
# August is SMA Awareness Month

## What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a motor neuron disease. The motor neurons affect the voluntary muscles that are used for activities such as crawling, walking, head and neck control, and swallowing. It is a relatively common "rare disorder": approximately 1 in 6000 babies born are affected, and about 1 in 40 people are genetic carriers.

## What Causes Spinal Muscular Atrophy?

SMA is caused by a missing or abnormal (mutated) gene known as survival motor neuron gene 1 (SMN1). In a healthy person, this gene produces a protein in the body called survival motor neuron (SMN) protein. In a person with mutated genes, this protein is absent or significantly decreased, and causes severe problems for motor neurons. As a child with SMA grows, it is difficult for his/her weakened muscles to keep up with the demands of daily activities. The resulting weakness can also lead to bone and spine changes that may cause breathing problems and further loss of function.

There are four types of SMA: Type I, II, III, and IV.

## Signs and symptoms:

The symptoms vary greatly depending on the SMA type involved, the stage of the disease, and individual factors and they commonly include:

- Areflexia (below normal or absent reflexes), particularly in extremities.
- Overall muscle weakness, poor muscle tone, limpness or a tendency to flop.
- Difficulty achieving developmental milestones, difficulty sitting/standing/walking.
- In infants: adopting of a frog-leg position when sitting (hips abducted and knees flexed).
- Loss of strength of the respiratory muscles: weak cough, weak cry (infants), accumulation of secretions in the lungs or throat, respiratory distress.
- Bell-shaped torso (caused by using only abdominal muscles for respiration).
- Clenched fists with sweaty hands.
- Head often tilted to one side, even when lying down.
- Fasciculations (twitching) of the tongue.
- Difficulty sucking or swallowing, poor feeding.
- Arthrogryposis (multiple congenital contractures).
- Weight lower than normal.

## Treatment:

There is no known cure for spinal muscular atrophy.

***Palliative care*:** Care is symptomatic. Main areas of concern are as follows:

- ***Orthopedics*** — Spine fusion is sometimes performed in SMA I/II patients once they reach the age of 8-10.
- ***Orthotics / Splints*** — Orthotic devices can be used to support the body and to aid walking.
- ***Respiratory care*** — Respiratory system requires utmost attention in SMA as once weakened it never fully recovers.
- ***Nutritional care*** — Difficulties in jaw opening, chewing and swallowing food might put patients with SMA at risk of malnutrition. It is suggested that patients with SMA reduce intake of fat and avoid prolonged fasting.
- ***Mobility*** — Assistive technologies may help in managing movement and daily activity and greatly increase the quality of life.
- ***Cardiology*** — Although heart is not a matter of routine concern, a link between SMA and certain heart conditions has been suggested.
- ***Mental health*** — SMA children do not differ from the general population in their behavior; their cognitive development can be slightly faster, and certain aspects of their intelligence are above the average. Despite their disability, SMA-affected people report high degree of satisfaction from life.

## Three Ways to Help During SMA Awareness Month:

August is SMA Awareness Month. Spinal Muscular Atrophy, or SMA, is a disease that most people don't know about. One of the biggest goals of "Fight SMA" is to make sure more people know what Spinal Muscular Atrophy is, how it impacts families and individuals, and how we are working to fight it. You can make more people aware of SMA by the following approaches:

- ***Tell everyone*:** This idea has the benefit of not costing a thing. If you have a child with SMA or you have SMA yourself, you probably find that people are curious. Do not ignore them. Tell them about the disease and what it does.

- ***Donate*:** Fighting a killer takes money, and while we understand that times are tight and it costs a lot to care for someone with SMA, a donation of any amount can help and also makes possible programs offering support to families battling SMA.

- ***Lobby your legislator*:** Our fight is for everyone who has been impacted by this disease, and we thank you for fighting with us.

## Reference Links:


- http://www.fightsma.org/blog/fightsma-articles/three-ways-to-help-during-sma-awareness-month/
- http://everydaychildhood.com/2011/08/august-is-sma-awareness-month-what-is-sma/
- http://www.fsma.org/FSMACommunity/understandingsma/WhatIsSMA/
- http://en.wikipedia.org/wiki/Spinal_muscular_atrophy#Treatment


**For any enquiry or assistance please contact:** wellness@medicaretpa.co.in