Spinal Muscular Atrophy (SMA) is a neuromuscular disease characterized by degeneration of motor neurons resulting in progressive muscular atrophy (wasting away) and weakness. The motor neurons affect the voluntary muscles that are used for activities such as walking, running, crawling, holding up your head and swallowing. SMA also affects muscles throughout the body with the most severely affected being proximal muscles (shoulders, hips, and back). Weakness of both legs and arms also occurs. Involvement of respiratory muscles (muscles involved in breathing and coughing) can lead to an increased tendency for pneumonia and other lung problems. Sensation and the ability to feel are not affected. Intellectual activity is normal and it is often observed that patients with SMA are unusually bright and sociable.

Quick Facts about Spinal Muscular Atrophy

- SMA is the number one genetic killer of children under the age of two.
- One in every 6,000 babies is born with SMA.
- SMA can strike anyone of any age, race or gender.
- One in every 40 people carries the gene that causes SMA.

Diagnosing Spinal Muscular Atrophy

In order to be diagnosed with SMA, symptoms need to be present. In most cases a diagnosis can be made by the SMN gene test, which determines whether there is at least one copy of the SMN1 gene by looking for its unique sequences (that distinguish it from the almost identical SMN2) in exons 7 and 8. In some cases, when the SMN gene test is not possible or does not show any abnormality, other tests such as an EMG or muscle biopsy may be indicated.

Types of Spinal Muscular Atrophy (SMA)

- Type I (Werdnig Hoffmann disease) is the most severe, and manifests in the first year of life. This type generally onsets quickly and unexpectedly after birth; babies diagnosed with Type I SMA do not generally live past one year of age without intensive respiratory support.
- Type II or intermediate SMA, describes those children who are never able to stand and walk, but who are able to maintain a sitting position at least some time in their life. The onset of weakness is usually recognized sometime between 6 and 18 months. It is known to vary; some patients gradually grow weaker over time, while others through careful maintenance avoid any progression.
- Type III (Kugelberg-Welander or Juvenile Spinal Muscular Atrophy) Symptoms typically appear between eighteen months and early adulthood. People with SMA Type III often exhibit difficulty walking, have mild muscle weakness and are at risk for respiratory infections. These patients have a normal life expectancy.
- Type IV, Adult Form of SMA: A less common form of SMA that afflicts adults and is characterized by a slower progression of symptoms that particularly affect walking. Symptoms typically emerge after age 35.

Sources: Wikipedia and Families with SMA