Spinal Muscular Atrophy (SMA)

Spinal muscular atrophy is a genetic disease that causes progressive muscle weakness; severe cases can limit a person's ability to move, eat and breathe.

Q: What is SMA?

SMA is a disease that affects nerve cells in the brain and spine that control movement and muscle activity. The main areas of the body affected include the trunk, legs, arms, face, chest, throat and tongue. Its symptoms most commonly emerge by early childhood. Depending on the severity of the disease, a child living with SMA may not be able to walk, stand or sit without support. In those cases, the ability to speak, swallow and breath without mechanical ventilation will also be limited.

Q: How common is SMA?

About 1 in 11,000 babies in the U.S. are born with SMA each year.

Q: What causes SMA?

SMA is caused by a mutation of the survival motor neuron gene 1 (SMN1) gene that produces most of the protein needed by the spinal cord nerves that control movement; without the protein, spinal nerve cells die and muscles become weak, leading to the most severe cases of SMA. A different survival motor neuron gene (SMN2) produces smaller amounts of this protein; children and adults with enough functioning copies of the SMN2 gene will typically have milder cases of SMA.

Q: What are the different types of SMA?

The five types of SMA were historically organized into a classification system based

on the age at which symptoms emerged and motor function ability.

- SMA Type 0: This is the rarest and most severe form of SMA and emerges prior to birth. Babies born with Type 0 experience comprehensive muscle weakness, including facial paralysis, and require support breathing and eating. Life expectancy for this form of the disease is typically one to six months.

- SMA Type 1: The most common form of SMA (also called Werdnig-Hoffman disease) typically emerges before a baby is six months old. Symptoms include severe muscle weakness and trouble breathing, coughing, and swallowing.

- SMA Type 2: This form of SMA emerges between 6 and 18 months. Children with SMA Type 2 experience muscle weakness mainly in the trunk and lower limbs, and may need to use assistive devices, such as braces, walkers or wheelchairs, to move.

- SMA Type 3: This form of SMA (also known as Kugelberg-Welander disease) emerges after 18 months. Children with SMA Type 3 may be prone to falling and have challenges running or climbing stairs.

- SMA Type 4: This rare form of SMA typically emerges in adulthood with mild symptoms of muscle weakness that do not affect milestones or life expectancy.

After the first treatment for SMA was introduced in 2016, the terms "non-sitter, sitter and walker" were also adopted by healthcare professionals and families to more accurately describe maximum motor function. This change was made to reflect the new milestone achievements and gains prompted by treatments that surpassed the original motor function predictions set forth within the historical classification system.

Q: What functions are not affected by SMA?

The disease does not affect mental function, vision and hearing, the ability to feel sensations, or bowel and bladder function.

Q: How is SMA diagnosed?

Symptoms of SMA may be similar to other neuromuscular conditions such as muscular dystrophy. Doctors will review family history, taking note of any motor difficulties and lack or reflexes, and order a genetic blood test to identify mutations or deletions of the SMN1 gene. Additional tests may include electromyography (to record the electrical activity of the muscles); nerve conduction velocity studies (to measure the nerve's ability to send an electrical signal0; or muscle biopsy.

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Symptoms of SMA may include:

- Facial muscle weakness and paralysis
- Low tone and weakness in torso
- "Floppy" and weak limbs
- Muscle twitching
- Muscle contractures (or stiffening of joints)
- Difficulty holding up head or sitting without support
- Difficulty swallowing
- Abnormal breathing patterns
- Scoliosis
- Weak legs and arms
- Difficulty running, climbing stairs or standing from seated position

Q: What is the treatment for SMA?

While there is no cure for SMA, breakthrough treatments have emerged in the last decade that are improving symptoms and extending life expectancy for children living with SMA.

Current FDA-approved treatments for SMA include:

- Risdiplam (Evrysdi) is designed to increase the production of SMN protein in the body. This medication, approved to treat people two months and older, is taken orally.
- Nusinersen (Spinraza) is designed to increase the production of the SMN protein in the body. This medication, approved to treat children and adults with SMA, is administered via lumbar puncture.
- Onasemnogene abeparovec-xioi (Zolgensma) is a gene therapy approved to treat children less than two years old living with SMA. This therapy, delivered via a one-time injection, is designed to replace the SMN1 gene function by delivering a new working SMN gene to the child's motor neurons.

- Onasemnogene abeparvovec-brve (Itvisma) is a gene therapy approved to treat people living with SMA who are two years of age and older. This therapy, delivered via one-time injection, is designed to replace the SMN1 gene function by delivering a new working SMN gene to the individual's motor neurons.

Q: What doctors will be involved with managing and treating SMA symptoms?

The complex health challenges stemming from SMA will require care from a team of healthcare professionals including:

- **Pediatric neurologists** are doctors who treat disorders of the brain and nervous system in children; they will likely perform the testing to diagnose SMA.
- **Physiatrists** are rehabilitative doctors who treat medical conditions related to the brain, bones, nerves and muscles, including SMA.
- **Pulmonologists** are doctors who care for people with respiratory issues; they will oversee and manage respiratory complications stemming from SMA.
- **Respiratory Therapists** provide tracheostomy and ventilator support, manage oxygen levels and secretions; they will help people with more severe cases of SMA who need breathing support.
- **Speech Language Pathologists** specialize in the anatomy and physiology of the upper airway; they will help support people with SMA who have trouble speaking and swallowing.

Q: What are some strategies for maintaining overall health while living with SMA?

Monitoring respiratory function is critical for individuals living with SMA; children with SMA Types 1 and 2 may need to visit a pulmonologist every three to six months to proactively monitor breathing issues. Doctors may recommend regular at-home use of cough assist machines, percussion vests and/or non-invasive ventilation (such as CPAP machines) by children and adults with SMA to help optimize lung function and prevent, or quickly treat, lung infections.

Physical therapy and low impact exercise can help those living with SMA build active and healthy lives. Walking, swimming, and stationary bicycling can help strengthen muscles, improve cardiovascular health and prevent depression. Range of motion and stretching exercises can help manage spasticity and stiff joints. Occupational therapists can help children and adults adapt to changing symptoms and choose devices such as

ramps, braces, walkers, and wheelchairs that help preserve mobility and independence.

Q: What other supports might help families manage a diagnosis of SMA?

Families who have received a new diagnosis of SMA may benefit from working with a psychologist or counselor to help process complicated emotions. Connecting with community members whose loved ones live with SMA can also provide a sense of emotional support, along with critical practical and life tips for managing symptoms and seeking treatments.

Cure SMA, a non-profit organization dedicated to fostering research and improving quality of life for people with SMA, has 36 volunteer chapters throughout the United States that provide families with a chance to meet one another. In addition, Cure SMA maintains a searchable database of hospitals across the country that offer diagnosis and treatments for children and adults living with SMA. Specialized clinics dedicated to SMA can be found in larger and sometimes offer support programs, along with treatments, for families and children affected by SMA.

Q: What is the life expectancy for people living with SMA?

Current life expectancy estimates for people living with SMA vary based on the type of disease: in general, the lower the classification, the more severe the symptoms. Children diagnosed with SMA Type 2, for example, may live into their mid-20's, while adults living with SMA Type 4 have a life expectancy similar to the general population. However, the development of new treatments throughout the last decade is expanding what has traditionally been possible for people with SMA, including by increasing life expectancy.

Q: Are there clinical trials for SMA?

Yes. Doctors across the country are working to identify biomarkers of disease progression and develop effective new therapeutics to preserve motor neurons and increase muscle function. To find current SMA trials, visit ClinicalTrials.gov, a searchable website for publicly and privately supported clinical studies maintained by the National Library of Medicine at the National Institutes of Health.

Sources: National Institute of Neurological Disorders and Stroke, Cure SMA, Muscular Dystrophy Association, Cleveland Clinic, Johns Hopkins Medicine, Children's Hospital of Boston, U.S. Food & Drug Administration.

Need to talk to someone?
Our Information Specialists are available to answer your questions.
Call toll-free 1-800-539-7309 Mon-Fri, 7 am - 8 pm EST.
Or schedule a call or ask a question online at
https://www.ChristopherReeve.org/Ask.

Resources for SMA:

Cure SMA
https://www.curesma.org/
50 E Commerce Drive, Suite 90
Schaumburg, IL 60173
Phone: 847-367-7620
Email: info@curemsa.org
Cure SMA is a non-profit organization dedicated to fostering research and improving quality of life for people with SMA.

Genetics and Rare Diseases Information Center (GARD)
https://rarediseases.info.nih/gov/diseases/7674/spinal-muscular-atrophy
Phone: 888-205-2311
GARD is a program of the National Institutes of Health that provides free access to information about genetic and rare diseases.

Muscular Dystrophy Association: Spinal Muscular Atrophy
https://www.mda.org/disease/spinal-muscular-atrophy
1016 W Jackson Blvd #1073
Chicago, IL 60607
Phone: 800-572-1717
ResourceCenter@mdausa.org
The Muscular Dystrophy Association is a non-profit organization dedicated to accelerating research, advancing care and advocating for families living with neuromuscular disease.

SMA Treatments and Programs in the U.S.

Cure SMA maintains a database of hospitals across the country that provide treatment and care for children and adults living with SMA Visit its website at https://www.curesma.org/find-a-treatment-center/ to find specialized SMA clinics in your region. Program locations include:

Boston Children's Hospital
https://www.childrenshospital.org/services/spinal-muscular-atrophy-program
300 Longwood Avenue

Boston, MA 02115
Phone: 617-919-6814

Children's Hospital of Los Angeles
https://www.chla.org/conditions/spinal-muscular-atrophy
4650 Sunset Blvd
Los Angeles, CA 90027
Phone: 323-361-2471

Children's Hospital of Philadelphia
https://www.chop.edu/conditions-diseases/spinal-muscular-atrophy-sma
3401 Civic Center Blvd
Philadelphia, PA 19104
Phone: 215-590-1719

Johns Hopkins Hospital
https://www.hopkinsmedicine.org/neurology-neurosurgery/specialty-areas/sma
200 North Wolfe Street
Rubenstein 2158
Baltimore, MD 21287
Phone: 410-955-4259

Phoenix Children's Hospital
https://phoenixchildrens.org/specialties-conditions/spinal-muscular-atrophy-children
1919 E. Thomas Road
Phoenix, AZ 85016
Phone: 602-933-5437

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