SMA Awareness & Activism

Spinal Muscular Atrophy (SMA) is a genetic neuromuscular disease that is characterized by muscle atrophy and weakness. The disease usually appears early in life and is currently the leading genetic cause of death in infants and toddlers.

What's the Cause?

SMA is caused by defects in the Survival Motor Neuron 1 otherwise known as the SMN1 gene that is in charge of encoding the SMN protein.

SMN Protein

The SMN protein is in charge of keeping our body healthy and helping nerve cells in the spinal cord survive. The nerve cells that are located in our spinal cord are responsible for muscle contraction and motor neurons. SMN is found in the cytoplasm of all animal cells and is also found in the nuclear gems.

Many neurogenetic disorders are caused by the mutation of the SMN1 gene. In Spinal Muscular Atrophy there is a loss or mutation of the SMN1 gene causing reduced SMN protein levels and a dysfunction of motor neurons.

Neurogenetic Disorders

Other Neurogenetic Disorders that are caused by mutations in genes like SMA include:

- Lateral sclerosis
- Huntington's disease
- And several others

Cure SMA Organization

Ways to Give

Donate

The SMA Cure Organization has a link on their website http://www.curesma.org/ where you can make a secure online donation.

Fundraise for Cure SMA

You can run your own fundraiser by planning any event you would like, or you can participate in an event that is already planned. There is also the opportunity to set up a donation page in honor of a birthday, anniversary, or personal celebration. If you would like to plan your own event or join an event that is already scheduled visit Cure SMA's website.

Merchandise

Cure SMA bracelets, t-shirts, and more are available on purchase on Cure SMA's website. All money raised from merchandise goes directly to the organization.

Diagnosis

Testing

SMA diagnosis must be confirmed through genetic testing.

SMA is usually diagnosed in one of three ways:

- Through genetic testing, after an infant or child shows symptoms of SMA
- Through a positive newborn screening result
- Through prenatal testing

If a doctor suspects SMA, they may:

- Order genetic testing through a blood sample
- Refer the patient to a neurologist who will perform an examination, and then order a genetic testing to confirm the diagnosis.

"Don't be ashamed of your story, it will inspire others."

-Unknown

SMA Facts

- SMA affects approximately 1 in 11,000 babies, and about 1 in every 50 Americans is a genetic carrier.
- SMA can affect any race or gender.
- There are four primary types of SMA – I, II, III, IV – based on age of onset and highest physical milestone achieved.
- Individuals of SMA have difficulty performing the basic functions of life, like breathing and swallowing.
- SMA does not affect a person's ability to think, learn, and build relationships with others.
- Cure SMA has the first-ever approved treatment that targets the underlying genetics of SMA and they are on the verge of several other breakthroughs.