

Spinal Muscular Atrophy

- Spinal muscular atrophy (SMA) is a genetic neuromuscular disorder characterized by the degeneration of motor neurons in the spinal cord (a type of motor neuron disease).
- The loss of motor neurons leads to progressive symmetrical muscle weakness and wasting of voluntary muscles as well as dysphagia (problems swallowing), dyspnea (problems breathing), and in severe cases, to death.
- Spinal muscular atrophy usually affects the muscles closer to the center of the body (proximal muscles) more than the muscles farther from the center (the distal muscles).
- Intellectual activity is normal and it is often observed that patients with SMA are unusually bright and sociable. Sensation and the ability to feel are not affected.
- SMA affects approximately 1 in 6000 to 1 in 10,000 live births and is the leading genetic cause of infant mortality. It is the second most common lethal, autosomal recessive disease in Caucasians (after cystic fibrosis).
- Four types of SMA are identified based upon varying clinical presentations:
 - Spinal Muscular Atrophy I (SMA I): (severe) Onset from birth to 6 months, child is severely affected (cannot sit without help) and with rapid mortality due to respiratory complications, life expectancy is less than 2 years (death may occur in the womb). Over 50% of SMA patients have this most severe form of the disease. Also called Werdnig-Hoffmann disease.
 - Spinal Muscular Atrophy II (SMA II): (intermediate) Onset from 6 to 18 months, child is seriously affected (but can sit without help) and shows varying degrees of muscle weakness. Life expectancy is variable.
 - Spinal Muscular Atrophy III (SMA III): (mild) Onset after 18 months, these children have less weakness and usually have a normal life expectancy. The disorder progresses slowly, with the ability to walk usually lasting until into adolescence. A wheelchair is often required later in life. Also called Kugelberg-Welander disease.
 - Spinal Muscular Atrophy Type IV: (adult onset) Onset after age 35, much less common than the other forms, it is characterized by a gradual onset and very slow progression.
- A number of secondary physical issues may arise, notably, bone and spinal deformities. As well, typically children with SMA have inborn errors of fatty acid and protein metabolism and many benefit from a special diet.
- Diagnosis: The first diagnosis of SMA is based on clinical observations and this is usually confirmed through genetic testing.

- Cause: A genetic mutation in the SMN1 gene that makes a protein called SMN1, disrupts normal protein production and there is not enough in the nerve cells to properly operate. The motor neurons die off and this in turn affects the transmission of nerve signals from the brain to the muscles. The muscles cannot function properly and they eventually shrink and die.
- In general, the severity of SMA corresponds to the amount of SMN protein available in cells. Why low levels of the SMN protein lead to the specific loss of motor neurons is a central question in SMA research.
- Genetic Testing: there is a widely available test that is both very specific and very accurate for SMA that detects problems with the SMN1 gene.
- Inheritance: SMA is inherited in an autosomal recessive manner. In order for a child to be affected by SMA, both parents must be carriers of the abnormal gene and both must pass the mutated gene on to the child. About 1 in every 30 to 60 Caucasians appear to be carriers of the SMA mutation.
- Treatments: There are currently no treatments for SMA that can halt or reverse the symptoms and muscle weakness. Each symptom displayed reflects an underlying issue and each needs to have a treatment plan developed to best address the concern on an individual basis.
- ANESTHESIA WARNING: A higher rate of complications is generally associated with general anesthesia in patients with neuromuscular disorders. Even mildly affected patients may have complications. Patients with SMA or a family history of SMA need to discuss this with their surgeons.

Neuromuscular disorders and what people commonly call muscular dystrophies are very complex and often devastating conditions. Some strike children, others do not display symptoms until well into adult life. Most are progressively disabling, getting worse over time and some are fatal, often in childhood. Some affect primarily muscles, others can affect a wide spectrum of health, including; personality and intellectual functioning, the eyes, the bones, digestion and diabetes to list just a few. There are over 100 different types of disorders and variations listed under this umbrella. Many disorders have genetic causes but in many cases, just how these genetic mutations cause the symptoms we see is not yet understood. Every day, medicine is making progress in understanding these disorders, but at the present time, there are more theories than facts. Sadly there are no direct treatments yet.

Please feel free to contact Muscular Dystrophy Canada if you have suggestions or questions about this page. Our website is: <http://www.muscle.ca/> or e-mail us at info@muscle.ca

Thank you.