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 affected[1]

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 distal[3] means that a baby will have difficulty lifting their arms and legs, but retain use of their hands and fingers[8]
- The baby's legs may seem weaker than their arms[3]
- In profound cases, the baby may have a frog-like posture when lying[1,9]

AREFLEXIA[2]

- Absent or reduced deep tendon reflexes are characteristic of SMA[1,2] and a critical part of the baby's exam in cases of hypotonia[9]
- 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 hammer[10]

HEAD LAG[11]

- 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 lag[12]
- 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 down[9]

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 FASCICULATION[1-3]

- A baby with SMA often will have tongue fasciculations, or twitching of the tongue, together with atrophy[1–3]

WEAK CRY & COUGH[3]

- A baby with SMA may have a weak cry[3,8]
- Weakness of the respiratory muscles can also cause severe difficulties with coughing[8]

REFER URGENTLY TO A PEDIATRIC NEUROLOGIST IF YOU SEE THE SIGNS[4,14]

#SIGNSOF SMA

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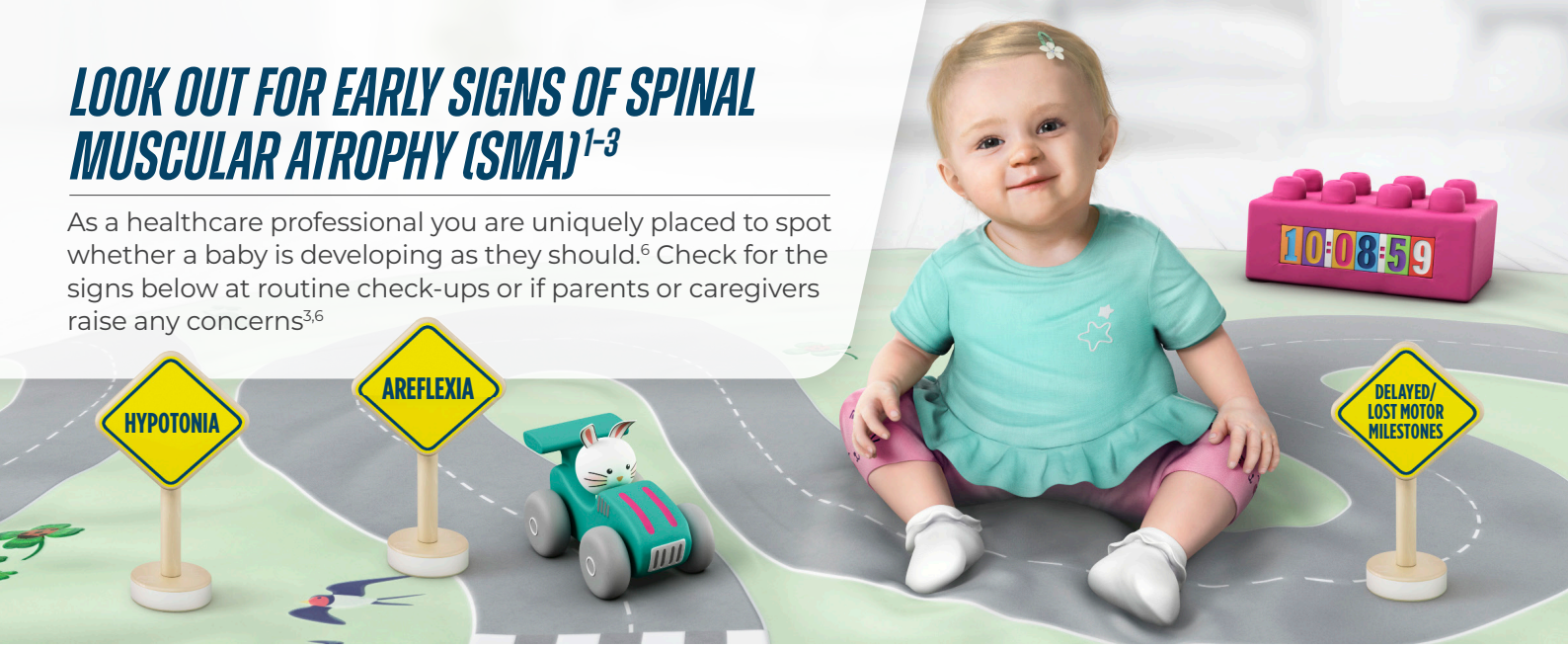
LOOK OUT FOR EARLY SIGNS OF SPINAL MUSCULAR ATROPHY (SMA)[1-3]

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 development[15]

HYPOTONIA[1]

- 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 SMA[2]
- Some weakness in the legs and arms may be present[1]
- The baby may have difficulty reaching for and picking up objects[16]
- The baby is unable to stand due to pronounced leg weakness, and unlikely to walk independently[1,3]

AREFLEXIA[2]

- Absent or reduced deep tendon reflexes are characteristic of SMA[2] and a critical part of the baby's exam in cases of hypotonia[9]
- 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 hammer[10]

FINE TREMOR[3]

- 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 present[17]

PROGRESSIVE SCOLIOSIS & JOINT CONTRACTURES[1-3,18]

- The baby may have more severe motion limitations in the lower extremities than upper[19]
- 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 SYMPTOMS[2]

- Restrictive lung disease can result from progressive intercostal muscle weakness[2] particularly if the baby also has scoliosis[1]
- Signs of restrictive lung disease include a reduced total lung capacity and forced vital capacity with preserved expiratory volume[20]

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 lost[2]

REFER URGENTLY TO A PEDIATRIC NEUROLOGIST IF YOU SEE THE SIGNS[4,14]

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