

EMPOWERING FAMILIES WITH INFORMATION AND INSPIRATION

Quest

MDA.ORG/QUEST ISSUE 4 • 2021

Mental Health Is for Everyone

Erasing stigma and finding support for mental health

FROM WHERE I SIT
MDA community members share their mental health journeys

DEMYSTIFYING DRUG PRICES
Why are new therapies so expensive?



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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

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✓ **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



Patient Information
EVRYSDI® (ev-RIZ-dee)
(risdiplam)
for oral solution

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.

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, 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.

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.

- 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.

Taking EVRYSDI

- 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.
 - 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:
 - 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.

Reusable Oral Syringes

- 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.

What are the possible side effects of EVRYSDI?

The most common side effects of EVRYSDI include:

- **For later-onset SMA:**
 - fever
 - diarrhea
 - rash
- **For infantile-onset SMA:**
 - fever
 - runny nose, sneezing, sore throat, and cough (upper respiratory infection)
 - diarrhea
 - lung infection
 - vomiting
 - rash

These are not all of the possible side effects of EVRYSDI. For more information, ask your healthcare provider or pharmacist.

Call your doctor for medical advice about side effects. You may report side effects to FDA at 1-800-FDA-1088.

How should I store EVRYSDI?

- 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).

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.

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1 DNA Way
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For more information, go to www.EVRYSDI.com or call 1-833-387-9734.

MDA is committed to transforming the lives of people affected by muscular dystrophy, ALS, and related neuromuscular diseases through innovations in science and innovations in care.

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A Good Time for Gaming

Mental health is at the forefront of the national conversation these days — you see it covered from various angles in all the major news outlets. But rarely does the mental health of those with disabilities get proper “airtime.” That’s why we decided to theme this issue of *Quest* on this deeply important topic.

As we all know, one key to mental health is the ability to develop and sustain meaningful connections. I’m proud to say we recently introduced an exciting new way for our community to connect and build camaraderie with others: MDA Let’s Play, our online gaming platform, where kids and adults can come together in a safe, inclusive virtual environment. Through Let’s Play, everyone can get in the game,

“talk smack” with their friends, and feel the thrill of slaying the dragon — the type of spirited fun that unites all gamers.

“MDA is a very important cause to me, and I am honored to have participated. I want to say thank you to Bennett, Puzzle, Dink, and all the others that run MDA Let’s Play. You do a fantastic job being inclusive, and that means the world to us.

— MDA Let’s Play participant

and join us soon for some video game mayhem that I can almost guarantee will put a smile on your face.

Sincerely,



Donald S. Wood, PhD

President and CEO

Muscular Dystrophy Association



Donald S. Wood, PhD

LET’S PLAY

Here’s how to get in on the gaming action.

Learn how to play with us: mda.org/lets-play
Join our team on Discord: discord.gg/MDALetsPlay
Watch the stream on Twitch: twitch.tv/mda_letsplay

Recently, we brought Let’s Play to a new level and recaptured our Labor Day prominence at the same time with the MDA Takes Vegas Stream-a-thon, an eight-hour event where our Let’s Play community welcomed celebrity gamers and even some professional athletes to join the fun. The event attracted 2.7 million unique viewers.

But perhaps the best way to communicate the impact of MDA Let’s Play is through the words of one of our gamers (see quote at left).

I hope you’ll check out mda.org/lets-play

My Promise to You

When I was diagnosed with spinal muscular atrophy (SMA) as an infant, no one would have guessed the very thing that was destined to make my life challenging would also fill my life with purpose.



MDA became a partner to me early on. As an MDA State Ambassador, I gave speeches to staff, volunteers, and corporate sponsors about how their support made a difference in my family's and my lives. I learned the power of sharing my story and saying, "Thank you." At MDA Summer Camp, I gained confidence and saw the possibility of independence and the power of community.

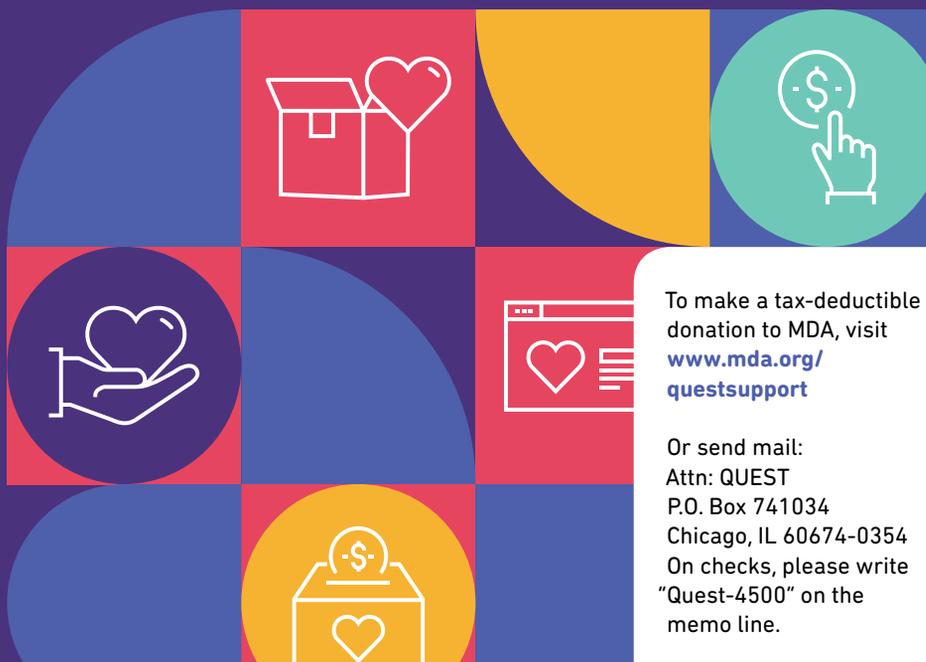
Being involved with MDA in my childhood taught me to use my voice to make a difference however I felt I could, which led me to become the motivational speaker and author I am today.

Now, in this full-circle moment, I am honored to serve MDA and all of you as editor-in-chief of the *Quest* family of content. My promise is to bring all that life and MDA have given me through the years and give that back to YOU in these pages, on the podcast, in the newsletter, and on the blog — because we have life to live and work to do. Let's get started!

Sincerely,
Mindy Henderson
 Director, *Quest* Editor-in-Chief
 Muscular Dystrophy
 Association

THANK YOU FOR SUPPORTING QUEST!

We're grateful to our generous donors who agree that it's important to empower and inform our community. For more than 25 years, *Quest* Magazine has been telling the stories of MDA's community. Donations to MDA help us continue to be a resource for people exploring independence, wellbeing, and research, whether they find us in print, online, or through our new podcast.



To make a tax-deductible donation to MDA, visit www.mda.org/questsupport

Or send mail:
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 P.O. Box 741034
 Chicago, IL 60674-0354
 On checks, please write "Quest-4500" on the memo line.

EXPLORE THE WHOLE FAMILY OF *QUEST* CONTENT AT MDA.ORG/QUEST.

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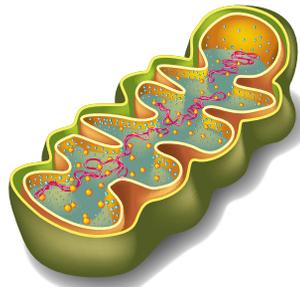


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Cover image: iStock.com/ Dumitru Ochievski

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MORE ONLINE

Supporting mental health is an important part of MDA's mission to transform the lives of people living with neuromuscular diseases. Visit MDA's new Mental Health Hub at mda.org/mental-health, and read the *Quest* online exclusive article on body positivity, "Embrace Your Body," at mda.org/quest.

WATCH THIS!
Check out this video featuring MDA's new theme song, "It's All About the Life We Live," and tell us what you think.





Let's Talk

The MDA Resource Center is available to provide one-on-one support. Here, Resource Center specialists answer *Quest* readers' questions.

How can I find a therapist who knows about mental health and living with neuromuscular disease?

—Jeffrey, New York

First, we applaud you for prioritizing your mental health. Because neuromuscular diseases are considered rare, it might be difficult to find a mental health professional who is familiar with a specific disease. We recommend looking for a therapist who has worked with people living with chronic health conditions. Consider asking if they are also willing to counsel your family members, as a neuromuscular disease can also affect loved ones in their own ways.

Contact the Resource Center to learn about resources for finding therapists and paying for mental health services.

Helpful resource: Visit MDA's Mental Health Hub at mda.org/mental-health.

When should I start talking to my grandson about his diagnosis and how his physical abilities could change?

—Katherine, Michigan

Every child is unique, and when they are ready to learn about their diagnosis varies. It's always best to meet your child at their individual level of understanding while being open and honest. Encourage your child to ask questions and come to you when they want to understand more.

When discussing their disease and its progression, focus on the positives of what they can do and how they can learn new ways of doing things if their abilities change. Encourage your child to meet others living with similar experiences, such as through MDA Summer Camp.

The social worker at your MDA Care Center can share more ways to talk with your child about their abilities.

+WE CAN HELP

Contact the Resource Center at [833-ASK-MDA1](tel:833-ASK-MDA1) or resourcecenter@mdausa.org.

Helpful resource: Find an MDA Care Center and learn about Care Center teams at mda.org/care/mda-care-centers.

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Continuing Our Commitment to Duchenne Muscular Dystrophy

Since 2014, BioMarin has explored various compounds and therapeutic approaches for Duchenne Muscular Dystrophy (DMD). We are excited to announce a new pre-clinical candidate, BMN 351. BMN 351 is a third-generation antisense oligonucleotide therapy targeted for exon 51 skipping. BMN 351 has not been approved for use or determined to be safe or effective. It is being investigated in non-human trials to determine if it can proceed to research trials in humans.

If results from the ongoing pre-clinical studies are supportive, BioMarin anticipates filing an investigational new drug application with regulatory authorities in the first half of 2022. This will allow research to begin with humans, at which time further information on the development program will be available.

BioMarin is a global pharmaceutical company with seven approved therapies and more than 20 years of experience in developing innovative medicines for rare genetic conditions.

For more information, visit our Patient Advocacy page at www.BioMarin.com

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PROGRESS NOW

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

New approvals

Octagam 10% Approved for Dermatomyositis

The US Food and Drug Administration (FDA) has approved Octagam 10% for the treatment of adult dermatomyositis.

The approval was based on the results of the ProDERM phase 3 clinical trial, which evaluated the long-term efficacy and safety of intravenous immunoglobulin (IVIg) for adults with dermatomyositis.

During the ProDERM trial, 95 participants were randomly assigned to receive either high-dose Octagam 10% or an inactive placebo every four weeks for 16 weeks. This was followed by a 24-week open-label extension phase, during which all participants received Octagam 10%. Response to treatment was measured using the 2016 American College of Rheumatology (ACR)/European League Against Rheumatism (EULAR) myositis response criteria.

During the initial 16-week study period, 78.7% of participants receiving Octagam 10% responded positively to treatment as compared to 43.8% of those receiving placebo. After switching to Octagam 10% in the extension period, the placebo group attained a similar response rate at week 40 as did the drug-treated participants at week 16. Common adverse effects experienced by participants included chills, difficulty breathing, fever, headache, increased blood pressure and heart rate, infusion site reactions, musculoskeletal pain, and nausea or vomiting.

Boxed warnings for Octagam 10% include risk for thrombosis (blood clots), kidney dysfunction, and acute kidney failure.

To learn more about Octagam 10%, visit octagamus.net.

Nexviazyme Approved for Pompe

The FDA has granted accelerated marketing approval to avalglucosidase alfa (Nexviazyme) for the treatment of individuals 1 year and older living with late-onset Pompe disease (LOPD). The second drug approved to treat Pompe disease, Nexviazyme is marketed in the United States by Sanofi Genzyme.

The FDA based its decision on encouraging results from the ongoing phase 3 COMET and phase 2 mini-COMET studies, which are both expected to conclude in 2024.

In the COMET trial, researchers are comparing avalglucosidase alfa to Lumizyme (alglucosidase alfa) in 100 previously untreated patients with LOPD, ages 3 years and older. Myozyme, also developed by Sanofi Genzyme, was approved by the FDA in 2006 for individuals with infantile-onset Pompe disease and was later marketed as Lumizyme for individuals with LOPD. The three-year mini-COMET study is evaluating the safety and effectiveness of avalglucosidase alfa in 22 children and adolescents with infantile-onset Pompe disease who previously failed to respond to Lumizyme treatment.

Data from the trials has shown that Nexviazyme, administered via intravenous (into a vein) infusion, has a safety and efficacy profile similar to Lumizyme, with improvements noted in respiratory muscle function, mobility, quality of life, and disease biomarkers.

Learn more at nexviazyme.com. For more information about the COMET and mini-COMET trials, visit clinicaltrials.gov and enter NCT02782741 or NCT03019406, respectively, in the “Other terms” search box.

Amyotrophic lateral sclerosis (ALS)

Tofersen Early Access Program

Individuals with ALS caused by mutation of the superoxide dismutase 1 gene (*SOD1*) who are experiencing rapid disease progression may now request to participate in part one of Biogen's early access program for the investigational therapy tofersen.

An antisense oligonucleotide (ASO) therapy, tofersen is administered as an intrathecal (into the spinal canal) injection and is designed to reduce the production of nonfunctional or altered SOD1 protein in cells. The drug is being evaluated for the potential treatment of SOD1-ALS in a phase 3 clinical study.

Data from a phase 1/2 study of tofersen showed that the drug was generally well-tolerated and that treatment with high-dose tofersen led to a decrease in SOD1 protein levels in participants' cerebrospinal fluid (the clear liquid that surrounds the brain and spinal cord). In addition, tofersen slowed functional decline of participants, as assessed by the ALS Functional Rating Scale Revised (ALSFRS-R), pulmonary function testing (slow vital capacity), and examination of muscle strength.

Physicians may submit requests for early access on behalf of eligible patients by emailing medicineaccess@clinigengroup.com. If results from the phase 3 study indicate that tofersen is safe and effective, and no further studies are required, the early access program will be opened more broadly to the SOD1-ALS population.

For more information about the tofersen early access program, visit biogen.com/en_us/access-programs.html.

Duchenne muscular dystrophy (DMD)

Pamrevlumab Phase 3 Trial Enrolling



Researchers at FibroGen are seeking ambulatory (able to walk) boys living with DMD to participate in a phase 3 clinical trial to evaluate efficacy of the investigational drug pamrevlumab (FG-3019) in combination with systemic corticosteroids to treat DMD.

Pamrevlumab is designed to decrease fibrosis (thickening or scarring) in the muscles, thereby protecting the heart and

lungs. This has the potential to improve muscle strength in people living with DMD.

Participants in the study will be randomly assigned to receive either pamrevlumab or an inactive placebo control,

administered by intravenous (in the vein) infusion, over the course of the study. Neither participants nor researchers will know who is receiving a placebo and who is receiving pamrevlumab. Participants will be involved for 60 weeks, with 29 visits scheduled every two weeks.

Participants will be evaluated for various outcome measures using tests such as the North Star Ambulatory Assessment (NSAA), four-stair climb velocity, 10-minute walk/run, time to stand, and time to loss of ambulation.

Individuals may not be eligible to participate if they are affected by another illness or are receiving another treatment that might interfere with the ability to undergo safe testing.

Travel support is available for study participants and families.

To learn more about inclusion/exclusion criteria, visit clinicaltrials.gov and enter NCT04632940 in the "Other terms" search box. To inquire about participation, contact Jessica Charpentier at 415-978-1346 or ielantos@fibrogen.com.

Investigational Drug Trial for DMD

Researchers at Astellas Pharma Inc. are seeking boys ages 8 to 16 living with DMD to participate in a phase 1b clinical trial and open-label extension to evaluate safety, tolerability, and preliminary efficacy of the investigational drug ASP0367 (MA-0211) to treat DMD.

ASP0367 is designed to increase the number and function of the mitochondria in cells, thereby increasing energy production. This could improve muscle health and function, reduce inflammation, and increase endurance in people living with DMD.



Ifetroban Trial Seeks Participants

This is a randomized, placebo-controlled study. Because it is double blind, neither participants nor researchers will know if participants are taking placebo or ASP0367. The drug or placebo will be administered as an oral tablet taken daily over the study period. A qualified home healthcare provider will evaluate participants for various outcome measures at approximately seven hospital visits and eight home visits, as well as through follow-up phone calls. The study will be followed by an open-label extension, during which all participants will be given ASP0367.

Individuals may not be eligible to participate if they are affected by another illness or receiving another treatment that might make testing less safe. Travel support is available for study participants and families.

To learn more about the study and inclusion/exclusion criteria, visit clinicaltrials.gov and enter NCT04184882 in the "Other terms" search box. To inquire about participation, contact Astellas Pharma Inc. at 800-888-7704 or astellas.registration@astellas.com.

Researchers at Cumberland Pharmaceuticals Inc. are seeking boys and men 7 years and older with DMD to participate in a phase 2 clinical trial of oral ifetroban to treat heart disease associated with DMD.

Ifetroban, administered as an oral capsule, is designed to reduce fibrosis (thickening and scarring) and fat deposits in the heart and may prevent heart disease and improve mortality in people living with DMD.

Participants will be randomly assigned to receive either low-dose or high-dose ifetroban or an inactive placebo during the study. Following the study, there will be an open-label extension period in which all eligible participants will have the opportunity to receive ifetroban. Participants will be involved for 12 months. Researchers will assess the effects of oral ifetroban on cardiac and respiratory function, muscle strength, daily activity, and quality of life.

Travel support is available for study participants and families.

To learn more about the trial, visit clinicaltrials.gov and enter NCT03340675 in the "Other terms" search box. To inquire about participation, visit fightdmdtrial.com or contact Ines Macias-Perez at 615-979-5778.

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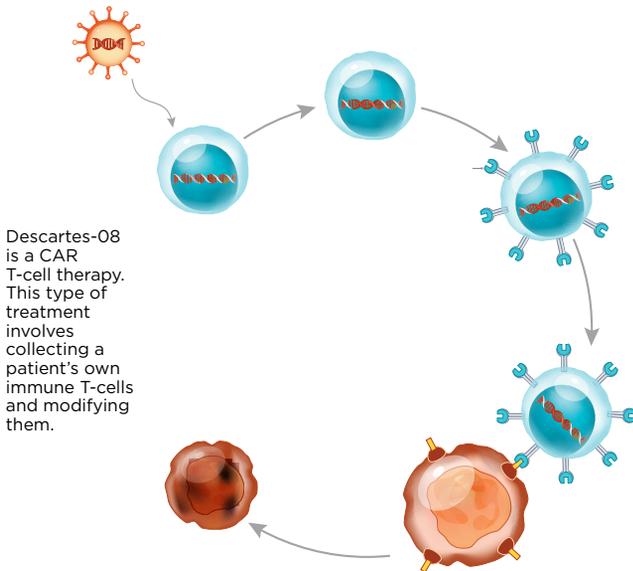
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Myasthenia gravis (MG)

Descartes-08 Study



Researchers at Cartesian Therapeutics are seeking adults living with generalized MG (gMG) to participate in a phase 1b/2a clinical trial to evaluate safety, tolerability, and manufacturing feasibility of the investigational drug Descartes-08 to treat gMG. Descartes-08 is designed to eliminate the abnormal plasma cells that cause gMG, potentially reducing the clinical symptoms of the disease.

All participants will receive up to six doses of Descartes-08, administered as an intravenous (in the vein) infusion, during the course of the open-label, dose-escalation study.

Participants will be evaluated for various outcome measures at approximately 10 doctor visits, using standard clinical assessment scales such as Quantitative Myasthenia Gravis (QMG), Myasthenia Gravis Quality of Life (MG QoL 15R), Myasthenia Gravis Activities of Daily Living (MG ADL), MG Composite, MGFA Class, and MG Post Intervention Status (MG PIS).

To be eligible for the study, individuals must be at least 18 years old, have a clinical diagnosis of gMG at the time of screening, be able to give written informed consent, and meet additional criteria.

Travel support may be available for study participants.

To learn more about the study and inclusion/exclusion criteria, visit clinicaltrials.gov and enter NCT04146051 in the "Other terms" search box. To inquire about participation, contact Adam Chowdhury at adam.chowdhury@cartesiantx.com.

Myotonic dystrophy (DM)

Trial for Excessive Sleepiness Treatment

Researchers at Harmony Biosciences are seeking adults living with DM, type 1 (DM1) to participate in a phase 2 clinical trial and open-label extension to evaluate safety and efficacy of the investigational drug pitolisant (Wakix) to treat excessive daytime sleepiness and other nonmuscular symptoms of DM1.

Wakix is designed to increase the amount of the chemical histamine in the brain, which is expected to promote wakefulness.

Study participants will be randomly assigned to receive either Wakix or an inactive placebo control, administered as an oral tablet, over the 17-week study. Following study completion, there will be an open-label



Treatment with pitolisant may decrease the excessive daytime sleepiness experienced by many people with DM1, among other improvements.

extension period in which all eligible participants will have the opportunity to receive Wakix.

To be eligible, individuals must be 18 to 65 years old at the time of enrollment, have a diagnosis of DM1 confirmed by genetic testing, and meet additional criteria.

Travel support is available for study participants and families.

To learn more about the study and inclusion/exclusion criteria, visit clinicaltrials.gov and enter NCT04886518 in the "Other terms" search box. To inquire about participation, email Harmony at clinicaltrials@harmonybiosciences.com, or call Ann Adey at 773-383-6258.

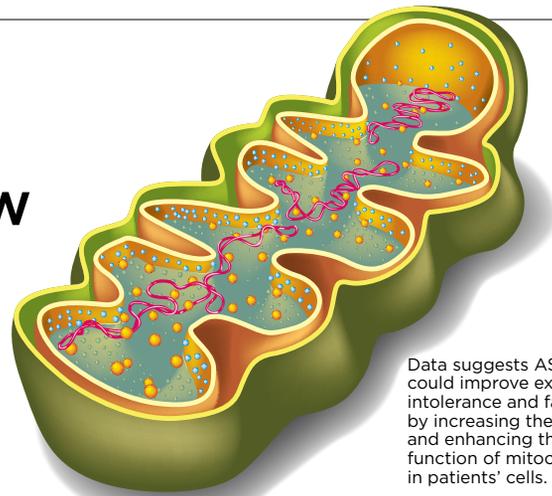
Primary mitochondrial myopathy (PMM)

Drug Trial Enrolling Now

Researchers at Astellas Pharma Inc. are seeking individuals 18 to 80 years old living with PMM to participate in a phase 2/3 clinical trial and open-label extension to evaluate safety, tolerability, and preliminary efficacy of the investigational drug ASP0367 (MA-0211) to treat PMM.

ASP0367 is designed to increase the number and function of cells' mitochondria (the "powerhouse" of the cells), thereby increasing energy production. This has the potential to improve muscle health and function, reduce inflammation, and increase endurance in people living with PMM.

Participants will receive the drug or placebo, administered as an oral tablet, taken daily during the study period. Participants will be evaluated for various outcome measures at approximately 12 hospital visits. Evaluations may include general health and functional exams, as well as muscle function tests with imaging markers. At-home tests using video recordings and a mobile app, remote interviews, and surveys may also be required.



Data suggests ASP0367 could improve exercise intolerance and fatigue by increasing the number and enhancing the function of mitochondria in patients' cells.

Individuals may not be eligible to participate if they are affected by another illness or receiving another treatment that might interfere with safe testing.

Travel support may be available for study participants and families.

To learn more about the trial and exclusion criteria, visit clinicaltrials.gov and enter NCT04641962 in the "Other terms" search box. To inquire about participation, contact Astellas Pharma Inc. at 800-888-7704 or astellas.registration@astellas.com and reference study 0367-CL-1201.

Images: iStock.com/lukaves; DrAfter123

Spinal muscular atrophy (SMA)

Share Your Decision-making Process



Researchers at Michigan Medicine at the University of Michigan are seeking caregivers of children living with SMA to participate in a survey about treatment decision-making.

Over the last five years, three treatments for SMA have been FDA-approved and are changing life expectancy and physical functioning for children with the disease. The goal of this survey is to increase understanding about how caregivers choose among SMA treatments for their children.

Participants must complete an online survey that will take approximately 10 minutes.

To be eligible, individuals must meet the following criteria:

- Be the parent or legal guardian of a child who is currently 0 to 18 years old with genetically proven SMA diagnosis
- Be over 18 years old

- Be able to read and respond to written questions in English, either independently or with assistance of a medical interpreter
- Reside in the United States

Individuals may not be eligible to participate if they are not involved in medical decision-making regarding the child's care.

If interested, contact the SMA Survey Research team at smasurvey@med.umich.edu.

The Latest on Mitochondrial DNA Depletion/Deletion Syndrome

A Q&A with Michio Hirano, MD



Michio Hirano, MD (fourth from right), and his research team gather with the Coiculescu family, whose sons received an experimental treatment for TK2 deficient myopathy through a compassionate use study.

Mitochondrial DNA (mtDNA) depletion/deletion syndrome (MDDS) is a group of genetic diseases in which the mtDNA inside cells cannot replicate correctly. Mitochondria are the powerhouses of cells, generating more than 90% of the energy in our cells. With fewer functioning mtDNA, cells fail to generate enough energy to function properly.

There are 30 different genes that can cause these conditions, which may appear anywhere from infancy through adulthood. Symptoms vary depending on the form of the disease but typically involve muscle weakness. Developmental delays and difficulty eating and breathing may also occur as the diseases advance. Until recently, there were no disease-modifying treatments for these conditions. Now, however, a drug therapy offers hope for at least one type of MDDS.

We talked with Michio Hirano, MD, who directs the H. Houston Merritt Center for Neuromuscular Diseases at Columbia University in New York City, about his lab's research into treatments for these diseases.

How do MDDS types differ?

The prognosis depends on which genes are involved. Even within a group of patients with a particular gene, there's variability depending on the severity of the mutation and other factors we don't fully understand. In general, the earlier the onset, the worse the prognosis.

WHAT IS COMPASSIONATE USE?

Also called expanded access, this is a potential pathway for a patient with an immediately life-threatening condition or serious disease to gain access to an investigational drug or medical device for treatment outside of clinical trials when no comparable or satisfactory options are available.

What therapies show promise in treating MDDS?

In the past, the only treatment for these conditions was dietary supplements (special vitamin mixtures called mito cocktails), exercise, and physical therapy. More recently, however, we've begun to identify potential therapies. One is for a form of MDDS that results from mutations in the thymidine kinase 2 gene (TK2), which causes TK2 deficient (TK2d) myopathy. It can begin any time between infancy and adulthood but usually begins before age 12. The prevalence of TK2d myopathy is unknown, but it's considered very rare.

We tested two naturally occurring molecules, deoxycytidine (dC) and deoxythymidine (dT), in a mouse model of TK2d and found that it delayed disease onset, prolonged lifespan, and restored mtDNA, as well as mitochondrial enzyme activities critical for generating cellular energy. Since then, we've given it to more than 40 children and adults in the United States and in other countries as part of a compassionate use study.

What we're seeing in patients is even more remarkable than what we saw in mice. Most show benefits — not only stabilization but also improvements in their condition. So this seems to be a disease-modifying drug.

The treatment is currently in phase 2 trials, with most of the patients who received it under compassionate use

now in that trial. A phase 3 study should begin in late 2021.

Are you also working on gene therapy?

We recently published a paper on gene therapy for TK2 deficiency, which is more effective in mice than the pharmacological (drug) therapy. What was even more striking is the combination of gene therapy plus pharmacological therapy was more effective than either alone.

We also think the pharmacological therapy may have relevance to other MDDS subtypes, and certainly the gene therapy will have relevance to an even larger number of these diseases.

What is your message for patients?

Number one is to get diagnosed accurately and to engage with physicians and get supportive care. And to have hope that we will be able to develop additional therapies for many more forms of MDDS and other neuromuscular diseases. I think this is a time for great optimism, and we look forward to additional breakthroughs for these diseases. [@](#)

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Read an expanded version of this interview and more articles on rare diseases at mda.org/quest/spotlight.



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TODAY IN DUCHENNE MUSCULAR DYSTROPHY

A CLOSE LOOK AT CORTICOSTEROIDS

BY: ARAVINDHAN VEERAPANDIYAN, M.D.

Aravindhan Veerapandiyam, MD is a Child Neurologist with specialized interest in neuromuscular disorders. He serves as Director to both the Comprehensive Neuromuscular Program and the PPMD Certified Duchenne Care Center at Arkansas Children's Hospital.

Considerations when selecting a therapy

For boys with Duchenne muscular dystrophy (DMD), corticosteroids work by reducing inflammation in the muscles; they can have an impact by improving strength and function. Over time, corticosteroids can help prolong ambulation by years, improve lung function, and potentially reduce the need for scoliosis surgery.

Although the benefits of corticosteroids are well established, it's important to work closely with your care team to decide which option is best. A child's unique functional and emotional needs may also play a role in this decision.

I have been taking care of boys with DMD for 6 years. I use both prednisone and EMFLAZA® (deflazacort) commonly in my practice for my boys with DMD. I choose a corticosteroid based on the individual needs of my patients. I've found that the available data on these medicines is representative of my own experiences.



There are two corticosteroids used to treat boys with DMD: prednisone and EMFLAZA. These medicines differ in clinical evidence, cost, availability, and side effect profile. It's important to learn more about each treatment option and tap into available resources for all the information.

To learn more about the functional and behavioral benefits of corticosteroids, speak to your healthcare professional.

Please see the Brief Summary of Information for EMFLAZA® (deflazacort) on the following page.

Indication and Important Safety Information

What is EMFLAZA® (deflazacort) used for?

Emflaza is a prescription medicine used to treat Duchenne muscular dystrophy (DMD) in patients 2 years of age and older.

When should I not take EMFLAZA?

Do not use if you have had hypersensitivity, including allergic reactions, to deflazacort or any of the inactive ingredients.

What warnings should I know about EMFLAZA?

- EMFLAZA can cause changes in endocrine function. Do not stop taking EMFLAZA, or change the amount you are taking, without first checking with your healthcare provider, as there may be a need for gradual dose reduction to decrease the risk of adrenal insufficiency and steroid “withdrawal syndrome”. Acute adrenal insufficiency can occur if corticosteroids are withdrawn abruptly, and can be fatal. A steroid “withdrawal syndrome,” seemingly unrelated to adrenocortical insufficiency, may also occur following abrupt discontinuance of corticosteroids. For patients already taking corticosteroids during times of stress, the dosage may need to be increased.
- There is an increased risk of infection when taking EMFLAZA. Tell the healthcare provider if the patient has had recent or ongoing infections or if they have recently received a vaccine. Medical advice should be sought immediately if the patient develops fever or other signs of infection. Patients and/or caregivers should be made aware that some infections can potentially be severe and fatal. Warn patients who are on corticosteroids to avoid exposure to chickenpox or measles and to alert their healthcare provider immediately if they are exposed.
- EMFLAZA can cause an increase in blood pressure and water retention. If this occurs, dietary salt restriction and potassium supplementation may be needed.
- There is an increased risk of developing a hole in the stomach or intestines in patients with certain stomach or intestine disorders when taking corticosteroids like EMFLAZA.
- EMFLAZA can cause severe behavioral and mood changes. Seek medical attention from the health care provider if any behavioral or mood changes develop.

- There is a risk of osteoporosis with prolonged use of EMFLAZA, which can lead to vertebral and long bone fractures.
- EMFLAZA may cause cataracts or glaucoma and a health care provider should monitor for these conditions if corticosteroid therapy is continued for more than 6 weeks.
- Immunizations should be up-to-date according to immunization guidelines prior to starting therapy with EMFLAZA. Live-attenuated or live vaccines should be administered at least 4 to 6 weeks prior to starting EMFLAZA. Live-attenuated or live vaccines should not be used in patients taking EMFLAZA.
- EMFLAZA can cause serious skin rashes. Seek medical attention at the first sign of a rash.
- Rare instances of anaphylaxis have occurred in patients receiving corticosteroid therapy, including EMFLAZA.

What should I tell my health care provider?

Tell the health care provider about all medical conditions, including if the patient:

- is pregnant or planning to become pregnant. EMFLAZA® (deflazacort) can harm your unborn baby.
- is breastfeeding or planning to breastfeed. EMFLAZA may appear in breastmilk and could affect a nursing child.

Certain medications can cause an interaction with EMFLAZA. Tell your healthcare provider of all the medicines you are taking, including over-the-counter medicines (such as insulin, aspirin or other NSAIDs), dietary supplements, and herbal products. Alternate treatment, dosage adjustment, and/or special test(s) may be needed during the treatment.

What are the side effects of EMFLAZA?

The most common side effects of EMFLAZA include facial puffiness or Cushingoid appearance, weight increased, increased appetite, upper respiratory tract infection, cough, frequent daytime urination, unwanted hair growth, central obesity, and colds. These are not all of the possible side effects of EMFLAZA. Call your doctor for medical advice about side effects.

To report an adverse event, please call 1-866-562-4620 or email at usmedinfo@ptcbio.com. You may also report side effects to FDA at 1-800-FDA-1088 or at www.fda.gov/medwatch.

Ment Health Is for Everyone

How are you? We're asked this question frequently, and we often respond, "Fine." But the truth is, many times we're not fine. Life is complicated. And for people living with a neuromuscular disease or caring for a loved one who is, there are a multitude of challenges to navigate.

Having a lot to deal with, feeling alone, and coping with medical trauma and loss are among the many life experiences that can affect mental health. It's important to know that no matter what you're feeling, you don't have to go through it alone.

It's OK not to feel OK

Lauren Presutti, MEd, EdS, LLMSW, is a psychotherapist and disability advocate who lives with congenital muscular dystrophy (CMD). Being a member of the neuromuscular disease community and using a power wheelchair since she was 5 gives her a firsthand perspective on how disability affects mental health.

"Many people with disabilities struggle with self-acceptance, confidence, relationships, self-advocacy, goal setting, body image, and empowerment," Lauren explains. "Largely because many non-disabled people in the world don't understand neuromuscular diseases, those who have one often feel like it's harder to fit in, life seems unfair and beyond their control, or their circumstances are defining who they are."

In addition, the progressive nature of many neuromuscular diseases means physical abilities change over time. An action as simple as raising a hairbrush to your head can be possible one day and not the next. Throughout life, grieving the loss of abilities is a common theme for people with neuromuscular disease. Adjusting to those losses creates a slew of feelings.

"Integrating a neuromuscular disease into your self-concept can be emotionally triggering, and it's often hard to navigate without the right support," Lauren says.

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*Erasing stigma and finding
support for mental health*

BY BARBARA TWARDOWSKI
AND JIM TWARDOWSKI, RN

Image: iStock.com/Ponno marifove_Maria

Let's talk about it

According to the National Association of Mental Illness (NAMI), 1 in 5 U.S. adults experiences a mental illness. That's more than 51 million people living with an anxiety disorder, depression, obsessive compulsive disorder, or other illness. Yet most people are reluctant to talk about their mental health struggles because they feel shame or fear judgment. This stigma prevents many people from seeking help.

People with mental health concerns and disabilities are fighting multiple stigmas at once.

"Disability stigmas and stereotypes are so powerful that most people and families affected by neuromuscular diseases tend to minimize the impact the disease is having on their mental health," Lauren says. "Our society is consumed with the idea that people with disabilities should be strong and inspirational all the time. That's a problem for our community because living with a neuromuscular disease is hard at times. We have to be able to safely acknowledge that and allow people to express their emotional pain without fear of receiving pity or being shamed in response."

It's becoming more common for disability activists to look beyond accessibility and talk about how issues like ableism, inspiration porn, and body positivity affect people in the disability community. (To learn more about body positivity, read



Lauren Presutti, MEd, EdS, LLMSW

"Embrace Your Body" at mda.org/quest.) In addition, more people are willing to speak out about their own mental health journeys on social media and other platforms. (Turn to page 32 to read three stories from our own community.)

Groups like NAMI, with their Stigma-Free campaign (nami.org/get-involved/pledge-to-be-stigmfree), are encouraging everybody in society to talk openly about mental health. "Mental health is just as important as physical health," is a mantra of the destigmatization movement.

It's not weak to ask for help

Whether or not you're ready to open up to the world, one of the best things you can do for your mental health is to talk with a professional. Both individuals and families affected by neuromuscular disease can benefit from having a safe space to express themselves without any judgment. Therapy can help you get through tough times.

"When to see a therapist is different for everyone. Seek a therapist as soon as you have an issue, or when you are ready," says Rhoda Olkin, PhD, distinguished professor of clinical psychology at the California School of Professional Psychology. The Centers for Disease Control and Prevention (CDC) offers this guideline: Contact your physician when

YOU ARE NOT ALONE

You don't have to navigate your neuromuscular disease journey by yourself. Use these resources to find mental health support.

American Psychological Association: Search for a psychologist by location or practice area. locator.apa.org

Child and Adolescent Psychiatrist Finder: The American Academy of Child and Adolescent Psychiatry offers an online tool to find psychiatric care for children. aacap.org

MDA Mental Health Hub: MDA has collected a number of resources to support your mental health needs. mda.org/mental-health

MDA Resource Center: Resource Center specialists can provide information on finding therapists and resources for paying for mental health services. 833-ASK-MDA1 or resourcecenter@mdausa.org

NAMI HelpLine: Volunteers answer questions and offer support Monday through Friday, 10 a.m.-10 p.m. ET. [800-950-NAMI \(6264\)](https://800-950-NAMI) or info@nami.org

National Suicide Prevention Lifeline: The Lifeline provides 24/7, free, and confidential support for people in distress. 800-273-8255 or suicidepreventionlifeline.org

Psychology Today: A database of therapists allows users to filter by ZIP code, specialty area, insurance type, and more. psychologytoday.com

Substance Abuse and Mental Health Services Administration (SAMSHA) National Helpline: This service provides referrals to local treatment facilities, support groups, and community-based organizations. [800-662-HELP \(4357\)](https://800-662-HELP)



your mental health adversely impacts daily living for at least 14 days in a 30-day period.

It's worth noting that there are differences between the mental health effects of early-onset and late-onset neuromuscular diseases. People diagnosed as young children tend to grow up feeling like it's hard to fit in. People diagnosed later generally go through a stressful period between the time when they first notice symptoms and get diagnosed. "Dealing with the psychosocial issues comes after they've figured out the medical concerns," Dr. Olkin says.



Rhoda Olkin, PhD

Someone who “gets” you

When you want to find a therapist, ask friends, family members, and your MDA Care Center team for recommendations. Consider whether you'd like to meet a therapist in person or via telemedicine, which allows you to have virtual sessions from the comfort of your home.

The “chemistry” between a therapist and client is crucial. Both parties need to feel comfortable. Before booking an appointment, ask if the therapist has any experience working with clients with disabilities or chronic conditions. While they might not be familiar with your neuromuscular disease, this experience might give them some understanding of your situation.

Additional questions you might want to ask a prospective therapist include:

- How long have you been a therapist?
- What are your areas of expertise?
- Do you have experience working with others who have issues like mine?
- What is the typical length of time a client is in therapy for my issue?
- Do you offer a free first visit?

Discuss financial concerns up front, such as: do they accept your insurance, do they take credit cards, and what is their policy regarding missed appointments.

What to expect in therapy

What is therapy like? “It should be a safe, genuinely supportive environment where you can focus entirely on the reasons you are seeking help,” Lauren says.

The first few sessions may feel like a game of 20 questions. Don't rush the process. Your therapist needs to collect a family history and understand what brought you to therapy. It's OK if you aren't ready to talk about something just yet. If the topic is too painful, the therapist should not push you to share. Trust develops over time.

Be prepared to be proactive about educating your therapist about your neuromuscular disease and how it impacts

your daily life. They might not be familiar with challenges such as caregiver problems, accessibility barriers, and transportation issues. And non-disabled therapists may not feel comfortable asking direct questions about your condition. Dr. Olkin shares that, in a recent survey of people with physical and visual disabilities, most therapists they saw did not feel free to say, “Tell me about your disability;” or “How much does your disability affect this current problem?”

Explaining your neuromuscular disease might be worth the effort. According to Lauren, for people with disabilities who struggle with interpersonal relationships, intimately connecting with a non-disabled therapist may be therapeutic in itself and a skill they will use in relationships outside of therapy sessions.

How long you go to therapy depends on the problem and the therapist's approach. Couples therapy might be 16 weeks, therapy for depression four to six months, and counseling to work through a specific life problem might be a few sessions. Every situation is unique, and there is great variability across therapists and presenting problems.

According to Dr. Olkin, you know therapy is working when you notice problems you started with begin to ease. That might mean feeling depression recede, anxiety becoming more manageable, or panic attacks stopping.

To get to that point, it's important to feel that your therapist treats you with empathy and has your best interests at heart. Keep in mind, you are under no obligation to stick with the first therapist you see. You might need to shop around to find a good fit.

“The relationship between therapist and patient is the most important factor in predicting positive outcomes,” Lauren says. “Check in with yourself throughout the process and truly assess whether you feel your time with the therapist is benefiting you.”

Ultimately, therapy isn't just about making you feel better in the moment, but about helping you understand how to cope with negative thoughts and unmanageable emotions when they occur. Let's face it: Life will still be complicated. But when you know you're not alone, that your mental health is important, and that you can talk about it — whether that's with friends and family or a professional — it becomes easier to face life's challenges. [Q](#)

Barbara Twardowski lives with Charcot-Marie-Tooth disease (CMT) and uses a power wheelchair. Jim, her husband, is a registered nurse. The couple writes about accessible travel, health, and lifestyle.

Why are new therapies for neuromuscular diseases so expensive?

BY LARRY LUXNER

DEMYSTI

Back in December 2016, when the US Food and Drug Administration (FDA) approved Spinraza — an intravenous therapy developed by Biogen to treat all types of spinal muscular atrophy (SMA) — parents, patient advocates, and even industry observers were shocked at its price: \$750,000 for the first year, then \$375,000 every year after, for the rest of a patient's life.

But then along came Zolgensma — a one-time-only gene therapy that targets the genetic root cause of SMA. AveXis, a subsidiary of Novartis at the time, announced that a single dose of its intravenous fluid would cost \$2,125,000, quickly earning Zolgensma the title of “world's most expensive drug.”

Whether that headline-grabbing label is fair is contentious because it's difficult to directly compare the cost of a one-off gene therapy to that of medications that must be taken regularly over a long period of time. But the debate over exorbitant drug prices is one that's being repeated throughout the rare disease community as more treatments are being developed for neuromuscular diseases ranging from SMA and Duchenne muscular dystrophy (DMD) to Gaucher disease and amyotrophic lateral sclerosis (ALS).

Behind the numbers

“With rare disorders, it takes between \$100 million and \$300 million to bring a new drug to market, and then another

\$100 million to launch it,” says Jeremy Levin, DPhil, MB BChl, who until this past June chaired the board of directors of the Biotechnology Innovation Organization (BIO), the world's largest biotech industry advocacy group.

Dr. Levin, former president and CEO of Israel's Teva Pharmaceuticals Ltd. — the world's top generic drugmaker — now heads New York-based Ovid Therapeutics, which is pursuing cures for a variety of rare epilepsies.

“Whenever you price something, you must take into account the hundreds of millions of dollars these tiny biotech companies spend in the hope they'll be successful,” he says. “Ovid spent in excess of \$100 million, and unfortunately our product, OV101 (gaboxadol) failed in Phase 3 trials for Angelman syndrome. However, we had the good fortune to demonstrate to the industry that you could cure this genetic condition. As a result, now 14 or 15 other companies are trying to find a cure, and all are investing comparable amounts of money.”

In Spinraza's case, the benefit was evident early on, says Doug Kerr, MD, PhD, who led Biogen's efforts on that project and is now chief medical officer at Generation Bio in Cambridge, Massachusetts.

“Spinraza had been in development since 2008. We ultimately moved it into testing patients in 2012, and it won FDA approval only four years later,” he says. “The reason it



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was so quickly approved was because there was such a clear benefit. The babies with SMA we treated would otherwise have progressed to ventilator dependence and death, but they didn't. We eventually developed a newborn screening test, and whenever a child was ultimately diagnosed with SMA, we offered this treatment. And if you treated these babies early enough — essentially at 3 weeks of life — most of them never got any disease symptoms at all.”

In the nearly five years since its FDA approval, more than 11,000 people worldwide have received Spinraza.

Getting to market

Success stories like Spinraza's come infrequently in the industry. Sharon Hesterlee, PhD, MDA's chief research officer, says about 50% of the costs of bringing a new drug to

market are “sunk costs” due to failure.

“Neuromuscular diseases are all rare, meaning market sizes are small, and sometimes just to make the numbers work from a commercial standpoint, they increase prices,” she says. “Another reason is that some of these technologies are relatively new and a little exotic. Gene therapies are expensive to develop and produce.”

Dr. Hesterlee says it's not clear if drug companies are adding a big profit margin when they price new therapies. “They're not very transparent,” she says. “But we do know they're compensating for the number of failures in other programs. What they're counting on is to get a couple of winners that will pay for the sunk costs that went into all the drugs that didn't make it.”

One particularly contentious drug is deflazacort (Emflaza), a steroid originally developed by Marathon and now marketed by PTC Therapeutics to treat boys with DMD. For years, US families had been paying \$1,200 or less annually out-of-pocket to import Emflaza from Europe. When the FDA approved the drug in 2017, its cost suddenly jumped to around \$70,000 a year.

“The company was able to pull together a lot of old data, combine it with new studies, and get it approved as a treatment specifically for DMD in the United States,” Dr. Hesterlee explains. “Now that it was a prescription drug for DMD, they could charge more for it, so they did.”

Support for orphan drugs

Most companies wouldn't invest in rare disease research in the first place if not for the Orphan Drug Act of 1983. The law's Orphan Drug Tax Credit (ODTC) allows sponsors with an orphan designation to collect tax credits for expenses



Sharon Hesterlee, PhD

RESOURCES FOR MEDICAL TREATMENTS AND THERAPIES

The national organizations below provide financial assistance for needs including medical treatments and therapies. Contact MDA's Resource Center ([833-ASK-MDA1](tel:833-ASK-MDA1) or resourcecenter@mdausa.org) for more information and help finding resources for specific medications.

CHIVE Charities
chivecharities.org/recipient

HealthWell Foundation
healthwellfoundation.org

Modest Needs
modestneeds.org/for-applicants

National Organization for Rare Disorders (NORD) RareCare
rarediseases.org/for-patients-and-families/help-access-medications/patient-assistance-programs-2

NeedyMeds
needymeds.com
Helpline:
[800-503-6897](tel:800-503-6897)

Patient Access Network (PAN) Foundation
panfoundation.org

Patient Advocate Foundation Co-Pay Relief
patientadvocate.org/connect-with-services/copay-relief

PhRMA's Medicine Assistance Tool
medicineassistance.org

The Assistance Fund
tafcares.org



Doug Kerr, MD, PhD

Photo: Larry Luxner

Image: iStock.com/ilyaliren

incurred conducting clinical trials of potential therapies for the indicated rare, or orphan, disease.

This tax credit lowers the cost of drug development. According to the National Organization for Rare Disorders (NORD), 33% fewer rare-disease drugs would be developed without the ODTC.

Before the Orphan Drug Act came into existence, the industry saw an average of one new rare-disease drug approval per year. But between 1983 and 2016, the FDA approved 451 orphan drugs for 590 rare disease indications. Yet the act is a perennial target by lawmakers in Congress who see it as a tax loophole for pharmaceutical companies.

In fact, orphan drugs account for only 11% of what Americans spend annually on prescription drugs.

A current provision of the Build Back Better Act, which was being considered by the US Congress at press time, seeks to remove the tax credit for all but the first approved orphan use of a new drug.

Dr. Hesterlee contends it would be a disaster for rare diseases if the tax credit is cut. “It motivates companies to develop drugs for orphan diseases, and in some cases, it’s not enough,” she says. “The vast majority of diseases are still too rare for companies to touch, even with the tax credit.”

Dr. Levin agreed that if this provision becomes law, it will be “very difficult, if not devastating” for the multitudes of innovative biotech startups that develop therapies for rare diseases.

“The vast majority of tax relief does not go to big pharmaceutical companies, but to small biotech companies,” Dr. Levin says. “Without the tax credit, it becomes incredibly onerous for a young company to raise the hundreds of millions of dollars required to market a drug.”

Dr. Levin adds that, rather than eliminate the ODTC, lawmakers should focus on how to reduce out-of-pocket expenses for people with rare diseases. “They need to ensure that insurance companies don’t have patients paying additional money for medicines that could cure them,” he says.

Fortunately, most health insurance companies now offer coverage of even expensive drugs like Zolgensma. “There are lots of hoops to jump through, but insurers’ familiarity with the drug helps,” Dr. Hesterlee says. “Drugmakers are getting better at laying the groundwork, so insurance companies aren’t getting caught flat-footed.”

Searching for a better model

The cost of new gene therapies will remain high until advances in production bring down prices significantly, according to Generation Bio’s Dr. Kerr.

For example, adeno-associated virus (AAV) vectors are the leading platform for delivering gene therapy into the body. “Manufacturing

AAV is very complicated and very expensive,” Dr. Kerr explains. That expense will add to the cost of gene therapies involving AAV until scientists develop more efficient manufacturing methods.

Dr. Kerr also points out that as long as gene therapies like Zolgensma cost \$2 million per patient, it won’t be possible to provide the drug to all who truly need it. “You’d relegate gene therapy to ultra-rare disorders, and we’d see rare examples of dramatic cures, but they would be unavailable to the world at large,” he says. “That’s not the world we want to see with gene therapy. Ultimately, manufacturing costs will go down, and these drugs will potentially be available to a much broader segment of the population. That is our model.”

In the meantime, there are programs to help people obtain medications and new therapies that would otherwise be out of their reach. (See Resources for Medical Treatments and Therapies on page 22.)

Some programs are sponsored by pharmaceutical companies. “There are lots of ways companies can help patients pay,” Dr. Hesterlee says. “They have support programs where they will help people defray the cost of medications. They also have whole armies of people who will help them navigate the insurance process.”

Ultimately, the industry’s goal — as well as MDA’s — is to get lifesaving and disease-modifying therapies to people who need them. “Rare-disease therapies must be accessible, whether that’s through reduced pricing, insurance coverage, or some combination that works,” Dr. Hesterlee says. [Q](#)



Jeremy Levin, DPhil, MB BChIR

Photo: Larry Luxner

Larry Luxner is a freelance journalist and photographer based in Israel. He writes frequently about rare diseases.



Did You Know?

The hospital where you receive treatment might have a program to pay for all or part of treatment, such as a Hill-Burton program. Hill-Burton is a federal law requiring any medical institution that used federal funds to build, expand, or renovate its facilities to provide a percentage of its services free or at a reduced cost. Find a list of Hill-Burton facilities around the country at hrsa.gov/get-health-care/affordable/hill-burton/facilities.html. Ask the financial coordinator at your MDA Care Center (mda.org/care/mda-care-centers) about any financial assistance programs available at that center.

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*These findings were not considered statistically significant. This means that because the two groups studied were not large enough, the results could have occurred by chance.

Not actual patients.

In a study to observe long-term effects of steroids, deflazacort was shown to delay disease progression by years



Loss of ability to stand from a lying-on-the-back position
—delayed by 2.1 years when compared with prednisone



Loss of ambulation (ability to walk)
—delayed by 2.7 years when compared with prednisone



Loss of hand-to-mouth function with retained hand function
—delayed by 2.7 years when compared with prednisone

STUDY INFORMATION

This study examined the long-term effects of glucocorticoids on milestone-related disease progression across the lifespan and survival in patients with Duchenne muscular dystrophy. Comparisons between deflazacort and prednisone are not included in the approved Prescribing information for deflazacort, as prednisone is not an approved treatment for Duchenne muscular dystrophy.

Please see the full outline of study information after summary of information for Emflaza.


Emflaza[®]
(deflazacort)

6 mg | 18 mg | 30 mg | 36 mg tablets
22.75 mg/mL oral suspension

Indication and Important Safety Information

What is EMFLAZA® (deflazacort) used for?

Emflaza is a prescription medicine used to treat Duchenne muscular dystrophy (DMD) in patients 2 years of age and older.

When should I not take EMFLAZA?

Do not use if you have had hypersensitivity, including allergic reactions, to deflazacort or any of the inactive ingredients.

What warnings should I know about EMFLAZA?

- EMFLAZA can cause changes in endocrine function. Do not stop taking EMFLAZA, or change the amount you are taking, without first checking with your healthcare provider, as there may be a need for gradual dose reduction to decrease the risk of adrenal insufficiency and steroid "withdrawal syndrome". Acute adrenal insufficiency can occur if corticosteroids are withdrawn abruptly, and can be fatal. A steroid "withdrawal syndrome," seemingly unrelated to adrenocortical insufficiency, may also occur following abrupt discontinuance of corticosteroids. For patients already taking corticosteroids during times of stress, the dosage may need to be increased.
- There is an increased risk of infection when taking EMFLAZA. Tell the healthcare provider if the patient has had recent or ongoing infections or if they have recently received a vaccine. Medical advice should be sought immediately if the patient develops fever or other signs of infection. Patients and/or caregivers should be made aware that some infections can potentially be severe and fatal. Warn patients who are on corticosteroids to avoid exposure to chickenpox or measles and to alert their healthcare provider immediately if they are exposed.
- EMFLAZA can cause an increase in blood pressure and water retention. If this occurs, dietary salt restriction and potassium supplementation may be needed.
- There is an increased risk of developing a hole in the stomach or intestines in patients with certain stomach or intestine disorders when taking corticosteroids like EMFLAZA.
- EMFLAZA can cause severe behavioral and mood changes. Seek medical attention from the health care provider if any behavioral or mood changes develop.
- There is a risk of osteoporosis with prolonged use of EMFLAZA, which can lead to vertebral and long bone fractures.

- EMFLAZA may cause cataracts or glaucoma and a health care provider should monitor for these conditions if corticosteroid therapy is continued for more than 6 weeks.
- Immunizations should be up-to-date according to immunization guidelines prior to starting therapy with EMFLAZA. Live-attenuated or live vaccines should be administered at least 4 to 6 weeks prior to starting EMFLAZA. Live-attenuated or live vaccines should not be used in patients taking EMFLAZA.
- EMFLAZA can cause serious skin rashes. Seek medical attention at the first sign of a rash.
- Rare instances of anaphylaxis have occurred in patients receiving corticosteroid therapy, including EMFLAZA.

What should I tell my health care provider?

Tell the health care provider about all medical conditions, including if the patient:

- is pregnant or planning to become pregnant. EMFLAZA® (deflazacort) can harm your unborn baby.
- is breastfeeding or planning to breastfeed. EMFLAZA may appear in breastmilk and could affect a nursing child.

Certain medications can cause an interaction with EMFLAZA. Tell your healthcare provider of all the medicines you are taking, including over-the-counter medicines (such as insulin, aspirin or other NSAIDs), dietary supplements, and herbal products. Alternate treatment, dosage adjustment, and/or special test(s) may be needed during the treatment.

What are the side effects of EMFLAZA?

The most common side effects of EMFLAZA include facial puffiness or Cushingoid appearance, weight increased, increased appetite, upper respiratory tract infection, cough, frequent daytime urination, unwanted hair growth, central obesity, and colds. These are not all of the possible side effects of EMFLAZA. Call your doctor for medical advice about side effects.

To report an adverse event, please call 1-866-562-4620 or email at usmedinfo@ptcbio.com. You may also report side effects to FDA at 1-800-FDA-1088 or at www.fda.gov/medwatch.

The following study support information is not included in the approved label for EMFLAZA® (deflazacort). Please talk to your healthcare professional if you have any questions.

MCDONALD 2018 STUDY SUMMARY (ABSTRACT)

Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study.

Background: Glucocorticoid treatment is recommended as a standard of care in Duchenne muscular dystrophy; however, few studies have assessed the long-term benefits of this treatment. This study examined the long-term effects of glucocorticoids on milestone-related disease progression across the lifespan and survival in patients with Duchenne muscular dystrophy.

Methods: For this prospective cohort study, male patients aged 2-28 years with Duchenne muscular dystrophy were enrolled at 20 centers in nine countries. Patients were followed up for 10 years. Comparisons were no glucocorticoid treatment or cumulative treatment duration of less than 1 month versus treatment of 1 year or longer with regard to progression of nine disease-related and clinically meaningful mobility and upper limb milestones. Kaplan-Meier analyses to compare glucocorticoid treatment groups for time to stand from supine of 5 s or longer and 10 s or longer, and loss of stand from supine, four-stair climb, ambulation, full overhead reach, hand-to-mouth function, and hand function. Risk of death was also assessed. This study is registered with **ClinicalTrials.gov**, number NCT00468832.

Findings: 440 patients were enrolled during two recruitment periods (2006-09 and 2012-16). Time to all disease progression milestone events was significantly longer in patients treated with glucocorticoids for 1 year or longer than in patients treated for less than 1 month or never treated (log-rank). Glucocorticoid treatment for 1 year or longer was associated with increased median age at loss of mobility milestones by 2.1-4.4 years and upper limb milestones by 2.8-8.0 years compared with treatment for less than 1 month. Deflazacort was associated with increased median age at loss of three milestones by 2.1-2.7 years in comparison with prednisone or prednisolone (log-rank). 45 patients died during the 10-year follow-up. 39 (87%) of these deaths were attributable to Duchenne-related causes in patients with known duration of glucocorticoids usage. 28 (9%) deaths occurred in 311 patients treated with glucocorticoids for 1 year or longer compared with 11 (19%) deaths in 58 patients with no history of glucocorticoid use (odds ratio 0.47, 95% CI 0.22-1.00).

Interpretation: In patients with Duchenne muscular dystrophy, glucocorticoid treatment is associated with reduced risk of losing clinically meaningful mobility and upper limb disease progression milestones across the lifespan as well as reduced risk of death.

McDonald CM, Henricson EK, Abresch RT, et al; CINRG Investigators. Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study. *Lancet*. 2018;391(10119):451-461.

A Special Summer

Camper Spotlight: Kendal Blankenship

Age: 12

Condition: Charcot-Marie-Tooth disease (CMT)

In-person or virtual: Virtual this year; in person two years ago

Favorite part of camp: “I really liked all the craft activities that came in our camp bundles. My mom helped me, and it was so much fun! Building Lego cars and towers was one of my favorite activities, outside of chatting with other campers.”

Advice for future campers:

“Try it! Don’t let your worries keep you from camp. Camp is so great, and the people make it extra special! My first year, I got the Rock Wall Climbing Award, and that was a lot of fun. You will make amazing friends and have memories to last for a lifetime!”

This summer, nearly 800 campers joined MDA’s Virtual Camp. Additions to last year’s successful model included daily Cabin Chat video calls, allowing campers to interact with each other, as well as Camp Supply and STEM Connections kits. These kits filled with project necessities and swag were mailed to campers before Virtual Camp started. All of these activities kept the campers busy. Here are a few fun facts:



Kendal Blankenship

- Campers around the nation engaged in 50 hours of video calls over six weeks.
- Campers chose among 26 unique activities.
- 90 volunteer “camp counselor” facilitators helped campers.
- 100% of campers said the Camp Supply kits were a great addition to Virtual Camp.

Start planning next summer’s camp adventure at mda.org/summer-camp.

Gaming for Good

MDA supporters and gamers united on Labor Day weekend for MDA’s inaugural Takeover Stream-a-thon, an eight-hour event filled with gaming, competitions, giveaways, and celebrity and guest appearances. Building on MDA’s history of innovative fundraising, the event provided opportunities for everyone — from beginners to advanced gamers — to get in the game for a good cause.

The gaming event originated from the HyperX Esports Arena Las Vegas at the Luxor Hotel and Casino and was live-streamed on the gaming platform Twitch. Internationally renowned gamers Alpharad and Terroriser, along with MDA Let’s Play host Beaniez, led the action. In one highlight, MDA National Spokesperson Nyheim Hines of the Indianapolis Colts and former Raider Anttaj “Taj” Hawthorne joined in, playing Fall Guy with Beaniez, Let’s Play community members, and Twitch creators.

Touching on the reason behind the event, Mission Moments highlighted MDA’s important work. In one such moment, Megan E. Jennings, aka Sybil Thorn, 31, who lives with spinal muscular atrophy (SMA), spoke with Beaniez

and thanked MDA for meeting the needs of the community. “Our entire lives, MDA has been there to support people like us, our families, friends, and significant others,” she said.

The Takeover Stream-a-thon brought in more than 3.1 million live views, 2.7 million unique viewers, and more than 50,000 active viewers.

Sponsors included Carnival Corporation & PLC, Las Vegas Aviators, Las Vegas Raiders Foundation, Inspire Brands: Buffalo Wild Wings Foundation and Arby’s Foundation, and Cytokinetics.



Want to get in the game? MDA Let’s Play hosts gaming events every week. Join the fun at mda.org/lets-play.



Canaday family



Image: KFox Photography

Boundless Support

The Canaday family may have moved to Indiana from New York only recently, but they already have rallied the support of their local community for this year's Muscle Walk — raising more than \$20,955 (so far) for MDA.

The family named their team LincDTogether to honor their son Lincoln, who lives with Duchenne muscular dystrophy (DMD). With help from friends and their church community, they had plenty of volunteers and 126 participants at the in-person walk on Aug. 7. Sponsors included a local Fitness Premier gym, Franciscan Hospital, KFox Photography, Strack & Van Til food market, and radio station Shine.FM.

"It was a great opportunity for Lincoln to see the support, and we all definitely felt it," says Bethany, Lincoln's mom. "It was more than we expected, and we were overwhelmed in the best possible way."

Thanks to a virtual participation option, supporters could walk for Lincoln no matter where they were.

"I think one of the blessings of having done the walk in multiple locations is that we have donations from all different states," Bethany says. "People who walked with us in New York could still support us virtually."

Bethany says that the initial hope MDA gave her fuels her reason for participating in Muscle Walk. "Especially when you first get a diagnosis, it can be unexpected and devastating," she says. "You can live in that devastation, or you can choose to be grateful for the day — and we want to highlight that."

Learn more at mda.org/muscle-walk.



From Print to Podcast

Get ready to add to your playlist — *Quest* has a podcast! Join *Quest* Editor-in-Chief Mindy Henderson as she talks with the MDA community about everything from receiving a diagnosis to traveling with a disability. Mindy, who lives with spinal muscular atrophy (SMA), brings her warmth and knowledge as an author and motivational speaker as she engages with the community.

"I hope our community will enjoy the conversations we're putting out there and will come away with new ideas, resources to explore, and a renewed sense of hope and camaraderie," Mindy says.

Listen now at mda.org/podcast.

STEM Connections

As part of MDA's STEM (science, technology, engineering, and math) Connections program, MDA representatives sat down with experts from General Motors (GM) on July 22 to discuss STEM education and careers.

The panel was co-moderated by MDA Ambassadors Justin Moy and Amanda Zurek, both of whom are pursuing careers in STEM-based fields, and it featured engineers, designers, and innovators from GM. Together, they discussed their experiences with STEM careers, how they are working to improve accessibility and address sustainability, and the importance of encouraging youth of all abilities and backgrounds to enter STEM-based fields.

"When you work in a STEM area, you are really trying to solve a problem or trying to meet a need," said VeRonica Mitchell, GM's Electric Vehicle Center of Expertise brand integration and business planning manager. "The input or insight that [people with various abilities and backgrounds] have to offer will be valuable in making that solution. I really encourage you to try and be open to this stuff."

View the entire expert discussion at youtube.com/watch?v=ho8XSPJWVlo.

Images: iStock.com/Magnilion; Vladyslava Yakovenko

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PRESYMPTOMATIC SMA, TREATED WITH SPINRAZA



NICOLE // AGE 45

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 **11,000+** who have been treated with SPINRAZA worldwide.*

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Thousands of adults have been treated with SPINRAZA worldwide*

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DAYS YEARS

There's someone from almost every age group who has taken SPINRAZA^{†‡§}

7+
YEARS

Safety and efficacy evaluated in the longest clinical trial in SMA to date[§]

*Based on commercial patients, early access patients, and clinical trial participants through December 2020.

†Includes clinical trial patients.

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

§Clinical studies of SPINRAZA included patients from 3 days to 16 years of age at first dose.

§Based on commercial patients in the US (including Puerto Rico) through December 2020.

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[®]
(nusinersen) injection
12 mg/5 mL

BYRON // AGE 15

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.

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

IMPORTANT FACTS ABOUT SPINRAZA[®] (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).

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Claiming My Courage

I had to face my son's diagnosis to take better care of my family and myself

BY JESSICA STEPHAN



Jessica Stephan and her son, who is now 9 years old.

There are few specific dates that stick in my memory. Sept. 21, 2016, is one of them. On that day, a phone call changed my family's lives forever. After two long years of visits with specialists and countless tests, there was finally a diagnosis for my 4-year-old son: centronuclear myopathy. In addition, his diagnosis was my diagnosis; he had inherited the mutation from me.

I knew in the back of my mind the call was coming. My father had lived with this disease for more than 30 years, but the reality of the diagnosis still caught me off guard. I was devastated and distraught.

My whole life, I never once heard my father complain or make excuses. It was something I took for granted until I was faced with the same reality for my son, and I was nowhere near as brave. This diagnosis filled my mind with endless questions and overwhelming guilt.

We want to make sure that our son has the opportunity to live as normal a life as possible, without physical restrictions.

The reality, though, is that he wears out faster than his peers. Trying to explain that to a 4-year-old was frustrating. As a parent, you do your best to stay positive. It's exhausting and takes a toll on your patience in all other facets of your life.

I cried daily on my drive to work, had insomnia from my anxiety, and was tired all the time. I realized I had to face this head on with someone who could help me walk through the journey of this diagnosis with a different perspective. It took two therapists to find one who resonated with me, and things got worse initially, but my hard work paid off when some of my symptoms eased. I still work regularly to manage my anxiety with the tools I learned in therapy.

As he got older, my son started asking questions, and it was time to explain his "special muscles" to him. He is very much his grandfather — kind, smart, and courageous. He has never complained through the tests, casting, and wearing leg braces every day. He has more discipline for his self-directed daily physical therapy than people four times his age.

My son is going to be fine, and so am I. I got the help I needed to process the trauma of the diagnosis and find some peace with my family's new reality. It was one of the hardest things I have ever been through, but I am better for it. My focus now is taking the best care of him and myself as possible. I want to be a role model — like my father was for me — of fortitude and grace in the face of a progressive disease. [Q](#)

Jessica Stephan, 41, lives in the Twin Cities of Minnesota with her husband and three children. She is an executive committee member for the Minnesota MDA Muscle Team and full-time Realtor®. You can find her at [linkedin.com/in/jessicastephan](https://www.linkedin.com/in/jessicastephan).

Joanna Buoniconti has known she would be a writer since she was 8 years old.



During my sophomore year of high school, in the midst of my parents' hostile divorce, I noticed that it was becoming harder to raise my right hand to my eye to apply makeup. I refused to acknowledge the weakness at first. I figured that if I kept forcing the muscles to work, it would prevent the inevitable muscle deterioration. But one of the cardinal rules of a chronic illness is that it never follows the logic you want it to. A few months later, I lost the ability to raise my right forearm completely. A couple of months after that, I could no longer reposition my right hand on my wheelchair controller.

It always followed the same pattern: I would notice the weakness, find ways to work around it, and then a short time later, I would wake up to find I no longer had that function. It was frustrating. It was physically and emotionally debilitating. And it plunged my mind into a darkness I had never experienced before.

However, it was also at this time that I began to use writing as an emotional salve. Allowing my emotions to venture out from the confines of my mind helped me to connect with others in ways that I never could have imagined. Since I was 8, I have known that I was born to write, but I did not fathom that the act of writing down my emotions would become akin to therapy for me.

Witnessing my body get weaker was a positively soul-sucking experience that, fortunately, came to an end in the fall of 2018, when I began a treatment that stopped my disease's progression. And while the treatment even helped me regain some strength, the fear that it could stop working has not faded from my mind.

But, in creating the opportunity to write about my anxieties, I have learned to make peace with my body, in all its stages, because it is the vessel that has allowed me to tell my story. And I appreciate it for that. [@](#)

Joanna Buoniconti, 22, was diagnosed with SMA when she was 9 months old. She began writing about living with a disability through a column in her local newspaper, the Daily Hampshire Gazette. She lives with her mother in Western Massachusetts.

Making Peace

Writing helps me process emotions about living with a progressive disease

BY JOANNA BUONICONTI

A common but rarely spoken truth within the neuromuscular disease community is that it is a continuous battle to deal with the emotional repercussions of watching the mobility you have dwindling, before your very eyes, as your disease progresses along its fated course.

For the first 15 years of my life, my strength was consistent. But my doctors had always warned me that spinal muscular atrophy (SMA) would dictate how my muscles deteriorated and that it would occur in stages. Once the deterioration began, it would enact a progressive downhill slope.

However, no one warned me that emotional turmoil could trip the wire and set in motion a cascade of events that I had spent my life narrowly avoiding.

Keeping a Sense of Humor

Acknowledging loss with laughter helps me maintain emotional balance

BY TODD KELLY

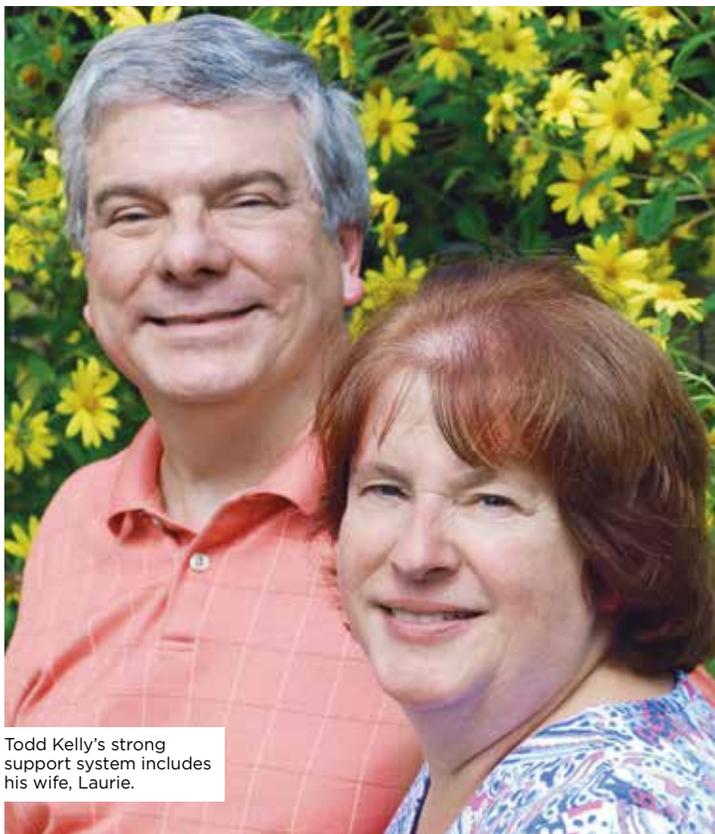
Many years ago, I helped my grandmother, then in her 90s, downsize from her apartment to a single room in an assisted-living wing. Surveying the brimming room, I tried to say something encouraging: “Grandmom, it’s like you’re going back to college.” Her response surprised me. “Actually,” she said, “I like to think of it as finishing school.” Dark humor indeed. I like to believe I’ve inherited her remarkable sense of humor (and verbal cleverness).

Years later, after my left foot inexplicably dropped in 2013, an electromyography revealed the most likely culprit: amyotrophic lateral sclerosis (ALS). I thought my previous bout with prostate cancer at age 41 might have prepared me emotionally for this. But that was different. There were steps to be taken, therapies to try, and a plan of action. I emerged from a whirlwind of activity cancer-free.

Who could anticipate facing another life-threatening disease? With ALS, there is no cure. Life expectancy from diagnosis to death is three to five years. Cancer had certainly propelled me into a period of introspection, but ALS was a total system shock. I was suffering from a form of post-traumatic stress disorder.

I was very fortunate to have such a strong support system in place. I had a loving wife and three wonderful children. I had caring colleagues and empathetic students at the high school where I had taught for the past 27 years. However, for the first time in my life, I required more; I needed professional psychiatric help.

I needed to explore the darkness and express my grief. My wife believed the best way to help me was to focus on the positives and stay optimistic. She wanted me to embrace the happy moments of each day and not dwell on the future. But I needed to acknowledge and anticipate the finish, as well.



Todd Kelly's strong support system includes his wife, Laurie.

A psychologist at the ALS Hope Foundation Clinic in Philadelphia counseled my wife and me and set me up with a local psychologist for several productive sessions.

I found the solution for me was keeping a tenuous emotional balance. In the past eight years, I have experienced moments of intense joy and profound sadness. Looking backwards, I’ve reconnected with former students who are now nurses, doctors, ministers, and counselors. Looking forward, I’ve found purpose in ALS advocacy and serving as a consumer reviewer for the Congressionally Directed Medical Research Program. I’ve maintained my sense of humor throughout this journey, although sometimes it is rather grim.

One day last summer, my wife came back into the house drenched in sweat. She had been weeding and pruning the front landscaping bed (a task that had been my purview for many years). “I hate that plant,” she said, referring to a creeping juniper that was thriving there. “I want to get rid of it,” she muttered. “Over my dead body,” was my response. She gave me an eye-roll and added, “OK then.”

To date, the shrub is still growing there. I’m not finished yet. [Q](#)

Todd Kelly, 58, and his wife, Laurie, live in Gilbertsville, Pennsylvania. Diagnosed with ALS in 2013, Todd is a former high school English teacher and drama director.

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actor portrayal



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Our photo contest winner is in his element



“

I was hospitalized about a month before this — also four times in 2020. I think those hard times inspired me.

— Leslie Crowley Jr.

Congratulations to our 2021 Lasting Impression Photo Contest winner Leslie Crowley Jr., 27, of Atlanta, Georgia. The photo shows Leslie, a former MDA Summer Camper who lives with Duchenne muscular dystrophy (DMD), in his natural element, with his music equipment behind him in his home studio. A lifelong music lover, Leslie produces music under the name J0K3 (find him on social media @jokemusic3).

“When this photo was taken, I had just gotten through a rough moment,” Leslie says. “I was hospitalized about a month before this — also four times in 2020. I think those hard times inspired me.”

This photo was taken on the first day of shooting a documentary that Leslie is making through his nonprofit iNeed2Go Inc. The documentary, which he is making with the help of friends, will tell the stories of Leslie and others who are living with disabilities. While filming, his friend, the musician, photographer, and filmmaker known as Trvy, stopped to snap a few photos.

“I wanted him to capture me in my natural state,” Leslie says. “That whole day was just spontaneous, and the photos were shot on the fly. Trvy also commented on how he admired my resilience.” [📷](#)

+MORE PHOTOS ONLINE

Congratulations to the runners-up in our reader photo contest: Leah Hover-Preiss of Wisconsin Dells, Wisconsin; Nicklaus James Shorten of Portland, Oregon; and David Leatherman of Cedar Rapids, Iowa. See their photos at mda.org/quest.

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