

Identifying, referring and supporting infants with Spinal Muscular Atrophy



This Good Practice Point aligns to the NMC 2022 Standards of Proficiency for SCPHN Health Visitors – in particular: Sphere of Influence C: C.HV2, C..HV5; Sphere of Influence D: D.HV1, D.HV2, D.HV3, D.HV6, D.HV8; and Sphere of Influence E: E.HV1. See the [Nursing and Midwifery Council website](https://www.nmc.org.uk/standards-of-proficiency-for-scpnhn-health-visitors/) for more details.

Spinal Muscular Atrophy (SMA) is a rare progressive inherited neuromuscular condition. Lower motor neurons degenerate, causing muscle wasting (atrophy) weakness. It may affect crawling and walking ability, arm, hand, head and neck movement, breathing and swallowing. See section “Signs of SMA”.

In general, the earlier the onset of symptoms, the more severe the condition. Once symptoms appear, damage has already been done. Motor neurons die, cannot be replaced and lost skills cannot be regained.

Early diagnosis and NHS-funded disease-modifying treatments are essential in preserving the remaining motor neurons, slowing disease progression, and can be life-changing

What causes SMA?

There are different forms of SMA with different genetic causes, this GPP will focus on 5q SMA which is the most common form. It is called 5q SMA because the gene that causes it is located on the 5th chromosome called ‘q’.

The cause of 5q SMA is that the *SMN1* gene is altered or missing. As a result, the body cannot produce enough Survival Motor Neuron (SMN) protein. This protein is essential for maintaining the nerve cells that transmit signals from the spinal cord to muscles. Without it there is rapid and irreversible damage to the nerve cells².

Approximately 1 in every 40 people carry the altered gene, around 1.7 million people in the UK and 4 babies a month are born with SMA¹. The condition only occurs if a child inherits two altered genes – one from each parent. When two carriers have children there is a one in four chance for **each pregnancy** that the child will inherit both altered copies of the *SMN1* gene and will have SMA¹.

A second gene also produces SMN protein². This is the *SMN2* gene, sometimes referred to as the SMA ‘back-up’

gene. The *SMN2* gene only produces some of the SMN protein and cannot fully make up for the altered *SMN1* genes in people who have 5q SMA. In general, people with fewer copies of the *SMN2* gene are likely to develop more severe SMA².

- For more about genomics and understanding genomic screening and support, please see iHV GPP eXtra – Understanding Genomics <https://bit.ly/4vaw4TY>.

Types of 5q SMA³

Before the new drugs for 5q SMA were developed, the ‘natural history’ of the condition 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 are achieved. **The severity and impact will vary from person to person, both within and between ‘Types’.** Each child and adult is affected differently.

More information on page 2

For additional resources see www.ihv.org.uk

The information in this GPP version was created on 21/05/2026.

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Table 1 ‘Natural History’ of SMA without treatment

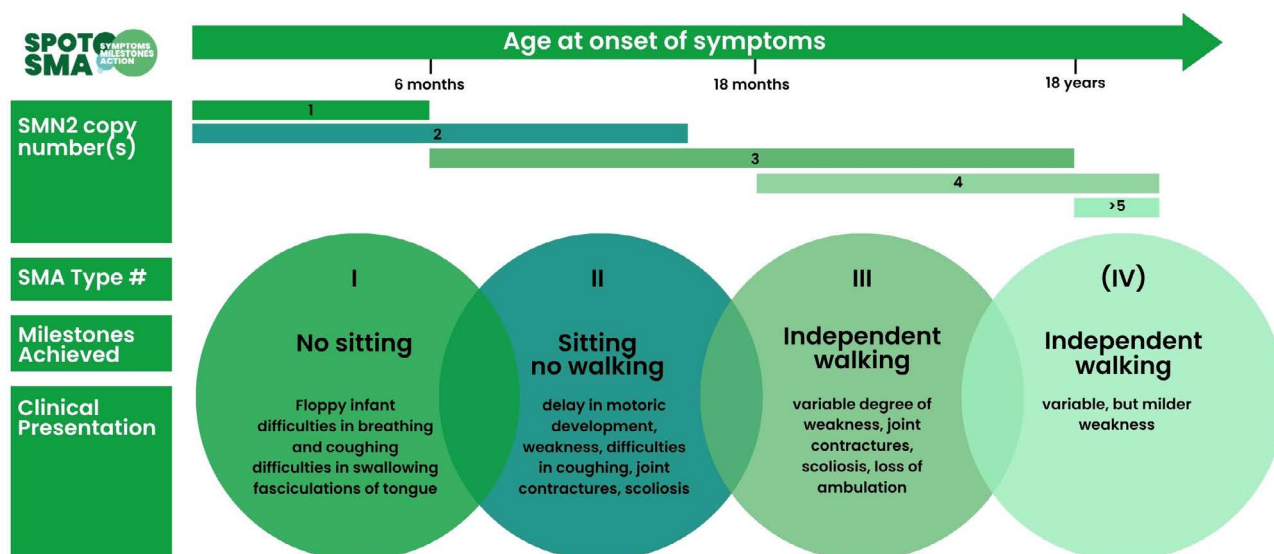


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Early identification

Listen to parents/carers, including grandparents, they know their babies and children best. ALWAYS take their concerns seriously.

NO baby is ‘lazy’. Don’t assume, always explore why they are not moving as expected.

DON’T ‘watch and wait’ – have the baby or child reviewed immediately.

Signs of SMA⁴

Age	Characteristics	Look for:
In babies aged about 0-6 months	Babies with early onset SMA are often ■ bright, alert and responsive ■ able to smile and frown	■ low muscle tone (hypotonia) and muscle weakness - a floppy baby ■ inability to support or lift their head ■ when lying on their back, inability to spontaneously kick with their legs into the air ■ inability to sit unsupported once around 5-6-months-old ■ difficulty rolling over ■ difficulty lifting their arms and legs but able to move their hands and fingers ■ ‘tummy breathing’ in absence of other illness ■ a weak cry and difficulties with breathing and coughing ■ difficulty feeding and gaining weight ■ tongue muscles that twitch (fasciculations) ■ may sound chesty from swallowing saliva ■ becoming extremely unwell with common colds.

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In infants aged about 6-18 months	Infants with symptoms at this age are usually bright and engaging	■ muscle weakness on both sides and closest to the centre of the body: » not sitting unsupported by age 7-8 months maximum. Difficulties lifting legs » difficulties moving arms, but hands and fingers less so » legs that are weaker than arms ■ loss of motor skills they had previously gained – such as crawling, rolling or sitting, e.g. if they had achieved sitting unsupported by 6-7 months, but are now having difficulties with this ■ possible slight tremor in their hand ■ a weak cough ■ gagging and/or coughing when feeding or eating ■ becoming extremely unwell with common colds ■ tongue muscles that twitch (fasciculations) ■ repeated upper respiratory infections and becoming extremely unwell with these.
In children aged about 18 months and older	Children with symptoms at this age are usually bright and engaging	■ muscle weakness on both sides and closest to the body » legs that are weaker than arms » difficulties with standing and walking » loss of physical ability e.g. to walk or climb stairs » falling more easily » needing help to get up when sitting on the floor » slight tremor in their hands.

REMEMBER: Symptoms can develop at any age in childhood

ACTION

If you think this could be SMA:

- make an **urgent** phone referral to the nearest paediatrician or [neuromuscular service](#)
 - choose the quickest route to having the baby/child seen urgently by a clinician
 - reassure, explain what is happening, what the concerns are and the next steps

How is SMA diagnosed?⁵

Once babies and children are referred for assessment, they will be carefully examined and have genetic testing to see if they carry the altered gene. If during pregnancy there is a concern due to previous SMA within a family and therefore a higher risk, a range of testing options are available depending on the specific situation of the family.

How is SMA treated?⁵

Collaboration between families and professionals to diagnose SMA and offer treatment is critical. Health visiting teams have an important role in this.

The NHS funds clinically effective disease-modifying drug treatments. Along with better care and management of the condition, this can change what motor milestones children may be able to achieve and greatly improve their general health.

It is important to recognise that the day-to-day life and management of SMA varies hugely, dependent on how early treatment, care, support and guidance was given. Each baby, child and family are unique, and it is important to understand their specific needs and work with the multi professional team to support them.

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Top tips for health visitors

- Listen to parent/carers concerns and act on them immediately
- Complete a full assessment to inform the referral
- Explain to parents/carers what is happening next and what to expect
- Explain your role and how they can get in contact with you
- Keep in contact with the family, continue to offer support during the referral and diagnosis processes. REMEMBER other support may be needed, such as for mental health, siblings, and day-to-day aspects of family life such as sleep, feeding, oral health and immunisations.
- Ongoing support following diagnosis will be required
- Connect families as needed with wider support, including referral and contact with [SMA UK Support Team](#), who offer a gateway to a range of services. The community support team offers a UK wide service for anyone affected by SMA, including support and information, a number of grant schemes and opportunities for family peer support via WhatsApp groups and events.
- Promote and use the SpotSMA (Spot the Signs, Measure Milestones, Action Treatment) short resource as an AIDE MEMOIR <https://smauk.org.uk/advocacy-and-campaigning/sma-awareness-campaigns/spot-sma/>

Further information

- SMA-UK: <https://smauk.org.uk/>
- NHS Spinal muscular atrophy (SMA): <https://www.nhs.uk/conditions/spinal-muscular-atrophy-sma/>
- NHS Wales Spinal muscular atrophy: <https://111.wales.nhs.uk/encyclopaedia/s/article/spinalmuscularatrophy/>
- NI direct Spinal muscular atrophy: <https://www.nidirect.gov.uk/conditions/spinal-muscular-atrophy>



References

¹SMA UK. (2026) What is 5q Spinal Muscular Atrophy?. Accessed 08/05/2026. Available at: <https://smauk.org.uk/support-information/about-sma/what-is-5q-spinal-muscular-atrophy/>

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³Schorling D et al. (2019) Advances in Treatment of Spinal Muscular Atrophy – New Phenotypes, New Challenges, New Implications for Care > J Neuromuscular Dis 7: 1-13. Accessed 08/05/2026. Available at: https://www.researchgate.net/publication/337118299_Advances_in_Treatment_of_Spinal_Muscular_Atrophy_-_New_Phenotypes_New_Challenges_New_Implications_for_Care?tp=eyJjb250ZXh0Ijp7ImZpcnN0UGFnZSI6Ii9kaXJlY3QilCJwYWdlIjoX2RpcmVjdCJ9fQ under the CC BY-NC 4.0 licence >

⁴SMA UK. (2024) Symptoms & Effects of 5q Spinal Muscular Atrophy. Accessed 08/05/2026. Available at: <https://smauk.org.uk/support-information/about-sma/symptoms-effects-5q-sma/>

⁵NHS. (2024) Spinal muscular atrophy (SMA). Accessed 08/05/2026. Available at: <https://www.nhs.uk/conditions/spinal-muscular-atrophy-sma/>

The production of this GPP, including expert clinical input and editorial coordination, was sponsored by SMA-UK.

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