Cure SMA Medical Provider Information Kit

About SMA

Spinal muscular atrophy (SMA) is a genetic neuromuscular disease characterized by weakness of the skeletal and respiratory muscles. SMA affects approximately 1 in 10,000 births, and about 1 in every 50 Americans is a genetic carrier. SMA is the leading genetic cause of death for infants.

SMA is often diagnosed on a clinical basis by how the child appears physically. The diagnosis may be suspected when children are noted to be weak or have a delay in their developmental milestones, have low muscle tone, difficulty holding their head up, rolling over, sitting independently, standing or walking later than would be expected. A thorough medical history is performed, and a primary care provider may order genetic testing through a blood sample. A child may be referred to a pediatric neurologist who will also perform an examination and order genetic testing.

Types of SMA

There are four primary types of SMA. The type of SMA is based on the age of onset and the highest physical milestone achieved. SMA type I is the most severe, and also the most common, representing over 60% of cases. It is usually diagnosed during an infant’s first six months. SMA type I is often fatal early on in life.

SMA type II is usually diagnosed between six months and two years of age. Individuals with SMA type II can typically sit up without help, but are unable to walk and require a wheelchair.

SMA type III is usually diagnosed between 18 months and three years of age, though it can be diagnosed as late as the teenage years. Individuals with type III are initially able to walk, though as they grow older, their mobility may become increasingly limited and they may eventually require a wheelchair. Although SMA is not generally regarded as a progressive disease, patients often lose function as muscles continue to weaken.

SMA type IV is very rare. It is usually diagnosed after age 35, and leads to mild motor impairment.

Genetics and Treatments

SMA is caused by the mutation in the survival motor neuron gene 1 (SMN1). In a healthy person this gene produces a protein, called survival motor neuron (SMN) protein that is critical to the function of nerves that control our muscles. Without it, the nerve cells cannot properly function and die, leading to debilitating and often fatal muscle weakness.

A second gene also has a role in producing SMN protein. This is survival motor neuron gene 2 (SMN2), often called the SMA “backup gene.” Most of the SMN protein produced by SMN2 lacks a key building block that is normally produced by SMN1. However, people with more copies of SMN2 often have a less severe form of SMA than those with fewer copies.

It must be noted, however, that the number of copies of SMN2 does not reliably predict what type of SMA an individual will have or how weak their muscles will become. The determination of the type of SMA is based upon the physical milestones achieved.
The SMA drug pipeline consists of 18 ongoing drug programs, six of which are currently in clinical trials. These 18 programs represent four different therapeutic approaches: correction of the SMN1 mutation; modulation of SMN2, to prompt that gene to make more protein or to make a complete protein; neuroprotection; and muscle protection. Many researchers believe that the most effective treatment may be a combination of nerve and muscle protectors to slow or stop disease progression, taken with treatments that address the underlying genetics.

Though no treatment has yet been approved, proper care can improve quality of life for those with SMA. Patients with SMA often have impaired cough, respiratory insufficiency, dysphagia, gastroparesis, constipation, and evolving orthopedic issues including scoliosis. Various types of equipment may be used, from respiratory support during sleep, e.g. BiPAP and mucus clearance devices, to gastrostomy tubes, to wheelchairs, and braces. Cognitive development is usually not affected. Usual primary care practice, especially care coordination, family support, as well as routine pediatric care immunizations, developmental surveillance, and monitoring of growth contribute to the overall well-being of this child and their family.

About Cure SMA

Cure SMA leads the way to a world without spinal muscular atrophy, the number one genetic cause of death for infants. We fund and direct comprehensive research that drives breakthroughs in treatment and care, and we provide families the support they need for today.

Cure SMA offers many documents and resources addressing the standard of care for patients with SMA. This packet contains the Consensus Statement on the Standard of Care for SMA, two documents addressing respiratory care for SMA, and “At a Glance,” a summary of the services that Cure SMA offers.

Cure SMA’s website, www.cureSMA.org, has many additional resources for both families and healthcare providers, including our SMA Care Series Booklets. Should any clinical issues arise, the members of the Medical Advisory Council are available to you. The MAC represents a number of disciplines, including neurology, pulmonology, critical care, rehabilitation medicine, orthopedics, physical therapy, nutrition, psychology, primary, and palliative care.

Contact Information

For specific questions about care protocols, the MAC, or medical professional educational opportunities for SMA, email Mary Schroth, MD, at mary@curesma.org. Dr. Schroth is Cure SMA’s medical professional education consultant.

To receive an informational packet, including copies of all seven Cure SMA Care Series Booklets, email infopack@curesma.org or call 800.886.1762.
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Resources Available

A number of resources for medical professionals are available on the Cure SMA website.

Our For Healthcare Providers section is a good place to begin.

http://www.curesma.org/support-care/for-healthcare-providers/

Our SMA Topics For Healthcare Providers page provides an overview of the issues related to SMA.

http://www.curesma.org/support-care/for-healthcare-providers/sma-topics-for-healthcare-providers/

On the Support & Care Publications page, you can download copies of all of our care series booklets.

http://www.curesma.org/support-care/care-publications/

Our Medical Issues page includes links to the complete Consensus Statement for Standard of Care in Spinal Muscular Atrophy, as well as a shorter version developed for families.

http://www.curesma.org/support-care/living-with-SMA/medical-issues/

Within this same section, we also have pages devoted to some of the most significant medical issues that individuals with SMA face: palliative care, breathing, orthopedics/musculoskeletal, nutrition, and equipment. Each of these pages also contains additional links that medical providers or their patients may find helpful.

If an individual has been diagnosed with SMA, we encourage you to point that individual and/or his/her parents to our For Newly Diagnosed section:

http://www.curesma.org/support-care/newly-diagnosed/

This will help them to connect with the resources we can offer to families directly.

If you aren’t sure where to find the resource you’re looking for, please feel free to reach out to us.

http://www.curesma.org/support-care
Cure SMA

AT A GLANCE

Spinal muscular atrophy (SMA) is a disease that robs people of physical strength by affecting the motor nerve cells in the spinal cord, taking away the ability to walk, eat, or breathe. It is the number one genetic cause of death for infants. SMA affects approximately 1 in 10,000 babies, and about 1 in every 50 Americans as a genetic carrier. SMA can affect any race or gender. But there’s great reason for hope. We know what causes SMA and what we need to do to develop effective therapies, and we’re on the verge of major breakthroughs that will strengthen our children’s bodies, extend life, and eventually lead to a cure.

RESEARCH

Since 1986, Cure SMA has led and invested in the research that has made today’s breakthrough possible. With deep connections and expertise in both the patient and research communities, we’re uniquely positioned to direct funds to where they can make the greatest difference as quickly as possible. Fifteen years ago, we had just two potential drugs in the beginning stages of preclinical discovery. Today we have 11, including seven now in clinical trials.

We have invested more than $500 million in research and have funded half of all the ongoing new drug programs for SMA.

FAMILY SUPPORT AND PATIENT CARE

We won’t stop moving toward a world without SMA, but until we have a treatment and cure, we’ll do everything we can to improve quality of life for children and families affected by the disease today:

Each year, we reach nearly 4,000 families through our informational packets, newly dispensed care packages, equipment pool, and more.

We also reach the healthcare community through our medical professional educational opportunities and care series toolkits.

CONFERENCE AND COMMUNITY

Our Annual SMA Conference brings together researchers, healthcare professionals, and families to network, learn, and collaborate.

The conference is the largest in the world focused specifically on SMA, and it attracts the top scientific and companies in the field.

Today, we have more than 12,000 members and supporters, with chapters active throughout the country. We host approximately 50 fundraising and awareness events annually.

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