



Spinal Muscular Atrophy Type 1: Spotting the signs & symptoms

What is Spinal Muscular Atrophy type 1?

For many parents and carers, seeing their baby grow and develop is an amazing time, providing ‘firsts’ that will become treasured memories. Many infants, but not all, develop so that over time they become able to do more advanced activities such as reaching out for a toy or taking their first steps. However, there are certain neuromuscular conditions (i.e. affecting the nerves and muscle function) that can have a big impact on a baby’s development, one of which is Spinal Muscular Atrophy (SMA). This resource is all about spotting the signs and symptoms of SMA and getting support as soon as possible.

- SMA is a rare disease affecting about 70 babies a year in the UK. SMA type 1 is the most severe form of SMA and it can, unfortunately, cause a baby or young child to die. Understanding your baby’s development is really important because the sooner a baby with SMA type 1 is diagnosed, support can be provided and the long term outcomes can be improved. There are three other types of SMA, and we talk about each one on page 2.
- This resource provides information on key movements, also known as motor developmental milestones, that most babies can do in the first few months of life. If a baby is not reaching these milestones, it is important to discuss this with your health visitor or GP. A baby with SMA type 1 may not be able to do some of the activities listed on page 2.

[More information on Page 2](#)

For additional Parent Tips see www.ihv.org.uk

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What is the cause of SMA?

- The root cause of SMA is a problem with a gene that is either faulty or completely missing. As a result, the body cannot produce enough Survival Motor Neuron (SMN) protein to maintain the nerve cells that transmit signals from the spinal cord to muscles, leading to rapid and irreversible damage.
- Approximately 1 in every 40-60 people has one faulty/missing gene, and the condition only occurs if a child inherits faulty/missing genes from both parents (this is a 1-in-4 risk if both parents are carriers). The body also has “back-up” genes that produce some working SMN protein, but not enough to entirely prevent the condition. People with fewer copies of the back-up gene are likely to develop more severe SMA (classified as type 1).

The different types of SMA you may see, with typical symptoms

	Type 1	Type 2	Type 3	Type 4
Age when symptoms may develop	< 6 months	6-18 months	After 18 months	Adulthood (after 21 years)
How motor development is affected*	Never able to sit or stand	Never able to stand or walk; may lose ability to sit independently	Able to stand and walk; may lose ability to walk when older	Gradually finds walking more difficult
Other symptoms	Feeding and breathing difficulties; curvature of the spine	Breathing problems, particularly while sleeping; curvature of the spine	Prone to chest/breathing infections; curvature of the spine; may develop difficulty with joints moving freely	Mild to moderate muscle weakness; shaking and twitching muscles
Life expectancy*	2 years	Adolescence or young adulthood	Most have a normal lifespan	Normal lifespan

* Effect on motor development and life expectancy are based on untreated SMA. Different treatment options are only approved/available on the NHS for certain types of SMA. Possible treatments should be discussed on a case-by-case basis with specialist healthcare professionals.

Observing your baby's movements

Looking to see if your baby can do the following helps check if your baby is developing as expected:

- 1 Raising their head when lying on their tummy**
You can easily check this milestone during ‘tummy time’ (this is when the baby is lying on their tummy). Tummy time is great for babies but only when they are awake. A baby should never be placed to sleep on their tummy. For more information, see **iHV Parent Tips - Safer sleep for your baby**: <https://bit.ly/3hYcQxl>
- 2 Kicking their legs and waving their arms when on their back**
When awake, their arms and legs should not be ‘floppy’
- 3 Reaching for things**
You can check this by putting an object or toy just out of reach on the floor or on their play mat

[More information on Page 3](#)

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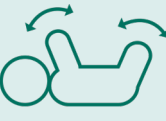
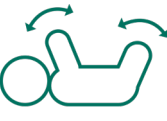



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This table (below) shows us at what age babies often become able to do activities, but each baby is unique and this can vary. Always talk to your health visitor if you have any worries about your baby's development.

1 month old	2 months old	3 months old
 <p>When lying on their front/tummy baby can briefly lift their head</p>	 <p>When lying on their tummy, baby can lift their head and move it from side to side – they may also use their arms to push off the ground</p>	 <p>When lying on their tummy, baby can lift their head and chest</p>
 <p>Baby can move their hands to their face and mouth</p>	 <p>When lying on their back, baby can move and wriggle their arms and legs. As development continues these movements will become smoother</p>	 <p>When lying on their back, baby waves their arms and kicks their legs</p>
 <p>Startle reflex – when baby is startled, such as a loud noise, they throw out their arms and spread their fingers</p>	 <p>Baby can briefly hold a toy that is placed in their hands</p>	 <p>Baby is beginning to reach out for toys and deliberately grasps and holds them</p>

For more information about development for most children by 3 months, please visit <https://www.think3at3months.co.uk>. You will also find videos showing a child's typical movements at 3 months, milestone cards, and a conversation guide to aid conversations with your GP about your child's development.

What to do if you have any worries about your baby

- Babies develop at different rates, so there may be nothing to worry about. However, if you have any concerns about your baby's movements or development, please don't delay - talk to your health visitor or see your GP as soon as you can.
- Health visitors are there to offer you support and consider your baby's development in partnership with you. Please feel able to discuss any concerns you have about your baby's developmental movement milestones openly with your health visitor.
- SMA is not routinely screened for in the UK, unless there is a family history of SMA. Many expert healthcare professionals and SMA advocacy groups hope that this will change, as this could enable earlier diagnosis and treatment, leading to improved outcomes. In the meantime, health visitors can help you as they are uniquely placed to spot the signs in a child's early development and can raise these with a GP as needed.

Further information:

- **NHS** - Spinal Muscular Atrophy: <https://bit.ly/37DC56u>
- **Think 3 at 3 months**: <https://bit.ly/3M8IFIB>

The following organisations can provide advice and support:

- **Muscular Dystrophy UK** - 0800 652 6352
- **Spinal Muscular Atrophy UK** - 01789 267520
- **Treat SMA** - 0300 800 0202
- **The Annabelle Rose Foundation for Spinal Muscular Atrophy** - 0300 8000 762

If a child receives an SMA diagnosis, parents can register for the UK SMA patient registry here: <https://bit.ly/3wlaMWR>

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