Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a disease that affects the nervous system. It damages the nerve cells that control how muscles work. As a result, muscles slowly weaken and waste away.

SMA is a genetic disease. A person can get it only if they inherit the gene for it from both parents.

Types of spinal muscular atrophy

SMA has four types. Three of them affect children. Each has different symptoms. And each starts at a different age.

- **Type 1.** This is known as Werdnig-Hoffman SMA. It's also called infantile-onset SMA. A child with this type often has symptoms right after birth. A baby may have trouble moving, swallowing, and feeding. The baby may not be able to hold up their head. A child with this severe form of SMA usually dies within 2 to 6 years. Children with type 1 often die from breathing problems.
- **Type 2.** This is called juvenile or chronic SMA. A child with this type is often diagnosed between ages 6 and 18 months. The main symptom is muscle weakness all over the body. This causes trouble walking and standing. Children with type 2 may need help moving around. They may need a walker or wheelchair. They often live into adulthood. But they're more likely to get respiratory infections.
- **Type 3.** This is a milder form. It can affect children up to their teen years. The most common symptoms include clumsiness, trouble walking and climbing steps, fine tremor, and muscle weakness. Children with type 3 often have trouble getting up from a seated position.
- **Type 4.** This is a less common, milder form. Symptoms may not start until after age 30. Most people with type 4 have a typical life span.

Diagnosing spinal muscular atrophy

SMA is often diagnosed after symptoms start and after testing is done. For instance, early signs of muscle weakness may point to the disease. Your child's healthcare provider will also do a physical exam and ask about your child's health history.

Other tests can confirm SMA. These include:

- **Blood tests.** These check for the gene that causes SMA.
- **Muscle biopsy.** The healthcare provider takes a sample of muscle tissue to look at under a microscope.
- **Electromyogram.** This test can find electrical problems in nerves and muscles.

Treating spinal muscular atrophy

SMA can't be cured. So finding it early is important. Treatment may depend on your child's age, their overall health, and the type of SMA they have.

Nusinersen is a medicine to treat children and adults with SMA. This medicine is given by shot (injection) around the spinal cord.

Children with SMA may have problems breathing and swallowing. This is because the disease affects the muscles that control these functions. Children with severe cases are especially at risk for these problems. They may need a breathing machine or nutritional care.

Weak arm and leg muscles can make it hard for a child to walk and stand. A wheelchair or walker can help keep a child independent. Physical therapy and exercise can help, too. They can make it easier to move and stay flexible.

Many children with SMA also have scoliosis. Scoliosis is a condition that causes the spine to curve. A healthcare provider may recommend special devices to help prevent this problem. Surgery may be needed to fix it.

A healthy diet can help manage SMA. But chewing and swallowing problems can make it a challenge to follow a healthy diet.

Exercise is important for overall health and well-being. But it must be done in a way that protects the joints. Check with your SMA specialist before starting an exercise program.

Talk with a healthcare provider who treats SMA if you have questions about diet or exercise. Another resource is the Muscular Dystrophy Association.