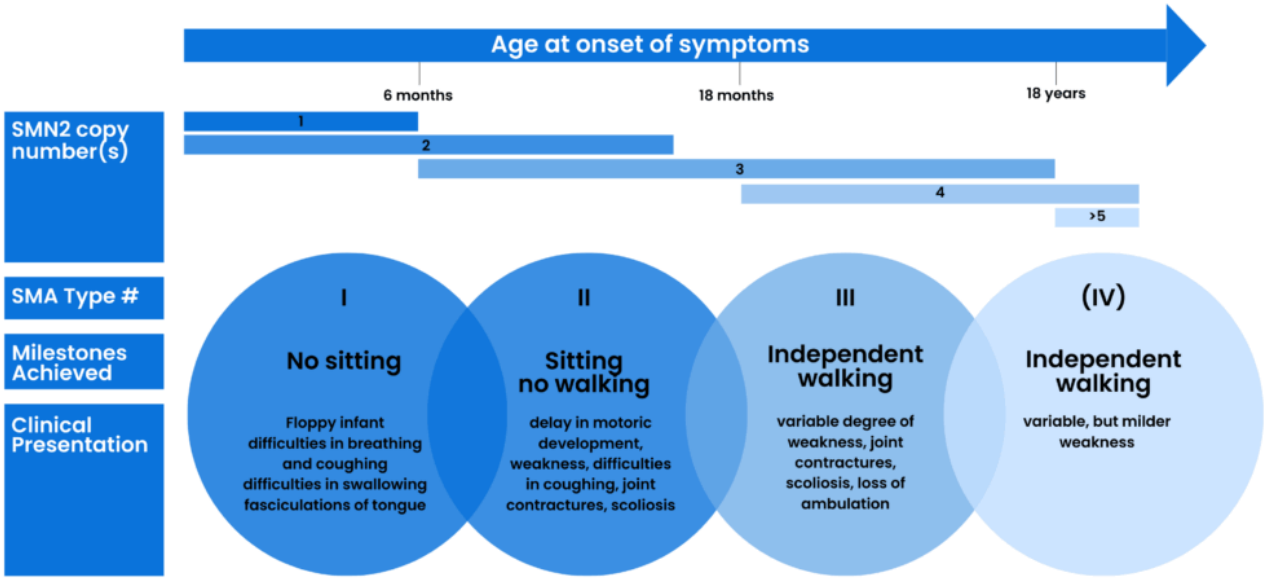
These ‘Types’ of SMA are based on the age that symptoms begin, and what physical milestones (e.g. sitting, standing, walking) are achieved. Clinicians agreed that there could be variation both within and between ‘Types’.

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. The Natural History of SMA Type 2: Symptoms and Effects

Your clinician will advise you if they would expect your child's SMA to follow the 'natural history' of SMA Type 2 if they do not have drug treatment. They will discuss the latest clinical trial and real-world evidence for these drug treatments. They will tell you what you might expect if your child is treated.

Our Guide to Drug Treatments > covers questions and topics that you may wish to discuss with your child's clinical team.

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

In the **natural history** of SMA Type 2, each child is affected differently. In general, infants are bright and engaging. However, their SMA causes:

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

Intellectual and sexual development[i] is not usually affected.

As children get older, the **natural history** of SMA would usually cause them to have:

- muscle weakness that may make it difficult to keep up with daily activities. For example, if someone had been able to crawl or roll, they may have lost this ability.

- weak breathing muscles, making it difficult to cough effectively and be more vulnerable to chest infections.
- muscles supporting the spinal column that are weak. Most children develop a sideways curvature of their spine[2] (scoliosis).
- reduced ability to move so that some joints may have become tight (contractures). This further restricts their range of movement.

The **natural history** can also weaken chewing and swallowing muscles. For some this means their tongue and shoulder muscles may twitch. They may also have a slight tremor in their hands[3].

Bladder and bowel control is not usually affected.

Major growth spurts, such as puberty, often create greater demands on weak muscles. SMA Type 2 can also mean that children and adults may become weaker after infections.

Most children, young people and adults living with SMA Type 2 need powered wheelchairs to maintain independent mobility. They also need help with daily tasks like:

- washing
- getting onto and off the toilet
- dressing and undressing.

Serious complications, like severe respiratory infections, can lower life expectancy[4]. But thanks to better healthcare, most people now live longer lives.

3. SMA Type 2 Today

Most clinical trials and studies of drug treatments for SMA Type 2 have focused on their effects on motor milestones gained in childhood. There is now more understanding of the importance of other outcomes that affect quality of life for children, young people and adults. This includes respiratory outcomes, fine motor skills and fatigue levels.

Changing Outcomes

Since 2019 in the UK, most children with SMA Type 2 will have started ongoing **Spinraza™ / Nusinersen** treatment. For adults, this possibility came later. More recently, ongoing treatment with **Evrysdi™ / Risdiplam** also became a possibility for children and adults.

Many children with SMA Type 2 have gained strength, movement and better health with treatment. Symptoms have stabilised for many adults, with some seeing improvements.

How SMA will impact the health and daily life of a treated person living with SMA Type 2 today – and how the natural history of their condition may change – is very individual. It will be influenced by factors including:

- the age at which symptoms first appeared,
- the severity of symptoms,
- how early any treatment was started and
- their individual response to treatment.

Though they can alleviate some symptoms, none of the drug treatments are a cure. For children and adults, they must be combined with the best supportive care and management of symptoms to ensure the best possible outcomes for each individual.

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 child's clinical team.

4. Related Information

- **Hearing your Child's diagnosis of SMA›**
- **Drug Treatments for Children who have 5q SMA ›**
- **Looking after your child who has had a recent diagnosis of SMA Type 2 – for parents and carers ›**
- **Living With SMA – information and ideas for daily living ›**
- **The Genetics of 5q SMA ›**

5. References

1. Montes J et al. (2009) Clinical outcome measures in spinal muscular atrophy, *J Child Neurol 24*: 968-978
2. Mercuri E, et al. (2012) Childhood spinal muscular atrophy: controversies and challenges. *Lancet Neurol 11*: 443-452.
3. Tsirikos AI & Baker ADL (2006) Spinal muscular atrophy: classification, aetiology, and treatment of spinal deformity in children and adolescents. *Curr Orthop 20*: 430-445.
4. Farrar MA et al. (2013) Pathophysiological insights derived by natural history and motor function of spinal muscular atrophy. *J Pediatrics 162*: 155-159.

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