Spinal Muscular Atrophy

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Spinal Muscular Atrophy Information Page.

Spinal Muscular Atrophy refers to a group of hereditary diseases that damages and kills specialized nerve cells in the brain and spinal cord (called motor neurons). Motor neurons control movement in the arms, legs, face, chest, throat, and tongue, as well as skeletal muscle activity including speaking, walking, swallowing, and breathing. The most common form of SMA is caused by an abnormal or missing gene known as the survival motor neuron gene 1 (SMN1), which is responsible for the production of a protein essential to motor neurons. This form of SMA has four types: • Type I, also called Werdnig-Hoffman disease or infantile-onset SMA, is usually evident before 6 months of age. The most severely affected children will have reduced movement and chronic shortening of muscles or tendons (called contractures). Other children may have symptoms including reduced muscle tone, lack of tendon reflexes, twitching, skeletal abnormalities, and problems swallowing and feeding. Without treatment, many affected children die before age 2 years. • SMA Type II is usually first noticed between the 6 and 18 months of age. Children can sit without support but are unable to stand or walk unaided. Children also may have respiratory difficulties. Life expectancy is reduced but most individuals live into adolescence or young adulthood. • SMA Type III (Kugelberg-Welander disease) is seen after age 18 months. Children can walk independently but may have difficulty walking or running, rising from a chair, or climbing stairs. Other complications may include curvature of the spine, contractures, and respiratory infections. With treatment, most individuals can have a normal lfespan. • Individuals with SMA Type IV develop symptoms after age 21 years, with mild to moderate leg muscle weakness and other symptoms.