

LOOK OUT FOR EARLY SIGNS OF SPINAL MUSCULAR ATROPHY (SMA)¹⁻³

SMA is a race against time, where fast diagnosis is vital because damage that occurs before treatment is irreversible^{4,5}. Check for the signs below at routine check-ups or if parents or caregivers raise any concerns^{3,6}.



SIGNS OF SMA: AGE 0-6 MONTHS

Very early signs of SMA are typically seen up to 6 months of age,^{1,2} often by age 3 months.^{2,7} Although babies may be symptomatic, they will remain alert and attentive; their cognition not affected¹



HYPOTONIA^{1,7}

- A baby with hypotonia is often described as 'floppy'⁸ due to weakness in their arms and legs^{1,7}
- Symmetrical weakness that is more proximal than distal³ means that a baby will have difficulty lifting their arms and legs, but retain use of their hands and fingers⁸
- The baby's legs may seem weaker than their arms³
- In profound cases, the baby may have a frog-like posture when lying^{1,9}



AREFLEXIA²

- Absent or reduced deep tendon reflexes are characteristic of SMA^{1,2} and a critical part of the baby's exam in cases of hypotonia⁹
- Evaluation of deep tendon reflexes can be achieved by close observation of the baby's response to brisk strikes of the tendon with a specialized hammer¹⁰



HEAD LAG¹¹

- If a baby seems unable to lift their head or has poor head control,^{1,3,8} the pull-to-sit test can be used to confirm head lag¹²
- A baby that is not developing typically will likely have their head lag behind their trunk, with their neck completely extended^{1,11-13}
- They may not lift their head above the line of their back when held horizontally face down⁹



DIFFICULTY BREATHING^{7,8}

- Weakness of the intercostal muscles with sparing of the diaphragm can give the baby a bell-shaped chest and paradoxical pattern of breathing, sometimes referred to as 'belly-breathing'¹



DIFFICULTY SWALLOWING^{1,3}

- Difficulties with sucking, feeding, or managing oral secretions (saliva) can suggest tongue and swallowing weakness typical in SMA^{1,3,11}
- In more progressed cases, a history of choking, recurrent aspiration, or slow or reduced growth may be present^{1,8,11}



TONGUE FASCICULATION¹⁻³

- A baby with SMA often will have tongue fasciculations, or twitching of the tongue, together with atrophy¹⁻³



WEAK CRY & COUGH³

- A baby with SMA may have a weak cry^{3,8}
- Weakness of the respiratory muscles can also cause severe difficulties with coughing⁸

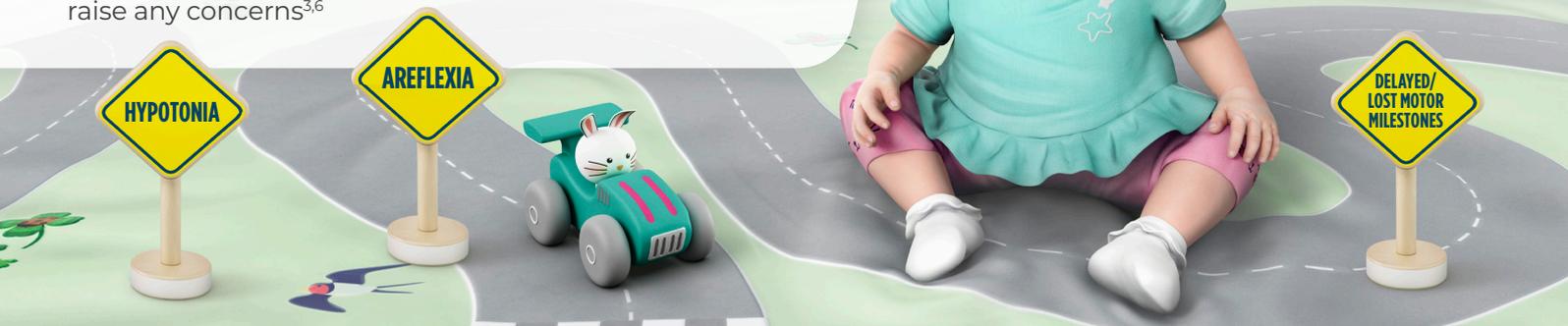
REFER URGENTLY TO A PEDIATRIC NEUROLOGIST IF YOU SEE THE SIGNS^{4,14}



#SIGNSOFSMA
SMA
signsofSMA.com

LOOK OUT FOR EARLY SIGNS OF SPINAL MUSCULAR ATROPHY (SMA)¹⁻³

As a healthcare professional you are uniquely placed to spot whether a baby is developing as they should.⁶ Check for the signs below at routine check-ups or if parents or caregivers raise any concerns^{3,6}



SIGNS OF SMA: AGE 6-18 MONTHS

Early signs of SMA are typically seen up to 18 months of age,¹ often by age 10 months.⁷ Although babies may be symptomatic, they will remain alert with normal speech development¹⁵



HYPOTONIA¹

- Reduced muscle tone and strength on examination, perhaps with a history of poor muscle tone in the first few months of life is a key sign of SMA²
- Some weakness in the legs and arms may be present¹
- The baby may have difficulty reaching for and picking up objects¹⁶
- The baby is unable to stand due to pronounced leg weakness, and unlikely to walk independently¹³



AREFLEXIA²

- Absent or reduced deep tendon reflexes are characteristic of SMA² and a critical part of the baby's exam in cases of hypotonia⁹
- Evaluation of deep tendon reflexes, can be achieved by close observation of the baby's response to brisk strikes of the tendon with a specialized hammer¹⁰



FINE TREMOR³

- When the baby extends their fingers or attempts to grip an object with their hands you may see a fine tremor^{3,17}
- Twitching of their shoulder muscles can also present¹⁷



PROGRESSIVE SCOLIOSIS & JOINT CONTRACTURES^{1-3,18}

- The baby may have more severe motion limitations in the lower extremities than upper¹⁹
- Progressive scoliosis, most likely C-shaped, contractures, particularly of the knee and ankle, and pelvic obliquity may also be observed^{1-3,18,19}



RESPIRATORY SYMPTOMS²

- Restrictive lung disease can result from progressive intercostal muscle weakness² particularly if the baby also has scoliosis¹
- Signs of restrictive lung disease include a reduced total lung capacity and forced vital capacity with preserved expiratory volume²⁰



DELAYED/LOST MOTOR MILESTONES^{2,3}

- Although the baby may have achieved milestones,¹⁻³ it is likely they were delayed.²
- Eventually a gradual decline in motor function is observed and some milestones, such as sitting unassisted or standing will be lost²

REFER URGENTLY TO A PEDIATRIC NEUROLOGIST IF YOU SEE THE SIGNS^{4,14}



#SIGNSOFSMA
SMA
signsofSMA.com

1. Kolb SJ and Kissel JT. *Neural Clin.* 2015;33(4):831-46. 2. Prior TW, Leach ME, Finanger E. *Spinal Muscular Atrophy*. 2000 Feb 24 [Updated 2019 Nov 14]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. *GeneReviews*[®] [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2020. 3. Wang CH, et al. *J Child Neurol.* 2007;22(8):1027-49. 4. Govoni A, et al. *Mol Neurobiol.* 2018;55(8):6307-18. 5. Stifani N. *Front Cell Neurosci.* 2014;8:293. 6. Qian Y, et al. *BMC Neurology.* 2015;15:217. 7. Pera MC, et al. *PLoS One.* 2020;15(3):e0230677. 8. SMA Europe (2020). Type 1. Available at: Available at: <https://www.sma-europe.eu/essentials/spinal-muscular-atrophy-sma-type-1/>. Date accessed: October 2020. 9. Leyenaar J, et al. *Paediatr Child Health.* 2005;10(7):397-400. 10. Zimmerman B, Hubbard JB. *Deep Tendon Reflexes (Stretch Reflexes)* [Updated 2020 Jul 3]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan. 11. Markowitz JA, et al. *JOGNN.* 2004;33:12-20. 12. Great Ormond Street Hospital for Children NHS. *Brief Developmental Assessment (BDA)*. Available at: <http://www.gosh.nhs.uk/file/1841/download?token=otvMwb9q>. Date accessed: October 2020. 13. Hammersmith Infant Neurological Examination (v07.07.17). Available at: https://bpna.org.uk/userfiles/HINE%20proforma_07_07_17.pdf. Date accessed: October 2020. 14. Mercuri E, et al. *Neuromuscul Disord.* 2018;28(2):103-15. 15. Shababi M, et al. *J Anat.* 2014;224(1):15-28. 16. Spinal Muscular Atrophy UK (2019). *Toys and play for babies and children who have spinal muscular atrophy - A professional's guide*. Available at: <https://smauk.org.uk/toys-and-play-a-guide-for-professionals>. Date accessed: October 2020. 17. SMA Europe (Type 2). Available at: <https://www.sma-europe.eu/essentials/spinal-muscular-atrophy-sma-type-2/>. Date accessed: October 2020. 18. Fujak A, et al. *BMC Musculoskelet Disord.* 2013 Oct 4;14:283. 19. Wang HY, et al. *Arch Phys Med Rehabil.* 2004;85(10):1689-93. 20. Martinez-Pitre PJ, Sabula BR, Cascella M. *Restrictive Lung Disease*. [Updated 2020 Jul 15]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan.

This material was developed by Novartis for educational purposes only.

ALL-UNB-20-0123 | Date of preparation: October 2020.