



SMA Fact Sheet

What is SMA?

SMA stands for **Spinal Muscular Atrophy**

SMA is a devastating, genetic disease. It is a leading genetic killer of infants and toddlers, with 50% of the most severely diagnosed cases resulting in death by the age of two. Children with a less severe form of SMA face the prospect of progressive muscle wasting, loss of mobility and motor function. But their minds are unaffected resulting in bright, intelligent children with varying degrees of physical impairment.

- SMA is currently incurable and often fatal
- 1 in every 6,000 births is affected by SMA
- 1 in every 40 people is a carrier of the defective gene that causes SMA
- the child of two carriers has a one in four chance of developing SMA
- SMA affects motor neurons in the spinal cord. This results in muscular weakness, leading to severe disability and the possibility of premature death

SMA is currently incurable

There are three types of SMA that affect children:

- **Type I** is the most common form and is the severest type. Onset typically occurs before 6 months of age. Weakness is severe and manifests in difficulties moving, eating, swallowing and breathing. The proximal muscles, or muscles closest to the trunk such as the neck, shoulder and pelvic girdle muscles, are most significantly involved. SMA Type I babies have floppy limbs and tongue fasciculations (flickering).
- **Type II** is less severe. Onset typically occurs between 6 and 18 months of age. These children are able to sit at some point in their lives but never achieve the ability to walk. The most significant weaknesses are manifest in their proximal muscles. Physical therapy and orthopaedic evaluations can monitor the progression of joint limb contractures and scoliosis (a curved or rotated spine) development. Providing appropriate exercises and bracing may slow progression. Self-initiated mobility is often attained with electronic wheelchairs. Respiratory health is a concern and should be monitored to avoid dangerous respiratory failure and chest infections.
- **Type III** is the mildest of the three types. Babies appear normal at birth, and diagnosis is generally made when they are over the age of two. Children with Type III range from those able to take a few steps to those who can ambulate throughout their home and community. As some children grow, their larger bodies and heavier weight make walking and other activities more difficult. A child may require a wheelchair to navigate their environment. Individual assessments can help create tailored stretching/exercise programs.

Bright young children are trapped within disabled bodies.



What causes SMA?

SMA is an autosomal recessive disease:

- “autosomal” diseases are inherited through non-sex chromosomes
- “recessive” genetic diseases only occur when genes from both parents are abnormal
- SMA is the most common autosomal recessive disease to cause infantile death and disability
- Most people have two Spinal Motor Neuron 1 (SMN1) genes
- 1 in 35 people has only one SMN1 gene. But those with just a single SMN1 gene still lead healthy, normal lives. They are SMA carriers
- When two SMA carriers have a child together, there is a 50% chance that the child will be a SMA carrier.
- When two SMA carriers have a child together there is a 1 in 4 chance that the child will have no SMN1 genes at all. They have been "deleted". This causes SMA.

How does SMA work?

In SMA, the absence of the SMN1 gene results in the production of low levels of a protein (SMN) necessary for the survival of motor neurons

- Motor neurons are the nerve cells that connect the brain and spinal cord to the muscles. Without them, muscles cannot be controlled and wither away
- Progressive loss of motor neurons in the spinal cord causes muscle atrophy
- Muscular atrophy of the trunk can lead to skeletal deformities. Muscular atrophy of the breathing muscles can lead to fatal respiratory problems. Muscular atrophy of the swallowing muscles can lead to difficulties eating and swallowing.

How many people have SMA?

In the UK **at any one time**, between 5,500 and 6,000 people have SMA

What is being done?

Rapid advances have been made since the SMN1 gene was first identified. Treatment pathways are under scrutiny:

- Mouse and fruit fly models have been developed which provide the means for high throughput screening. High throughput screening is a means of testing many thousands of possible treatment compounds using robotics and computers. This electronic means of testing large number of chemicals has significantly speeded up the process which allows us to find out if a particular compound has any therapeutic value for SMA.



- Early clinical trials are taking place
- Numerous research projects are underway.

The Good News

Although there is no cure, advances in supportive therapies have led to increased longevity and improved quality of life, proving that the disease progress is amenable to change.

- Type I: Advanced nutritional and respiratory support has been shown to improve the longevity of children with Type I, with up to 50% of children living past 2 years old.
- Type II: Advances in respiratory support and management of scoliosis, a curved or rotated spine, have contributed to healthier lives with improved quality of life.
- Type III: Assistive devices and tailored programs have the capacity to increase independence and minimize disability.

Continued aggressive research efforts hold the key to a future treatment

The SMA Trust is dedicated to finding that cure

For more information see www.smatrust.org