Spinal Muscular Atrophy

What is Spinal Muscular Atrophy?

Spinal muscular atrophy (SMA) is a hereditary neuromuscular disorder that causes severe muscle weakness and progressive loss of voluntary muscle control. Muscles that control breathing, swallowing, head and neck control, walking, and crawling are the most severely affected. There are different types of spinal muscular atrophy that range in severity and age of onset. In spinal muscular atrophy type 1 (the most common type), muscle weakness begins in the first few months of life, infants are unable to sit without support and they have difficulty breathing and swallowing. Life expectancy is typically less than two years. In milder types of SMA (types 2 and 3), symptoms can develop in adolescence or adulthood and may include muscle weakness, tremors, and twitching. Intelligence is not affected.

How Common is Spinal Muscular Atrophy?

The incidence of spinal muscular atrophy is approximately 1 in 10,000 live births. The general population carrier rate is approximately 1 in 50. However, specific carrier frequency estimates differs by ethnic group.

What Causes Spinal Muscular Atrophy?

SMA is a hereditary disorder caused by a change, or mutation, in both copies of the Survival Motor Neuron 1 gene (SMN1), which causes these genes to work improperly or not work at all. Individuals with SMA have two non-working copies of this gene. SMA is inherited in an autosomal recessive manner. This means that, in most cases, both parents must be carriers of a mutation in one copy of SMN1 to have a child with SMA. If the mother and father are both found to be SMA carriers, there is a 1 in 4, or 25%, chance in each pregnancy for both partners to pass on their SMA gene mutations to the child, who will then have SMA. Males and females have an equal chance of being affected with SMA.

Is Spinal Muscular Atrophy Treatable?

There is currently no cure for SMA. Treatments are supportive to help manage symptoms and improve the quality of life.

What Does it Mean to be a Carrier of Spinal Muscular Atrophy?

Carriers are usually healthy and do not have symptoms of the condition themselves, but do have an increased chance of having a child affected with SMA.

Resources

Claire Altman Heine Foundation: www.clairealtmanheinefoundation.org

Families of SMA: http://www.fsma.org.hk/
Citations