



# CURE SMA

CARE SERIES BOOKLET

GUIDE FOR PARENTS AND CAREGIVERS

WHAT YOU NEED TO KNOW AND  
DO ABOUT AN SMA DIAGNOSIS



Make today a  
breakthrough.

## Dear Parent or Caregiver,

You are likely receiving this guide because you have learned that your baby has spinal muscular atrophy, or SMA. This is a rare genetic condition that people often do not know much about until a family member or friend has a child diagnosed with it.

**This guide is intended to provide you with a foundation for understanding SMA. Here are the most important things to know:**

- Treatment is available.
- You need to act quickly.

Do not wait for signs of SMA. The best time to get treatment is before you see symptoms. If you wait until you notice the muscle weakness, which is the hallmark of SMA, your child will have already lost some function that may never be regained.

It is important to contact your doctor or other healthcare provider immediately, learn about your options, and decide if your child needs to start medicine right away or if they can be monitored for a time.

We realize it can be a challenge to act fast, perhaps before you fully understand your child's illness. You can do this. Your child's health depends on it.

And we can help. We are a nonprofit advocacy group that focuses on SMA. Contact us for information, guidance, and support.



**Phone: 800.886.1762**

**Email: [info@curesma.org](mailto:info@curesma.org)**

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## WHAT YOU SHOULD DO NOW



If your baby was diagnosed through a newborn screening test, contact your pediatrician or other healthcare provider, and share your test results if you have not already. Say it's urgent that your baby get an appointment.



You may already have confirmation that your child has SMA, or you may be waiting for a blood test to confirm the diagnosis. If you are waiting for a blood test, it may take one to two weeks to get the results. While you are waiting, continue to care for your baby as you did before you knew SMA was a possibility. You should not do anything differently to care for your child during this time, unless they show any unusual signs or symptoms.



Ask your healthcare provider for a referral to a specialist. Often, though not always, you will be referred to a pediatric neurologist, a doctor who specializes in diseases of the nervous system in children.



Get more information for you and your baby's healthcare providers. A companion piece to this brochure for medical professionals is available from Cure SMA. To receive a copy or to ask any other questions, call Cure SMA at 800.886.1762, or email [info@curesma.org](mailto:info@curesma.org).



# WHAT IS SMA?

SMA affects the cells in the spinal cord that send signals to the muscles to work. When these special cells, called motor neurons, don't function properly, muscles become very weak. People with SMA may have difficulty walking, eating, and even breathing because of muscle weakness.

While children with SMA have limited physical function, they are not impaired intellectually. Children with SMA can think, learn, and build relationships with other people.

## SMA TYPES

You may hear people refer to SMA “types.” Before newborn screening for SMA, patients were often diagnosed only after symptoms appeared and were then categorized into four main types depending on the age when symptoms first appeared and the highest physical milestone achieved. Now that newborns can be diagnosed before they have symptoms, types may not always be used in diagnosing SMA. It may still be useful, nonetheless, to be familiar with them.

### TYPE 1

The most severe and most common form of SMA, with symptoms (such as being “floppy” or having low muscle tone) appearing within six months after birth. Babies with Type 1 cannot perform tasks like rolling over or sitting on their own and, if untreated, often die by 2 years old.

### TYPE 2

Onset of symptoms (such as delay in development of motor skills and muscle weakness) between 6 and 18 months of age. Children with SMA Type 2 can typically sit up without help, but they're never able to stand, and they require wheelchairs.

### TYPE 3

Diagnosis after 18 months old but usually in early childhood. Children with SMA Type 3 are able to walk on their own but may lose that ability.

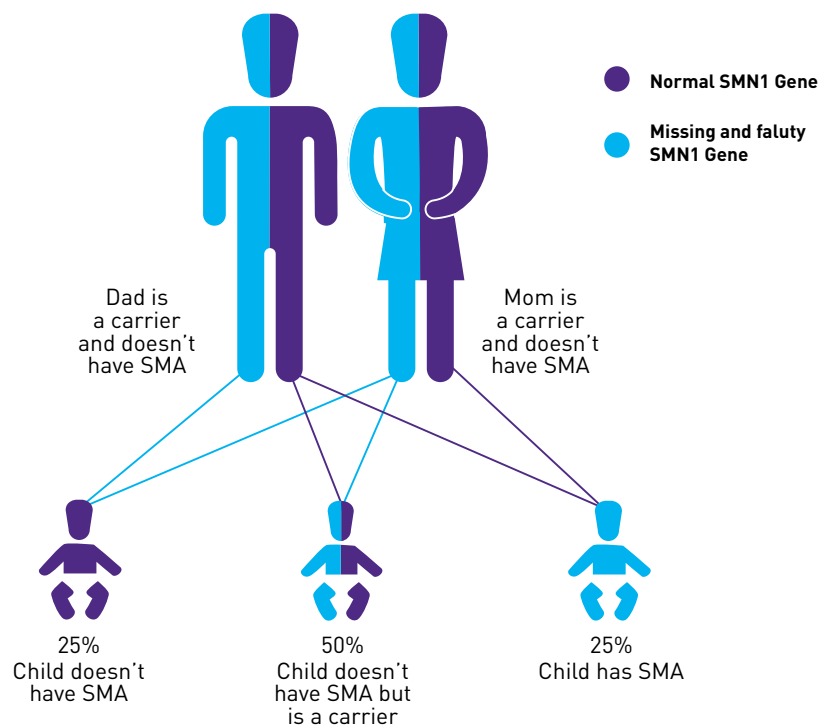
### TYPE 4

The adult form of SMA and the rarest. Symptoms (such as muscle weakness, tremors, or twitching) are relatively mild. People with SMA Type 4 typically live a full life with some support.

# WHAT CAUSES SMA?

SMA is an inherited disease, one that is passed from parents to children. It is caused by a missing or faulty gene—the survival motor neuron gene 1, or SMN1. In healthy people, the SMN1 gene produces a protein called SMN protein, which is important to motor neurons (special nerve cells that communicate with muscles). Without the protein, motor neurons don't work right and eventually die, leading the muscles to weaken and die.

## HOW SMA IS PASSED DOWN



SMA affects about one in 11,000 babies. About one in every 50 Americans is a carrier of the disease—meaning they can pass it down to their children, even though they are not ill themselves. SMA can affect any race or gender.

Babies usually receive two copies of the SMN<sub>1</sub> gene—one from each parent. A baby with SMA received missing or faulty SMN<sub>1</sub> gene copies from both parents.

Most children with SMA have two parents who do not have the disease but who carry it. These parents have one functioning SMN<sub>1</sub> gene, but their second copy of the gene is missing or malfunctioning.

Having one child with SMA does not lower the chances of having another child with the condition. Two carriers have a one-in-four chance in each pregnancy of having a child with SMA. The illustration shows possible gene combinations that carrier parents can pass down each time they have a child.

# DIAGNOSING

## SMA

Some states began testing newborns for SMA in 2018 by checking a small amount of blood at birth for the presence of SMN1 genes. Parents of babies who are missing two SMN1 genes receive notice that their babies likely have SMA. Following the identification of SMA on a newborn screening test, doctors may order a blood test to confirm the diagnosis. It can take one to two weeks to get results from this test.

## IMPORTANCE OF EARLY TREATMENT

Liliana Grace was born with SMA. She began treatment with SPINRAZA through a drug study when she was 12 days old. Now 2, she has reached every developmental milestone expected for a child her age. “Do not wait to seek treatment. I don’t know how to stress it enough.”

—Denise, mother of Liliana Grace



The addition of SMA to newborn screening tests has greatly improved the chance that a baby with the condition will be diagnosed early. The screening tests allow parents to talk to their healthcare providers about treatment for SMA before their babies have symptoms. This is the best way to prevent serious, and even life-threatening, problems.

Some of the symptoms of SMA, such as delays in physical development, are similar to the symptoms of other conditions. Without a newborn screening test, doctors and other healthcare providers may need to do multiple tests to rule out other possible causes of those symptoms before determining that the symptoms are caused by SMA. Because more common causes for delays in physical development are usually considered first, SMA is not always quickly diagnosed when symptoms appear.

In addition to newborn screening tests and confirmation tests to determine that a baby has SMA, additional tests may be conducted to estimate how serious a case of SMA a child has and to determine the best course of treatment.

# PREDICTING THE SEVERITY OF SMA

Several factors impact the seriousness of SMA. One that can be tested for is the presence of a second survival motor neuron gene, the SMN2 gene. It's similar to SMN1 and serves as a partial backup to SMN1. SMN2 does not produce the same amount of protein as SMN1. The amount of protein it produces is not enough to keep the motor neurons healthy. So even with SMN2 making some SMN protein, motor neurons can still die and muscles can become weak.



The number of copies of the SMN2 backup gene varies from person-to-person. In people with SMA, the number of copies of SMN2 is connected to how serious their illness is. Generally, the more copies of SMN2 the better, because more SMN protein means the motor neurons are getting more of what they need to send signals to the muscles. Some treatments currently available for SMA focus on getting SMN2 to produce more protein.

The number of SMN2 copies a baby has will help determine whether a doctor recommends immediate treatment or monitoring for a time. If a baby has more than four copies of SMN2, the doctor may recommend that their condition be carefully monitored before beginning treatment. However, these cases are very rare. Usually the doctor will recommend that treatment begin as soon after birth as possible.



# TREATING SMA

There are two ways of treating SMA that researchers are studying. One way of treating SMA is to address the underlying disease cause by targeting the gene and increasing the amount of SMN protein in the body. These approaches are called “SMN-based” or “SMN-enhancing” approaches.

**Additional treatments for SMA are being tested in clinical trials. Information on available treatments and open clinical trials changes often, so it’s best to check the Cure SMA website at:**

<https://www.curesma.org/clinical-trials/>

**Or, talk to your healthcare provider about current options before making a final decision about treatment.**

Currently, three SMN-enhancing treatments for SMA are approved by the U.S. Food and Drug Administration (FDA). Evrysdi (risdiplam) is a small molecule taken daily by mouth or through a g-tube that causes the SMN2 gene to make more complete SMN protein. Spinraza (nusinersen) is an injection that targets the SMN2 gene, causing it to make more complete protein. Zolgensma (Onasemnogene abeparvovec-xioi) is a gene therapy that replaces the function of the missing or mutated SMN1 gene.

Other types of therapies are in clinical trials or at even earlier stages of research, and these may become available in the future. Some of these approaches are targeting “non-SMN” approaches, focusing on other systems, pathways, and processes in the body that are affected by SMN protein. Examples of such approaches are drugs that increase muscle strength or motor neuron function.

## Early Is Better

Early treatment offers the best chance that a child stays as healthy as possible.

Without enough protein from the missing or faulty SMN1 gene or the backup SMN2 gene, motor neurons die quickly. Without any treatment, babies with the most severe cases of SMA lose 90% of their motor neurons by the time they are 6 months old.

Once lost, motor neurons cannot be replaced. The body does not generate new motor neurons, and none of the treatments available or being researched will do so either.

That means that the best treatment may be before a baby shows signs of being sick. The goal is to save the greatest possible number of motor neurons so a baby’s muscles develop properly.





# LIVING WITH SMA

With newborn screening, early diagnosis, and early treatment, many children with SMA may be able to live healthier lives. Nonetheless, they may occasionally require help with physical needs and daily activities.

## Food and Nutrition

Getting proper nutrition can be a challenge for some children with SMA because weak muscles make it difficult to chew, swallow, and digest food.

Your healthcare provider may refer you to a nutritionist, who may recommend changes to your child's diet, depending on individual needs. These can include soft foods that are easy to swallow or low-fat foods that do not aggravate acid reflux caused by weak muscles around the stomach or esophagus.

## Adaptive Equipment

A variety of supportive and assistive devices exist to help with daily challenges caused by muscle weakness. Ask your doctor or check the Cure SMA website for options.

# BREATHING AND COUGHING

People with SMA can have trouble breathing fully or coughing strongly enough to clear their airways. This is because the muscles between the ribs, called intercostal muscles, are very weak. This can be a problem, especially when a child has a virus that affects the lungs or upper airways, including the nose and throat.

## Common recommendations include:

- Referral to a pulmonologist (a doctor who specializes in lung problems), who can monitor your child's breathing capacity.
- A cough assist device to help children clear mucus, phlegm, and other secretions from their airways.
- Breathing support, perhaps at night or when a child is sick. Bi-level positive airway pressure, or BiPAP, is a way to provide noninvasive support through a mask.

# NEWBORN SCREENING REGISTRY

## The Cure SMA Newborn Screening Registry (NBSR)

is an online Registry established to help our SMA community (including affected individuals, families, clinicians, and researchers) learn more about SMA, better manage symptoms over time, and develop new treatments.

**We invite you to participate by going to the NBSR website at [www.curesma.org/nbsr](http://www.curesma.org/nbsr) and following the instructions to provide Cure SMA with information about your child.**

The NBSR is a program of Cure SMA. Cure SMA is the sole guardian of NBSR and its material. NBSR information can be used to improve clinical care and to support new therapy development. Registries in other diseases also have a long history of success in moving research and clinical care forward.

Visit the NBSR portal at [www.curesma.org/nbsr](http://www.curesma.org/nbsr) to receive additional information or register your child.



# NOTES

Here’s a space to jot down the questions you have, as well as notes from your appointment

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**We recommend taking this guide with you when you visit your baby’s healthcare provider. A separate, corresponding guide written for doctors and other healthcare providers is also available at [www.curesma.org/care-series-booklets/](http://www.curesma.org/care-series-booklets/). Otherwise, call Cure SMA at 800.886.1762, or email [info@curesma.org](mailto:info@curesma.org).**



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