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WINTER 2021/2022

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TIPS TO MANAGE SMA

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SPECIALISTS TO SEE

Nicholas E. Johnson, MD, shares the types of health care professionals who can help your SMA issues.

- **Neuromuscular neurologist.** This is typically the leader of your team at a multidisciplinary clinic.
- **Pulmonologist** to manage lung problems.
- **Orthopedic surgeon** to evaluate and manage scoliosis (spine curvature).
- **Physical therapist** to deal with strength issues.
- **Genetic counselor** to help you understand your diagnosis.
- **Speech therapist** to monitor sleep and feeding.
- **Dietitian** to help you get good nutrition.



“JUST AS IT’S IMPORTANT TO CARE FOR YOUR CHILD’S PHYSICAL WELL-BEING, CARING FOR YOUR PSYCHOLOGICAL WELL-BEING IS ALSO VERY IMPORTANT. DON’T FORGET TO TAKE CARE OF YOURSELF, TOO.”

—MATTHEW M. HARMELINK, MD,
DIRECTOR, PEDIATRIC
NEUROMUSCULAR PROGRAM,
CHILDREN’S WISCONSIN IN MILWAUKEE

MANAGING YOUR SPINAL MUSCULAR ATROPHY

TIPS AND TREATMENTS FOR EVERYDAY LIVING WITH YOUR SYMPTOMS

By Rachel Reiff Ellis

Reviewed by Michael W. Smith, MD, WebMD Chief Medical Director

Although spinal muscular atrophy (SMA) doesn’t yet have a cure, there are many treatments to help manage it. SMA treatments focus on easing your symptoms, preventing complications, and helping you live more comfortably. And these therapies are improving constantly.

“With new disease-modifying therapies available, the goal of treatment should be to slow the progression of the disease and therefore improve quality of life,” says Nicholas E. Johnson, MD, associate professor and division chief of neuromuscular and vice chair of research in the Department of Neurology at Virginia Commonwealth University in Richmond.

Here are some of the ways to do that:

IMPROVE YOUR MOVEMENT

Several FDA-approved medications are now available for SMA. Typically, you get these in one infusion dose. They work directly on cells in the spinal cord and can improve motor function.

“The natural progression of SMA across all ages is slow loss of the ability to use arms, legs, and even the trunk,” Johnson says. “It’s critical to be on a therapy to help prevent this, whether it’s gene therapy, antisense oligonucleotides, or splicing inhibitors.”

In addition, orthopedic braces, wheelchairs, and physical and occupational therapies can help improve your movement and help you function better throughout the day.

CHECK YOUR SPINE

Nearly all children with SMA develop scoliosis (curvature of the spine) at some point. This can cause problems such as hip dislocation, balance issues, and even trouble with breathing.

You may need a brace or even surgery to help correct the curve in your spine. This can help lessen pain, ease your breathing, and allow you to use your arms better.

SUPPORT BREATHING

The level of breathing help you need will depend on how severe your SMA is. “For patients with milder symptoms, they may not need any respiratory treatments,” says Matthew M. Harmelink, MD, director of the pediatric neuromuscular program at Children’s Wisconsin in Milwaukee.

For those who do, he says, the options range from a mask and machine that provides air pressure (BiPAP) to a permanent tube in the throat connected to a machine that breathes for you (tracheostomy and ventilator). There are also medications that can help with breathing.

Additionally, there are devices that can

help you cough up mucus to help prevent infection in your lungs and airways and to keep oxygen flowing to your tissues. “As the disease progresses, the ability to cough up bad bacteria or viruses worsens,” Johnson says. You can treat this by supporting the pressure in your lungs, particularly at night.

EAT WELL

A proper diet can help you grow, maintain a healthy weight, breathe better, prevent illness, and even improve your motor function.

If weak swallowing muscles get in the way of your eating, you may need a feeding tube that goes through your nose into your stomach (called an NG tube) or directly into your stomach (called a gastrostomy tube, or G-tube). See a specialist if you’re having trouble getting the nutrition you need.

“A dietitian can work with you to

modify which foods you eat if you have increasing difficulty swallowing,” Johnson says. You can also see a speech therapist for help strengthening the muscles used for swallowing.

CONSERVE ENERGY

The muscle weakness of SMA can often make it feel like you’re walking around with full medieval chain mail on your body. Fatigue is a common issue. Harmelink says it’s helpful to budget your energy like you would your money.

“Think about how and when you’ll need your energy during the day,” he says. “If you have a big event or meeting in the afternoon, for example, take it easy in the morning.”

Exercise done right can also help boost your mood and energy levels. Talk to your doctor about what type of movement could work for you to get your heart pumping and muscles moving in a positive way.



Watch a video on **Treatments for Spinal Muscular Atrophy** at [WebMD.com/treatsma](https://www.webmd.com/treatsma).



Evrysdi in action

Proven to make a difference
in infants, children, and
adults 2 months and older
with spinal muscular
atrophy (SMA)

1,400+
people in the US with SMA
are taking Evrysdi, including
people up to 83 years old**

* Number of people taking Evrysdi since August 2020 (approval) through June 2021.

† Clinical studies of Evrysdi did not include people aged 65 and older to determine whether they respond differently from those who are younger.

What is Evrysdi?

Evrysdi is a prescription medicine used to treat spinal muscular atrophy (SMA) in adults and children 2 months of age and older.

It is not known if Evrysdi is safe and effective in children under 2 months of age.

Important Safety Information

- Before taking Evrysdi, tell your healthcare provider about all of your medical conditions, including if you:
 - are pregnant or plan to become pregnant. If you are pregnant, or are planning to become pregnant, ask your healthcare provider for advice before taking this medicine. Evrysdi may harm your unborn baby.
 - are a woman who can become pregnant:
 - Before you start your treatment with Evrysdi, your healthcare provider may test you for pregnancy. Because Evrysdi may harm your unborn baby, your healthcare provider will decide if taking Evrysdi is right for you during this time
 - Talk to your healthcare provider about birth control methods that may be right for you. Use birth control while on treatment and for at least 1 month after stopping Evrysdi
 - are an adult male planning to have children: Evrysdi may affect a man's ability to have children (fertility). If this is of concern to you, make sure to ask a healthcare provider for advice
 - are breastfeeding or plan to breastfeed. It is not known if Evrysdi passes into breast milk and may harm your baby. If you plan to breastfeed, discuss with your healthcare provider about the best way to feed your baby while on treatment with Evrysdi
- **Tell your healthcare provider about all the medicines you take**, including prescription and over-the-counter medicines, vitamins, and herbal supplements. Keep a list of them to show your healthcare provider and pharmacist when you get a new medicine

✓ Evrysdi helped infants with Type 1 SMA achieve a key motor milestone and delayed disease progression†

41% of infants (7/17) sat without support for at least 5 seconds after 12 months, as measured by the BSID-III gross motor scale

90% of infants (19/21) at 12 months and 81% of infants (17/21) at 23 months were alive and able to breathe without permanent support§

✓ Evrysdi significantly improved or maintained motor skills in adults and children with Type 2 and 3 SMA*

Motor function improved after 12 months (average 1.36-point increase on the MFM-32 scale with Evrysdi vs average 0.19-point decrease without Evrysdi)

- 1.55-point estimated improvement versus placebo on the MFM-32 scale at 12 months (95% CI: 0.30, 2.81; $P=0.0156$)||

✓ Evrysdi is designed to help make and maintain more SMN protein

✓ The safety of Evrysdi is being studied in more than 450 people, from 2 months to 60 years old, with Type 1, 2, or 3 SMA‡

✓ The first and only medication to treat SMA with at-home dosing

‡ The efficacy and safety of Evrysdi was established in 2 main studies. FIREFISH is a 2-part, open-label study of Evrysdi in 62 infants aged 2-7 months with Type 1 SMA. SUNFISH is a 2-part study of Evrysdi in 231 children and adults aged 2-25 years with Type 2 and 3 SMA. A third study, JEWELFISH, is an ongoing safety study of Evrysdi in 174 infants, children, and adults aged 1-60 years with Type 1, 2, and 3 SMA previously treated with approved and investigational SMA medications.

§ Permanent support was defined as having a tracheostomy (a surgery where a tube is inserted in the front of the throat into the windpipe) or more than 21 days of either noninvasive ventilation support (16 or more hours a day) or being intubated (a procedure where a breathing tube is inserted down the throat and into the windpipe) to help with breathing, in the absence of an acute reversible event.

|| This 95% CI (confidence interval) means that we are 95% confident that the actual average change in MFM-32 with Evrysdi will be between 0.30 and 2.81 points higher than with placebo.

BSID-III stands for the Bayley Scales of Infant and Toddler Development-Third Edition.

MFM-32 stands for the Motor Function Measure-32 Items.

SMN stands for survival motor neuron.

Important Safety Information (continued)

- You should receive Evrysdi from the pharmacy as a liquid that can be given by mouth or through a feeding tube. The liquid solution is prepared by your pharmacist. If the medicine in the bottle is a powder, **do not use it**. Contact your pharmacist for a replacement
- Avoid getting Evrysdi on your skin or in your eyes. If Evrysdi gets on your skin, wash the area with soap and water. If Evrysdi gets in your eyes, rinse your eyes with water
- The most common side effects of Evrysdi include:
 - For later-onset SMA: fever, diarrhea, rash
 - For infantile-onset SMA: fever, diarrhea, rash, runny nose, sneezing, sore throat, and cough (upper respiratory infection), lung infection, constipation, vomiting

These are not all of the possible side effects of Evrysdi. For more information on the risk and benefits profile of Evrysdi, ask your healthcare provider or pharmacist.

You may report side effects to the FDA at 1-800-FDA-1088 or www.fda.gov/medwatch. You may also report side effects to Genentech at 1-888-835-2555.

Please see accompanying brief summary for additional Important Safety Information.

Talk with your doctor about Evrysdi
or visit **www.Evrysdi.com/Go** to learn more

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Patient Information EVRYSDI® [ev-RIZ-dee] (risdiplam) for oral solution	
What is EVRYSDI? <ul style="list-style-type: none"> EVRYSDI is a prescription medicine used to treat spinal muscular atrophy (SMA) in adults and children 2 months of age and older. It is not known if EVRYSDI is safe and effective in children under 2 months of age. 	Reusable Oral Syringes <ul style="list-style-type: none"> Your pharmacist will provide you with the reusable oral syringes that are needed for taking your medicine and explain how to use them. Wash the syringes per instructions after use. Do not throw them away. Use the reusable oral syringes provided by your pharmacist (you should receive 2 identical oral syringes) to measure your or your child's dose of EVRYSDI, as they are designed to protect the medicine from light. Contact your healthcare provider or pharmacist if your oral syringes are lost or damaged. Once transferred from the bottle to the oral syringe, take EVRYSDI right away. Do not store the EVRYSDI solution in the syringe. If EVRYSDI is not taken within 5 minutes of when it is drawn up, EVRYSDI should be thrown away from the reusable oral syringe, and a new dose should be prepared.
Before taking EVRYSDI, tell your healthcare provider about all of your medical conditions, including if you: <ul style="list-style-type: none"> are pregnant or plan to become pregnant. If you are pregnant, or are planning to become pregnant, ask your healthcare provider for advice before taking this medicine. EVRYSDI may harm your unborn baby. are a woman who can become pregnant: <ul style="list-style-type: none"> Before you start your treatment with EVRYSDI, your healthcare provider may test you for pregnancy. Because EVRYSDI may harm your unborn baby, you and your healthcare provider will decide if taking EVRYSDI is right for you during this time. Talk to your healthcare provider about birth control methods that may be right for you. Use birth control while on treatment and for at least 1 month after stopping EVRYSDI. are an adult male planning to have children: EVRYSDI may affect a man's ability to have children (fertility). If this is of concern to you, make sure to ask a healthcare provider for advice. are breastfeeding or plan to breastfeed. It is not known if EVRYSDI passes into breast milk and may harm your baby. If you plan to breastfeed, discuss with your healthcare provider about the best way to feed your baby while on treatment with EVRYSDI. 	What are the possible side effects of EVRYSDI? The most common side effects of EVRYSDI include: <ul style="list-style-type: none"> For later-onset SMA: <ul style="list-style-type: none"> fever diarrhea rash For infantile-onset SMA: <ul style="list-style-type: none"> fever runny nose, sneezing, sore throat, and cough (upper respiratory infection) constipation diarrhea lung infection vomiting rash <p>These are not all of the possible side effects of EVRYSDI. For more information, ask your healthcare provider or pharmacist.</p> <p>Call your doctor for medical advice about side effects. You may report side effects to FDA at 1-800-FDA-1088.</p>
Tell your healthcare provider about all the medicines you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements. Keep a list of them to show your healthcare provider and pharmacist when you get a new medicine.	
How should I take EVRYSDI? See the detailed Instructions for Use that comes with EVRYSDI for information on how to take or give EVRYSDI oral solution. <ul style="list-style-type: none"> You should receive EVRYSDI from the pharmacy as a liquid that can be given by mouth or through a feeding tube. The liquid solution is prepared by your pharmacist. If the medicine in the bottle is a powder, do not use it. Contact your pharmacist for a replacement. Avoid getting EVRYSDI on your skin or in your eyes. If EVRYSDI gets on your skin, wash the area with soap and water. If EVRYSDI gets in your eyes, rinse your eyes with water. 	How should I store EVRYSDI? <ul style="list-style-type: none"> Store EVRYSDI in the refrigerator between 36°F to 46°F (2°C to 8°C). Do not freeze. Keep EVRYSDI in an upright position in the original amber bottle to protect from light. Throw away (discard) any unused portion of EVRYSDI 64 days after it is mixed by the pharmacist (constitution). Please see the Discard After date written on the bottle label. (See the Instructions for Use that comes with EVRYSDI).
Taking EVRYSDI <ul style="list-style-type: none"> Your healthcare provider will tell you how long you or your child needs to take EVRYSDI. Do not stop treatment with EVRYSDI unless your healthcare provider tells you to. For infants and children, your healthcare provider will determine the daily dose of EVRYSDI needed based on your child's age and weight. For adults, take 5 mg of EVRYSDI daily. <ul style="list-style-type: none"> Take EVRYSDI exactly as your healthcare provider tells you to take it. Do not change the dose without talking to your healthcare provider. Take EVRYSDI 1 time daily after a meal (or after breastfeeding for a child) at approximately the same time each day. Drink water afterwards to make sure EVRYSDI has been completely swallowed. Do not mix EVRYSDI with formula or milk. If you are unable to swallow and have a nasogastric or gastrostomy tube, EVRYSDI can be given through the tube. If you miss a dose of EVRYSDI: <ul style="list-style-type: none"> If you remember the missed dose within 6 hours of when you normally take EVRYSDI, then take or give the dose. Continue taking EVRYSDI at your usual time the next day. If you remember the missed dose more than 6 hours after you normally take EVRYSDI, skip the missed dose. Take your next dose at your usual time the next day. If you do not fully swallow the dose, or you vomit after taking a dose, do not take another dose of EVRYSDI to make up for that dose. Wait until the next day to take the next dose at your usual time. 	Keep EVRYSDI and all medicines out of the reach of children.
	General information about the safe and effective use of EVRYSDI. Medicines are sometimes prescribed for purposes other than those listed in a Patient Information leaflet. Do not use EVRYSDI for a condition for which it was not prescribed. Do not give EVRYSDI to other people, even if they have the same symptoms you have. It may harm them. You can ask your pharmacist or healthcare provider for information about EVRYSDI that is written for health professionals.
	What are the ingredients in EVRYSDI? Active ingredient: risdiplam Inactive ingredients: ascorbic acid, disodium edetate dihydrate, isomalt, mannitol, polyethylene glycol 6000, sodium benzoate, strawberry flavor, sucralose, and tartaric acid.
	Genentech <i>A Member of the Roche Group</i> EVRYSDI® (risdiplam) Distributed by: Genentech, Inc. A Member of the Roche Group 1 DNA Way South San Francisco, CA 94080-4990 EVRYSDI is a registered trademark of Genentech, Inc. M-US-00007143(v2.0) ©2021 Genentech, Inc. All rights reserved.
	For more information, go to www.EVRYSDI.com or call 1-833-387-9734.



ASSISTIVE AIDS

THESE DEVICES HELP IMPROVE EVERYDAY LIVING WITH SPINAL MUSCULAR ATROPHY

By Rachel Reiff Ellis

Reviewed by Michael W. Smith, MD, WebMD Chief Medical Director

Assistive devices are pieces of equipment you or your child can use to stay as functional as possible while dealing with the symptoms of spinal muscular atrophy (SMA). These devices can help you do things like shower, get dressed, walk, speak, breathe, swallow, and cough.

You may wonder if using an assistive device will hold you back in some way, but Nassim Rad, MD, physical medicine and rehabilitation specialist and co-director of the UW Medicine MDA-ALS Center in Seattle, says the opposite is true.

“Many people are resistant to assistive devices because they fear they’re ‘giving up’ or they believe in the ‘use it or lose it’ mentality,” she says. “But using assistive devices helps you save energy to engage in *more* activities while also keeping you safe and reducing your risk of falls or infection.”

Some of the tools you can use include:

DEVICES TO HELP YOU MOVE

Because muscle weakness is a hallmark symptom of SMA, there’s a wide range of mobility devices. These aids help with sitting, standing, walking, or generally moving around. Aids you might use:

- Cane
- Walker
- Manual or power wheelchair
- Adaptive strollers (for a child with SMA)
- Bath chairs

“Many people will start with one device and over time need to transition to another form as their disease progresses,” Rad says.

A few accessibility changes around your house can help with mobility issues, too. “You might need to alter bathrooms and showers to accommodate raised toilet seats, grab bars, and shower/bath chairs,” Rad says.

DEVICES TO HELP YOU BREATHE AND EAT

Respiratory (breathing) problems are the leading cause of illness in people with SMA. These devices can help prevent infection as well as make you or your child more comfortable. You may use these devices for help coughing, swallowing, and breathing:

- **Bilevel positive airway pressure (BiPAP) machine**—a ventilator that helps get more air in as you breathe, especially when you’re sleeping.
- **Cough assist machines**—face masks hooked to a tube and machine that

pushes air in and out of lungs.

- **Standers**—devices that hold you in a standing position to help with digestion, circulation, and breathing.
- **Feeding seat**—a special foam seat to position a child for eating.

The level of breathing or eating help you or your child needs will depend on how severe the SMA symptoms are.

ASK THOSE IN THE KNOW

Many other incredible tools are available for everyday functioning such as robotic arms, eye gaze computers, and wheelchairs you can power with head movements. Talk with your doctor so you can figure out what you need when.

“Technology is constantly advancing, so it’s important to check in with your provider and discuss functions that are becoming harder for you,” Rad says. “They can refer you to physical therapy, occupational therapy, and speech therapy to evaluate for assistive devices you might not even know exist.”

THE IMPORTANCE OF MENTAL HEALTH CARE

TENDING TO YOUR EMOTIONAL NEEDS WHEN YOU HAVE—OR CARE FOR SOMEONE WITH—SMA

By Rachel Reiff Ellis

Reviewed by Michael W. Smith, MD, WebMD Chief Medical Director

Because spinal muscular atrophy (SMA) takes such a physical toll on those who have it, it can be easy to ignore mental and emotional well-being. But your mental and physical health are very much tied together.

Anxiety, depression, grief, body image issues, and even substance abuse are common for people with SMA. Noticing and dealing with them is an important part of whole-body wellness.

EMOTIONAL TOLL

Typical issues that can impact your mental well-being when you have SMA include:

- Making difficult treatment choices
- Worry about future loss of function
- Lack of sleep
- Stress
- Thoughts about death
- Limitations on social activities
- Finances

Kids with SMA may especially grieve their loss of independence once they reach the age where separation from family would typically happen.

“Just when you’re aching to get free, you can’t,” says Thomas O. Crawford, MD, professor of neurology and co-director of the Muscular Dystrophy Association Clinic at Johns Hopkins Medicine in Baltimore. “It’s a distortion of normal development.”

SOURCES OF SUPPORT

Connecting with others who understand SMA and its impact on daily life can bring you comfort and help you feel less alone. Online communities are invaluable for rare conditions like SMA, since you may not live close to others with the condition.

“It’s hard to get people physically together, so we now have these strong virtual communities coming together and forming groups,” Crawford says.

SIGNS OF MENTAL DISTRESS

Anxiety and depression can be hard to spot when you're experiencing them. Here are some symptoms you might have:

- Bouts of nervousness or feeling tense
- Concerns that something "bad" will happen
- Trouble concentrating
- Fast breathing or rapid heart rate
- Problems sleeping or sleeping more than usual
- Feeling sad or hopeless
- No interest in things you used to enjoy
- Irritability or unexplained anger
- Low energy levels

A big benefit of SMA support groups, especially for kids, is seeing others who are thriving with the condition. "One of my 14-year-old patients who had decided she'd never be able to do much met a young lady through an SMA group who had a baby, and it changed her whole life outlook," Crawford says. Positive representations of others with SMA can foster hope and encouragement.

Professional therapy is another avenue to help you cope with mental struggles. There are many different therapy methods, including interpersonal therapy and cognitive behavioral therapy. You'll get the most out of this kind of help if you

find a therapist you connect with who understands your issues.

"Remember, the purpose of life is not to do therapy. The purpose of therapy is to do more life," Crawford says.

CAREGIVER STRESS

If you're a loved one caring for someone with SMA, you also bear an emotional and mental stress load. A study of parents of kids with SMA found that not only was stress high for them, but also that stress went up with the severity of their child's disease.

"Caregiver burnout" is a state of emotional, physical, and mental exhaus-

tion that often happens when you're the primary support person for someone with SMA. You care for yourself when you:

- Find your own "support person" to talk to
- Tap into respite care resources such as home health
- Join a caregiver support group
- Know and name your limits

It's common to feel guilty about taking time for yourself when your loved one depends on you, but by investing in your mental well-being, you're better prepared to take care of them.

IT TAKES A VILLAGE

**WHEN YOU OR YOUR CHILD LIVES WITH
A RARE DISEASE, YOU WILL NEED HELP.
LEARN HOW TO FIND IT.**

By Sonya Collins

Reviewed by Michael W. Smith, MD, WebMD Chief Medical Director

The outlook for people with spinal muscular atrophy has improved dramatically in the last 5 years. During that time, the FDA approved several medications to treat this rare genetic disorder. These drugs have given renewed physical strength and abilities to babies, kids, and adults with spinal muscular atrophy.

“The babies that we see are doing things that we never in our wildest dreams thought would be possible,” says Richard S. Finkel, MD, who leads the Center for Experimental Neurotherapeutics at St. Jude Children’s Research Hospital in Memphis, TN. “But these drugs are not a cure, so we need to make sure they still get the support and care they need.”

Spinal Muscular Atrophy (SMA) affects just 1 in 6,000 to 1 in 10,000 people.

When you live with a rare disease—or you are the parent of a child who has a rare disease—it’s normal to feel lonely. You need support. Here’s how you can get it.

START WITH AN SMA CARE CENTER

Depending on when they start treatment and the severity of the condition, children and adults with spinal muscular atrophy could need a wide range of care. This could include neurology, breathing and feeding support, physical and occupational therapy, orthopedic care, and other treatments.

“We recommend being seen in a multidisciplinary clinic, where you see not only a neurologist, but a pulmonary specialist, a feeding specialist, a physical

therapist, perhaps a rehabilitation doctor, orthopedic doctors,” Finkel says. “We can try to anticipate what they will need in the future and stay ahead of the curve.”

The Cure SMA Care Center Network includes hospitals and clinics around the country that offer multidisciplinary team-based care for spinal muscular atrophy. You can find the nearest care center on the CURE SMA website (curesma.org/sma-care-center-network).

But you don’t have to live near a world-class hospital to get great specialized care. Though a local pediatrician or family doctor may see very few cases of spinal muscular atrophy during the course of their career, specialists in other regions often consult with community doctors through telehealth.

“We can educate the pediatrician, a





ASK YOUR DOCTOR

Here are some questions that might help guide your visit.

1. What is the future outlook for me/my child?
2. What are my chances of having another child with this condition?
3. What changes or symptoms should I look out for?
4. What treatments am I/is my child eligible for?
5. Which treatment is best for me/my child?
6. Where can I go to get the expertise I need?

local physical therapist,” Finkel says. “It may be that the physical therapist in my clinic does a Zoom call with the physical therapist back home. They can talk about what the goals are, what the strategy is, and what to do.”

FIND YOUR PEER NETWORK

Maybe the best source of support is other people like you—whether you’re a teen or adult living with spinal muscular atrophy or the parent of a child with the condition. Social media has been a guiding light for people affected by spinal muscular atrophy. Parents, caregivers, and people living with SMA take to Facebook and other online platforms to exchange ideas and help each other solve problems.

“Parents come to me and say, ‘I heard this from another parent on Facebook, do you think it will work for my child?’” Finkel says. “So, I am learning from Facebook, too.”

SMA Foundation connects people affected by this condition with patient advocacy groups around the world. CURE SMA has local chapters in 36 states. The organization also hosts an annual summit where doctors and scientists, parents and caregivers, and people living with spinal muscular atrophy gather to connect and learn from each other.

As people with spinal muscular atrophy now achieve things that even their doctors haven’t seen before, it’s more important than ever to compare notes with others who are a little further along in their journey.

BE A PART OF YOUR OWN CARE TEAM

While you might lean heavily on others for support at first, you will soon be offering support to others. And you’ll realize that you are the expert in your or your child’s care.

Finkel often sees parents who become so skilled at caring for their children that, when they come to the hospital, they show the health care providers what to do for their child. He encourages all parents and caregivers to become proactive members of their care team and not passive recipients of care.

“We want to educate parents and make them part of the care team, so they can become effective advocates for their child.”

FACTS & STATS

By Sonya Collins

Reviewed by Michael W. Smith, MD, WebMD Chief Medical Director

up to
25,000

Estimated number of children in the U.S. who have spinal muscular atrophy (SMA).

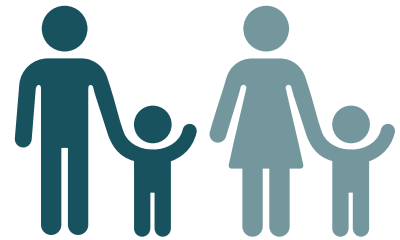


1

Number of copies of a mutated VAPB gene that a child must inherit



from their parents in order to have adult-onset Finkel type spinal muscular atrophy.



1 in 2

Chance that a person with Finkel type spinal muscular atrophy will pass the condition on to a child.

1 in 6,000 to 1 in 10,000

Number of people who are born with spinal muscular atrophy.



2

Number of copies of a mutated SMN1 gene a person must inherit from their parents in order to have spinal muscular atrophy. (A child must inherit a mutated gene from both the mother and the father.)



Up to \$196,429

Average annual cost of SMA type 1—the most common and severe type of SMA. Cost depends on severity of symptoms and the age at which they start.



1 in 4



Chance that a child will be born with spinal muscular atrophy when both parents carry the gene.

Up to \$82,474

Average annual cost of SMA types 2, 3, and 4—less severe forms of the condition that develop later than type 1.

1 in 40 to 1 in 50



Number of people in the U.S. (about 6 million) who carry the gene for spinal muscular atrophy.

SOURCES: National Human Genome Research Institute, SMA Foundation, National Organization for Rare Diseases, *Orphanet Journal of Rare Diseases*