

Infantile spinal muscular atrophy

Nerve cells called motor neurones link muscle fibres to the nervous system. The instruction to contract a muscle is sent from the brain along the spinal cord and through the motor neurones to the muscle fibres. Spinal muscular atrophies are inherited disorders characterised by the deterioration of these motor neurones. As a result, the muscles don't receive messages from the nervous system. This lack of movement causes the muscles to weaken and wither away. Motor neurones in the brain stem may also be affected. The brain stem sits on top of the spinal cord and oversees the functioning of the face, jaw, tongue, eyes and throat. Spinal muscular atrophies are categorised by the age of onset. The symptoms of infantile spinal muscular atrophy (infantile SMA) appear before the age of two years, and are sometimes present at birth. A child with infantile SMA rarely survives beyond the age of three years. There is no cure.

Different categories according to age

Spinal muscular atrophies (SMA) are categorised by the age of onset. The categories include:

- **Infantile SMA** - also known as Type 1 SMA or Werdnig-Hoffman disease
- **Intermediate SMA** - Type 2 SMA
- **Juvenile SMA** - also known as Type 3 SMA or Kugelberg-Welander disease
- **Adult onset SMA** - Type 4 SMA.

Symptoms

Infantile SMA is the most severe form. The symptoms include:

- Muscle weakness
- Poor muscle tone
- Weak cry
- Limpness or a tendency to flop
- The legs tend to be weaker than the arms
- Feeding difficulties
- Increased susceptibility to respiratory tract infections
- Developmental milestones, such as lifting the head or sitting up, can't be reached.

The onset of symptoms

Generally speaking, the child's survival depends on the age of onset of the condition. The earlier the symptoms appear, the shorter the expected life span. In some cases, the baby stops moving in the later stages of its mother's pregnancy. Sometimes, the symptoms don't appear for days, weeks or even months after birth. The onset can be sudden and dramatic, or very gradual. However, once the symptoms appear, the child quickly deteriorates.

Faulty genes

Infantile SMA is caused by a faulty gene. If a couple already has a child with the disorder, each of their subsequent children has a 25 per cent chance of inheriting the disorder too. Genetic counselling is available for these couples.

Pneumonia is a common complication

A child with infantile SMA is prone to respiratory infections. Pneumonia is a type of lung infection where the smallest airways, called the alveoli, are blocked with mucus and secretions. In healthy people, pneumonia can be simply treated with antibiotics. However, a child with infantile SMA is already in a weakened and vulnerable state. Pneumonia is the cause of death in the majority of cases.

There is no cure

Infantile SMA is a fatal disorder and there is no cure. Treatment can only ease any associated complications. For instance, since a child with infantile SMA is prone to respiratory infections and pneumonia, treatment focuses on trying to maintain the child's lung function and health. Usually, a team of professionals - paediatricians, physiotherapists, neurologists, and respiratory physicians and therapists - work together to help improve the child's quality of life.

Where to get help

- Your doctor
- Paediatrician
- Muscular Dystrophy Association Tel. (03) 9320 9555 or 1800 656 632

Things to remember

- Infantile spinal muscular atrophy is an inherited condition.
- The nerve cells that service the muscles don't work properly, causing muscle weakness and withering.
- A child with infantile spinal muscular atrophy rarely lives beyond three years of age.
- There is no cure.

Want to know more?

Go to [More information](#) for support groups, related links and references.

This page has been produced in consultation with, and approved by:

Muscular Dystrophy Association

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