Spinal Muscular Atrophy

Spinal Muscular Atrophy (SMA) Types I, II, III and IIV belong to a group of hereditary diseases that cause weakness and wasting of the voluntary muscles in the arms and legs of infants and children. The disorders are caused by an abnormal or missing gene known as the survival motor neuron gene (SMN1), which is responsible for the production of a protein essential to motor neurons. Without this protein, lower motor neurons in the spinal cord degenerate and die.

The type of SMA (I, II, III or IV) is determined by the age of onset and the severity of symptoms. Type I (also known as Werdnig-Hoffman disease, or infantile-onset SMA) is evident at birth or within the first few months. Symptoms include floppy limbs and trunk, feeble movements of the arms and legs, swallowing difficulties, a weak sucking reflex, and impaired breathing. Type II (also known as juvenile SMA, intermediate SMA, or chronic SMA, has an onset between 6 and 18 months. Legs tend to be more impaired than arms. Children with Type II are usually able to sit without support if placed in position. Some may be able to stand or walk with help. Type III (also called Wolhlfart-Kugelberg-Welander disease, or mild SMA) can begin as early as the toddler years or as late as adolescence. Children can stand alone and walk but may have difficulty getting up from a sitting position. Type IV develops after age 21 and the individual has mild to moderate leg muscle weakness.

Source: NINDS Spinal Muscular Atrophy Information Page
https://www.ninds.nih.gov/health-information/disorders/spinal-muscular-atrophy?search-term=spinal%20muscular%20atrophy

Websites

Cure SMA (formerly Families of SMA)
http://www.curesma.org/
925 Busse Road
Elk Grove Village, IL 60007
Toll-free: 800-886-1762
E-mail: info@curesma.org

Cure SMA is dedicated to promoting and supporting research, helping families cope through informational programs and support, and educating the public and the medical community about SMA. The organization has 29 chapters serving all but 3 states. Offers publications such as Understanding Spinal Muscular Atrophy, Breathing Basics, and Caring Choices.

Never Give Up/Gwendolyn Strong Foundation
https://nevergiveup.org/
27 W. Anapamu St., Suite 177
Santa Barbara, CA 93101
Phone: 805-203-0334
Email: info@nevergiveup.org
Gwendolyn Strong Foundation offers advocacy and assistance to families of SMA children and also continues to offer practical family support for the overwhelming expenses of life with a disability, helping give families greater independence. The Foundation is building an inclusive playground in Santa Barbara, CA.

NINDS: Spinal Muscular Atrophy booklet
https://catalog.ninds.nih.gov/publications/spinal-muscular-atrophy

SMA Foundation
https://smafoundation.org/
970 W. Broadway, Suite E
PMB 140
Jackson, WY 83001
Phone: 646-253-7100, 877-FUND-SMA (Toll-free)
E-mail: info@smafoundation.org
The SMA Foundation promotes the development of a treatment for SMA. The Foundation funds research and fosters information exchanges and alliances among pharmaceutical companies, biotech, academia, non-profits, clinicians, patients, government/regulators, and the media.

Consensus Statement for Standard of Care in Spinal Muscular Atrophy
https://journals.sagepub.com/doi/abs/10.1177/0883073807305788
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