

A quick guide to recognising the red flags of SMA



Spinal muscular atrophy (SMA) and other paediatric neuromuscular disorders (NMDs) are an important cause of infant morbidity and mortality that should not be missed. Clinical suspicion warrants an urgent specialist referral – in most cases, patients experience superior outcomes with timely diagnosis and early intervention (including prompt initiation of drug therapy, where relevant).

1. Does the infant/child have one* or more of these signs?

Signs	Clinical observations
Muscle weakness	Reduced anti-gravity movement of arms and legs Not easily lifting arms and legs from hips and shoulders (proximal)
Hypotonia	Floppy-weak; feels as if they will 'slip out of your hands' when held by underarms 'Frog leg' posture when lying on back
Poor head control	Unable to lift head when lying on belly (if age >3 months) Head lag when pulled to sit (if age >5 months)
Reduced or absent tendon reflexes	Diminished response at knee and elbow
Bright eyed and smiling	Appears to be alert and responsive; not visibly dysmorphic

Source: Adapted from Kolb S, Kissel J. Spinal muscular atrophy. *Neurol Clin* 2015;33(4):831–46.
*Bright eyed and smiling should only elicit concern when accompanying another sign.

**For more information, including a CPD activity,
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2. Is there a delay in achieving, or regression of, motor milestones?

Gross motor milestones [†]	Red flags by age				
	6 months	9 months	12 months	18 months	2 years
	Not holding head up well when prone or in supported sitting	Not rolling Not sitting without support Not moving (eg creeping, crawling) Not taking weight on legs	Not moving (eg crawling, 'commando crawling', bottom shuffle) Not pulling to stand	Not standing independently Not attempting to walk without support	Not able to walk independently Not able to walk up and down stairs holding on

Source: Adapted from Child Development Program and Brisbane North Primary Health Network. Red flags early identification guide. Brisbane North, Qld: Child Development Program, 2016. Available at www.childrens.health.qld.gov.au/wp-content/uploads/PDF/red-flags.pdf [Accessed 14 January 2020].

3. Do parents report these types of concerns?

Attribute	Examples of words and phrases that may be used by parents
Overall concern	'He doesn't seem to be hitting milestones anywhere close to when his brother did or when other kids his age do – he's just not where he should be.' 'Something just isn't quite right.'
Hypotonia and weakness	'He's floppy like a rag doll and just seems weak all over.' 'She can't reach out with her arms or pull herself up from the floor.'
Head control	'She struggles to lift her head.' 'He can't hold his head up or do "tummy time".'
Movement	'She's slow in crawling and won't put any weight on her legs.' 'He has an awkward, funny-looking walk.' 'She doesn't move as much as other kids her age and seems to tire very easily.'

Source: Adapted from National Task Force for Early Identification of Childhood Neuromuscular Disorders. Signs of Weakness by Parent Report. US: National Task Force for the Early Identification of Childhood Neuromuscular Disorders, 2020. Available at www.childmuscleweakness.org/know-the-signs/signs-of-weakness-by-parent-report [Accessed 13 January 2020].

If the answer is YES to questions 1 AND 2 and/or 3, query SMA or other NMD and refer urgently. Where possible, refer the patient to a paediatric neurologist or tertiary paediatric neuromuscular clinic in the first instance, indicating the urgency of your referral. If access to sub-specialty care is limited, refer urgently to a general paediatrician for further assessment, stating that you are concerned about a possible SMA diagnosis.

Resources for healthcare professionals

Visit bewaretherare.com.au for useful clinical resources, including a video library and an RACGP-accredited CPD (Cat. 2) Activity suitable for general practitioners, practice nurses, obstetricians, fertility specialists, general paediatricians and maternal child health nurses.

[†]Aid suitable for children who were born on or after 38 weeks' gestation. If born prior, use adjusted age for milestones.

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