Spinal Muscular Atrophy

What is SMA?

- Spinal muscular atrophy is a degenerative problem that affects the motor nerves, resulting in muscle wasting and weakness.
- Spinal muscular atrophy occurs in approximately one in 6,000 -10,000 live births.
- Is indiscriminant of age, sex, race
- 1 in every 40 people carry the gene that causes SMA
- 7.5 million Americans are carriers

What causes spinal muscular atrophy?

- SMA is an autosomal recessive disease. This means that two abnormal copies of the gene, one inherited from each parent, are necessary to have the condition.
- Boys and girls are affected with equal frequency.
- A child who inherits only one abnormal gene copy is a carrier, and is not a risk to develop symptoms. A child of two carriers has a ¼ chance of developing SMA
- A gene called survival motor neuron (or SMN) is found to have an abnormal area (called a deletion) in over 95 percent of cases of SMA. Symptomatic individuals of all ages can be tested through DNA studies typically done from a blood sample.

What are the symptoms of spinal muscular atrophy?

- Spinal muscular atrophy is sometimes difficult to diagnose, as symptoms can resemble other conditions or medical problems. Each child may experience symptoms differently. There are several types of spinal muscular atrophy based on symptoms and age of onset.

Type I (Werdnig-Hoffman)

- This is the most severe type of SMA, and unfortunately, the most common. Symptoms may be present at birth or develop within the first few weeks or months after birth.
    - Infants have difficulty holding up their head, sucking, feeding, swallowing, and often move very little.
    - The legs are more severely affected than the arms.
    - The muscles of the chest which help to expand the lungs are affected, and the chest may appear small or "bell-shaped".
    - They have a weak cough, and are prone to respiratory infections. Complications from breathing problems often lead to death or dependence on some form of respiratory support by 2 to 3 years of age.
    - The tongue may demonstrate "worm-like" movements, and they may demonstrate a tendency to choke while feeding.

Type II (intermediate form)

- This form of SMA most commonly becomes evident in children between 6 months to 2 years of age.

- They may show delays in acquiring motor skills such as rolling, sitting or crawling. They are unable to walk independently without support. They typically have generalized muscle weakness and may require braces, walkers, or a wheelchair for assistance.
- Life-expectancy varies greatly in this group of children, since they demonstrate a very wide range in degree of weakness. However, complications commonly include weakness of chest muscles involved in breathing, resulting in a weak cough and tendency for pneumonia.
- Scoliosis develops in virtually all children at some point, and they are prone to bone fractures. Children who are unable to bear weight often develop hip dislocation. Contractures of the muscles and joints can limit function over time.
- Children in this group may also demonstrate difficulties in swallowing and chewing, and require close monitoring of nutrition.
- Many children survive well into adulthood.

Type III (Kugelberg-Welander)

- This form of SMA most commonly becomes evident in children between 2 and 17 years of age.
    - Delays in motor development, difficulty walking, trouble getting up from the floor, mild muscle weakness, and frequent falls.
    - Fatigue can be a significant problem, which limits the ability to walk long distances. A tremor involving the hands is common.
    - Scoliosis is frequent in later childhood.
    - Respiratory muscle involvement is much less often a problem, and difficulty swallowing is rare.

Type IV

- This form of SMA includes those individuals who don't develop symptoms of weakness until they reach adulthood.
    - Usually, this results in muscle weakness predominantly affecting the legs, and manifests as a walking disability.
    - The symptoms of spinal muscular atrophy may resemble other problems or medical conditions, and can be confused with other muscle or nerve conditions, including muscular dystrophy, myopathy, other spinal muscular atrophy variants or even forms of amyotrophic lateral sclerosis (ALS).
    - Another closely related condition, known as spinobulbar muscular atrophy (Kennedy's disease), can also present in late childhood or adulthood.

How is spinal muscular atrophy diagnosed?

- The diagnosis of spinal muscular atrophy may be suspected if you or your child demonstrate specific symptoms or demonstrate signs on examination that are consistent with the pattern of weakness seen in this disorder.
- During the physical examination, your child's physician will obtain a complete medical history, and he/she may also ask if there is a family history of any medical problems.
- Diagnostic tests which can help to confirm the diagnosis of spinal muscular atrophy include:
    - ***Blood tests:*** Genetic testing can confirm a suspected diagnosis in most cases. Sometimes the initial genetic test is negative, and additional genetic testing or other testing may be needed to confirm a diagnosis.
    - ***Electromyogram (EMG):*** A test that measures the electrical activity of a muscle or a group of muscles. An EMG can detect abnormal electrical muscle activity due to diseases and neuromuscular conditions.
    - ***Muscle biopsy:*** A small sample of the muscle is removed and examined to determine and confirm a diagnosis or condition.
    - ***MRI or imaging studies:*** Imaging studies of the brain or spine to help rule out other conditions

Management & Treatment

- No specific pharmaceutical therapies which have been confirmed which can either extend lifespan or increase strength in SMA subjects
- The identification of compounds that can increase SMN protein (higher levels reduce severity of SMA) in cells from SMA subjects and in genetic animal models of SMA is promising.
- Proactive management strategies to optimize lung function, physical mobility and nutrition can help preserve motor function, improve quality of life and extend survival, particularly in more severely affected SMA infants and young children.
- Close work with parents, especially in milder cases of SMA, in order to anticipate problems and pursue management aggressively to optimize outcomes.
- Because of the tremendous variability in severity of muscle weakness, an individualized approach is often necessary. Often, respiratory, nutritional and even physical rehabilitation interventions are reactive rather than proactive.
- Treatment for the intermediate or milder forms of SMA should focus on preserving mobility and minimizing respiratory complications, particularly restrictive pulmonary disease or respiratory compromise due to progressive scoliosis.

Resources:

Families of SMA: Research. Support. Hope.
http://www.fsma.org/FSMACommunity/UnderstandingSMA/

Spinal Muscular Atrophy FAQ
http://www.smafoundation.org/faq

Facts About SMA
http://www.fightsma.org/index.php?sma-guidebook

Facts About SMA: From the Muscular Dystropy Association
http://www.mda.org/publications/fa-sma-qa.html

Genetic Conditions: Spinal Muscular Atrophy
http://ghr.nlm.nih.gov/condition=spinalmuscularatrophy