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# Quest

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**Lindsey Baker**

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PUBLICATION MANAGEMENT



# Keeping Our Spirit Strong

When I began writing this, my first *Quest* column as editor-in-chief, I thought I'd be telling a different story. I was excited to share scenes from MDA's 2020 Clinical & Scientific Conference, plus what we had in store for our programs.

And then, as the novel coronavirus and COVID-19 spread, our world changed.

MDA has changed with it. The risks posed by gathering groups of people made it impossible to host our conference in March, or plan to open MDA Summer Camps this year. Our team, across the country, moved work from offices into their homes.

But we're working. That hasn't changed. Our mission remains strong. We're still pushing for research and treatments, still advocating, still educating and informing in these very pages.

*Quest's* mission is to bring you news and stories that highlight both the big picture and the everyday reality of living with neuromuscular disease. We know this breadth of information is vital — I know, as an adult living with Charcot-Marie-Tooth disease (CMT). I know personally how crucial it is for *Quest* to be a robust, relevant, continually evolving part of our community's conversation on how we can thrive, even in the most uncertain of times.

*Quest* has always been a free resource for MDA families. And each year, the cost of printing and mailing the magazine increases. We've relied only on advertising revenue to cover these costs, even as advertising has become scarcer. We're dedicated, especially now, to bringing *Quest*, at no cost, to your door, alongside new, expanded communication through a regular e-newsletter and digital formats.

But we need help.

If what you read in *Quest* is valuable to you, I urge you to make a donation to MDA today to help us continue to seek out information and inspiration we can share with you. In this issue, we're taking the focus inside, with tips on how to cope with fear — and even have some fun — as we weather this storm together. We're here with you — and your support can help us continue to be.

Please, give to MDA and *Quest* online or by mail. And read on. The stories may be different, but the spirit is the same, and strong.

Sincerely,

*Lindsey Baker*  
Quest Editor-in-Chief and General Manager  
Muscular Dystrophy Association



Lindsey Baker

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### ADVOCACY IN UNCERTAIN TIMES

MDA's advocacy team continues to amplify the voices of those in the neuromuscular disease community. This is even more important now, when lawmakers and regulators are making critical decisions that affect our community.

Learn how MDA advocates for the neuromuscular disease community and how you can join us at [mda.org/advocacy](https://mda.org/advocacy).



MDA-056



## **EMFLAZA® (deflazacort) AVAILABILITY DURING CORONAVIRUS (COVID-19) PANDEMIC**

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# New Ways to Learn



## WE WANT TO HEAR FROM YOU

What is important to you when it comes to education? What information would be most helpful to you? Your feedback will help the MDA Engage team shape future events. Please email [mdaengage@mdausa.org](mailto:mdaengage@mdausa.org) with your suggestions.

Perhaps now, more than ever, we believe in the power of community. At MDA, we've seen firsthand how important it is to learn about the neuromuscular diseases that affect our community and build relationships with families going through similar experiences.

Because of COVID-19, MDA is making all MDA Engage events virtual for the remainder of 2020. If you are an individual living with a neuromuscular disease,

we invite you and your loved ones to join MDA Engage education events taking place online.

### +FIND AN EVENT

For a complete list of MDA Engage events, visit [mda.org/engage](http://mda.org/engage). Here, you can register for upcoming events and view videos from past Engage events and webinars.

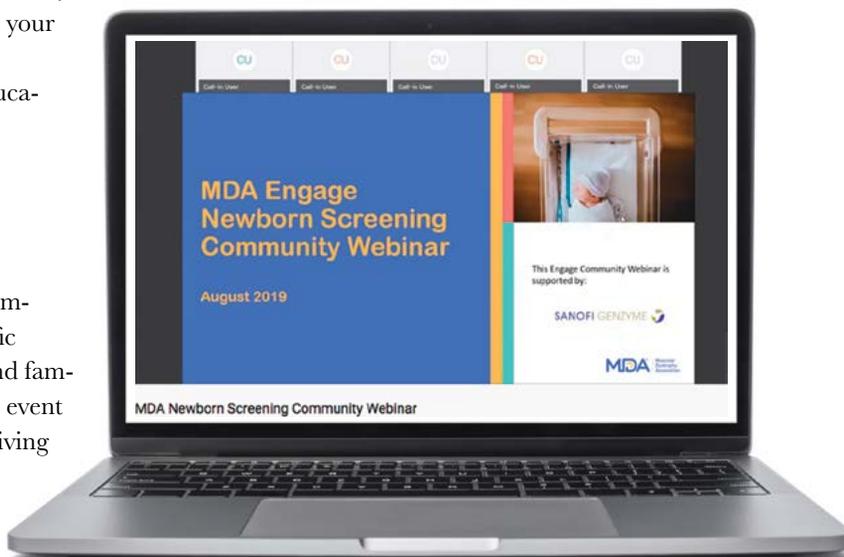
### Seminars and Symposia

MDA Engage Community Education Seminars and Disease Specific Symposia are events empowering individuals and families with knowledge and resources. Each online event will feature experts in the field and individuals living with neuromuscular disease. They will cover topics such as clinical care best practices, research updates, and more.

### Webinars

MDA Engage Community Webinars are online education sessions geared for those living with neuromuscular disease and their support network. Each webinar is led by experts in the field. Webinars are offered live and on-demand.

View MDA Engage webinars at [mda.org/engage](http://mda.org/engage).



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LIVING WITH MG



## The CHAMPION MG STUDY

Alexion is currently recruiting patients with anti-acetylcholine antibody receptor positive generalized myasthenia gravis (MG) 18 years of age or older for a Phase 3 study of ravulizumab-cwvz, called the CHAMPION MG Study. The study will assess ravulizumab-cwvz, compared to placebo, on the improvement of MG symptoms (MG activities of daily living). Participants may continue on their current medicines\*, as long as they are stable, and after a 26-week study treatment period all participants can receive ravulizumab-cwvz for an additional follow up period of up to 2 years. For more information and to learn if you are eligible for the CHAMPION MG Study, please contact [ClinicalTrials@alexion.com](mailto:ClinicalTrials@alexion.com) or go to [MGCHAMPION.com](http://MGCHAMPION.com).

\*Except for other complement inhibitors, rituximab, chronic Plasma Exchange or Intravenous Immunoglobulin



# PROGRESS NOW

Tracking research updates and breakthroughs that help accelerate treatments and cures across MDA diseases

## **Amyotrophic lateral sclerosis (ALS)**

### **Phase 3 Radicava Study Seeks Participants**



Researchers at Mitsubishi Tanabe Pharma Development America are looking for individuals with ALS to participate in a phase 3 study evaluating the safety of oral edaravone (Radicava). Intravenous (IV) infusion of Radicava is approved by the US Food and Drug Administration (FDA) to treat ALS. It has been shown to slow the decline of physical function. Radicava is thought to act as a free radical scavenger and prevent oxidative stress damage to neurons.

All participants will receive an oral suspension of Radicava. The study will consist of eight visits over the course of one year. Throughout the study, participants will continue to see their regular doctor for routine care.

---

To learn more about eligibility requirements or to inquire about participation, visit [clinicaltrials.gov](https://clinicaltrials.gov) and enter NCT04165824 in the “Other Terms” search box.

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### **FDA Approves Tiglutik for Feeding Tube**

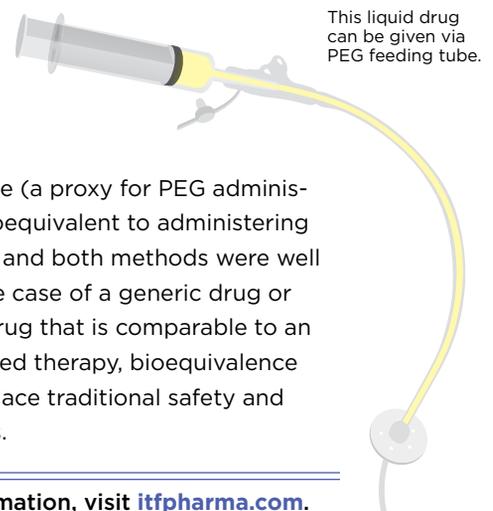
The FDA approved expanded labeling for ITF Pharma’s Tiglutik, the liquid form of the tablet riluzole (Rilutek), to include administration via percutaneous endoscopic gastrostomy (PEG) feeding tube for the treatment of ALS. While Rilutek has been available for more than 20 years, this expanded labeling will enable individuals with ALS who not only have difficulty swallowing but also require a PEG feeding tube to take Tiglutik.

The approval of expanded labeling for Tiglutik was based on a study that showed that dosing Tiglutik via

intra-gastric tube (a proxy for PEG administration) was bioequivalent to administering the drug orally, and both methods were well tolerated. In the case of a generic drug or reformulated drug that is comparable to an existing approved therapy, bioequivalence testing can replace traditional safety and efficacy studies.

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For more information, visit [itfpharma.com](https://itfpharma.com).

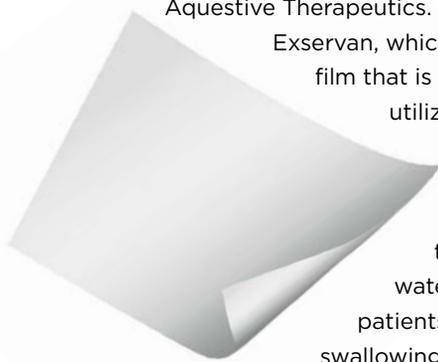


Images: iStock.com/serezny; muzon; pattarawit; Katerina Sisperova; romma; Enis Aksoy

## Amyotrophic lateral sclerosis (ALS)

# New Rilutek Formulation Dissolves on the Tongue

The FDA approved riluzole oral film (Exservan) for the treatment of ALS. Exservan is an oral film formulation of Rilutek, which has been available in tablet form to treat ALS for more than 20 years. Exservan will be made available in the United States and will be marketed by Aquestive Therapeutics.



The thin oral film may be an easier way to take riluzole for patients who have trouble swallowing.

Exservan, which consists of a thin film that is placed on the tongue, utilizes the company's "PharmFilm" technology. The dissolving oral film can be taken twice daily without water, making it easier for patients who have difficulty swallowing pills or liquids — a common symptom of ALS.

Studies performed by Aquestive demonstrated that Exservan is almost identical to Rilutek in how it is absorbed, distributed, and metabolized in the body. The company also performed studies to assess patients' ability to swallow Exservan.

For more information, visit [aquestive.com](https://aquestive.com).



## Duchenne muscular dystrophy (DMD)

# FDA Approves Vyondys 53 Exon-Skipping Therapy

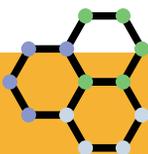
Last December, the US Food and Drug Administration (FDA) conditionally approved golodirsen (Vyondys 53) for the treatment of DMD in patients amenable to skipping exon 53. It is the second exon-skipping, disease-modifying drug to treat DMD, the most common childhood form of muscular dystrophy. It is estimated that up to 8% of patients with DMD have mutations amenable to treatment with Vyondys 53.

The drug is administered by intravenous (IV) infusion. Vyondys 53 will be made available in the United States and marketed by Sarepta Therapeutics.

The 25 boys with DMD who participated in the pivotal phase 1/2 clinical trial conducted in Europe were evaluated for a total of 144 weeks. Trial results showed proper exon skipping and an increase in dystrophin production in participants' skeletal muscles. Based on these results, the FDA found it reasonably likely that Vyondys 53 would be beneficial to those who were treated. The continued approval of Vyondys 53 may be contingent on confirmation of a clinical benefit in a post-marketing confirmatory trial (ESSENCE), which is currently enrolling and expected to conclude by 2024.

For more information about the ESSENCE trial, visit [clinicaltrials.gov](https://clinicaltrials.gov) and enter NCT02500381 in the "Other Terms" search box.

## Researchers Speak



When clinicians and researchers could not gather in person in March, MDA found an innovative way to bring them together for important interdisciplinary learning through a virtual presentation of the 2020 MDA Clinical & Scientific Conference's Clinical Trials session. More than 1,200 clinicians and scientists registered for this intensive one-day session to hear researchers and industry experts present key findings from promising studies and updates on research that has the potential to advance discovery and drug development across multiple neuromuscular diseases.

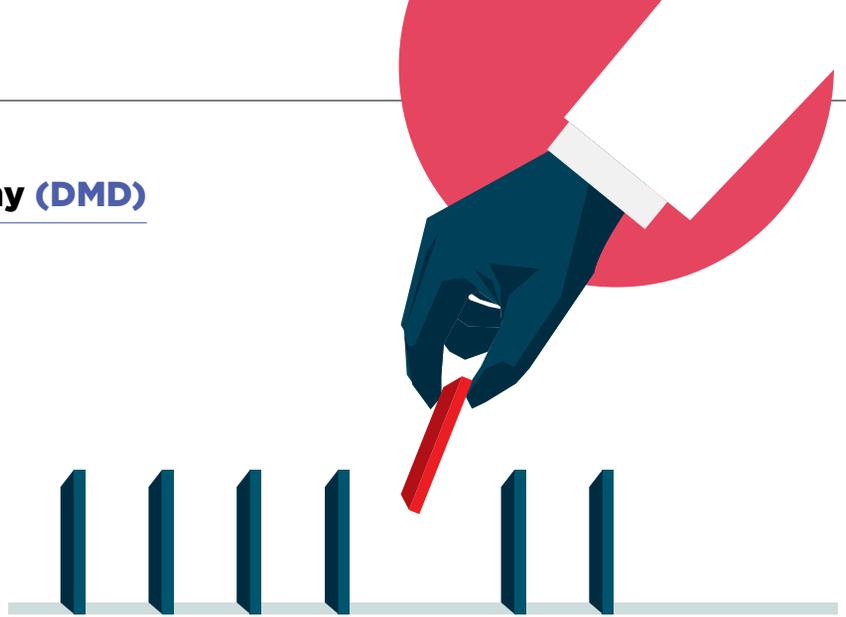
Read highlights from the Clinical Trials session in "MDA'S 2020 Clinical & Scientific Conference Trial News and Updates" at [mda.org/quest](https://mda.org/quest).

## Duchenne muscular dystrophy (DMD)

# FDA Considers New Exon-Skipping Therapy

The FDA accepted NS Pharma's New Drug Application (NDA) for viltolarsen, an investigational therapy for the treatment of DMD in patients amenable to skipping exon 53. An anticipated decision is expected in the third quarter of 2020.

If approved, viltolarsen would be the third disease-modifying therapy for DMD and another option for the treatment of DMD in patients amenable to exon 53 skipping. Recently, the FDA conditionally approved Vyondys 53, the first disease-modifying therapy for this patient population.



The submission included positive results from a phase 2 study and its long-term extension in the United States and Canada, and from phase 1 and phase 1/2 studies performed in Japan.

To learn more about the completed phase 2 trial, visit [clinicaltrials.gov](https://clinicaltrials.gov) and enter NCT02740972 in the "Other Terms" search box.

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## Friedreich's ataxia (FA)

# MDA Awards MVP Grant to Help Develop Gene-Replacement Therapy

MDA awarded Venture Philanthropy (MVP) funding totaling \$1,076,232 to AavantiBio. This award will help advance the biotechnology company's phase 2 clinical trial of a gene-replacement therapy for FA.

MVP is MDA's drug development program, which is focused on funding the discovery and clinical application of treatments and cures for neuromuscular disorders.

AavantiBio co-founders Manuela Corti, PT, PhD, assistant professor of pediatrics at the University of Florida (UF), Gainesville, and Barry Byrne, MD, PhD, associate chair of pediatrics and director of the Powell Gene Therapy Center at UF, started working with the FA community five years ago and founded AavantiBio with the goals of developing an effective treatment for FA and improving the lives of people living with FA.

"This is a great opportunity for AavantiBio, and we're

thankful to MDA for their generous contribution," Dr. Corti says.

"We hope to strengthen our collaboration as we work together on this project."

Drs. Corti and Byrne plan to include adults and children who have FA in the clinical trial, which is expected to begin this year.



Read more at [mda.org](http://mda.org); enter AavantiBio in the search box on the homepage.

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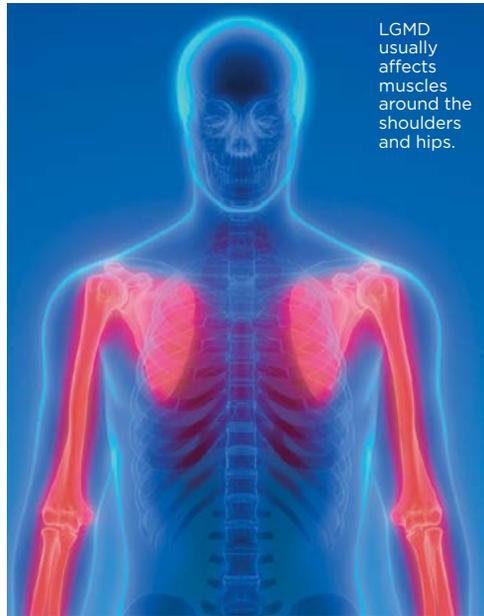
## Limb-girdle muscular dystrophy (LGMD)

# Phase 3 Emflaza Trial Seeks Participants

Researchers at PTC Therapeutics are looking for individuals with LGMD type 2I (LGMD2I) to participate in a phase 3 study evaluating the efficacy of deflazacort (Emflaza), which researchers hope may reduce inflammation in muscles and potentially lead to improved muscle strength and motor function.

The placebo-controlled study will last one year, with approximately six clinic visits that will involve quality-of-life questionnaires, functional tests, and laboratory tests.

To be eligible to participate, candidates must meet several



criteria, including having a genetic diagnosis of LGMD2I with a confirmed mutation in the fukutin-related protein gene (FKRP). Travel support is available.

To learn more about eligibility requirements or to inquire about participation, contact Mary Frances Harmon at **866-562-4620** or [medinfo@ptcbio.com](mailto:medinfo@ptcbio.com). To find out more information about the study, visit [clinicaltrials.gov](http://clinicaltrials.gov) and enter NCT03783923 in the "Other Terms" search box, or visit [clinicaltrials.ptcbio.com/en/trial-details/?id=PTCEMF-GD-004-LGMD](http://clinicaltrials.ptcbio.com/en/trial-details/?id=PTCEMF-GD-004-LGMD).

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## Spinal muscular atrophy (SMA)

# FDA Accepts New Drug Application for SMA Therapy

The US Food and Drug Administration (FDA) accepted Genentech's New Drug Application (NDA) for risdiplam, an investigational therapy for the treatment of SMA. An anticipated decision is expected by Aug. 24, 2020.

The company's NDA submission includes data from 12 months of the initial dose-finding parts of the company's pivotal FIREFISH and SUNFISH trials, as well data from the confirmatory second part of SUNFISH. Previously, the FDA granted Orphan Drug and Fast Track designations to risdiplam in January 2017 and April 2017, respectively.

Risdiplam is an investigational SMN2-splicing modifier designed to help the SMN2 gene produce more SMN protein, which is deficient in SMA patients. Risdiplam's development is part of a collaboration between Genentech (a Roche company), PTC Therapeutics, and the SMA Foundation. Risdiplam is being studied in a broad clinical trial program in patients with SMA ranging in age from newborn to 60 years old.

To learn more about the FIREFISH, SUNFISH, JEWELFISH, and RAINBOWFISH trials, visit [clinicaltrials.gov](http://clinicaltrials.gov) and enter NCT02913482, NCT02908685, NCT03032172, and NCT03779334 in the "Other Terms" search box.



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# Pompe Disease Treatment Is Moving Forward

A Q&A with Priya Sunil Kishnani, MD, MBBS

**P**ompe disease (also called acid maltase deficiency) is a rare, inherited glycogen storage disease that affects the muscles, particularly the heart and skeletal muscles. It results from mutations in a gene that carries instructions to make the enzyme acid alpha-glucosidase (GAA), also called acid maltase, which plays a role in the body's ability to process and break down complex sugars (glycogen). With insufficient GAA, glycogen builds up in and damages muscle cells.

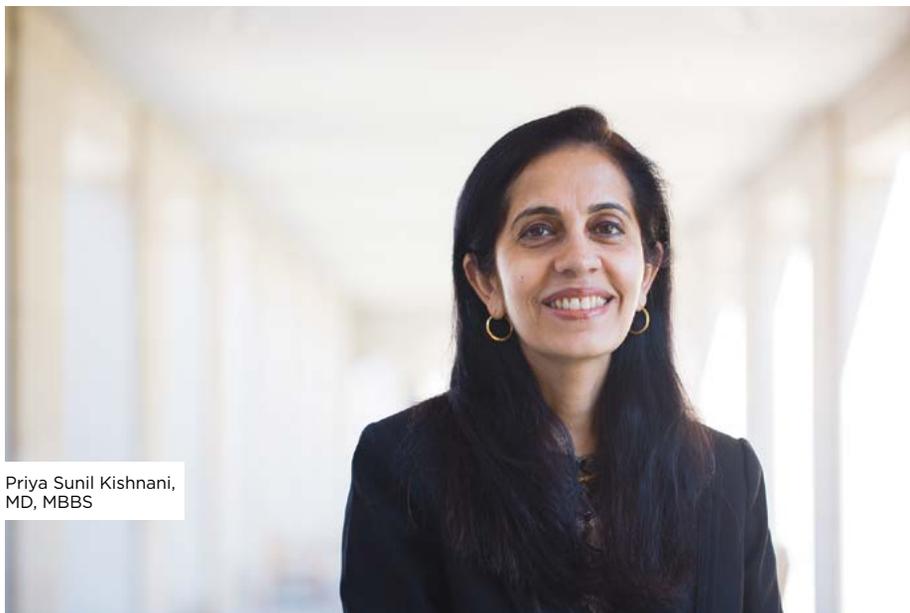
Pompe affects an estimated 1 in every 40,000 births, although the incidence is increasing as more newborns are screened for the disease.

There are two types: infantile-onset and late-onset (juvenile or adult).

We spoke with Priya Sunil Kishnani, MD, MBBS, the Chen Family Distinguished Professor of Pediatrics at Duke Medical Center in Durham, N.C., about the latest in Pompe treatment and research.

## What is it like to live with Pompe?

That's a multipronged answer, because Pompe involves a spectrum of disease. The infantile form requires intervention starting right after birth. It requires not only



Priya Sunil Kishnani,  
MD, MBBS

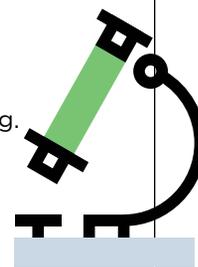
enzyme-replacement therapy (ERT), which is lifesaving, but also multidisciplinary support with occupational therapy, physical therapy, speech therapy, and multiple specialists, including cardiologists and pulmonologists.

Families have to adjust to the fact that their child is living with a lifelong chronic condition with lifelong interventions. But I see families embrace this new way of life and find a new normal.

The rest of the disease spectrum is often called late-onset Pompe disease; however, these patients can present as early as the first year of life to as late as the sixth decade.

## GETTING REAL WITH DRUG DEVELOPERS

MDA is preparing for our first Pompe disease patient-focused drug development (PFDD) meeting. This meeting will provide individuals with Pompe and their families an opportunity to inform the US Food and Drug Administration (FDA), drug developers, and other key stakeholders about the daily experiences of living with Pompe disease and how they view the benefits and risks of treatments. Look for updates at [cqcengage.com/mda/pfddmeeting2020?0](https://cqcengage.com/mda/pfddmeeting2020?0).



Interventions still need to be started early — not just ERT, but also nutrition, physical therapy, and other specialty care.

### **How have you seen Pompe care evolve from no treatment to the potential of gene therapy?**

I've been working in the field of Pompe disease for close to 30 years and have been part of the journey from understanding the natural history of the disease to the excitement of next-generation treatments, including what we call second-generation ERTs, as well as gene therapy.

### **What are the benefits and limitations of current ERTs?**

There are limitations to the current therapy, including access to the treatment (it costs about \$300,000 a year). And while it is very good at clearing glycogen in the heart, we don't find it as efficient in targeting skeletal muscle. Thus, we see glycogen building up again in skeletal muscles or not being cleared in some of the muscle fibers. Over time, patients tend to plateau, or even decline.

We've learned that starting treatment early, before there is any damage, helps. That's why it's so important that Pompe be added to state newborn screening panels. (As of February 2020, just 20 states and the District of Columbia screened for Pompe.)

The other challenge is immunogenicity, which is an antibody response to the enzyme that can be deadly. However, we have developed immune tolerance induction protocols that are largely successful, especially in the highest-risk babies who have two mutations and make no GAA enzyme in their own bodies. As such, when they receive ERT (Myozyme/Lumizyme), the body treats it as a foreign protein and makes antibodies to the ERT. By treating these babies with agents that trick the immune system, we are able to prevent an antibody response and now have some long-term survivors in their teens.

Nutrition, exercise, and physical therapy are all important, but there are often access issues to obtaining these services. We also see a psychosocial aspect of the illness, with children often feeling some social isolation.

### **What are the most recent advances in treatment and clinical research?**

Second-generation ERTs in phase 3 clinical trials provide a more efficient delivery method and better clearance of



glycogen in skeletal muscle. They may also have better benefits than the current ERT (Myozyme/Lumizyme) at a lower dose. There is also a gene therapy in phase 1 clinical trials and several others that are close to starting clinical testing.

There is a lot of interest in the field in moving forward.

We are also trying to better understand the earliest features and manifestations of late-onset Pompe disease, as now we are identifying babies who will have the late-onset type through newborn screening. We are investigating if starting people early on the right dose of ERT could impact outcomes. Early evidences suggests it can, but there is still work to be done.

Another good thing is that the lessons from Pompe disease are guiding clinical development for other rare muscle diseases, like Duchenne muscular dystrophy (DMD) and spinal muscular atrophy (SMA). These learnings are complementary and enable us to move the field forward at a better pace than if we were studying these in isolation.

We are so grateful to our patients with Pompe who are always available to allow this continued learning. [@](#)

“

We've learned that starting treatment early, before there is any damage, helps. That's why it's so important that Pompe be added to state newborn screening panels.



**“I’ll always remember the day when a one-time dose gave him a second chance at life.”**

**Natalie, mother of Eli**

ZOLGENSMA® (onasemnogene abeparvovec-xioi) is a prescription gene therapy used to treat children less than 2 years old with spinal muscular atrophy (SMA). ZOLGENSMA is given as a one-time infusion into a vein. ZOLGENSMA was not evaluated in patients with advanced SMA.



Eli, treated at ~4 months and pictured at 5 years, has SMA Type 1.

To learn more, talk to your child’s doctor about the **one-time-only dose** and visit [ZOLGENSMA.com](http://ZOLGENSMA.com).

**Indication and Important Safety Information**

**What is ZOLGENSMA?**

ZOLGENSMA is a prescription gene therapy used to treat children less than 2 years old with spinal muscular atrophy (SMA). ZOLGENSMA is given as a one-time infusion into a vein. ZOLGENSMA was not evaluated in patients with advanced SMA.

**What is the most important information I should know about ZOLGENSMA?**

- ZOLGENSMA can cause acute serious liver injury. Liver enzymes could become elevated and may reflect acute serious liver injury in children who receive ZOLGENSMA.
- Patients will receive an oral corticosteroid before and after infusion with ZOLGENSMA and will undergo regular blood tests to monitor liver function.
- Contact the patient’s doctor immediately if the patient’s skin and/or whites of the eyes appear yellowish, or if the patient misses a dose of the corticosteroid or vomits it up.

**What should I watch for before and after infusion with ZOLGENSMA?**

- Viral respiratory infections before or after ZOLGENSMA infusion can lead to more serious complications. Contact the patient’s doctor immediately if you see signs of a possible viral respiratory infection such as coughing, wheezing, sneezing, runny nose, sore throat, or fever.
- Decreased platelet counts could occur following infusion with ZOLGENSMA. Seek immediate medical attention if a patient experiences unexpected bleeding or bruising.

**What do I need to know about vaccinations and ZOLGENSMA?**

- Talk with the patient’s doctor to decide if adjustments to the vaccination schedule are needed to accommodate treatment with a corticosteroid.
- Protection against respiratory syncytial virus (RSV) is recommended.

**Do I need to take precautions with the patient’s bodily waste?**

Temporarily, small amounts of ZOLGENSMA may be found in the patient’s stool. Use good hand hygiene when coming into direct contact with bodily waste for 1 month after infusion with ZOLGENSMA. Disposable diapers should be sealed in disposable trash bags and thrown out with regular trash.

**What are the possible or likely side effects of ZOLGENSMA?**

The most common side effects that occurred in patients treated with ZOLGENSMA were elevated liver enzymes and vomiting.

**The safety information provided here is not comprehensive. Talk to the patient’s doctor about any side effects that bother the patient or that don’t go away.**

**You are encouraged to report suspected side effects by contacting the FDA at 1-800-FDA-1088 or [www.fda.gov/medwatch](http://www.fda.gov/medwatch), or AveXis at 833-828-3947.**

**Please see the Brief Summary of the Full Prescribing Information on the next page.**



## IMPORTANT FACTS ABOUT ZOLGENSMA® (onasemnogene abeparvovec-xioi)

### USE

ZOLGENSMA is a prescription gene therapy used to treat children less than 2 years old with spinal muscular atrophy (SMA).

- ZOLGENSMA is given as a one-time infusion into the vein.
- ZOLGENSMA was not evaluated in patients with advanced SMA.

### WARNINGS

#### Acute Serious Liver Injury and Elevated Liver Enzymes

- ZOLGENSMA can cause acute serious liver injury. Liver enzymes could become elevated and may reflect acute serious liver injury in children who receive ZOLGENSMA.
- Patients will receive an oral corticosteroid before and after infusion with ZOLGENSMA and will undergo regular blood tests to monitor liver function.
- Contact the patient's doctor immediately if the patient's skin and/or whites of the eyes appear yellowish, or if the patient misses a dose of the corticosteroid or vomits it up.

**Decreased platelet counts** could occur following infusion with ZOLGENSMA. Caregivers should seek immediate medical attention if a patient experiences unexpected bleeding or bruising.

### OTHER IMPORTANT INFORMATION

**Patients should be tested for the presence of anti-AAV9 antibodies** prior to infusion with ZOLGENSMA.

**Vaccination** schedule should be adjusted where possible to accommodate treatment with an oral corticosteroid. Caregivers should talk with the patient's doctor to decide if adjustments to the vaccination schedule are needed during corticosteroid use. Protection against respiratory syncytial virus (RSV) is recommended.

**Viral respiratory infections** before or after ZOLGENSMA infusion can lead to more serious complications. Contact the patient's doctor immediately if you see signs of a possible viral respiratory infection such as coughing, wheezing, sneezing, runny nose, sore throat, or fever.

**Temporarily, small amounts of ZOLGENSMA may be found in the patient's stool.** Use good hand hygiene when coming into direct contact with bodily waste for 1 month after infusion with ZOLGENSMA. Disposable diapers should be sealed in disposable trash bags and thrown out with regular trash.

### COMMON SIDE EFFECTS

**The most common side effects** that occurred in patients treated with ZOLGENSMA were elevated liver enzymes and vomiting.

**These are not all the possible side effects. Talk to the patient's doctor about any side effects that bother the patient or that don't go away.**

### QUESTIONS?

To learn more, talk to your doctor and you can visit [www.ZOLGENSMA.com](http://www.ZOLGENSMA.com) for Full Prescribing Information.

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# Coping

## THROUGH A

# Crisis

*Experts weigh in on the keys to resilience in a stressful world*

BY BARBARA  
TWARDOWSKI

Fifteen years ago, my family lived through Hurricane Katrina in Louisiana. At the time, our son was a freshman in high school. Falling pine trees and rain destroyed half of our home. Rebuilding took seven months, and we lived in the chaos during the process.

At the time, I was fairly independent. Because I have Charcot-Marie-Tooth disease (CMT), I used a small power wheelchair and drove a wheelchair-accessible van.

Fast forward to today: I no longer drive, and I need assistance with showering, transferring to a bed, and dressing. I'm often home alone, because my husband, a nurse who works in a hospital, is my caregiver, and our son lives across the country.

Again, we're living through a different kind of destruction. While our home is intact and secure, the danger of the novel coronavirus has gripped the nation and changed our way of life.

### Fear is normal

Functioning in the world often feels like a precarious balancing act. For those of us with a neuromuscular disease, disruptions are difficult and require continually adapting and finding new ways to cope. Whether you are dealing with a pandemic or another stress-producing event, how you respond affects your physical, mental, and emotional well-being.

"Regardless of the type of crisis — whether it is a pandemic, a hurricane, a job loss, or the loss of a loved one — anything out of the ordinary is going to rock your boat a little bit when you have a chronic medical condition," says Teri Brister, PhD, LPC, director of information and support at the National Alliance on Mental Illness (NAMI).

"Two big causes of fear are encountering things that are unknown and feeling out of control," Dr. Brister says. These

feelings are normal. One of the best ways to respond is to take control of what you can. For example, take stock of your prescription medications and request a refill for anything you are running low on.

"There's a whole other layer of fear because you have a health issue," Dr. Brister says.

Worrying about your caregiver becoming ill is a legitimate fear. Working on solutions might be scary or present new challenges, but creating an action plan can help you regain a sense of control.

### Coping strategies

Every person needs an individualized strategic plan for coping with stressors.

"One simple technique is to acknowledge that you are feeling anxious and that it's a completely appropriate response," says Sarah Clark-Stoney, MSW, LSW, a social worker at the Children's Hospital of Philadelphia. "Dismissing these feelings can allow anxiety to compound."

A good coping strategy is to engage in a project that is meaningful to you. Think of things you like to do that you've been putting off, and experiment with outlets that take your mind off stressing. For example, immerse yourself in a good book, listen to music, participate in chair yoga, complete a jigsaw puzzle, or write in a journal.

Your fears are not irrational, and you are not going to make them go away. But instead of dwelling on "what ifs,"

### +GET SUPPORT

The National Alliance on Mental Illness (NAMI) provides mental health support and resources. Call the NAMI HelpLine Monday through Friday, 10 a.m.-6 p.m. ET, at **800-950-NAMI (6264)** or email [info@nami.org](mailto:info@nami.org).

Dr. Brister suggests, ground your thoughts by thinking about a person who is important in your life or a place where you feel relaxed and safe. Call upon these images when you feel distressed.

### Combating isolation

Having a network of friends, family, and neighbors who support you is crucial. If a crisis affects a large group of people, portions of that network might crumble. Be willing to ask for and seek help when you need it. For example, join a social media group such as Nextdoor to connect with people and businesses in your community that are offering support. If you belong to a place of worship, let them know you need assistance. You can also ask the MDA Resource Center about support organizations in your area. (Call 833-ASK-MDA1.)

### HELP CHILDREN HANDLE STRESS AT HOME



When the world goes topsy-turvy, the family is a child's support system. Here are tips for keeping home a calm, safe place.

- **Encourage open discussion.** Don't discourage kids from asking questions, and provide age-appropriate answers. Parents should never dramatize a situation; children emulate their parents.
- **Stick to a routine.** Following a regular schedule as much as possible gives kids the comfort of knowing what's coming next.
- **Be consistent.** Children's anxiety can manifest as misbehavior or physical symptoms, such as tummy aches. Continue to maintain house rules and set limits on kids' behaviors. "Consistency and structure are like a security blanket for your child during times like these," says Sarah Clark-Stoney, MSW, LSW, a social worker at the Children's Hospital of Philadelphia.
- **Connect with peers.** Just like adults, kids need social interaction. When you can't meet in person, schedule virtual chats with friends and family.
- **Engage with nature.** If you can't get outside, try growing herbs or a tomato plant in a pot. Or observe the birds in your backyard and try to identify them using an app.
- **Cherish your time together.** Extended time at home presents an opportunity to introduce new family rituals, such as a family video journal, or to try something new, like sleeping in a tent in the backyard.

Even if you don't live alone, you might feel isolated during times of social distancing. Make a point to speak with a family member or friend every day. Stay in touch with members of your social groups virtually using Facebook, Zoom, or Skype. Join in on MDA's Game Night, which is a wonderful way for kids and teens to interact online in a safe environment. (See page 21.)

"It's essential that we find a way to get connected. We get support from each other," Dr. Brister says.

### When to seek a professional

"When you need to consult a therapist looks a little different for everybody," Dr. Brister says. "Do you want to stay in bed and pull the covers over your head? Let's be real: Everybody has days like that from time to time. However, if that has gone on for several days or a week, it's a good idea to talk with somebody."

Says Sarah, "We can feel as though we are trapped when forced to stay inside. Restore some calm by looking to nature. Sit by an open window or take an activity outdoors. Connecting with nature can be very restorative to your mental health."

Finding this connection can offer a much-needed mental break. "Many of us are deeply connected to our technology, especially now. We're working from home, streaming our shows, texting our friends, and video chatting," Sarah says. "When you go outside, allow yourself to take a break from technology. Try to be present and mindful, focusing on the sounds and sights of nature."

### Manage media intake

Communication, particularly news, is important. However, if you can't stop watching television or checking your news feed because you are afraid of missing the latest information, anxiety is controlling you instead of you controlling the anxiety.

Limit how much time you spend reading and watching media. Be especially mindful of what children might overhear.

During a crisis, everyone feels afraid and anxious. These are normal responses.

"Make yourself get up, make yourself go outside, and make yourself do something," Dr. Brister says. "Surviving any catastrophic situation depends upon your resilience." 

Barbara Twardowski and her husband, Jim, a registered nurse, live in Louisiana. Together, they write about accessible travel, disability issues, and health topics.

### +EMERGENCY RESOURCES

MDA wants to give you the knowledge and resources to cope with any emergency. For COVID-19 resources, visit [mda.org/covid19](https://mda.org/covid19). For emergency preparedness resources, visit [mda.org/emergency-resources](https://mda.org/emergency-resources).

# Game On!



On March 21, MDA launched Game Night, a weekly Saturday night event designed to gather online gamers and families for regular connection (and a little healthy competition).

It couldn't have been better timed.

MDA began developing its online gaming platform, MDA Let's Play, in 2019. The idea was simple: connect the online gaming community and the MDA community — which already overlap — to grow a new network of support. And now, as families have found themselves practicing social distancing from friends, neighbors, and family, there's no better time to go online for a bit of fun together in support of MDA's cause — and maybe to build the most amazing Minecraft village in history while they're at it.

*MDA Let's Play  
takes family  
fun online*

BY LINDSEY  
BAKER





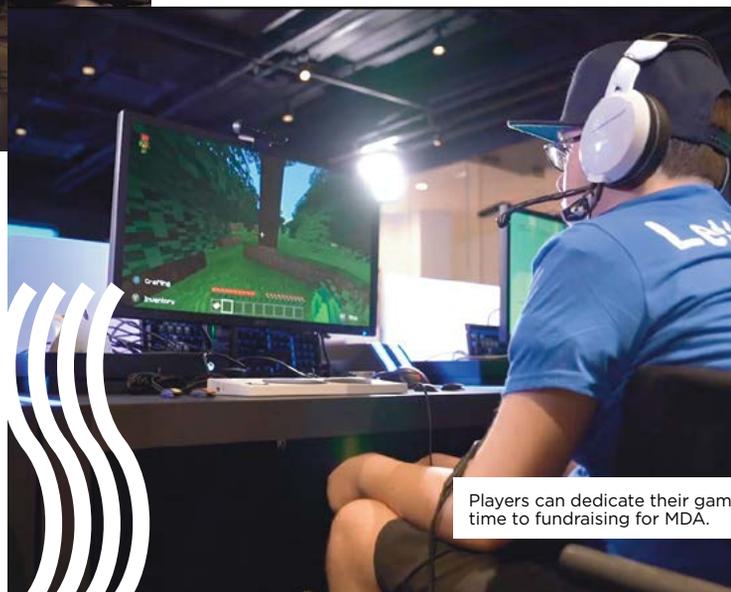
### Watch and play

Developed through a partnership with 4Ever Wild Productions, the centerpiece of MDA Let's Play is a custom channel on Discord, a group messaging platform popular with the online gaming community. People can join the Discord channel and log in to play with individuals, families, and supporters of the MDA community any time. Each Saturday at 7 p.m. ET, Game Night will feature several exclusive opportunities to play games like Minecraft or Fortnite with the community.

"Today, young people are gaming," says Larry Lieberman, 4Ever Wild's executive producer. "One-hundred sixty-five million Americans play video games. They come in all ages and styles, all across the country. MDA Let's Play is a place where anybody connected in any way to the MDA community can join and play games together and know it's a safe place for kids and adults, patients or families, volunteers, camp counselors, regional directors, fire fighters, Harley-Davidson owners, you name it. It's a place where folks can just reach out on Discord and start a game any time with anyone."

For those who don't game — or who are learning how to — Let's Play also has a channel on Twitch, a live-streaming platform where gamers share their gameplay in real time. Viewers can watch Let's Play Game Night on Twitch if they aren't quite ready to get in the game themselves — and of course, too, if they're experienced gamers getting a feel for the competition.

The Let's Play Twitch channel also features celebrity gamers and fundraising events. Earlier this year, Twitch influencer Wade Barnes, aka LordMinion777, hosted a six-hour



“



Knowing that you're not alone is the greatest, most powerful part of this — that you're not alone in your healthcare, you're not alone in your quest for research and new innovation, and you're not alone at play.

—Larry Lieberman

game-a-thon that raised more than \$23,000 to support MDA's mission.

Individual players can dedicate their game time to fundraising for MDA, as well. After registering at [mdaletsplay.org](http://mdaletsplay.org), gamers can create fundraising game-a-thons on their own social channels, including Twitch, YouTube, and Mixer.

### Creating positive community

Each week, individuals and families who've joined the Let's Play Discord channel will receive updates about that week's Game Night — what games the community will be playing and other announcements. Once players log into Discord, they can choose what game they'd like to play and they'll be taken to a private online room to game with other Let's Play players.

Both the Discord game rooms and the Twitch streaming channel have chat features where players and viewers can interact, and both have moderators who keep experiences safe, by blocking inappropriate language and behavior, and

engaging, by fielding questions and offering encouragement and guidance. Moderators are available outside of Game Night, too, to help players learn new games, pass on feedback, and reply to messages.

As of April, almost 800 players had joined the Let's Play Discord channel. Larry says he's hoping for 1,000 registered players by May. As the community grows, he says, members will help determine which games are played. Eventually, he hopes both to bring in volunteers from the MDA community to help moderate Let's Play channels and to work with moderators to build E-sports teams within the Let's Play community to participate in online tournaments.

And as supporters view, join, and begin to learn about MDA and the research it funds and shares worldwide, individuals and families impacted by neuromuscular disease can give back by making connections and sharing their stories — and their mad Jackbox skills.

“Just joining the community empowers everyone who's involved,” Larry says. “Knowing that you're not alone is the greatest, most powerful part of this — that you're not alone in your healthcare, you're not alone in your quest for research and new innovation, and you're not alone at play. This is an



## LET'S PLAY!

Online: [mdaletsplay.org](http://mdaletsplay.org)  
On Discord: [discord.gg/VracthK](https://discord.gg/VracthK)  
On Twitch: [twitch.tv/mda\\_letsplay](https://twitch.tv/mda_letsplay)



enormous community, and we aren't able, especially in this current life we're living, to connect enough with people who understand. Just by joining this community on Discord and following along on Twitch, every single person in the MDA community can get involved and learn and be touched.” 

Lindsey Baker is editor-in-chief of *Quest*.

### ADVERTISEMENT

## Pompe Pediatric Study Now Enrolling

The ZIP study will evaluate how an investigational co-administration treatment regimen of ATB200 and AT2221 may work for adolescents (12 through 17 years of age) with late-onset Pompe disease, who have either received Enzyme Replacement Therapy (ERT) (ERT-experienced) or have never received ERT (ERT-naïve) previously.

The safety and efficacy of the investigational agents ATB200 and AT2221 has not been established, and they are not approved for use in any country.

For more information, please visit [pompestudy.com/zip](http://pompestudy.com/zip) or [clinicaltrials.gov](https://clinicaltrials.gov) (Identifier: NCT03911505).



Now, more than ever, we're spending a lot of time at home. While the continuing threat of the novel coronavirus makes this the safest place to be, home is also a place where frustrations can multiply when trying to accomplish everyday tasks.

We chatted with experts and people living with neuromuscular diseases to get tips on how to ease some common struggles so you can truly feel the comforts of home.

# 7 Ways to Make Life at Home Easier

*How to master your tasks and practice self-care*

BY DONNA ALBRECHT



Image: iStock.com/polligonchik

## 1 Use your equipment.

When you're staying in, putting on equipment that supports posture or stability can seem like too much of a bother. However, slumping in a chair instead of wearing a body jacket compromises your ability to breathe.



Wearing leg braces at home helps Neenah Williams avoid falls.

Neenah Williams, who lives with facioscapulohumeral muscular dystrophy (FSHD), recently started wearing her leg braces around the house daily, instead of only when she's going out. "Wearing braces, even though I can still walk without them, is the smartest thing I have done," she says. "They give me my mobility back, and I don't have to constantly be afraid of tripping or hurting myself."

## 2 Find kitchen helpers.

Christy Horner, an occupational therapist at the MDA/ALS Care Center at

Vanderbilt University Medical Center, often tells her clients about common household items that can be repurposed to aid with food prep. "I advise patients to use pizza cutters instead of knives if they have the use of only one hand," she says. "And a hanger with pants clips can be used to keep a cookbook open; hang it on an upper cabinet handle for easy reading."

If gathering your ingredients is a challenge, Christy has solutions for that, too: "For a person who does not have a strong grip, tie a loop around the fridge handle to open the door with your forearm. In addition, tongs can be used as an inexpensive reacher."

## 3 Hack your meds.

Medications aren't always dispensed in the dose you need, and cutting a pill with a knife can give uneven results. Even pill cutters don't always help. Leah Leilani, who lives with mitochondrial myopathy (MM), says, "I find that certain medications either crumble easily, or I may need



Leah Leilani uses a razor blade to cut pills.

## DELIVERY IS THE NEW NORMAL

To adapt with the times, many businesses have found ways to take their services online, or bring them right to your door. Here are some things you can do without leaving the house.

- **Manage your money.** Nearly all transactions you used to conduct at the bank can now be done online, including depositing checks, paying bills, and applying for loans. Money apps like Venmo allow you to pay right from your smartphone. And budgeting apps like Mint can help you save more and spend less.
- **Run errands.** The Taskrabbit app connects you with "taskers" who can run errands, do pickup and delivery, and even help out in the yard or around the house. Currently, they're focusing on contactless tasks to help maintain social distancing.
- **Shop for food.** Type "grocery store delivery" into your search engine and you'll find a range of merchants, from local grocers to Target and Amazon, that deliver fresh food and pantry staples. If you're hankering for fruits and veggies, search for a community-supported agriculture (CSA) farm subscription service in your area. You'll not only get seasonal produce delivered, but you'll also support local farmers.
- **Travel.** When the world's tourist destinations closed, many of them went digital. Choose a Monterey Bay Aquarium web cam to watch frolicking sea otters or drifting jellies ([montereybayaquarium.org/animals/live-cams](http://montereybayaquarium.org/animals/live-cams)), or take an online tour of the Louvre ([louvrefr/en/visites-en-ligne](http://louvrefr/en/visites-en-ligne)). Check out Google Arts & Culture ([artsandculture.google.com](http://artsandculture.google.com)) for a collection of art and architecture from around the world.

two-thirds of a tablet, and a pill cutter can't help in these cases." But she has a solution: "In these situations, a simple razor blade is best to use."

## 4 Care for your hair.

For people with facial muscle weakness, like Dani Liptak, who lives with FSHD, washing hair can be a pain — literally. Dani can't always close her eyelids enough to keep the shampoo out. She tried baby shampoo, but it didn't clean her hair well. So she came up with alternative ways of washing her hair.

"I started using a shampoo with tea tree oil, and I installed a shower chair that allows me more accessibility to my head, so I can prevent the shampoo from getting in my

eyes,” Dani says. “I also use my kitchen sink once a week, because it is easier to lean my head in and control the sink’s hose and spout.”

## 5 Stay active.

When you’re staying home all day, it’s easy to be inactive. But according to physical therapist Polly Swingle, owner of The Recovery Project in Michigan, protecting your strength helps you maintain use of your hands and arms. Polly advises people with neuromuscular diseases to do daily range-of-motion exercises with their arms, moving the shoulder, elbow, wrist, and hand for 10 to 20 repetitions. If you are unable to do that on your own, you can have someone help you. Along with stretching the arm muscles, this helps expand the chest for better breathing.

Physical activity has the added benefit of improving your heart health and increasing blood flow to your brain. Christy notes that chair exercises can provide these benefits with less strain than standing ones. A search for “seated exercise” on YouTube offers a range of chair exercise videos that you can follow along with on your own or with a partner.

Christy emphasizes that exercises should be fun. “It should feel good, definitely not painful,” she says.

## 6 Do breathing exercises.

In her work with MDA Care Centers, Polly has seen that many people with neuromuscular diseases are particularly concerned with maintaining breathing muscles. To do that, it’s important to do breathing exercises. For example, once a day, take a moment to inhale through the nose and exhale slowly through the mouth, like blowing out a candle, for 10 to 20 repetitions.

Want to make a game of breathing exercises? Put cotton balls on a table and blow through a straw to move them around. Place an object on the table that serves as a goal and earn a point every time you get a cotton ball to your target.

## 7 Fall-proof your home.

Healing from a fall may cause some loss of strength that can’t be recovered. Christy advises modifying your home with fall prevention in mind. Grab bars, bed rails, and easy-grip door-knobs are inexpensive and easy to install, and they can make your home a safer space. [Q](#)

Donna Albrecht is a freelance writer who lives near San Francisco with her husband and border collie. She is the author of “Raising a Child Who Has a Physical Disability” (Wiley).

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**MDA**

## The Confidence to Succeed

One student works toward a better future for others with neuromuscular disease

Former Pennsylvania MDA Ambassador Shelby VanVliet is a familiar face on campus at the University of Pittsburgh. The neuroscience major is featured on a campaign poster for a campus-wide diversity and inclusion campaign, a joint effort between the school's offices of Diversity and Inclusion and Disability and Resources.

After getting involved in the campaign, Shelby, who lives with titin myopathy muscular dystrophy, joined a chancellor's advisory committee on disability to discuss accessibility issues on campus.

"It's an older campus, and I want to help make it more accessible to other students who are in wheelchairs," Shelby says. Specifically, she advised the committee on making campus sidewalks more user-friendly.

After she graduates in 2022, she plans to pursue a doctorate in computational neuroscience, and then begin a career in medical research for neuromuscular diseases.

Whether advocating for accessibility or furthering medical research, Shelby credits MDA with giving her the confidence and independence she has today. Throughout



Shelby VanVliet and her roommates attend an MDA fundraiser.

her childhood she participated in MDA Ride for Life and Fill the Boot, went to MDA Summer Camp, and attended many MDA events.

"MDA helped me gain the confidence I needed to move five hours away from my family and pursue my dream of becoming a medical researcher," she says.

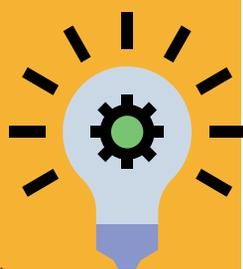
Learn how you can get involved with the MDA community at [mda.org/get-involved](https://mda.org/get-involved).

## Committed to a Cure

Big Apple Stores, owned by CN Brown Company, a CITGO marketer, have proudly partnered with MDA for 34 years, contributing to MDA's cause through Shamrocks, bowling tournaments, casual day campaigns, MDA golf benefits, and many other fundraising initiatives. Thank you to our partner for these accomplishments.

**\$311,864**  
raised in 2019

**\$2.3 million+**  
raised in the last 10 years



**BIG APPLE**



## 4 Ways to Fundraise Online

There are many ways to contribute to MDA, even in the age of social distancing. Here are a few ideas to get you started:

- 1 Hold a birthday fundraiser for MDA on Facebook. To create one, visit [facebook.com/MDAOrg](https://facebook.com/MDAOrg) and click on "Fundraisers."
- 2 Do good while getting solo time outside. Ask family and friends to pledge a dollar for every mile you walk, run, or roll in a month. Get started at [mda.org/yourway](https://mda.org/yourway).
- 3 Honor a loved one by collecting donations in their name for MDA. Create a fundraising page at [mda.org/yourway](https://mda.org/yourway).
- 4 Join MDA's Let's Play gaming platform, where gamers help transform lives. Learn more on page 21, or go to [mdaletsplay.org](https://mdaletsplay.org).



ADVERTISEMENT

THIS  
MOMENT  
COMES  
FROM WITHIN

ASHLEY // AGE 7  
LATER-ONSET SMA, TREATED WITH SPINRAZA



STEPHEN // AGE 38  
LATER-ONSET SMA, TREATED WITH SPINRAZA

Individual results may vary based on several factors, including severity of disease, initiation of treatment, and duration of therapy.

Victories are personal for the **10,000+**  
who have been treated with SPINRAZA worldwide.\*

For US individuals taking SPINRAZA:

40%

>40% of patients taking  
SPINRAZA are adults\*

3-80  
DAYS YEARS

Has treated SMA in patients  
3 days<sup>†</sup> to 80 years old<sup>‡§</sup>

90%

>90% of patients who started  
SPINRAZA remain on treatment<sup>‡</sup>

\*Based on commercial patients, early access patients, and clinical trial participants through December 2019.

<sup>†</sup>Includes clinical trial patients.

<sup>‡</sup>Based on commercial patients in the US (including Puerto Rico) through December 2019.

<sup>§</sup>Clinical studies of SPINRAZA did not include sufficient numbers of subjects aged 65 and over to determine whether they respond differently from younger patients.

#### INDICATION

SPINRAZA® (nusinersen) is a prescription medicine used to treat spinal muscular atrophy (SMA) in pediatric and adult patients.

#### IMPORTANT SAFETY INFORMATION

**Increased risk of bleeding complications** has been observed after administration of similar medicines. Your healthcare provider should perform blood tests before you start treatment with SPINRAZA and before each dose to monitor for signs of these risks. Seek medical attention if unexpected bleeding occurs.

**Increased risk of kidney damage, including potentially fatal acute inflammation of the kidney**, has been observed after administration of similar medicines. Your healthcare provider should perform urine testing before you start treatment with SPINRAZA and before each dose to monitor for signs of this risk.

**The most common side effects of SPINRAZA include** lower respiratory infection, fever, constipation, headache, vomiting, back pain, and post-lumbar puncture syndrome.

These are not all of the possible side effects of SPINRAZA. Call your healthcare provider for medical advice about side effects. You may report side effects to FDA at 1-800-FDA-1088.

**Before taking SPINRAZA**, tell your healthcare provider if you are pregnant or plan to become pregnant.

**Please see full Prescribing Information on SPINRAZA.com.**

This information is not intended to replace discussions with your healthcare provider.



**SPINRAZA**<sup>®</sup>  
(nusinersen) injection  
12 mg/5 mL

**CAMERON // AGE 4**

EARLY-ONSET SMA, TREATED WITH SPINRAZA

Individual results may vary based on several factors, including severity of disease, initiation of treatment, and duration of therapy.

[Learn more at SPINRAZA.com](https://www.spinraza.com)

**IMPORTANT FACTS ABOUT SPINRAZA<sup>®</sup> (nusinersen)****USES**

SPINRAZA is a prescription medicine used to treat spinal muscular atrophy (SMA) in pediatric and adult patients.

**WARNINGS**

**Increased risk of bleeding complications** has been observed after administration of similar medicines. Your healthcare provider should perform blood tests before you start treatment with SPINRAZA and before each dose to monitor for signs of these risks. Seek medical attention if unexpected bleeding occurs.

**Increased risk of kidney damage, including potentially fatal acute inflammation of the kidney**, has been observed after administration of similar medicines. Your healthcare provider should perform urine testing before you start treatment with SPINRAZA and before each dose to monitor for signs of this risk.

**COMMON SIDE EFFECTS**

- **The most common side effects of SPINRAZA include** lower respiratory infection, fever, constipation, headache, vomiting, back pain, and post-lumbar puncture syndrome (headache related to the intrathecal procedure).
- Serious side effects of complete or partial collapse of a lung or lobe of a lung have been reported.

**Talk to your healthcare provider about any side effect that bothers you or that does not go away.**

**OTHER INFORMATION**

SPINRAZA is a medication that should be administered as an injection into the lower back (a procedure called intrathecal injection) by, or under the direction of, an experienced healthcare professional.

**Before taking SPINRAZA**, tell your healthcare provider if you are pregnant or plan to become pregnant.

**QUESTIONS?**

The risk information provided here is not comprehensive. To learn more, talk about SPINRAZA with your healthcare provider or pharmacist. The FDA-approved product labeling can be found at [www.SPINRAZA.com](https://www.spinraza.com) or 1-844-4SPINRAZA (1-844-477-4672).

**MANUFACTURED FOR**

Biogen, Cambridge, MA 02142



# All in the Family

*Why we advocate for families like ours*

BY JAIME ZELAYA

Within our family of five, my daughter and I have a neuromuscular disease called scapulothoracic spinal muscular atrophy (SPSMA). I was born with this rare motor neuron disease, and our precious Leah inherited the same disease; however, this is not what makes us unique. What makes our family uncommon is our relentless dedication to accomplishing tasks collectively.

## The call

Even though I had wonderful parents and a supportive sister, I faced many challenges growing up with SPSMA. I experienced discrimination, bullying, and ignorance. These challenges felt like mountains of disabling fear and clouds of stunning dread that affected my thoughts and emotions from my early childhood. In times of doubt and pain, my family guided me to resources that fortified my emotional and physical development. Among those resources were MDA Care Centers, where I had surgery and physical therapy. Their medical care helped me understand

and accept my disability. Nonetheless, as a young adult, I drifted away from MDA.

In 2000, I met Bevsi, my wonderful wife. Bevsi accepted me for who I was while embracing my disease with unconditional love. She also encouraged me to seek a deeper understanding of my disease

through genetic counseling. After much hesitation, I completed genetic testing and learned the name of my disease for the first time. With this knowledge, I began to realize the

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I began to realize the importance of advocating for those, like me, who need representation.



The Zelaya family (clockwise from left): Bevsi (amazing mom), Jaime (advocate dad), Josiah (kind big brother), Nehemiah (curious younger brother), and Leah (compassionate middle sister)

importance of advocating for those, like me, who need representation. I was empowered and felt an overwhelming desire to help those who cannot help themselves. At the same time, I didn't know which direction to take to fulfill the calling.

## The mission

In 2007, our second child, Leah, was born. She was dazzling and wonderful, yet already faced with a host of challenges; she was born with SPSMA. From infancy and throughout her toddler stage, Leah faced difficult hurdles, such as multiple surgeries and arduous examinations. Her doctors informed

us her physical development would be affected by the severity of her disease. These assessments made us feel discouraged and afraid, yet they strengthened our family's faith and taught us to persevere and solidify our calling. In particular, as Leah grew older, she shared my desire to aid people with special needs in the neuromuscular disease community. Simply put, we esteemed others before our own needs.

To pursue our calling, we turned to the same resource that helped me develop as a person when I was young. MDA gave our family the opportunity to connect with other families facing similar hardships and obstacles. By engaging with an array of families, we were able to amplify our voice in advocating for their needs in our New York City communities.

### The fulfillment

With a few years of local advocacy under our belts, our family was ready and able to take the next step. In 2017 and 2019, MDA invited us to advocate for families like ours in our nation's capital. The five of us went to Washington, DC, to meet with the US senators and representatives from New York. We teamed up with other families and visited the offices of Chuck Schumer, Kirsten Gillibrand, Yvette Clarke, and more. During these meetings, we shared our experiences as people

with everyday physical challenges, and we asked senators to support legislation on newborn screening, healthcare reform, accessible air travel, and the Orphan Drug Act of 1983.

In Washington, DC, MDA saw the vital role our family played in advocating for families living with neuromuscular disease. We exhibited the power of a unified voice coupled with the love we have for each other. In addition, Leah's unique talent for public speaking has given her the opportunity to become New York State's MDA ambassador for 2020.

Advocacy has given Leah the confidence to flourish in other aspects of her life. She is part of a nonprofit organization called Dancing Dreams, where she learns to express herself through adaptive dance. She is also an adaptive skier, and she is making her acting debut in an upcoming feature film called "Marry Me," starring Jennifer Lopez and Owen Wilson.

Although we have been given many challenges to overcome, our family understands our calling, embraces the mission, and pursues the fulfillment to advocate for others and make the impossible possible by keeping it "all in the family." 

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Jaime Zelaya, 45, lives in Brooklyn, N.Y., with his family. With their help, he has been speaking about his journey to help others overcome bullying and discrimination.



## Support MDA and Quest Now

For more than 25 years, Quest Magazine has been telling the stories of MDA's community. With your donation to MDA, we can continue to educate and engage families and the physicians and researchers who help them with the latest news on neuromuscular disease research, health and wellness, mobility, travel, advocacy, and everyday thriving through Quest and our complementary educational channels.

Thank you for helping to keep our community informed at this most crucial time.

To make a tax-deductible donation to MDA, visit [www.mda.org/questsupport](http://www.mda.org/questsupport) or send mail:  
Attn: QUEST, 161 N. Clark St.,  
Suite 3550  
Chicago, IL 60601

On checks, please write "Quest-4500" on the memo line.

**Quest**

# Fashion Forward

*Taylor Torrez's designs check all the boxes*

When it comes to adaptive clothing, Taylor Torrez is setting out to prove that functional and fun aren't mutually exclusive.

Taylor, 22, lives with spinal muscular atrophy (SMA). For as long as she can remember, she's been frustrated by having to choose between comfortable, accessible clothing or clothing that looks good.

"I don't get the independence in clothing that I would like," says the North Carolina resident. "One day it just hit me: Why don't I design my own clothing line?"

Taylor, who recently graduated with a bachelor's degree in social work, started by conducting a short survey within a Facebook group of women with disabilities and her own friends in wheelchairs.

"I used those findings to see what similarities we had," she says. "It was pretty much all the same: Clothes aren't stretchy enough, clothes don't fit, and jeans are impossible to get on.

We all wished we had nicer clothing options, because clothing that is easy to get on isn't always the cutest."

Working with a pattern maker and seamstress in Asheville, N.C., Taylor's first prototype was a pair of leggings featuring extra stretch, reinforced horizontal loops on the sides and in the back — easy to slip fingers into and pull up — and

“One day it just hit me: Why don't I design my own clothing line?”



Taylor Torrez is wearing a jacket she designed with adaptive touches. Below left: A model shows Taylor's logo gear.



light elastic at the waist to keep them up. The waistband also offers higher coverage in back for those who are seated, like wheelchair users.

"It's just the slightest adjustments that make the biggest difference," she says.

Her next piece is a cheetah print moto jacket with thoughtful touches, like a silky inner lining so it's easy to slip on, and a magnetic zipper for one-handed zipping. She's funding her samples through sales of T-shirts, hats, and stickers that feature the logo of her fashion line, "The Reasons Why."

For now, Taylor is pursuing both her fashion line and her career in social work, hoping to work with children. "I'm a big believer that everything happens for a reason," she says. [Q](#)

## + LIFE STORY

Read stories from the MDA community at [strongly.mda.org](http://strongly.mda.org). If you'd like to share your story with Quest, email us at [quest@mdausa.org](mailto:quest@mdausa.org).

There are many questions about

**ALS**

You can help find answers.

## The National ALS Registry

is a program that allows people with ALS to fight back and help defeat the disease.

We are working towards a better future for people living with ALS by:

**Collecting** and analyzing data

**Striving** to better understand the disease

**Helping** researchers find possible risk factors

Your participation can make a difference.

Ask us about the Registry today.  
For more information,  
call (800) 232-4636 or visit [cdc.gov/als](https://cdc.gov/als).



U.S. Department of Health and Human Services  
Centers for Disease Control and Prevention  
Agency for Toxic Substances and Disease Registry





## The CHAMPION ALS Study

The CHAMPION ALS study is a clinical trial seeking to enroll people living with ALS. The purpose of the study is to assess the safety and efficacy of ravulizumab-cwvz compared to placebo in adults with ALS.

You may be eligible to participate if you have been diagnosed with ALS, are at least 18 years of age, and have had ALS symptoms for up to 3 years.

For more information about participating in the CHAMPION ALS study, visit [alschampion.com](http://alschampion.com) and talk to your doctor.

This information is intended as educational information for patients. It does not replace a doctor's judgment or clinical diagnosis. The medication being studied in the CHAMPION ALS study is not FDA approved for use in ALS.

[www.ALSCHAMPION.com](http://www.ALSCHAMPION.com)

**5**CHAMPION  
ALS-308