

UNOVARTIS

LOOK OUT FOR EARLY SIGNS OF SPINAL MUSCULAR ATROPHY (SMA)

All babies develop differently, it is important to keep track of how your baby is progressing. Developmental delays and other symptoms can be signs of SMA, a rare genetic disease which requires urgent medical attention. To ensure your baby's progress is on track, it's important to know what to look out for.⁴

If you feel something isn't right with your baby, or your baby is displaying any of the signs of SMA, trust your instincts and speak to your doctor.

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SPOTTING THE SIGNS OF SMA: AGE 0-6 MONTHS

All babies develop differently, but developmental delays and other symptoms can be signs of spinal muscular atrophy (SMA), a rare genetic disease which requires urgent medical attention. To ensure your baby's progress is on track, it's important to know what to look out for.⁴

AGE O-6 MONTHS



POOR HEAD CONTROL⁴

Babies may experience difficulty moving their head from side to side or lifting it up while lying on their back, being held, or lying on their tummy. ^{2,5} They might rest their head on one side with little movement.



WEAK LEGS & ARMS^{2,3}

Babies may exhibit limited arm and leg movement while lying on their back, such as rarely kicking their legs or moving their arms away from their body. They may also struggle to lift their arms and legs, appear floppy or weak, and have legs in a 'frog leg' position. Parents or caregivers may also notice that their baby's limbs feel limp when held.^{3,6}



FAST BELLY BREATHING³

Parents or caregivers may observe that their baby's breathing is rapid even at rest, and lacks deep breaths in or out. The baby's chest may appear bell-shaped, and their belly may move more noticeably than their chest while breathing.³ This is most easily seen when the baby is lying on their back without clothes on the upper half of their body.



DIFFICULTY FEEDING/ SWALLOWING 3,5

Due to muscle weakness, babies may experience difficulty sucking or swallowing during feeding,^{3,4} which may lead to choking and make weight gain challenging.⁵ Parents or caregivers may also notice slow feeding as the baby struggles to swallow.5

Remember, even babies with SMA remain alert, responsive and smiley. They'll appear happy, not in distress, which makes it less obvious that there might be a problem. If you notice any of the above symptoms, do not hesitate to reach out to your baby's doctor.





WEAK CRY & COUGH ⁵

Parents or caregivers may notice that their baby's cry sounds weak and difficult to hear. Babies with weakness may also experience difficulty coughing and clearing mucus from their chest, resulting in weak coughs and chest sounding congested.⁵







SPOTTING THE SIGNS OF SMA: AGE **6-18 MONTHS**

Seeing your baby develop is an amazing journey, and it's important as they grow to continue to keep track of their progress. Signs of SMA can be seen any time before 10 months, and up to 18 months of age.

AGE 6-18 MONTHS



WEAK LEGS & ARMS⁴

Babies may appear to have floppy arms and legs, like they lack strength, with more noticeable weakness in their legs.^{3,4} They may have difficulty pushing up to their elbows and maintaining the position while lying on their tummy. They may also show slow, fatigued movements when trying to reach or grab toys. When held, babies may struggle to support themselves using their legs.4



STRUGGLES TO SIT UNSUPPORTED^{2,3}

Babies may struggle to sit still for extended periods and may lean forward or to one side. ²⁻⁴ Initially, this lean may be slight, but as they continue to sit, they may find it increasingly challenging and require further assistance to maintain the position.4



CAN'T ROLL OVER³

Babies may rock from side-to-side but struggle to roll fully onto their side or tummy when lying on their back. Over time, these movements may become even increasingly difficult for them.4



SHAKING HANDS 3,5

Look out for small tremors or shakes in the fingers or hands when your baby holds their arms out. ³ The shaking may be subtle and challenging to notice.

Remember, babies with SMA remain alert, responsive and smiley. They'll appear happy, not in distress, which makes it less obvious that there might be a problem. But if you feel something's wrong, don't hesitate to see your doctor





SLOW/LOST PHYSICAL DEVELOPMENT ⁵

Babies may be slow to achieve developmental milestones such as sitting, standing, or walking without support. ²⁻⁴ Although they may reach these milestones, they may lose the ability to do so later.4





SUSPECT SMA? SEEK MEDICAL ADVICE TODAY



TALK TO YOUR DOCTOR

Stores .

Spinal muscular atrophy (SMA) is a disease which progresses quickly, so every day matters. Noticing signs early is key, because the sooner a baby can receive care from their doctor, the better. If you feel something isn't right with your baby, or your baby is displaying any of the early signs of SMA, trust your instincts and speak to your doctor.



baby's doctor.

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Make an appointment with your



Be prepared

It's natural to feel anxious when talking to your doctor about your baby's development or health. To help you prepare for the discussion, follow these tips:

- Note down key signs and concerns you have about your baby's development.
- Keep diary of the changes you notice and the dates they occur.
- It can be helpful to show your doctor any videos you may have which show lost motor milestones or regression over time, or examples of other signs.
- Beforehand, write down any questions you may have.



Discuss your concerns

When speaking to your doctor, the more information you can give them, the better equipped they'll be to provide the best advice or point you in the right direction. Ask your doctor about next steps and what to expect.



BOOK A DOCTOR'S VISIT TODAY IF YOU SEE THE SIGNS

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| NOTE Note down key signs and developmental concerns you have to aid conversations with your doctor | |
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UNDERSTANDING SMA

SMA is a rare genetic disease in which nerves are lost causing weakness of the muscles. This occurs because nerves that control muscle movement, called motor neurons, deteriorate and stop working.⁸⁻¹²

Motor neuron

Brain In babies with SMA, signals for muscle movement are still generated.

Individuals with SMA have a genetic fault that causes their motor neurons in the spinal cord to stop working, deteriorate, and eventually die. This is because signals from the brain are not carried to the muscles.

Muscle

When the muscles do not receive signals to move, they become increasingly weaker, also known as atrophy.

HOW DO YOU GET SMA?

SMA is typically inherited, although occasionally it is caused by a random error in the SMN1 gene⁸⁻¹²

- Children inherit two copies of the SMN1 gene, one from each parent¹⁹
- People with one healthy and one faulty SMN1 gene are known as carriers, they normally show no signs of SMA¹⁹
- If both parents are carriers there is
- effectively a 25% chance the baby will inherit and develop SMA¹⁹

Holds a defective and a healthy

copy of the SMN1 gene¹⁹

Half of the child's genetic

information comes from the

mother and half from the father

O Carrier

SMA affects approximately 1 in 12,000 live births and can impact any race or gender.^{1,13}

WHAT CAUSES SMA

- SMN protein is vital for motor neurons to be able to work properly. SMN stands for 'survival motor neuron⁸
- Without SMN protein, motor neurons in the spinal cord stop working and muscles become weaker¹²

Missing or faulty SMN1 gene

- SMN protein is made in the body from the SMN gene¹³⁻¹⁵
- In SMA, the main SMN gene known as SMN1 is faulty or missing^{13,16,17}
- There is a second SMN gene known as SMN2 that acts more
- like a 'back-up' and only produces small amounts of SMN protein^{8,13,18}



In SMA, not enough SMN protein is being produced and the motor neurons stop working causing the muscles to become weaker^{13,14}





The child has inherited

a healthy copy of the SMN1

gene from each parent



Carrier

The child has inherited a healthy copy and a defective copy of the SMN1 gene and so becomes a carrier of SMA





Muscle atrophy

Insufficient amounts of functional SMN protein (about 10%)







More than 1 in 58 people are carriers of the disease mutation.14



Holds a defective and a healthy copy of the SMN1 gene¹⁹

Carrier



Affected by SMA

The child has inherited a defective copy of the SMN1 gene from each parent and so develops SMA

Affected by SMA

UNDERSTANDING MORE ABOUT YOUR BABY'S PROGRESS

Motor (or developmental) milestones are a measure of the physical progress your baby makes as they grow.

Seeing your baby grow and develop is a fascinating experience -from birth onwards you will have plenty of exciting 'firsts' to look forward to

The figure below illustrates typical development patterns in the first few months of life, so you can see how your baby is progressing. Every baby is unique and the exact age individual babies reach these milestones may differ.



are no longer able to achieve milestones that they previously could, speak to your doctor immediately

TALK TO YOUR DOCTOR IF YOU'RE CONCERNED ABOUT YOUR CHILD'S DEVELOPMENT



4 months old







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