This guide is intended for healthcare professionals only.
Designed and funded by Novartis Gene Therapies
in partnership with the SMA Community.

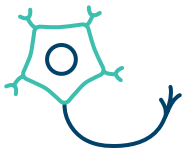
THINK 3 FOR SMA

SMA FACTS AND FIGURES

Spinal muscular atrophy (SMA) is a rare, inherited monogenic disease characterised by lower motor neuron degeneration and muscle weakness.[1,2] It is a devastating and often fatal disease that predominantly first presents in babies and young children.[3]

There are various types of SMA classified into 4 phenotypes (Types 1 to 4)[2] that range in severity and age of onset:[3,4]

SMA Type 1 is the most severe form of SMA with symptoms starting within the first 2 months[3]



Left untreated, 95% of motor neurons are lost by 6 months, with devastating and irreversible effects[1,5]

If left untreated, SMA Type 1 leads to permanent ventilation or death before the age of two years in about 90% of children affected[6]

SMA affects around **1 IN 10,000** live births and can impact any race or sex[3]

Around **70** births are affected by SMA per year in the UK*[7–9]

Approximately **40** of those births are **SMA TYPE 1***[7–9]

Approximately **1 IN 40–60** people is an SMA carrier[3,5]

Untreated, SMA is the **2ND** most common fatal autosomal recessive disorder (a genetic condition where you inherit faulty chromosomes from both parents) after cystic fibrosis, and is the leading genetic cause of infant death[10,11]

EARLY DIAGNOSIS AND INTERVENTION CAN IMPROVE OUTCOMES FOR INFANTS WITH SMA

Infants with SMA Type 1 may appear normal at birth, meaning early signs and symptoms can easily be attributed to late development. However, chances of survival can be increased with early diagnosis and intervention.[5,12,13]

*According to 2018 national birth statistics for England, Scotland, Wales and Northern Ireland.

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TEST FOR 3 SIGNS OF POSSIBLE SMA

1 Head lag[12,14]

Head lag can be seen when baby is being held or when placed on their stomach for tummy time.

2 Hypotonia (floppiness)[15–17]

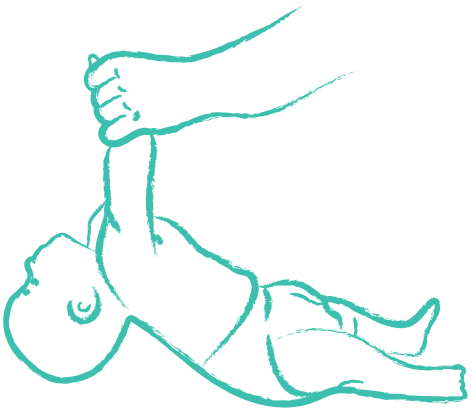
A baby with hypotonia is often described as 'floppy.' They will likely feel limp when held and may be unable to move their arms, legs or neck. Hypotonia may be particularly visible when the baby is held prone.

3 Inability to reach[12,18]

A parent or caregiver may notice that a baby is not raising their arms or reaching for things. When laying baby on their back, hold a toy above them to see if they reach for it.

PULL-TO-SIT TEST:[19]

The pull-to-sit test on babies is a key tool to see how strong a baby's muscles are and can be used to identify head lag and hypotonia. Start with the baby lying flat on their back – grasp their hands and slowly pull them into the sitting position



WHAT TO LOOK OUT FOR:

A typical developing baby will hold their head in line with their trunk, begin to brace their arms and hip flexion is initiated

A baby that is not developing typically will likely have their head lag behind their trunk, little or no brace in their arms resulting in the shoulders pulling forward and no bend in their hips. If this is seen, urgently refer to a paediatric neurologist

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