Spinal Muscular Atrophy: Frequently Asked Questions

What is Spinal Muscular Atrophy?

Spinal muscular atrophy (SMA) is a condition that causes muscle weakness and atrophy (when muscles get smaller.) SMA can affect a child's ability to crawl, walk, sit up, and control head movements. Severe SMA can damage the muscles used for breathing and swallowing.

There are four types of SMA. Some show up earlier and are more severe than others. All types of SMA need ongoing treatment by a medical care team.

What are the symptoms of SMA?

SMA is characterized by a progressive loss of muscle control, muscle movement, and increased muscle weakness. SMA type 1 patients generally start to show symptoms early in life. The disease becomes more severe over time.

People with SMA may progressively lose, or never acquire the ability to walk, stand, sit, or even move. Normal growth and development can place additional stress on already weakened muscles. Further, SMA patients often develop respiratory illnesses, and bone and/or spinal deformities, which may require surgical treatment.

What is the cause of SMA type 1?

SMA is caused by a problem with a gene called Survival Motor Neuron 1 (*SMN1*). The SMN1 gene is the main gene responsible for creating a protein called SMN protein. SMA is a result of reduced levels of this protein.

SMN protein plays a critical role in the survival of special types of nerve cells in the spinal cord called motor neurons. Motor neurons are responsible for controlling muscle movement. When there is not enough SMN protein produced, these neurons become sick and have trouble sending signals to various muscles. This lack of SMN protein makes muscles become smaller and weaker over time.

How is SMA type 1 inherited?

SMA is an autosomal recessive disease, caused by a deletion or mutation in the SMN1 gene. This means for a person to have SMA, both copies of their SMN1 gene (one inherited from their mother and the other from their father) are defective.

Having one normal SMN1 copy, and one defective copy will allow a person to produce enough SMN protein to prevent any symptoms of SMA. Such individuals with one normal and one defective SMN1 copy are called *carriers*.

How common is SMA?

Approximately 1 in 50 Americans, or about 6 million people, is a carrier of SMA. Carriers do not have SMA symptoms, but they have or '*carry*' mutations in the SMN1 gene. Thus, parents could potentially pass it along to their offspring. If both parents are carriers of the defective SMN1 gene, each of their children has a 1 in 4 chance (25%) of having the disease, or 1 in 2 chance (50%) of being a carrier.

One in every 6,000 to 10,000 children is born with this disease. SMA is believed to affect as many as 10,000 to 25,000 children and adults in the US, making it one of the most common rare diseases. The number of people living with the disease is comparable to other rare diseases such as Cystic Fibrosis (CF) and Duchenne muscular dystrophy (DMD).

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What type of SMA affects newborns?

Type 1, Acute Form of SMA (**Werdnig-Hoffmann**): About 60% of patients with SMA are born with this form of the disease. Newborns typically exhibit limited movement and have difficulty holding their heads straight, feeding, and swallowing. Reduced strength in the chest muscles often results in labored breathing with the chest appearing sunken. The progressive weakening of the muscles leads to respiratory infections and lung collapse. Without treatment, death occurs around the age of two years***.***

How is SMA diagnosed?

Infants with SMA generally appear normal at birth, with symptoms developing as early as a few months after they are born. Symptoms of SMA often overlap with other neuromuscular diseases. Consequently, a diagnosis of SMA can be challenging.

A diagnosis of SMA is usually made by a pediatric neurologist and confirmed by a blood test, which is designed to identify defects in the SMN1 gene.

What treatments exist for SMA?

In the past, many infants with SMA lived only into early childhood. Today those odds are improving. Gene replacement therapy can extend the lives of babies with SMA types 1 and 2. It also helps people with late-onset disease.

The first FDA-approved prescription medicine for SMA in pediatric and adult patients was nusinersen, also known as Spinraza®. This drug is delivered directly to the central nervous system (CNS) where motor neuron loss begins. After initial loading doses, the drug is given 3 times a year.

Zolgensma® (onasemnogene abeparvovec) and Evrysdi® (risdiplam) are forms of gene therapy that treat the genes involved in SMA. The SMN1 and SMN2 genes give the body instructions for making a protein that helps with controlling muscle movement.

Getting care from a team of medical specialists helps many children with SMA live well into adulthood. This is especially true when the disease is diagnosed in an older child. The care team may include:

- a neurologist - a medical specialist of the nervous system
- a geneticist - a medical expert in the variation of inherited characteristics
- a dietitian - an expert on diet and nutrition
- a respiratory therapist - cares for patients who have trouble breathing

Physical therapy and occupational therapy can also help.

Looking Ahead

Learning that a child has SMA can be hard for any family. It is important to remember that this complex condition affects every child differently.

For information and advice, speak with your child's primary health care provider and members of your child's specialist team. You can connect with online resources and support groups such as:

https://www.babysfirsttest.org/newborn-screening/conditions/spinal-muscular-atrophy

https://www.curesma.org/

https://smafoundation.org/