



Spinal Muscular Atrophy

Spinal Muscular Atrophy (SMA) Types I, II, and III 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, or III) 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 Wohlfart-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.

Source: NINDS Spinal Muscular Atrophy Information Page
http://www.ninds.nih.gov/health_and_medical/disorders/sma.htm

Websites

<http://www.curesma.org/>

Families of SMA (FSMA)

925 Busse Road

Elk Grove Village, IL 60007

Phone: 847-367-7620, 800-886-1762 (Toll-free, US and Canada)

E-mail: info@fsma.org

FSMA 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.

<http://www.fightsma.com/>

FightSMA/Andrew's Buddies Corporation

1807 Libbie Avenue, Suite 104

Richmond, VA 23226

Phone: 804-515-0080

FightSMA raises awareness and funding for SMA research.

<http://www.smafoundation.org/>

SMA Foundation

888 Seventh Avenue, Suite 400

New York, NY 10019

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.

<http://www.hopeandlight.org/>

Hope and Light Foundation: Benefiting Children with SMA

1355 Cortez Lane NE

Atlanta, GA 30319

Phone: 678-591-8331

E-mail: info@HopeandLight.org

The Hope and Light Foundation funds research to cure SMA and provides information for families.

<https://smaregistry.iu.edu/>

International Spinal Muscular Atrophy Patient Registry

410 West Tenth Street

HS 4000

Indianapolis, IN 46202-3002

Phone: 866-482-0248

Email: smareg@iupui.edu

The Registry functions as a liaison between patients and families interested in participating in research and researchers interested in studying SMA.

<http://jcn.sagepub.com/content/22/8/1027>

Consensus Statement for Standard of Care in Spinal Muscular Atrophy

This page presents the abstract for an article appeared in the August 2007 issue of *Journal of Child Neurology*. A PDF of the full article can be accessed for free from the site or by going directly to <http://jcn.sagepub.com/content/22/8/1027.full.pdf+html>. The page also lists other articles that cited the consensus statement article.

<http://www.treat-nmd.eu/sma/overview/>

Treat NMD Neuromuscular Network: Spinal Muscular Atrophy

Institute of Genetic Medicine
University of Newcastle upon Tyne
International Centre for Life
Newcastle upon Tyne
NE1 3BZ
United Kingdom
Phone: +44 (0)191-241-8605

This site provides information on SMA to patients and families, as well as doctors and researchers. Topics covered include research, standards of care, patient registries, patient organizations, and other resources.

Chat Rooms

<http://groups.yahoo.com/group/SMAfriends/>

Yahoo! Groups: Spinal Muscular Atrophy Friends

The following books and videos are available for free loan from the PRC library. For more information, please see www.paralysis.org and click *Borrow from Our Lending Library* under PRC Quick Links.

Books

- Allen, Susan and Trina Allen. **Living with Spinal Muscular Atrophy: The True Story of Kassidy Jade Sears**. New York: iUniverse, 2006.
Written by Kassidy Jade Sears' mother and grandmother.
- Baldwin, Helen. **The Jeffrey Journey**. Victoria, Canada: Trafford Publishing, 2003.
CD enclosed. Her baby was born with spinal muscular atrophy.
- Barzach, Amy Jaffe and Sandy Tovray Greenberg. **Accidental Courage, Boundless Dreams**. West Hartford, CT: Aurora Publishing, 2001.
Barzach's son Jonathan died at nine months old from SMA. As a way to heal, the family made a local playground accessible for children with disabilities. Out of this, Boundless Playgrounds, a national non-profit organization was born.
- **Facts About Spinal Muscular Atrophy**. Tucson, AZ: Muscular Dystrophy Association, 2003.
27-page booklet
- Gearhart, Elizabeth Jamsa with Victor Alvarez and Mary Cloud. **VICTORious Life**. McAllen, TX: Knowing Press, 2006.

Story of a young Latino man with spinal muscular atrophy.

- heBGG. **Solving the SMA Puzzle: Complications of SMN Protein Upregulation.** iUniverse, 2007.
Author's daughter has SMA.
- Napolitano, Linda. **Without Laughter and Music, Shoot Me.** Baltimore, MD: PublishAmerica, 2004.
Napolitano has spinal muscular atrophy.
- Wischnowsky, Dave. **Northern IlliNOISE: Tales of a Territory.** Xlibris, 2004.
Includes a chapter on Mark Wiebe, a teen with SMA.
- Zoern, Stacy. **I Like to Run Too: Two Decades of Sitting—A Memoir of growing up with a physical disability.** Chesterfield, MO: Science & Humanities Press, 2006.
Zoern has spinal muscular atrophy.

Videos

- **39 Pounds of Love.** HBO/Cinemax Documentary Films, 2005.
Ami Ankilewitz was diagnosed with an often fatal form of Spinal Muscular Atrophy that severely limits his physical growth and movement. He works in Israel as a 3D animator and travels to the U.S. looking for the doctor who predicted his early demise. He is now 34 years old.
- **Normal for Us: The Miller Twins.** Portland, OR: Oregon Public Broadcasting, 2002. (56 minutes)
Documentary that follows the Miller twins from youth to teenage years. The girls are quadriplegic as a result of spinal muscular atrophy. Their father builds them special wheelchairs so they can maneuver independently around the family farm.

The information contained in this message is presented for the purpose of educating and informing you about paralysis and its effects. Nothing contained in this message should be construed nor is intended to be used for medical diagnosis or treatment. It should not be used in place of the advice of your physician or other qualified health care provider. Should you have any health care related questions, please call or see your physician or other qualified health care provider promptly. Always consult with your physician or other qualified health care provider before embarking on a new treatment, diet or fitness program. You should never disregard medical advice or delay in seeking it because of something you have read in this message.