

***LOOK OUT FOR EARLY SIGNS OF  
SPINAL MUSCULAR ATROPHY (SMA) <sup>1-3</sup>***

**WEAK  
LIMBS**

**DIFFICULTY  
FEEDING**

**FAST BELLY  
BREATHING**

**POOR  
HEAD  
CONTROL**

**WEAK  
CRY**





# 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.<sup>4</sup>

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 baby's doctor.

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READ ON TO SEE HOW TO SPOT EARLY SIGNS OF SMA



# 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.<sup>4</sup>



## AGE 0-6 MONTHS



### POOR HEAD CONTROL<sup>4</sup>

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.<sup>2,5</sup> They might rest their head on one side with little movement.



### WEAK LEGS & ARMS<sup>2,3</sup>

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.<sup>3,6</sup>



### FAST BELLY BREATHING<sup>3</sup>

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.<sup>3</sup> This is most easily seen when the baby is lying on their back without clothes on the upper half of their body.



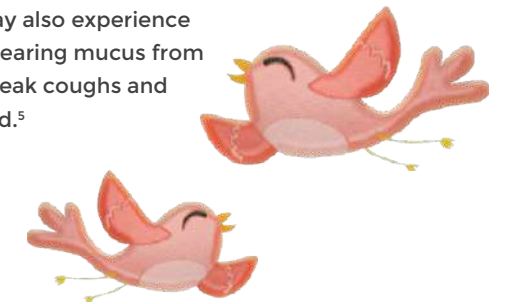
### DIFFICULTY FEEDING/ SWALLOWING<sup>3,5</sup>

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



### WEAK CRY & COUGH<sup>5</sup>

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.<sup>5</sup>



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 doctor.





# 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 & ARMS<sup>4</sup>**

Babies may appear to have floppy arms and legs, like they lack strength, with more noticeable weakness in their legs.<sup>3,4</sup> 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.<sup>4</sup>



### **STRUGGLES TO SIT UNSUPPORTED<sup>2,3</sup>**

Babies may struggle to sit still for extended periods and may lean forward or to one side.<sup>2-4</sup> 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.<sup>4</sup>



### **CAN'T ROLL OVER<sup>3</sup>**

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.<sup>4</sup>



### **SHAKING HANDS<sup>3,5</sup>**

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



### **SLOW/LOST PHYSICAL DEVELOPMENT<sup>5</sup>**

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



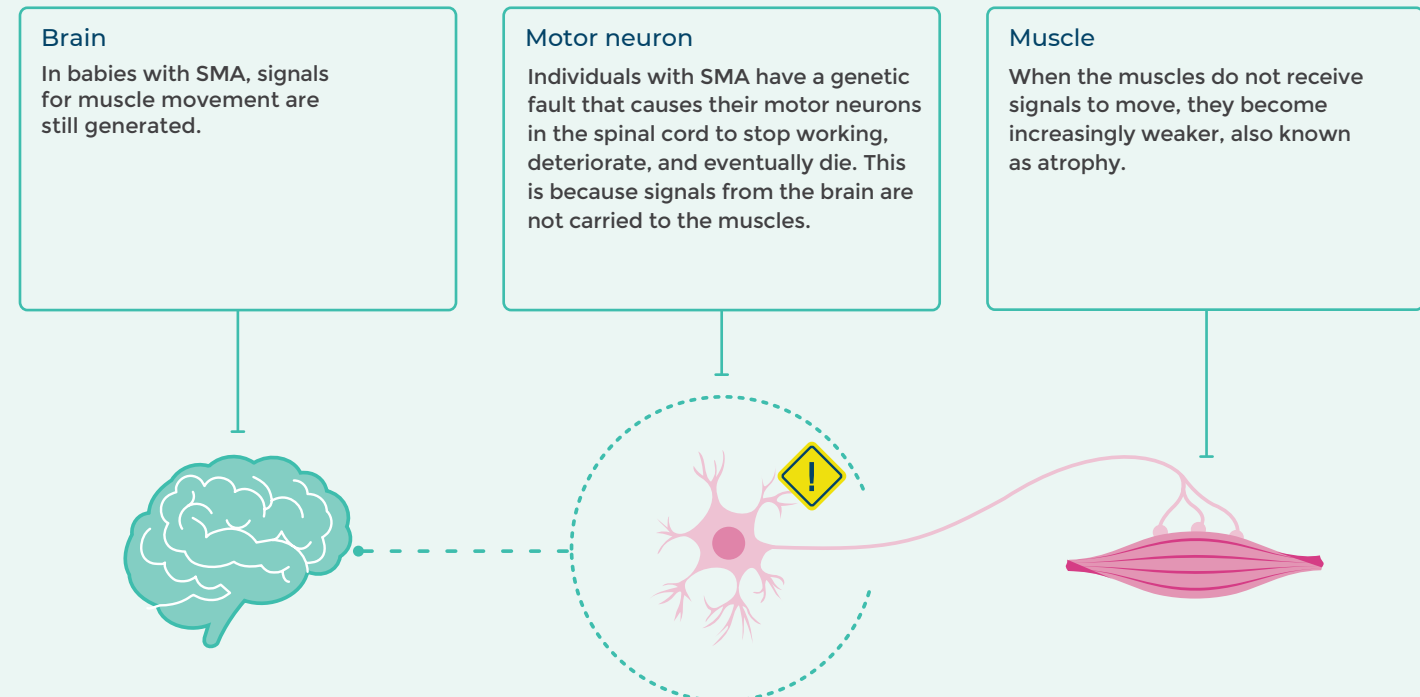
Remember, babies with SMA remain alert, responsive and smiley. They'll appear happy, not in distress, which makes it less obvious that there might be a problem. But if you feel something's wrong, don't hesitate to see your baby's doctor.





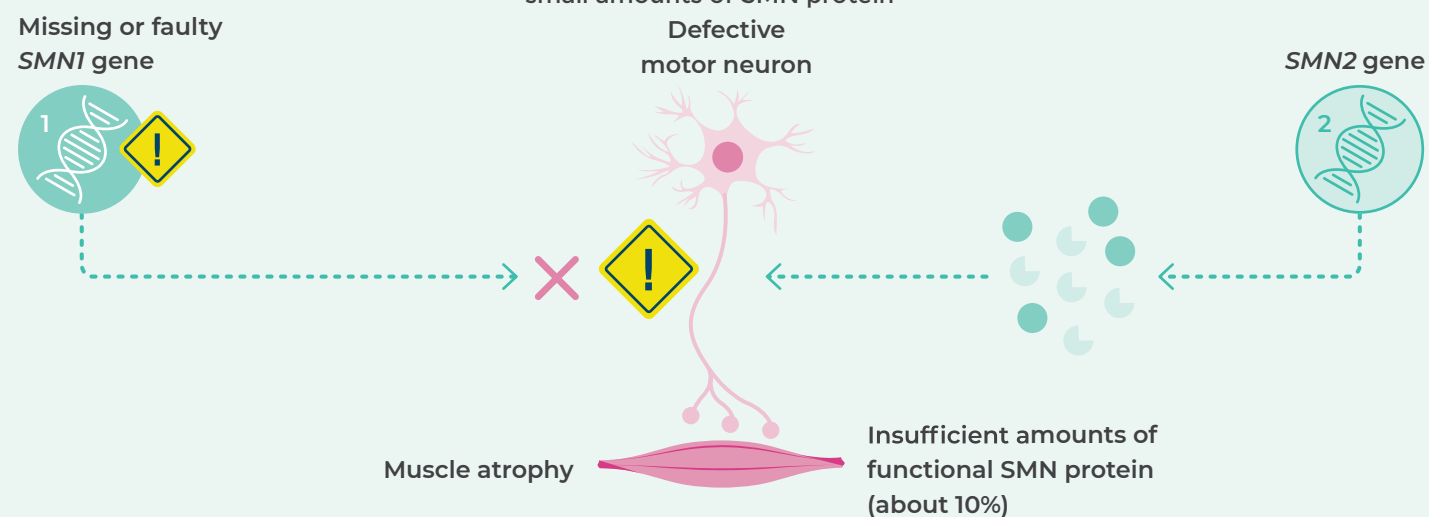
# UNDERSTANDING SMA

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.<sup>8-12</sup>



## WHAT CAUSES SMA

- SMN protein is vital for motor neurons to be able to work properly. SMN stands for 'survival motor neuron'<sup>8</sup>
- Without SMN protein, motor neurons in the spinal cord stop working and muscles become weaker<sup>12</sup>
- SMN protein is made in the body from the *SMN* gene<sup>13-15</sup>
- In SMA, the main *SMN* gene known as *SMN1* is faulty or missing<sup>13,16,17</sup>
- There is a second *SMN* gene known as *SMN2* that acts more like a 'back-up' and only produces small amounts of SMN protein<sup>8,13,18</sup>
- In SMA, not enough SMN protein is being produced and the motor neurons stop working causing the muscles to become weaker<sup>13,14</sup>



## HOW DO YOU GET SMA?

SMA is typically inherited, although occasionally it is caused by a random error in the *SMN1* gene<sup>8-12</sup>

- Children inherit two copies of the *SMN1* gene, one from each parent<sup>19</sup>
- People with one healthy and one faulty *SMN1* gene are known as carriers, they normally show no signs of SMA<sup>19</sup>
- If both parents are carriers there is effectively a 25% chance the baby will inherit and develop SMA<sup>19</sup>



SMA affects approximately 1 in 12,000 live births and can impact any race or gender.<sup>13</sup>



More than 1 in 58 people are carriers of the disease mutation.<sup>14</sup>

## ♀ Carrier

Holds a defective and a healthy copy of the *SMN1* gene<sup>19</sup>

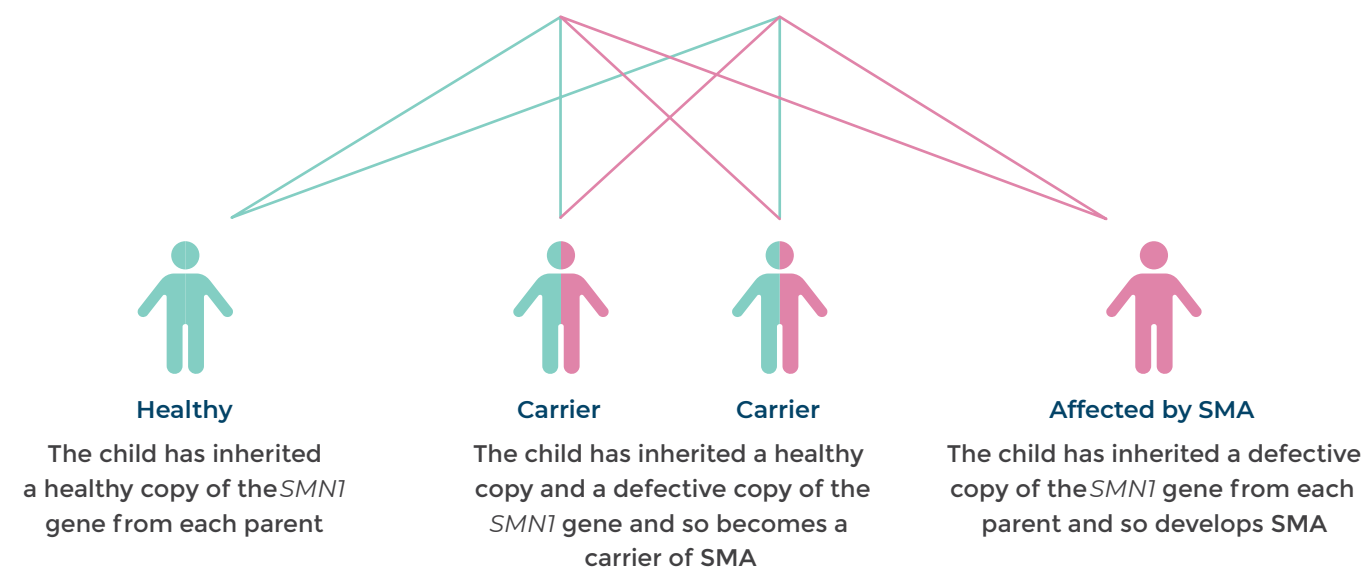


## ♂ Carrier

Holds a defective and a healthy copy of the *SMN1* gene<sup>19</sup>



Half of the child's genetic information comes from the mother and half from the father



● Healthy

● Carrier

● 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.



1 month old	2 months old	3 months old
<p>When lying on their tummy, baby can briefly lift their head<sup>22</sup></p>	<p>When lying on their tummy, baby can lift their head and move it side-to-side – may also use arms to push off of the ground<sup>22,23</sup></p>	<p>When lying on their tummy, baby can lift their head and chest<sup>22</sup></p>
<p>Can move hands to their face and mouth<sup>22</sup></p>	<p>When lying on their back, baby can move and wriggle their arms and legs. As development continues, the movements will become smoother<sup>22,23</sup></p>	<p>When lying on their back, baby waves their arms and kicks their legs<sup>22</sup></p>
<p>Startle reflex – when baby is startled, such as a loud noise, they throw out their arms and spread their fingers</p>	<p>Can briefly hold a toy that is placed in their hands<sup>23</sup></p>	<p>Beginning to reach out for toys and deliberately grasps and holds items</p>

If you notice your baby hasn't reached any of these milestones within the expected age range, or they are no longer able to achieve milestones that they previously could, speak to your baby's doctor immediately.

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



4 months old	6 months old	9 months old
<p>When lying on their tummy they can hold their head straight and look around<sup>22,23</sup></p>	<p>Beginning to sit up without help</p>	<p>Crawls on their hands and knees<sup>22,23</sup></p>
<p>When lying on their back they rock from side-to-side<sup>22</sup></p>	<p>Rolls from side-to-side and rolls over front-to-back<sup>22,23</sup></p>	<p>Can pull themselves up to stand whilst holding onto something<sup>22,23</sup></p>
<p>Pushes up to elbows when lying on their tummy<sup>22</sup></p>	<p>Beginning to support their weight on their legs and stand with help</p>	<p>Can stand whilst holding on to something<sup>22</sup></p>
<p>They grab and shake toys in their hands<sup>22,23</sup></p>	<p>Starts to crawl on their hands and knees<sup>22,23</sup></p>	<p>Beginning to take a few steps whilst holding onto furniture for support<sup>22</sup></p>
12 months old		
<p>Can stand alone without help<sup>22,23</sup></p>	<p>Begins to take a few steps alone<sup>22,23</sup></p>	<p>Can get into the sitting position alone and sits well<sup>22,23</sup></p>
18 months old		
<p>Can walk well alone<sup>22,23</sup></p>	<p>Starts to walk upstairs and run<sup>22,23</sup></p>	<p>Beginning to feed themselves with a spoon, and drink with a cup<sup>22</sup></p>

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