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Quest

MDAQUEST.ORG ISSUE 3 • 2023

The Buddy System

How to empower kids to make friends in school

JUST ASK
We all need help sometimes

MEDICAL EMERGENCIES
Know what to do



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MOVING AHEAD with

Evrysdi® (risdiplam)

1,800+

people in the US with spinal muscular atrophy (SMA) are taking Evrysdi, including people up to 84 years old**

*Number of people taking Evrysdi as of May 2022. Evrysdi approved in August 2020.

†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 children and adults.

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, as Evrysdi may harm your unborn baby. Ask your healthcare provider for advice before taking this medicine
- are a woman who can become pregnant:
 - Before you start your treatment with Evrysdi, your healthcare provider may test you for pregnancy
 - 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
 - **Pregnancy Registry.** Talk to your healthcare provider right away if you become pregnant while taking Evrysdi. Ask about registering with the Evrysdi Pregnancy Registry, which was created to collect information about your health and your baby's health. Your healthcare provider can enroll you in this registry by calling 1-833-760-1098 or visiting www.evrysdipregnancyregistry.com
- are an adult male. Evrysdi may affect a man's ability to have children (fertility). Ask a healthcare provider for advice before taking this medicine
- are breastfeeding or plan to breastfeed. It is not known if Evrysdi passes into breast milk and may harm your baby

Studied in the **most inclusive** clinical study program in SMA^{‡§}

- ✓ For newborns to adults with SMA — **later-onset, infantile-onset, and presymptomatic SMA**
- ✓ Designed to help the body **make more SMN protein**
- ✓ Safety profile that has been studied in **more than 490 people** from newborns to adults
- ✓ Oral treatment that **can fit into your day**



[‡]Studies included individuals with a broad range of physical ability, including those with and without the ability to walk, with and without scoliosis (mild to severe), with and without prior disease-modifying treatment (evaluated for safety).

[§]The efficacy and safety of Evrysdi was established in 3 main studies. SUNFISH is a 2-part, placebo-controlled study in 231 adults and children aged 2 to 25 years with Type 2 or 3 SMA. FIREFISH is a 2-part, open-label study in 62 infants aged 2 to 7 months with Type 1 SMA. RAINBOWFISH is an ongoing, open-label study in 26 newborns younger than 6 weeks (at first dose). These newborns were genetically diagnosed with SMA and had not yet shown symptoms (presymptomatic SMA). A fourth study, JEWELFISH, is an ongoing, open-label safety study in 174 people aged 1 to 60 years with Type 1, 2, or 3 SMA that was previously treated with approved or investigational SMA medications.

Important Safety Information (continued)

Tell your healthcare provider about all the medicines you take.

You should receive Evrysdi from the pharmacy as a liquid. 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, and sore throat (upper respiratory infection); lung infection (lower respiratory infection); constipation; vomiting; cough

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.

If you cannot afford your Evrysdi medication, visit MySMASupport.com for financial assistance 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 children and adults.

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.
 - **Pregnancy Registry.** There is a pregnancy registry for women who take EVRYSDI during pregnancy. If you become pregnant while receiving EVRYSDI, tell your healthcare provider right away. Talk to your healthcare provider about registering with the EVRYSDI Pregnancy Registry. The purpose of this registry is to collect information about your health and your baby's health. Your healthcare provider can enroll you in this registry by calling 1-833-760-1098 or visiting <https://www.evrysdipregnancyregistry.com>.
- 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, including your 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 or other healthcare provider. 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 syringe(s) 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 syringe(s) provided by your pharmacist (you should receive 1 or 2 identical oral syringes depending on your prescribed daily dose) 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 syringe(s) are lost or damaged.
- When 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, and sore throat (upper respiratory infection)
 - constipation
 - diarrhea
 - lung infection (lower respiratory infection)
 - vomiting
 - rash
 - cough

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.
 - If necessary, EVRYSDI can be kept at room temperature up to 104°F (up to 40°C) for a combined total of 5 days. EVRYSDI can be removed from, and returned to, a refrigerator. The total combined time out of refrigeration should not be more than 5 days.
- 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) or if EVRYSDI has been kept at room temperature (below 104°F [40°C]) for more than a total combined time of 5 days. Discard EVRYSDI if it has been kept above 104°F (40°C). Please see the Discard After date written on the bottle label. (See the **Instructions for Use** that comes with EVRYSDI).

Keep EVRYSDI, all medicines and syringes 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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EVRYSDI® (risdiplam)

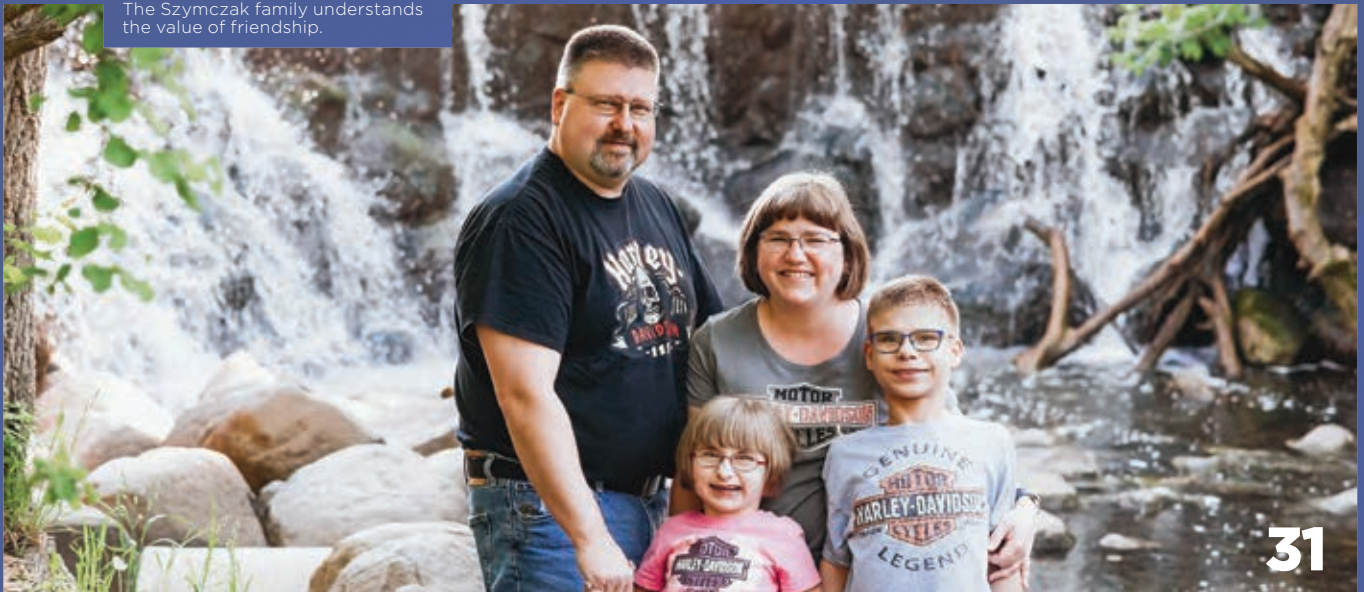
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For more information, go to www.EVRYSDI.com or call 1-833-387-9734.

The Szymczak family understands the value of friendship.



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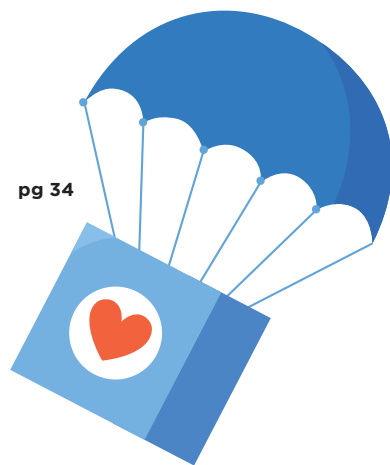
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LASTING IMPRESSION

MDA advocates returned to Washington.

The Power of Community

At MDA, our mission is to empower people with neuromuscular disease to live longer, stronger, more independent lives. As MDA approaches its 75th year of mission-driven research, care, and advocacy, Quest magazine is instrumental in keeping our diverse community informed, engaged, and connected. Knowledge is a catalyst for change, and it is why we have thoughtfully curated articles and interviews that explore the significance of friendships and the triumphs we can achieve when we band together.

As you enjoy this issue, I encourage you to embrace the concept of community and cultivating meaningful connections with others. Discover inspiration in the stories of individuals who are fearlessly embracing opportunities. Celebrate accomplishments, both big and small, and believe in the transformative power of our community to redefine what it means to live with a disability. By expanding our community and raising our voices, we are shaping a world where everyone is recognized, valued, and empowered to live life on their terms.

When we initiate and engage in meaningful conversations within the MDA community, we witness incredible achievements. Over the past 15 years, we have celebrated the approval of 20 new drugs, including three groundbreaking genetic treatments: one for spinal muscular atrophy (SMA), one for people with the *SOD-1* gene mutation for amyotrophic lateral sclerosis (ALS), and another for boys living with Duchenne muscular dystrophy (DMD). MDA's unique Gene Therapy Support Network has evolved as a trusted resource for families and medical professionals, providing guidance to inform treatment decisions.

Together, we are also influencing regulatory and legislative decisions on access to treatments, access to safe and reliable air transportation for people in power wheelchairs, and increased funding for ongoing neuromuscular and genetic research.

Thank you for connecting with our community. Let us continue to collaborate, uplift one another, and construct an inclusive world abundant with opportunities for longer, stronger, more independent lives.



Donald S. Wood, PhD
President and CEO
Muscular Dystrophy Association



Donald S. Wood, PhD

+ENTER THE PHOTO CONTEST

Share a photo of a meaningful moment for you or a loved one with a neuromuscular disease, and it could be selected to appear in a future issue of Quest magazine. Submit your photo at SurveyMonkey.com/r/QuestPhotos or scan this QR code.



Muscular Dystrophy Association is the #1 voluntary health organization in the United States for people living with muscular dystrophy, ALS, and related neuromuscular diseases. For more than 70 years, MDA has led the way in accelerating research, advancing care, and advocating for the support of our families. MDA's mission is to empower the people we serve to live longer, more independent lives.

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You also can sign up or change your personal information by calling the MDA Resource Center at **833-ASK-MDA1**.

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Burn Boot Camp Brought the Energy



From assisting with parking to leading the warm-up, Burn Boot Camp volunteers brought the energy during the MDA Muscle Walk of Houston.

Stoked at Arizona Golf



Each year MDA community sponsors gather for a day of golf at the MDA Golf Classic in Scottsdale, AZ. Thank you to our committee and Dutch Bros AZ volunteers.

End ALS with MDA Night of Hope



In 17 years, the gala committee has raised over \$11 million for ALS research. The committee, sponsors, and donors fund research initiatives and the MDA/ALS Care Center at Emory Healthcare.

Hitting the links with SUNDT Golf



Thank you, SUNDT Foundation for your continued commitment to the MDA through the Mike Gaines Charity Golf Tournaments. All your efforts are greatly appreciated.

Washington D.C. Tribute Tour



From left: Diana Bharucha-Goebel, M.D.; Paul Robertson, Founder FFMD; R. Rodney Howell, M.D.; Jeni Stepanek, Ph.D. Janet Woodcock, M.D. not pictured.

Throughout MDA's history, local volunteers and communities have been at the heart of MDA's mission-focused efforts. In June, MDA honored the legacy and life's work of five individuals who have been tireless in their efforts to support the patients and families we serve.



Pictured Above: Janet Woodcock, M.D. and Stanley H. Appel, M.D.

In honor of Lou Gehrig Day, MDA hosted a Tribute Tailgate with the Washington Nationals baseball team to recognize ALS Awareness Month and the fight to End ALS with MDA.



Pictured Above: Washington Nationals' mascot Screech and Donald S. Wood, PhD



Wishing You Success on Your Journey



While it can be said that we each have our unique barriers in life to overcome, growing up with spinal muscular atrophy (SMA) influenced me in some important ways. Making friends at school, navigating social settings, and getting comfortable asking for help as I worked to be as independent as possible were some of the things that took extra time and intention for me to become good at. Those life experiences helped inspire some of the content in this issue of Quest magazine.

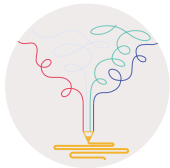
As the kids and some adults in our community go back to school this fall, I hope the tips for making friends at different ages and stages in life (page 28) and asking for help (page 34) will be useful, whatever phase of your journey — or your kids’ journey — you find yourself in.

We also took the liberty of exploring another resource for life’s journey: palliative and hospice care (page 24). As I read this article, I was surprised to learn all I didn’t know about these modes of care — and how much I wish I’d known sooner.

Regardless of where you are in life, here’s to our journey, to finding beauty in each phase, to connections, and to all of our success!

*Mindy Henderson, Senior Director and Editor-in-Chief, Quest Media
Muscular Dystrophy Association*

ICONS TO WATCH FOR THROUGHOUT QUEST



QUEST MAGAZINE ONLINE



QUEST BLOG



QUEST PODCAST



DISEASES A-Z CONTENT LIBRARY



DID YOU KNOW?

MDA Let’s Play brings the community together for video games, trivia, art, and good times.

Using the online platform Discord, MDA Let’s Play provides a safe and welcoming environment for everyone in the community to enjoy entertainment through gaming and activities while socializing with others who can truly understand their unique situations. Join events like Saturday Game Night and Wednesday Up Late, or check out daily streamed content.

Learn more at mda.org/lets-play.



By popular demand, we launched the new Quest Media inclusive product guide.

This guide is designed to help you find the products you need to live a more independent, stylish, fun, and all-around great life. Each product is picked by an MDA Ambassador who shares exactly how it helps them in their daily lives. We hope you find products here that enhance your life, too.

Check out the inclusive product guide at MDAquest.org.



Raising Our Voices

MDA's Advocacy team ensures the neuromuscular disease community is heard in Washington

Every day, MDA's advocacy team works relentlessly to champion the issues important to those living with neuromuscular diseases. Along with promoting access to care, disability policy, and accelerating drug development, the team also organizes and empowers MDA's grassroots advocacy network, made up of members of the neuromuscular disease community.

Quest spoke with MDA's Vice President of Public Policy and Advocacy, Paul Melmeyer, and MDA's Director of Advocacy Engagement, Mark Fisher, to learn more about MDA's Advocacy Program and the team's essential work.

Why is advocacy important to MDA and its mission?

MDA's mission is to empower the people we serve to live longer, more independent lives, and the organization couldn't fulfill that mission without an advocacy team. Changes to public policy, particularly at the federal government level, can help our community access care, thrive, and lead the life they want. To achieve those policy changes, we advocate for those with neuromuscular diseases and work to ensure that public policy decisions support them.

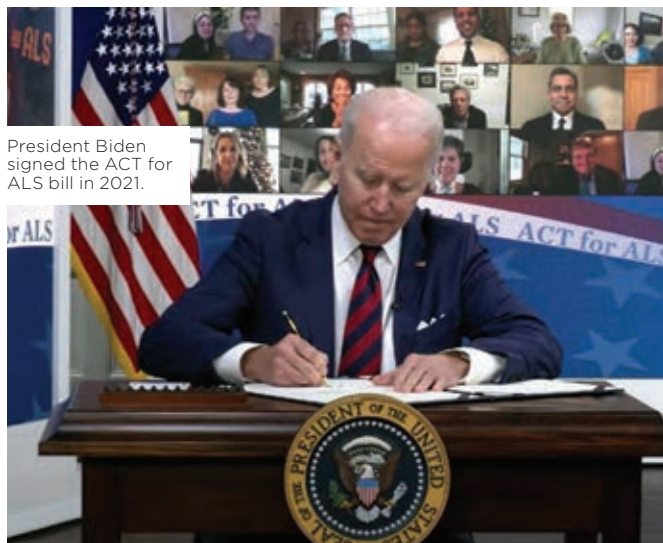
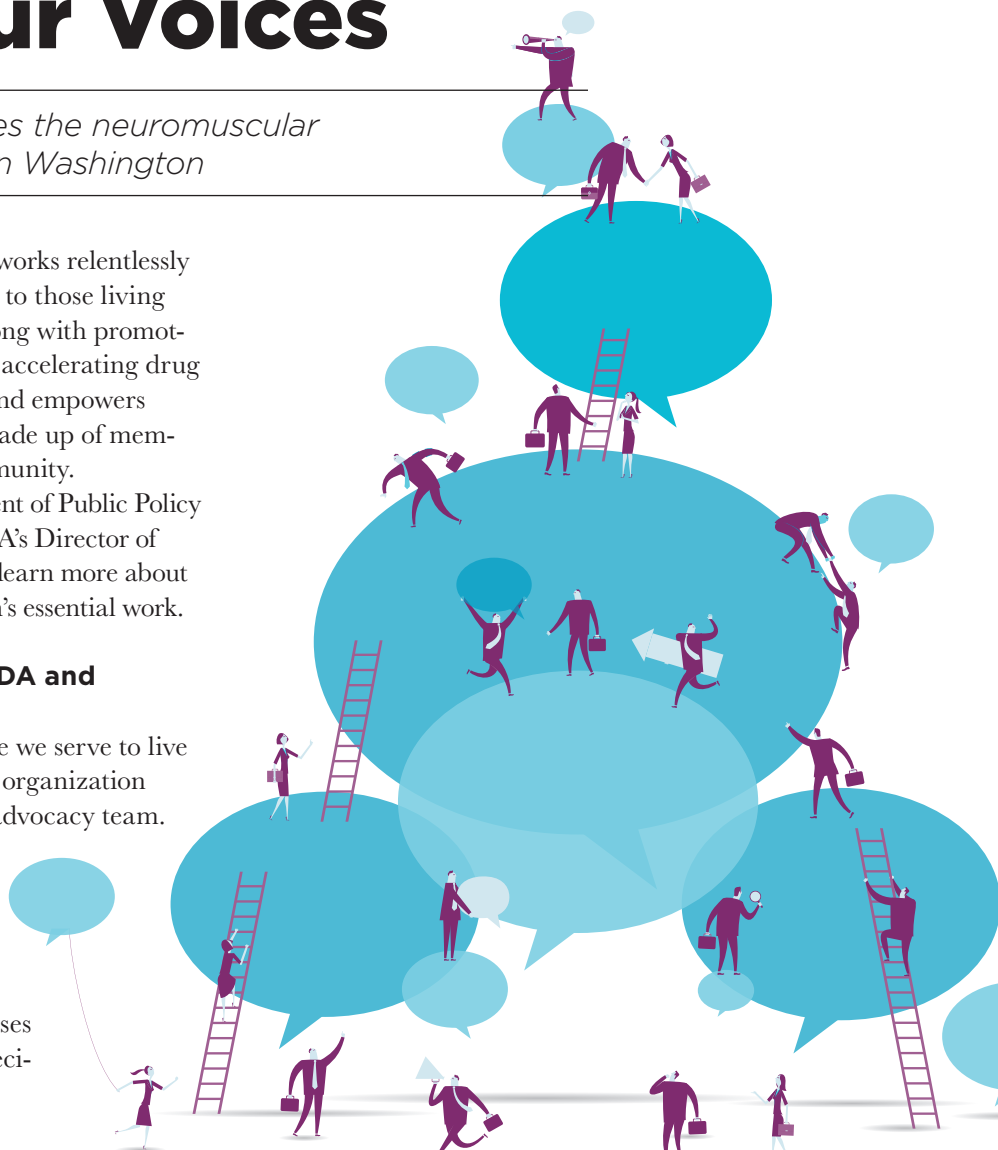
What are people surprised to learn about MDA's advocacy?

People are sometimes surprised that organizations like MDA have an advocacy program and registered lobbyists on our team. We primarily work at the federal level

because those decisions affect everybody with neuromuscular diseases across the country. But we do get involved at the state level if a state is making impactful decisions, such as those regarding public health, that may serve as a precedent for other states.

Lastly, grassroots advocates sometimes ask if their voices matter anymore. The answer is, yes! Members of Congress still need to hear from you. Your voice makes a difference.

Advocacy Win: Access to Care
 The MDA advocacy team and grassroots advocacy volunteers' efforts led to Medicare prescription drugs being capped at \$2,000 per year in out-of-pocket costs, starting in 2025.





Paul Melmeyer, MDA's VP of Public Policy and Advocacy, represents MDA at the White House.

Can you tell us more about the advocacy team's three focus areas: access to care, disability policy, and accelerating therapeutic development?

For access to care, we try to influence policy changes that would increase access and remove barriers to the health-care that the neuromuscular disease community faces.

This includes making clinicians, specialists, therapists, and medical equipment more easily

accessible. We also work to reverse or prevent policy changes that would create barriers, such as health insurance plan limitations, and try to remove challenges caused by geographic barriers.

For our focus on disability policy, we try to knock down the societal barriers preventing those with neuromuscular disabilities from living their best lives. This includes obsta-

cles and discrimination that make travel, employment, and education less accessible for someone living with a disability. Currently, one of our focus areas is making air travel more accessible. We are also working to increase

Advocacy Win: Access to Care

With the help of other organizations, MDA's advocacy team and grassroots volunteers successfully advocated for the Centers for Medicare and Medicaid Services to begin covering seat elevation systems for most power wheelchairs. Previously, individuals had to pay out of pocket for these expensive systems.



TRANSPORTATION CHAT

Listen to a conversation with advocates pushing for advances in accessible air travel and transportation policies at MDAQuest.org/podcast/accessibility-DOT.

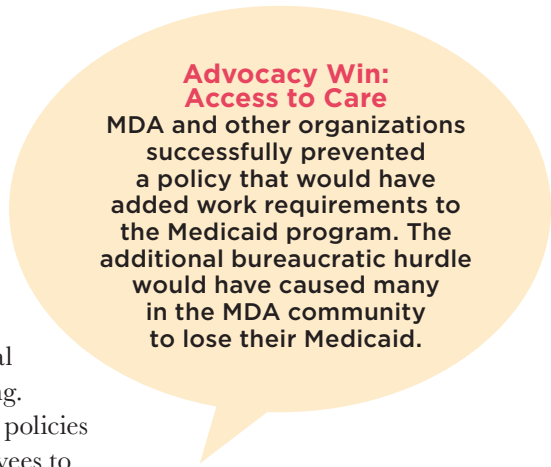
employment opportunities by ensuring that those living with neuromuscular diseases can continue to receive vital federal benefits while working. We also advocate for policies that empower employees to hire and retain individuals with disabilities.

Our third focus area, accelerating therapeutic development, is important because there are still far too few therapies approved by the US Food and Drug Administration (FDA) available to our community. So, we work with Congress and the FDA to ensure that more and better therapies become available to our community more quickly.

There are so many issues the advocacy team could get involved in. How do you decide where to focus your efforts?

The list of things we want to work on is never-ending, and it is a continuous challenge to prioritize our efforts. To ensure we put our time and resources where they will be most impactful and best serve those with neuromuscular diseases, we look at the different advocacy opportunities in front of us and evaluate each opportunity based on several factors, including:

- The issue's importance to and impact on the neuromuscular disease community
- If any other advocacy groups are working on the issue
- How urgent the issue is
- How much of an impact MDA could make



Advocacy Win: Access to Care

MDA and other organizations successfully prevented a policy that would have added work requirements to the Medicaid program. The additional bureaucratic hurdle would have caused many in the MDA community to lose their Medicaid.



Michael Lewis (left), MDA's Director of Disability Policy, meets with Rep. Jim Langevin of Rhode Island.

From the grassroots perspective, we look for the best opportunities for volunteers to weigh in. We know our volunteers have busy lives, and we want to give them opportunities that are meaningful to them.

How does the team advocate for these critical issues?

Our team goes to Capitol Hill in Washington, DC, and discusses these issues with policymakers, using compelling facts, statistics, and policy arguments to explain why we promote or disagree with a particular change. We also hold virtual and phone meetings with members of Congress; submit written comments and testimony to lawmakers, committees, and government agencies; and work with coalitions to achieve all our public policy goals. It's a challenging and fun job. Every day is different. The legislative process is unpredictable, so we must be flexible and ready to react to anything they decide to do.

Combining our efforts with the voices from the grassroots advocacy network lets policymakers know how a policy affects someone personally, and it's an extremely powerful combination.

What should the MDA community know about the grassroots advocacy network?

MDA's grassroots network is a place for the community to have their voices heard on MDA's advocacy issues. Our volunteer advocates are engaged in several issues, like ensuring that vital public insurance programs, like Medicare and Medicaid, stay strong and well funded; supporting efforts to strengthen and modernize the newborn screening program; and improving accessibility in air travel and other transportation.

Grassroots volunteers email, call, or tweet their members of Congress, and our website allows them to do that with just a couple of clicks of the mouse (visit mda.org/advocacy).

Advocacy Win: Disability Policy
After many years of advocacy from MDA and other disability organizations, restrooms in single-aisle airplanes will become more accessible. Grassroots advocates helped by emailing and calling the DOT to advocate for this.

We also encourage folks to contact government agencies, such as the Department of Transportation (DOT), which makes decisions about air travel. Some volunteers visit their members' of Congress

Advocacy Win: Therapy Development
Last year, several FDA programs were reauthorized and reformed, making clinical trials more diverse and accelerating new treatments, particularly gene therapies.

offices for in-person meetings, both in their districts and in Washington, DC. To effect change, the best thing someone can do is meet with their member of Congress or staff in person, and we can provide that opportunity.

To educate the community, the Advocacy team offers monthly webinars on topics such as specific policies we are trying to change, our work with the FDA, or even issues like voting and employment rights. We hope more people will be comfortable taking action after participating in a webinar. We also have opportunities for volunteers to write about an advocacy issue on our blog.

Collaboration is a crucial part of MDA's advocacy mission. Can you tell us more about this?

We know we can't do it all on our own, so we look to work with others within and outside the neuromuscular disease community to further the mission. For example, if we're working on an access to care issue, we may work with another nonprofit advocacy organization that focuses on a different disease with similar access challenges. This has been a successful approach.

In addition to working with outside organizations, the Neuromuscular Advocacy Collaborative, led by MDA, convenes other neuromuscular disease advocacy organizations that tend to focus on a specific neuromuscular disease community. This includes organizations focused on Duchenne muscular dystrophy (DMD), spinal muscular atrophy (SMA), amyotrophic lateral sclerosis (ALS), limb-girdle muscular dystrophy (LGMD), and others. Doing this brings together 20 to 25 organizations' voices, rather than only MDA's. Together, we work on a shared set of public policy changes that can benefit the entire neuromuscular disease community. [Q](#)

Advocacy Win: Access to Care
With the help of our grassroots volunteers, the advocacy team successfully encouraged CMS to give LGMD its own diagnostic codes, which could shorten the time to diagnosis, help clinicians deliver more precise medical care, improve clinical trials, and increase future access to targeted treatments.

+BE HEARD

Add your voice to MDA's Grassroots Advocacy Network. Sign up at MDA.org/advocacy.



VYVGART® Hytrulo

(efgartigimod alfa and
hyaluronidase-qvfc)

Subcutaneous Injection
180 mg/mL and 2000 U/mL vial

Picture your life in motion

GENERALIZED MYASTHENIA GRAVIS

*isn't canceling this
weekend getaway*

IMPORTANT SAFETY INFORMATION

What is the most important information I should know about VYVGART® HYTRULO (efgartigimod alfa and hyaluronidase-qvfc)?

VYVGART HYTRULO may cause serious side effects, including:

- **Infection.** VYVGART HYTRULO may increase the risk of infection. The most common infections for efgartigimod alfa-fcab-treated patients were urinary tract and respiratory tract infections. More patients on efgartigimod alfa-fcab vs placebo had below normal levels for white blood cell counts, lymphocyte counts, and neutrophil counts. The majority of infections and observed lower white blood cell counts were mild to moderate in severity. Your healthcare provider should check you for infections before starting treatment, during

treatment, and after treatment with VYVGART HYTRULO. Tell your healthcare provider if you have any history of infections. Tell your healthcare provider right away if you have signs or symptoms of an infection during treatment with VYVGART HYTRULO such as fever, chills, frequent and/or painful urination, cough, pain and blockage of nasal passages/sinus, wheezing, shortness of breath, fatigue, sore throat, excess phlegm, nasal discharge, back pain, and/or chest pain. If a serious infection occurs, your doctor will treat your infection and may even stop your VYVGART HYTRULO treatment until the infection has resolved.

- **Undesirable immune reactions (hypersensitivity reactions).** VYVGART HYTRULO and efgartigimod alfa-fcab can cause the immune system to have

undesirable reactions such as rashes, swelling under the skin, and shortness of breath. Hives were also observed in patients treated with VYVGART HYTRULO. In clinical studies, the reactions were mild or moderate and occurred within 1 hour to 3 weeks of administration, and the reactions did not lead to VYVGART HYTRULO discontinuation. Your healthcare provider should monitor you during and after treatment and discontinue VYVGART HYTRULO if needed. Tell your healthcare provider immediately about any undesirable reactions to VYVGART HYTRULO.

Before taking VYVGART HYTRULO, tell your healthcare provider about all of your medical conditions, including if you:

- Have a history of infection or you think you have an infection.

NOW AVAILABLE! VYVGART Hytrulo is a new, FDA-approved subcutaneous injection for adults with anti-AChR antibody positive gMG

VYVGART Hytrulo usually takes

30 to 90 seconds to inject

and is given at an infusion center, doctor's office, or at home*†

*For at least 30 minutes after your injection, a healthcare professional will monitor you for reactions.

†In some cases, VYVGART Hytrulo may also be given at home by a trained nurse.



Learn about the effectiveness and safety of VYVGART Hytrulo



Scan the QR code or visit
[GetStartedonVYVGARTHytrulo.com](https://www.vyvgart.com/GetStartedonVYVGARTHytrulo.com)

AChR=acetylcholine receptor; gMG=generalized myasthenia gravis | Visit [VYVGART.com/glossary](https://www.vyvgart.com/glossary) for a glossary of terms.

- Have received or are scheduled to receive a vaccine (immunization). Discuss with your healthcare provider whether you need to receive age-appropriate immunizations before initiation of a new treatment cycle with VYVGART HYTRULO. The use of vaccines during VYVGART HYTRULO treatment has not been studied, and the safety with live or live-attenuated vaccines is unknown. Administration of live or live-attenuated vaccines is not recommended during treatment with VYVGART HYTRULO.
- Are pregnant or plan to become pregnant and are breastfeeding or plan to breastfeed.

Tell your healthcare provider about all the medicines you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements.

What are the common side effects of VYVGART HYTRULO?

The most common side effects of efgartigimod alfa-fcab-treated patients were respiratory tract infection, headache, and urinary tract infection. Additional common side effects of VYVGART HYTRULO are injection site reactions, including rash, redness of the skin, itching sensation, bruising, pain, and hives.

These are not all the possible side effects of VYVGART HYTRULO. Call your doctor for medical advice about side effects. You may report side effects to the US Food and Drug Administration at 1-800-FDA-1088.

What is VYVGART HYTRULO?

VYVGART HYTRULO is a prescription medicine used to treat a condition called generalized myasthenia gravis, which causes muscles to tire and weaken easily

throughout the body, in adults who are positive for antibodies directed toward a protein called acetylcholine receptor (anti-AChR antibody positive).

Please see the full Prescribing Information for VYVGART HYTRULO at [VYVGARTHytrulo.com/PI](https://www.vyvgart.com/PI) and talk to your doctor.

Please see brief summary on next page.

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US-ESC-22-00021 V1 06/2023

<p>Important Information about VYVGART® HYTRULO (efgartigimod alfa and hyaluronidase-qyvc); Rx only.</p> <p>The risk information provided here is not comprehensive. To learn more, talk about VYVGART HYTRULO with your healthcare provider. The US Food and Drug Administration (FDA)-approved product labeling can be found by visiting www.VYVGARTHYTRULO.com/PI or calling 1-833-VYVGART (1-833-898-4278).</p>	<p>right away if you have signs or symptoms of an infection during treatment with VYVGART HYTRULO such as fever, chills, frequent and/or painful urination, cough, pain and blockage of nasal passages/sinus, wheezing, shortness of breath, fatigue, sore throat, excess phlegm, nasal discharge, back pain, and/or chest pain. If a serious infection occurs, your doctor will treat your infection and may even stop your VYVGART HYTRULO treatment until the infection has resolved.</p>	<p>What are the common side effects of VYVGART HYTRULO?</p> <p>The most common side effects of efgartigimod alfa-fcab-treated patients were respiratory tract infection, headache, and urinary tract infection. Additional common side effects of VYVGART HYTRULO are injection site reactions, including rash, redness of the skin, itching sensation, bruising, pain, and hives.</p> <p>These are not all the possible side effects of VYVGART HYTRULO. Call your doctor for medical advice about side effects. You may report side effects to the FDA at 1-800-FDA-1088</p>
<p>What is VYVGART HYTRULO?</p> <p>VYVGART HYTRULO is a prescription medicine used to treat a condition called generalized myasthenia gravis, which causes muscles to tire and weaken easily throughout the body, in adults who are positive for antibodies directed toward a protein called acetylcholine receptor (anti-AChR antibody positive).</p>	<ul style="list-style-type: none"> • Undesirable immune reactions (hypersensitivity reactions). VYVGART HYTRULO and efgartigimod alfa-fcab can cause the immune system to have undesirable reactions such as rashes, swelling under the skin, and shortness of breath. Hives were also observed in patients treated with VYVGART HYTRULO. In clinical studies, the reactions were mild or moderate and occurred within 1 hour to 3 weeks of administration, and the reactions did not lead to VYVGART HYTRULO discontinuation. Your healthcare provider should monitor you during and after treatment and discontinue VYVGART HYTRULO if needed. Tell your healthcare provider immediately about any undesirable reactions to VYVGART HYTRULO. 	<p>What are the effects of VYVGART HYTRULO on other drugs?</p> <p>The use of VYVGART HYTRULO with medications that bind to a receptor called the human neonatal Fc receptor (FcRn) may reduce the effectiveness of these medications. Tell your healthcare provider about all the medicines you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements.</p>
<p>What is the most important information I should know about VYVGART HYTRULO?</p> <p>VYVGART HYTRULO may cause serious side effects, including:</p> <ul style="list-style-type: none"> • Infection. VYVGART HYTRULO may increase the risk of infection. The most common infections for efgartigimod alfa-fcab-treated patients were urinary tract and respiratory tract infections. More patients on efgartigimod alfa-fcab vs placebo had below normal levels for white blood cell counts, lymphocyte counts, and neutrophil counts. The majority of infections and observed lower white blood cell counts were mild to moderate in severity. Your healthcare provider should check you for infections before starting treatment, during treatment, and after treatment with VYVGART HYTRULO. Tell your healthcare provider if you have any history of infections. Tell your healthcare provider 	<p>Immunization</p> <p>Discuss with your healthcare provider if you have received or are scheduled to receive a vaccine (immunization) and if you need to receive age-appropriate immunizations before initiation of a new treatment cycle with VYVGART HYTRULO. The use of vaccines during VYVGART HYTRULO treatment has not been studied, and the safety with live or live-attenuated vaccines is unknown. Administration of live or live-attenuated vaccines is not recommended during treatment with VYVGART HYTRULO.</p>	<p>What information should I know about VYVGART HYTRULO and pregnancy and breastfeeding?</p> <p>There are no available data on the use of VYVGART HYTRULO or efgartigimod alfa containing products during pregnancy. There is no information regarding the presence of efgartigimod alfa or hyaluronidase, from administration of VYVGART HYTRULO, during breastfeeding. Talk to your doctor if you are pregnant or plan to become pregnant and are breastfeeding or plan to breastfeed.</p>
		<p>Can VYVGART HYTRULO be used in children?</p> <p>The safety and efficacy in children (pediatric patients) have not been established.</p>





Defining Moments

Life experiences taught Ken Yorgan not to underestimate himself

BY REBECCA HUME

Ken Yorgan has worked hard — and taken a few chances — to build a life he loves and a career with the US Army. He found his path to success by embracing the support of encouraging mentors and ignoring false limitations set by others.

A can-do perspective

The 59-year-old is one of three children in his family born with spinal muscular atrophy (SMA). He began using a wheelchair in third grade, and his parents were strong advocates for his inclusion. They taught him that he could do anything he set his mind to. Ken spent his childhood swimming, playing stickball, and attending MDA Summer Camp.

Ken recalls his fifth-grade teacher, Mr. Griffin, including him in soccer games at recess, bolstering Ken’s belief that he was as capable as other children. In contrast, his sixth-grade teacher refused to allow him to participate in recess in an effort to keep him “safe.” This experience taught him never to allow someone else’s mindset to act as a barrier in his own life.

Ken graduated from high school early and moved from Texas to Pennsylvania to live with his aunt and uncle. They both encouraged him to try things outside his comfort zone, from tubing down the biggest waterslide at an amusement park to living on campus at college.

Independence at college

The Office of Students with Disabilities at Edinboro University (now PennWest Edinboro) provided Ken with a slew of resources, including access to personal care attendants, meal aides (one of whom is now his wife, Susan), accessible transportation, and an accessible dorm room on a floor modified for students with disabilities.



Ken Yorgan has lived in Orlando since he fell in love with the city on a Florida vacation.

Ken studied speech communications and worked at the college radio station. “College was an amazing experience,” he says. “Those four years were the most fun and formative years of my life.

Meeting other people with disabilities and learning their stories was an opportunity to grow and learn more about myself as well.” Ken embraced the opportunity to connect with new friends and even started a wheelchair football team.

His parents taught him that he could do anything he set his mind to. Ken spent his childhood swimming, playing stickball, and attending MDA Summer Camp.



Ken meets a group of Orlando Magic fans.



After graduating, Ken worked with an organization that connected potential employers and individuals with disabilities. Later, he accepted a position designing ads and writing commercials for a large RV, motorcycle, and car dealership in Pennsylvania. Although Ken thrived creatively in his role, a Florida vacation changed the path of his quest for success.

Changing plans and taking risks

After falling in love with the city of Orlando during a vacation, Ken quit his job, packed his car, and drove to Florida with no plan and only \$3,000 in his pocket. Ken now drives a modified van, but at the time he drove a car without modifications and was able to transfer

Ken met his wife, Susan, in college.



independently using a portable wheelchair. He lived in that car for two weeks while searching for an apartment and going to job interviews.

Ken chose to tell potential employers about his wheelchair before interviews, both to ensure that interview locations were accessible and to discern any negative assumptions. “I wouldn’t want to work for an employer who hesitated to hire me because of my wheelchair,” Ken says. “But I also have the opportunity to change some of those biases in the workforce. People might have a preconception, but when you get in front of them and do your job well, most of that goes away.”

In Florida, Ken worked as a cruise travel agent for more than 10 years, bringing confidence, charisma, and creativity to his role. Ready for a change in the early 2000s, he went back to school for an associate degree in IT.

A new career

A friend working as a contractor for the Army informed Ken of an open position at the IT help desk. Ken interviewed and accepted the role. Over the years, Ken pursued every training and certification available to him. He worked his way up to his current role as the Network Administrator for the US Army’s simulation training program.

In his role, Ken provides email and server support to more than 1,300 people and oversees system security. The Army provides some accessibility accommodations for him, such as a lanyard with a push button for automatic doors. For high-security areas that require badge clearance, Ken contacts someone in the office to assist him with the keypad and door.

Living the life you want

Ken believes that having people in his life who encourage him to challenge himself is a key motivating factor in his success. But he’s also motivated by a desire to prove doubters wrong. “Never underestimate the power you have,” Ken says. “Believe in yourself. You can do it — and don’t let anyone tell you that you can’t.” [Q](#)

Rebecca Hume is Senior Specialist and Writer for Quest Media.

Progress NOW

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

New approvals

FDA Approves Treatment for ALS

The US Food and Drug Administration (FDA) granted accelerated marketing approval to Biogen's tofersen (Qalsody) for the treatment of amyotrophic lateral sclerosis (ALS) associated with a mutation in the superoxide dismutase 1 (*SOD1*) gene (SOD1-ALS). This is a familial form of ALS.

Qalsody is the fourth approved therapy to treat a form of ALS and the first therapy to target a genetic cause of ALS. Accelerated marketing approval allows a drug that treats an unmet medical need to receive approval before it has gone through all the required human testing.

Qalsody is a type of drug known as an anti-sense oligonucleotide (ASO). It is administered intrathecally (through spinal injection) and acts by reducing the production of SOD1 protein in cells.

The approval of Qalsody is supported by the efficacy and safety data from the phase 3 VALOR trial and an open-label extension study. In the trial, the drug missed the primary endpoint of producing ALS functional changes, but it met the secondary endpoint of lowering levels of plasma neurofilament light (NFL), a biomarker of nerve damage, as well as SOD1 protein levels. The clinical benefits of Qalsody will continue to be studied in the phase 3 ATLAS study.



For more information on the ATLAS study, visit ClinicalTrials.gov and enter "NCT04856982" in the "Other terms" search box. To learn more about Qalsody, visit QalsodyHCP.com.

First Gene Therapy Approved for DMD



After extending their review period, the FDA granted accelerated approval to SRP-9001 (ELEVIDYS), Sarepta Therapeutics' treatment for ambulatory patients ages 4 to 5 with Duchenne muscular dystrophy (DMD).

DMD is caused by mutations in the dystrophin gene on the X chromosome, which result in little to no production of dystrophin protein. This protein is necessary for maintaining the structural integrity of muscle cells. Progressive muscular degeneration and weakness are symptoms of DMD. Over time, these symptoms can affect heart and lung functions.

CLINICAL TRIAL TERMS TO KNOW



Double-blind:

Neither researchers nor participants know which participants are taking the drug or placebo.

Multiaim: Comparing several different experimental treatments against a common control group within a single study.

Multicenter: The trial is completed at more than one site.

Randomized: Participants are assigned at random to groups taking the drug or placebo.

ELEVIDYS is a gene replacement therapy administered via one-time intravenous infusion. By introducing a smaller dystrophin gene (mini-dystrophin) into the muscle tissue of boys with DMD, ELEVIDYS partially compensates for the absence of a functional dystrophin gene and addresses the underlying genetic defect.

The FDA granted accelerated approval primarily based on data that demonstrated

expression of micro-dystrophin in patients treated with ELEVIDYS. Sarepta has committed to completing the phase 3 EMBARK study as the post-marketing confirmatory clinical trial.

For more information on SRP-9001, visit ClinicalTrials.gov, and enter “NCT04626674” in the “Other terms” search box.

New approvals

FDA Approves Injectable gMG Treatment



The FDA approved Vyvgart Hytrulo for the treatment of generalized myasthenia gravis (gMG) in adults who are anti-acetylcholine receptor (AChR) antibody positive (ab+).

Abnormal antibodies interfere with nerve-muscle communication in MG, a chronic autoimmune illness that results in crippling and potentially fatal muscle weakening. Nearly 85% of people with MG progress to gMG, which affects muscles throughout the entire body, within 24 months.

Vyvgart Hytrulo is the first subcutaneous (under the skin) injectable treatment for gMG approved by the FDA. The FDA initially authorized Vyvgart for use in the US and Europe in 2021 as an intravenous (IV) infusion given by a medical practitioner over four weeks in an infusion facility. The updated formulation is now available for a single subcutaneous injection (1,008 mg fixed dose) at home or in a doctor’s office.

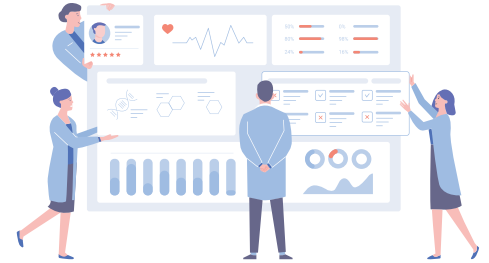
Until a few years ago, gMG had no approved therapy options, but now there are four. Vyvgart Hytrulo offers a new form of treatment and gives people with MG hope that more progress is to come.

Learn more about Vyvgart and Vyvgart Hytrulo at vyvgart.com.

Amyotrophic lateral sclerosis (ALS)

Phase 1/2 Study Enrolling

Researchers at Biogen are seeking individuals with ALS to participate in a phase 1/2 clinical trial (ALSPIRE) to evaluate the safety, efficacy, and pharmacological properties of the investigational therapy BIIB105 to treat ALS.



Most people with ALS have toxic clusters of a protein called TDP-43 in their brain and spinal cord cells. These clusters can lead to the death of cells in the brain and spinal cord. A protein called ataxin-2 (ATXN2) is thought to be involved in clumping TDP-43 proteins together, helping form these toxic TDP-43 clusters. The investigational drug, BIIB105, is designed to reduce the level of ataxin-2 protein in the brain and spinal cord cells, which may help reduce toxic TDP-43 clusters. Treatment with BIIB105 is being evaluated for the potential to reduce damage to motor neurons in the brain and spinal cord and ultimately slow down ALS disease progression.

In this phase 1/2 double-blind, randomized, placebo-controlled study, the drug will be administered by lumbar puncture into the spine. Participants enrolled in the study will participate for a duration of 29-41 weeks, with 19 clinic visits. This will be followed by an open-label extension (where all participants will receive the

investigational drug) of approximately 120 weeks, with 31 clinic visits.

Assistance with travel and accommodations and reimbursement for study-related expenses is available for eligible participants. Criteria to be eligible for the ALSPIRE study include:

- Having a laboratory-supported clinical diagnosis of ALS
- Having no known presence or family history of mutations in the superoxide dismutase 1 (*SOD1*) or fused in sarcoma (*FUS*) genes

- Meeting screening parameters for respiratory function, coagulation, etc., as set by study leads
- Having a caregiver who can provide accurate information about the participant's cognitive and functional abilities at screening

To learn more, visit AlspireStudy.com/MDA, or contact the study sponsors at the US Biogen Clinical Trial Center at **866-633-4636** or ClinicalTrials@biogen.com.

New Ways to Communicate

In the last four months, three communication devices designed for ALS patients have received "Breakthrough Device" designation from the FDA. The FDA grants breakthrough designation to transformative medical devices that may offer more effective diagnosis or treatment of life-threatening or irreversibly debilitating conditions. The goal of the program is to expedite the approval process to provide patients and healthcare providers with timely access to groundbreaking technologies.

Currently, the Breakthrough Device designation is held by fewer than 800 devices, of which about 140 are intended for neurological disorders and fewer are specific for ALS treatment. Conditions like ALS can cause severe motor impairment, affecting the ability of patients to communicate. Though many patients may have intact, highly active brains, some may struggle to speak or use a computer, and they may rely on gaze-tracking systems.

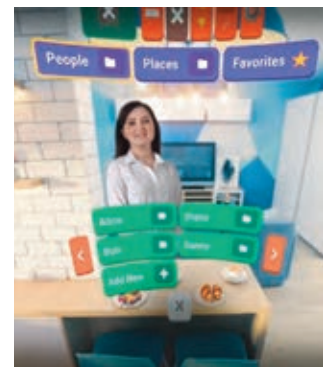
The FDA granted Breakthrough Device designation to the following products:

Speech Vitals - ALS: Developed by Aural Analytics, Inc., Speech Vitals — ALS is a software application that collects speech samples from patients via app-based tasks that measure speaking rate, articulation, and other metrics. These metrics are then analyzed and used to assess speech-motor conditions. The app can be deployed on any device, at-home or in-clinic, and can be embedded into provider and



healthcare system applications to integrate into a clinician's everyday practice. This app is available to participants of qualified clinical trials. Learn more at AuralAnalytics.com.

Cognixion ONE Axon: This wireless visor device was designed by Cognixion for people with ALS and other paralyzing conditions who can no longer communicate using standard gaze-tracking devices due to loss of eye muscle function. Using a brain-computer interface and integrating predictive language models powered by artificial intelligence (AI), Cognixion ONE provides suggestions of words in agreement with users' intentions. These suggestions can be selected and communicated via sound and visibly displayed on the visor, helping people with severe motor impairments communicate independently and interact with their surroundings. Learn more at one.cognixion.com.



Connexus Direct Data Interface (DDI): Paradromics's Connexus DDI is an assistive communication device that translates brain signals into speech and movement in real time, helping to restore social connection and enable independent patient engagement with technology. In addition to the Breakthrough Designation, Paradromics secured \$33 million in funding from Prime Movers Lab and additional investors to enable the development of the technology and launch the first-in-human clinical trial for the device. Learn more at paradromics.com.

Myasthenia gravis (MG)

Clinical Trial Seeks Participants



Researchers at Alexion AZ Rare Disease are seeking adults living with generalized myasthenia gravis (gMG) to participate in a phase 2 clinical trial (Expand Study) to evaluate the safety and efficacy of ALXN2050 (vemircopan) to treat gMG. Vemircopan is being evaluated for its ability to improve how easily people with gMG can perform activities of daily living.

In the double-blind study, participants will be randomly assigned to receive either vemircopan or an inactive placebo control. The total trial duration for each participant will be

approximately 125 weeks and will consist of the following periods: screening, primary evaluation, extended treatment, and open-label extension. Approximately 10 weekly visits will be required in the screening and primary evaluation period. Following the treatment period in which some participants may receive placebo, all participants will receive the investigational medication during an open-label extension period.

The drug/placebo will be administered orally (by mouth) twice a day. The effects of vemircopan will be evaluated using a number of tests and procedures, such as: medical history reviews, physical exams, vital sign measurements, electrocardiograms, and blood and urine sample collections.

Travel and other resources are available for eligible participants. Criteria to be eligible:

- Being at least 18 years old
- Having a confirmed diagnosis of myasthenia gravis (MG)
- Being on a stable dose of medication, if currently receiving treatment for MG

To learn more, visit [ExpandStudy.AlexionClinicalTrials.gov](https://www.expandstudy.com), or email the study coordinator at PatientAdvocacy@alexion.com.

Phase 3 Trial of Combination Therapy

Enrollment is ongoing at more than 100 sites in multiple countries for Regeneron's phase 3 NIMBLE trial assessing the investigational combination therapy of pozelimab and cemdisiran to treat adults with gMG. The trial is recruiting approximately 235 adults who have symptomatic gMG and present with abnormal antibodies in the blood that attack muscle cell proteins, specifically anti-acetylcholine receptor (anti-AChR) antibodies or anti-lipoprotein receptor-related protein-4 (anti-LRP4) antibodies. The combination therapy is designed to improve the connection between nerves and muscles in people with gMG, and is being evaluated for its ability to reduce muscle weakness.



During this randomized, double-blind trial, the drug or a placebo control will be administered subcutaneously (under the skin). The total trial duration for participants will be approximately 172 weeks, and participation will require regular clinic visits over the study period.

Criteria to be eligible to participate in the NIMBLE trial include:

- Being at least 18 years old

- Having a documented diagnosis of gMG with serologic tests or positive results during screening for anti-AChR antibodies or anti-LRP4 antibodies
- Having a myasthenia gravis activities of daily living (MG-ADL) score of ≥ 6 at screening

To learn more, visit [ClinicalTrials.Regeneron.com](https://ClinicalTrials.gov) and search for “generalized myasthenia gravis” in the “Condition” or “Keyword” field.

Spinal muscular atrophy (SMA)



Phase 3 Trial Seeks Participants

Researchers at Biohaven Pharmaceuticals, Inc., are seeking individuals living with SMA who are ambulatory (able to walk) or nonambulatory on a stable regimen of disease-modifying therapy to participate in a phase 3 clinical trial (RESILIENT). The trial will evaluate the safety and efficacy of the investigational drug taldefgrobep alfa to treat SMA. Taldefgrobep alfa is being developed as a potential therapy to improve muscle mass and function in people with SMA.

This study is a multicenter, randomized, double-blind, placebo-controlled study. It will be followed by an optional open-label extension in which all participants may receive the drug. The drug/placebo will be administered weekly by subcutaneous (under the skin) injection. The total study duration for participants will be 48 weeks and will include in-person clinic visits every 12 weeks.

Criteria to be eligible for the RESILIENT trial include:

- Having SMA confirmed by a genetic diagnosis of 5q-autosomal recessive SMA, as well as SMN2 copy number
- Receiving treatment with an SMA disease-modifying therapy, including risdiplam and/or nusinersen, and/or a history of onasemnogene abeparvovec-xioi, and remaining on that same treatment regimen throughout the trial

To learn more, email the study coordinator at Jackie.Marin@BiohavenPharma.com.

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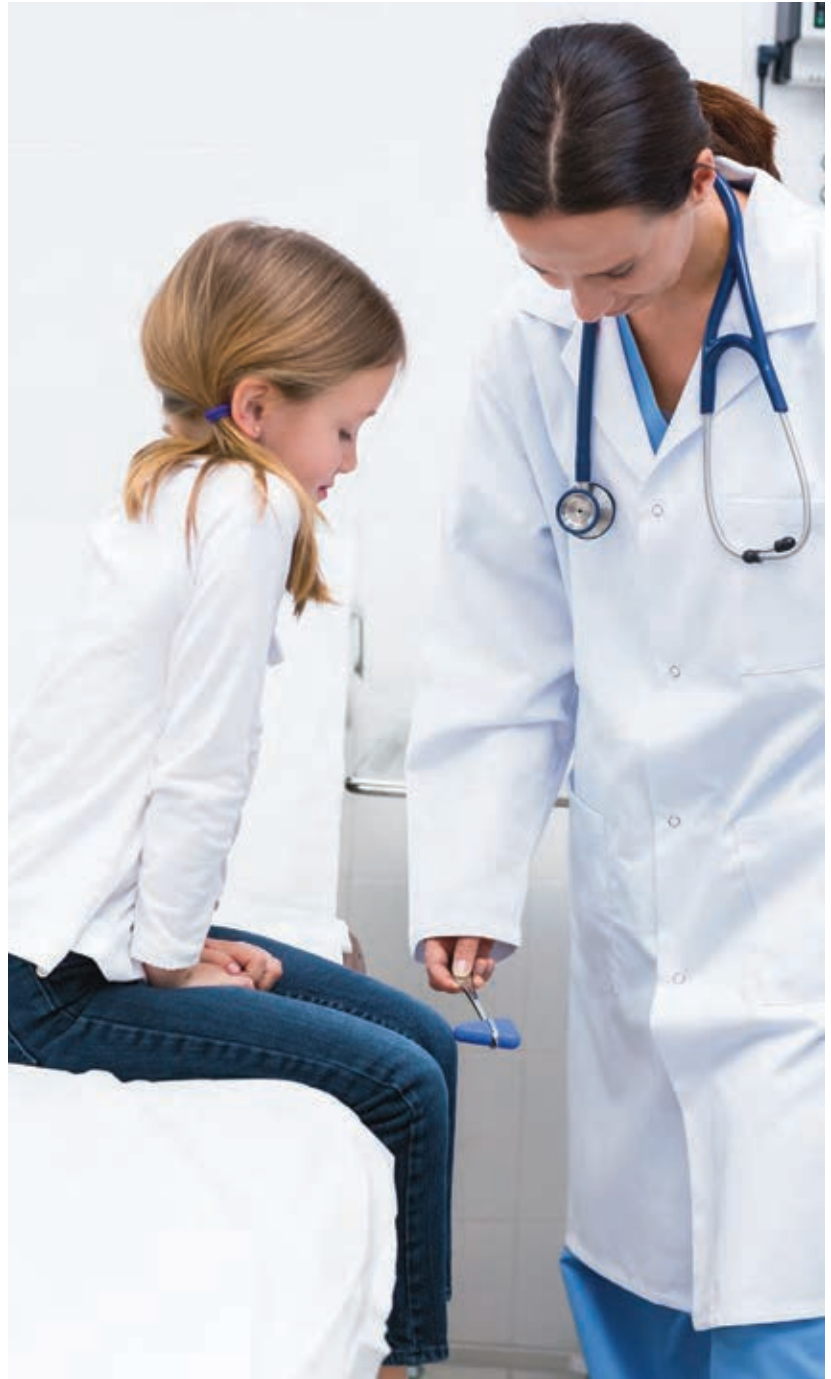
Advances in the Treatment of Friedreich Ataxia

A Q&A with Barry Byrne, MD, PhD, and David Lynch, MD, PhD

Friedreich ataxia (FRDA; sometimes called FA) is a neuromuscular disorder that affects about one in 50,000 people or about 8,000 people in the United States. The symptoms usually appear in childhood but can first appear in adults up to their 40s.

The primary symptoms include muscle weakness in the arms and legs, difficulty with balance and walking, and loss of sensation in the arms and legs, all of which grow worse over time. Many people with FRDA also experience heart problems.

Quest talked with two physicians who treat and conduct research into FRDA. Barry Byrne, MD, PhD, is the Associate Chair of Pediatrics and Director of the Powell Gene Therapy Center at the University of Florida. David Lynch, MD, PhD,



The primary symptoms include muscle weakness in the arms and legs, difficulty with balance and walking, and loss of sensation in the arms and legs.



DISEASES A-Z

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is Professor of Neurology at Perelman School of Medicine at the University of Pennsylvania and a neurologist at Children's Hospital of Philadelphia.

What are the first signs of FRDA?

Dr. Lynch: It usually starts with ataxia — meaning difficulty with balance — most commonly between the ages of 5 and 15. About 20% of people present after age 15, and maybe 1% or 2% before age 5. A few people are identified because they have scoliosis in their early teenage years. A small number of people are identified when they are very young because they have symptoms,

including chest pain and shortness of breath, indicative of hypertrophic cardiomyopathy, which is a thickening of the heart that makes it difficult to pump blood.

What causes the disorder?

Dr. Lynch: FRDA is a genetic disease. It's autosomal recessive, so each parent is a carrier of one frataxin (*FXN*) gene mutation, but the parents do not have the disease. A child gets one mutated gene from each parent, and the mutation leads to a failure to produce sufficient levels of the protein frataxin.

Dr. Byrne: Frataxin is important for the proper function of mitochondria, which produce energy for cells to function. The deficit in energy production means that some cells do not function normally and some cells die. When neurons die, they're not replaced. In the heart, muscle cells enlarge to compensate for the weakness that comes from the loss of frataxin. This leads to more severe complications in patients in their 20s and older, when they experience heart rhythm abnormalities and early mortality. Before that, many early-onset patients lose their ability to walk.

The US Food and Drug Administration (FDA) recently approved a new treatment for FRDA. What should people know about it?

Dr. Lynch: Omaveloxolone goes by the brand name Skyclarys. In double-blind clinical trials, this drug improved people's neurologic function and appeared to slow the progression of the disease. It's been approved for people ages 16 and older with FRDA.

Dr. Byrne: This drug is specifically designed to influence some of the metabolic abnormalities associated with the frataxin mutation and is intended to preserve the neurons' function. We expect the FDA to extend the treatment

to even younger individuals over the next few years.

Are there any new treatments on the horizon?

Dr. Byrne: There are some other approaches that are meant to increase the level of frataxin protein. They're in the very early stages, so there's not extensive data on those approaches yet. New drug therapies may have additional benefit when used in combination. The approach I am most excited about is to restore frataxin expression by gene therapy.

Dr. Lynch: One medicine, PTC 743, had some positive results recently in its clinical trial, but it didn't reach the primary endpoint. We will see whether it moves forward.

It was most effective at reducing fatigue, which is a big issue with FRDA. There is also a drug from Design Pharmaceuticals whose basis is to turn the defective gene back on so that people make normal levels of frataxin again. It's in early trials at this point, but it's very positive. We have good data to suggest that approach may be useful in the long-term.

How do people manage the challenges of living with FRDA?

Dr. Byrne: Besides the gait abnormality, patients may have secondary problems, such as difficulty with speaking and coordinating language. Some patients develop diabetes and problems with vision and hearing loss. It's a complex set of problems, but amazingly, the patient community with FRDA is extraordinary and extraordinarily resilient. They remain very optimistic about their current care and future options to improve their quality of life. [Q](#)



Barry Byrne, MD, PhD



David Lynch, MD, PhD

It usually starts with ataxia — meaning difficulty with balance — most commonly between the ages of 5 and 15.



LISTEN IN

The neuromuscular disease treatment landscape is constantly changing. Listen to a conversation with a neurologist about innovations and advances at [MDAQuest.org/podcast/neuromuscular-care](https://mdaquest.org/podcast/neuromuscular-care).

It's Time to Get Real about Becker Muscular Dystrophy

Health conditions, especially rare diseases, are not relative.

Each is difficult and complicated in its own right and affects individuals in uniquely hard ways. People living with Becker muscular dystrophy, or Becker, are no exception. Individuals with Becker often have the severity of their condition minimized relative to other neuromuscular diseases. For too long, individuals living with Becker, as well as their families and caregivers, have been de-prioritized and marginalized because the condition is mistakenly perceived as less severe than some other forms of muscular dystrophy. The reality is that Becker is a devastating disease.

Becker muscular dystrophy is a serious health condition.

Becker results in skeletal muscle damage, which drives muscle loss and disease progression resulting in difficulty in walking and doing everyday activities. Some individuals living with Becker also experience heart disease, which may lead to the need for a heart transplant or early death. Emerging data from studies that characterize the disease progression of Becker show that age does not predict when disease progression will begin, but the onset of muscle weakness does. Once muscles start to weaken, usually first experienced in the legs, the decline is irreversible.

"I was told 'you're lucky you don't have Duchenne.' It's frustrating that you live longer, but you are constantly going downhill, and you have to live with this long-term and the financial aspects that go along with that."

- individual living with Becker

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SPONSORED CONTENT

Because Becker is a genetic disease, the disease process begins at birth...

...although symptoms may not be immediately obvious. From a patient reported registry of individuals with Becker, the average age of symptom onset is 7 years old. As boys and men get older, Becker significantly impacts the skeletal muscles and the heart. In some cases respiratory function may be involved, and learning difficulties may be present. The impacts of Becker include muscle weakness, impaired mobility, fatigue, muscle cramps or pain, and may shorten an individual's lifespan.

It is time to recognize Becker muscular dystrophy as a distinct and serious health condition requiring active, ongoing management,

rather than a mild or less serious form of another disorder. Those affected by Becker are often in high school or college when challenges and health concerns arising from the diagnosis begin their relentless advance. Others may be husbands and fathers, caring and providing for their own families when their health concerns advance. As they face the myriad of mobility challenges and serious health concerns caused by Becker, it is unlikely that those living with the disease feel "fortunate" or "relieved" that they don't have a more serious form of muscular dystrophy.

We must all come together.

As patients, advocates, researchers, and healthcare providers in the neuromuscular space, **it is time to focus on the Becker muscular dystrophy community of children, young people, and adults who suffer from this disease.** It is time to recognize the need for Becker-specific standardized care guidelines including early diagnostics, focused advocacy, and patient and family support programs and services. Research is advancing and the community is learning more. Let's work together to ensure that those who live with the impact of Becker have the resources, education, and support to thrive.

For more information about Becker muscular dystrophy, please visit us at

**www.edgewisetx.com/patients-families/community-and-resources
or write us at info@edgewisetx.com**

Common Symptoms

of Becker Include:



Arm weakness



Difficulty walking that gets worse over time



Toe walking



Hard getting out of chair



Muscle pain and/or spasms



Frequent falls

Other Symptoms

of Becker Include:



Cardiomyopathy



Fatigue



Learning Differences



Breathing Problems

"There's a lot of things you gotta consider once you have to start to use mobility devices. You have to consider where you are going, if it's accessible, a lot of things you didn't have to consider before. So that's probably been the biggest hurdle for me recently... It's just one of those things you have to just learn to be okay with because sometimes the world is just not built for people that are disabled..."

- adult living with Becker



Healthcare That Supports Your Journey

You might want to bring in palliative care earlier than you think

BY EMILY BLUME



When you're preparing for a journey, you want to equip yourself with gear that will make your trip comfortable and help you make the most of your time. The same is true of a neuromuscular disease journey. Palliative care can be incredibly beneficial, but it is often overlooked on this type of journey.

What is palliative care?

Palliative care focuses on relieving and preventing symptoms, reducing stress, and helping people have the highest quality of life possible. The effort to relieve symptoms can range from treating pain and depression to addressing breathing difficulty or trouble sleeping. Palliative care can be, and often is, provided alongside

disease-directed or curative treatments for managing a neuromuscular disease.

Palliative care and hospice care

While the two types of care have much in common, they're not the same. Palliative care is the overarching umbrella under which hospice falls. Hospice care is a type of palliative care that focuses on quality of life in the final stages of life. However, palliative care is appropriate at any age and stage of a serious illness.

"We say that, in palliative care, the emphasis is on helping someone live as well as possible for as long as possible," explains Lauren Treat, MD, a child neurologist and hospice and palliative medicine doctor.

Hospice can come into play when patients want to prioritize comfort-focused above life-prolonging therapies. Sometimes this shift in philosophy occurs because of how the disease progresses or because the side effects of disease-directed treatments become too much.

Don't delay care

You may start palliative care immediately following a neuromuscular disease diagnosis, or if there is a significant shift in disease progression or symptoms. Studies show patients with appropriate pain and symptom management tend to live longer and enjoy a better quality of life than those without. In addition, palliative care can help normalize the involvement and support families will provide as they face a daunting road ahead.

Dr. Treat compares using palliative care to wearing a seat belt — it doesn't make you more or less likely to reach your destination, but it does make you feel more secure on the road and more capable of handling bumps and turns along the way.

Usually, referrals for palliative care don't come soon enough, and families miss out on months, or even years,

of support from a palliative medicine team.

Experts suggest asking your neuromuscular disease specialist — or any doctor you see regularly — if you might benefit from palliative care. Doctors may hesitate to bring it up because of misconceptions about palliative care.

“Patients get very worried that someone is ‘giving up on them,’ and that’s not the case,” explains Dr. Treat. Palliative care can coincide with pursuing disease-directed therapies for a neuromuscular disease. It is intended to augment treatment and help an individual’s care team



“We say that, in palliative care, the emphasis is on helping someone live as well as possible for as long as possible.”

—Lauren Treat, MD

	PALLIATIVE	HOSPICE
TIME FRAME	Any time after diagnosis	Final stages of life
TREATMENT	Symptom management and quality of life in conjunction with disease-directed treatment of illness	Complex system management and quality of life (no disease-directed treatment)
LOCATION	In healthcare facilities or at home	In healthcare facilities or at home
CARE TEAM	Doctors, nurses, pharmacists, therapists, social workers, dietitians, and chaplains	Doctors, nurses, therapists, social workers, dietitians, chaplains, and volunteers
INSURANCE	Medicare, Medicaid, and many private insurers cover some services.	Medicare covers. Medicaid coverage varies by state. Many private insurers provide some coverage.



“I always hope we will preserve people’s joy and happiness, balancing safety and comfort.”

—Ambereen Mehta, MD, MPH

understand what’s most important to them and their family. It’s not indicative that people are giving up or think the family is making a wrong decision. If an individual or family member worries about this, Dr. Treat encourages them to speak with their care team about palliative care and how it fits into their treatment plan.

Turning to hospice

Because neuromuscular diseases are progressive, they could involve a long period of palliative care before transitioning to hospice care when the disease reaches an advanced stage. This time usually comes when disease-directed therapies or procedures no longer fit

an individual’s goals and values for quality of life. “Then, we invoke a conversation about what would it be like to focus purely on comfort and help that person get as many good days as possible for as long as possible,” Dr. Treat says.

The timetable for hospice care can vary, but for insurance coverage, it begins in what is expected to be the last six months of life.

Family benefits

Palliative care emphasizes caring for the family unit, including everyone affected by the illness.

Ambereen Mehta, MD, MPH, Associate Professor of Palliative Medicine at the Johns Hopkins School of Medicine, works closely with patients with amyotrophic lateral sclerosis (ALS), so she knows that palliative care can be as crucial for the family as it is for the patient.

Palliative care providers can help families prepare for the changes the disease will bring, particularly for those who take on caregiving responsibilities. This help could include breaks for the caregiver or ensuring family members get quality time with their loved one. Finally, providers can help family members understand their loved one’s wishes and care goals, even those that might be difficult, such as a choice to not prolong life by artificial means.

Dr. Treat says the families receiving palliative care are often its greatest champions. “So many families come to my clinic and say, ‘I should have been doing this earlier,’ or, ‘It’s been so meaningful to have the support for some challenging times in my life.’”

While palliative care and hospice care differ in where they enter the neuromuscular disease journey, they share the goal of giving individuals as much time as possible while living as well as possible — and with dignity. “I always hope we will preserve people’s joy and happiness, balancing safety and comfort,” Dr. Mehta says. [@](#)

Emily Blume is a journalist living with myotonic dystrophy (DM) in Washington State.



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What is SKYCLARYS?

SKYCLARYS is used for the treatment of Friedreich ataxia in adults and children 16 years of age and older.

It is not known if SKYCLARYS is safe and effective for use in children younger than 16 years of age.

Before taking SKYCLARYS, tell your healthcare provider about all of your medical conditions, including if you:

- ▶ have liver problems.
- ▶ have a history of heart problems, including heart failure.
- ▶ have a high level of fat in your blood (high blood cholesterol).
- ▶ are pregnant or plan to become pregnant.
 - It is not known if SKYCLARYS will harm your unborn baby.
 - Women who use hormonal birth control should use another form of birth control such as a non-hormonal intrauterine system or an extra non-hormonal birth control such as condoms while using SKYCLARYS and for 28 days after stopping SKYCLARYS.
- ▶ are breastfeeding or plan to breastfeed. It is not known if SKYCLARYS passes into your breast milk. Talk to your healthcare provider about the best way to feed your baby if you take SKYCLARYS.

Tell your healthcare provider about all the medicines you take, including prescription and over-the-counter medicines, vitamins, and herbal supplements such as St. John's Wort.

Taking SKYCLARYS with other medicines can cause serious side effects.

SKYCLARYS may affect the way other medicines work, and other medicines may affect how SKYCLARYS works.

Know the medicines you take. Keep a list of them to show to your healthcare provider and pharmacist when you get a new medicine.

How should I take SKYCLARYS?

- ▶ Take SKYCLARYS exactly as your healthcare provider tells you to take it.
- ▶ Take SKYCLARYS capsules on an empty stomach at least 1 hour before eating.
- ▶ Swallow SKYCLARYS capsules whole. Do not open, crush, or chew.
- ▶ If you miss a dose, then you should skip the missed dose and take the next dose at the regular time the next day. Do not double your next dose or take more than the prescribed dose.

What should I avoid while taking SKYCLARYS?

Do not drink grapefruit juice or eat grapefruit. These may change the amount of SKYCLARYS in your blood.

What are the possible side effects of SKYCLARYS?

SKYCLARYS may cause serious side effects, including:

- ▶ **increase in blood liver enzymes.** Some people taking SKYCLARYS have had an increase in the level of liver enzymes in their blood. Your healthcare provider will do liver function tests

- before you start taking SKYCLARYS
- every month for the first 3 months after starting your treatment with SKYCLARYS
- during certain times as needed while taking SKYCLARYS

If your liver enzymes increase, your healthcare provider may change your dose during treatment, stop treatment for some time, or completely stop treatment with SKYCLARYS.

- ▶ **increase in a blood protein called B-Type Natriuretic Peptide (BNP).** BNP tells how well your heart is working. Your healthcare provider will check your BNP levels before your treatment with SKYCLARYS. Tell your healthcare provider if you have signs and symptoms of your heart not working well such as too much fluid in your body (fluid overload). Signs and symptoms may include:

- sudden weight gain (3 pounds or more of weight gain in 1 day, or 5 pounds or more of weight gain in 1 week)
- swelling in your arms, hands, legs, or feet (peripheral edema)
- fast heartbeat (palpitations)
- shortness of breath

If you have symptoms of fluid overload that is considered a side effect of SKYCLARYS, your healthcare provider may stop treatment with SKYCLARYS.

- ▶ **changes in cholesterol levels.** Increases in low density lipoprotein cholesterol (LDL-C) or bad cholesterol and decreases in high density lipoprotein cholesterol (HDL-C) or good cholesterol have happened during treatment with SKYCLARYS.

Your healthcare provider will check your cholesterol levels before and during your treatment with SKYCLARYS.

The most common side effects of SKYCLARYS include:

- ▶ increased liver enzymes (ALT/AST)
- ▶ headache
- ▶ nausea
- ▶ stomach pain
- ▶ tiredness
- ▶ diarrhea
- ▶ muscle pain

These are not all the possible side effects of SKYCLARYS. Call your doctor for medical advice about side effects.

You are encouraged to report negative side effects of prescription drugs to the FDA. Visit www.fda.gov/medwatch or call 1-800-FDA-1088. You may also report side effects to Reata Pharmaceuticals, Inc., at 1-800-314-3934.

General information about the safe and effective use of SKYCLARYS

Medicines are sometimes prescribed for purposes other than those listed here. Do not use SKYCLARYS for a condition for which it was not prescribed. Do not give SKYCLARYS to other people, even if they have the same symptoms you have. It may harm them.

For additional information about SKYCLARYS, the full Prescribing Information, and the Patient Product Information, go to [SKYCLARYS.com](https://www.SKYCLARYS.com).

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The Buddy System

How to develop your child's ability to make friends in school

BY CLAIRE SYKES

Making friends in school isn't always easy for kids at any age, especially those with neuromuscular diseases and physical disabilities. But with the right mindset and some guidance, they can form healthy and fulfilling social ties.

The benefits of buddies

Positive peer relationships offer emotional support and a sense of belonging, and help boost self-esteem and social and academic skills. They also can lessen negative thoughts, anxieties, depression, and loneliness, and shield against bullying. Plus, they're just plain fun.





Josh LyBrand (right) with his family, including Ethan (center)

“If I’m having a rough day, I can tell a friend, and that makes my day a lot better.”

—Ethan LyBrand

“You need friends you can talk to and not feel judged, who are on your side and there for you,” says Josh LyBrand of Decatur, Alabama, a special education middle school teacher. His son, Ethan, 14, lives with Duchenne muscular dystrophy (DMD). “If I’m having a rough day, I can tell a friend, and that makes my day a lot better,” Ethan says. The kids he confides in also give him confidence. “They don’t see my wheelchair first. This makes me act more positively, and then I share that positivity with others,” he says.

Friends are also important for Holly Szymczak, 15, and her brother Josh, 18, who live with limb-girdle



Abby Loch (center) with her parents

muscular dystrophy (LGMD) near Milwaukee, Wisconsin. “My friends push me in my chair and give me treats,” Holly says. Josh also gets help from friends: “They help me carry my backpack. They care about me,” he says. Their mother, Denise, adds, “Their friends include them in activities and help them if they see them struggling.”

Overcoming obstacles

Areti Vassilopoulos, PhD, a pediatric psychologist at the MDA Care Center at Yale School of Medicine, points out that growing up with a neuromuscular disease presents unique challenges that can affect kids’ abilities and opportunities to make friends. “Most kids progress in a typically expansive way, while those with neuromuscular diseases can face increasing social challenges,” she says. “Medical appointments and hospital stays can make them miss school and fall behind.”



Siblings Holly (left) and Josh (right) Szymczak with their parents

Photo: Meg McKenzie

In some cases, developmental delays play a role. Abby Loch, 16, who is a manifesting carrier of DMD, is “very chatty, fun, and loving,” says her mother, Melissa, of Apple Valley, Minnesota. “But the social and academic gap between her and her peers has gotten larger. It’s been hard for her to find true friends who are nonjudgmental and meet her where she is.”

The degree of difficulty for a kid to make friends “depends on the type and severity of their disease, their personality, how they view friendships and the challenges with them,” says Natalie Truba, PhD, a psychologist at the MDA Care Center at Nationwide Children’s Hospital in Columbus, Ohio.

A child’s disease is out of your control, but you can influence their perceptions of it and how it impacts them. Techniques to help a child make friends vary with their

age and stage in life. Here is a guide to adjusting your approach as they progress through school, keeping in mind that each child develops at their own rate.

Elementary school: Build a foundation

“There’s so much adult scaffolding in elementary school,” Dr. Truba says. “Prescribing friendships to kids and teaching them the skills to be good humans are two

“My friends push me in my chair and give me treats.”

—Holly Szymczak



SCHOOL SUPPORT

Learn about resources that will help you, your child, and your school navigate the accommodations and supports your child needs at [MDAQuest.org/navigating-school-supports-with-mda](https://www.mdaquest.org/navigating-school-supports-with-mda).



MAKE SCHOOL DAYS EASIER

These programs and resources help kids and families navigate school and make friends.

Access Workshops

Access to Education: K-12 is an on-demand virtual workshop covering common considerations for K-12 educational planning, including 504 plans and IEPs. This is one in a series of workshops addressing access barriers at mda.org/AccessWorkshops.

K-12 Education Resources

MDA's print-ready educational materials include guides to teach educators about neuromuscular diseases and an overview of the IEP process. Visit mda.org/education and click on "K-12 Education Resources."

Let's Play

Join MDA's gaming community for a fun, interactive way to get to know others in an inclusive environment. To learn more or join one of our weekly game-a-thon events, visit mda.org/lets-play.

National Resource Center

Get support finding information and resources about neuromuscular diseases, services, activities, and more. Resource Center staff are available Monday to Friday, 9 a.m. to 5 p.m. CT, and are typically able to answer questions within one to two business days. Call **833-ASK-MDA1** or email ResourceCenter@mdaUSA.org.

Peer Connections

This program helps folks in the MDA community — including individuals living with neuromuscular disease, caregivers, parents, spouses, and siblings — build connections with each other. To request a connection, contact MDA's Resource Center at **833-ASK-MDA1** or ResourceCenter@mdaUSA.org.

important aspects of childhood learning that are carried into adulthood."

Experts recommend starting early, taking children to playgrounds and inviting their friends to playdates. Together, read books about friendship and role-play social situations, such as asking someone to play or share a toy.

Middle school: Let them figure it out

Get middle-schoolers involved in after-school activities, clubs, and MDA Summer Camp to meet other kids. They may struggle with rejection, conflict, and awkwardness at times, but don't be too quick to rescue them. "Children need to learn on their own, or they could be less prepared to navigate friendships as adults," Dr. Truba says. "Instead, help them cultivate the attitude that building new skills may feel uncomfortable for a while but will grow easier with practice."

You can remain involved by keeping an open dialogue with your child. "Truly listen so they really feel heard before you offer advice," Dr. Vassilopoulos says.

By middle school, kids turn more to their peers than parents, but they may still need your support to handle others' curiosity about their differences. Dr. Vassilopoulos suggests they practice a response such as, "I have trouble moving my legs and arms, and I see doctors for that," or "It's hard for me to talk about it, but you can look up 'muscular dystrophy' on the internet."

When asked about his wheelchair, Ethan simply says, "My legs don't work like they used to," and that works.



Areti Vassilopoulos, PhD



Natalie Truba, PhD

High school: Follow their interests

Older kids' hobbies and pursuits deepen and change. Support them. For example, Josh has made friends in school by joining the golf team and finding other kids who enjoy going to movies.

In this stage, social dynamics also become more complex as kids solidify their own identities and give friendships greater importance. "Kids are learning more



from experience about navigating conflict, fitting in, and falling out of peer groups,” Dr. Vassilopoulos says. They may need adults less, but you can always be there for them.

10 tips for making friends

Experts and MDA community members offer their top tips to make friends:

- 1. Love yourself.** That’s the advice Abby offers younger kids. “It took me a long time to get where I am with that,” she says. “Now, when I look in the mirror, I think of four things I love about myself.”
- 2. Welcome others.** “Try not to let fear of rejection get in the way,” Dr. Vassilopoulos says. Instead, be curious, open, and willing to reach out to others. Holly starts with a simple greeting. “I say hi first, and then they say it back,” she says.
- 3. Share similar interests.** Holly has made friends in her ballet class and bowling. Ethan enjoys social video games and acting in local children’s theater plays.
- 4. Be yourself.** “I don’t try to change how I act to fit in. If you’re true to yourself, you’ll have more true friends,” Ethan says.
- 5. Ask questions.** When Abby talks about a new friend, her parents encourage her to think through some questions: How does this person make me feel? Why do I want them as a friend? “We’re helping to foster positive friends for her and steer her from those who aren’t,” Melissa says.
- 6. Choose wisely.** A good friend is open, honest, and responsive. “We explain to Ethan to notice how people also treat others,” Josh says. Ethan adds, “If they aren’t just as positive and kind to others, hanging out with them isn’t worth it to me.”



Experts recommend starting early, **taking them to playgrounds** and inviting other kids over for playdates. Together, read books about friendship and role-play social situations.

- 7. Be a good friend.** “I’m kind and respectful and there for the people around me,” says Ethan. For Abby, “being that ‘shoulder’ for someone makes me happy knowing I’m making someone else happy,” she says.
- 8. Sharpen your social skills.** Dr. Truba suggests kids attend MDA Summer Camp or volunteer somewhere like a nursing home. This provides practice in a welcoming setting.
- 9. Know what’s right for you.** We don’t all need the same level of social interaction. “Some kids want to see multiple friends many times a week, and for others, a couple of friends once a month is enough,” Dr. Vassilopoulos says.
- 10. Accept change.** At all stages of life, friendships have their ups and downs. “Whether for a reason, a season, or a lifetime, all friendships are valuable,” Dr. Vassilopoulos says. Melissa guided Abby in this when she felt sad about a friend who got a boyfriend and had less time for her. “I explained to Abby that she can be supportive, and it can still be a good friendship,” she says. And, there’s always the possibility of making a new friend.

Words of wisdom

Adults pass on their wisdom, but kids have some, too.

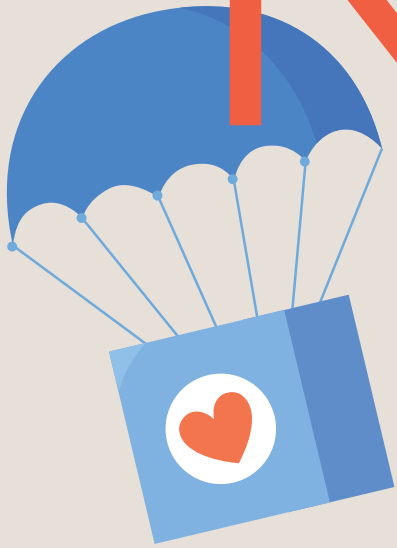
Josh offers timeless advice: “It’s what’s on the inside that counts when you make friends, not the outside,” he says. And we can all learn from Ethan: “Go with the flow, be kind to everybody, and try your best,” he says. “That’s what will get you through life and help you have healthy relationships.”

+MORE THAN FUN

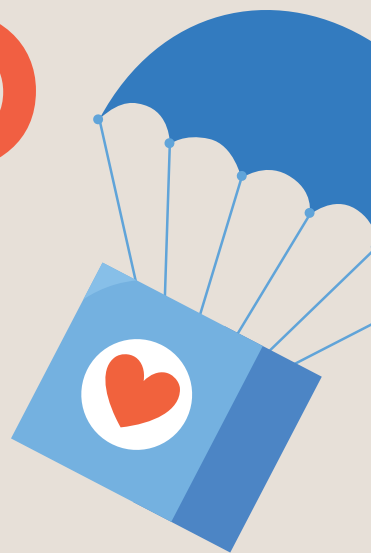
Participating in extracurricular activities helps kids build skills and make friends while having fun. Read about how to get kids involved at [MDAQuest.org/after-school-activities-for-all](https://mdaquest.org/after-school-activities-for-all).

Claire Sykes is a freelance writer in Portland, Oregon, who covers health and the human side of science.

Need a



Hand? Just Ask



*Overcome
embarrassment
with asking for
help in daily life*

BY ANDREW
ZALESKI



For Jess Hetzel, a 22-year-old living with spinal muscular atrophy (SMA), getting assistance on everyday tasks is a normal part of life. Her friends and family know she might need a hand using the bathroom or opening a door. It's so ordinary for Jess that now she tells people she's meeting for the first time that they shouldn't be alarmed by her power wheelchair and need not feel awkward if she asks for a drink of water.

"I prefer to tell people what I need at the beginning of the friendship. It makes my life easier and less stressful when I'm hanging out with them," she says.

But Jess wasn't always this forward. As a teenager, she struggled when it came to asking people for help and recalls not enjoying going to parties or eating out with friends.



Jess Hetzel explains what she needs at the beginning of a friendship.

“The fact that I needed help to eat was just very embarrassing for me,” Jess says. “I had to rework my mindset and how I view disability and my life in general. And practice reminding myself that I am not a burden.”

We all need help

Needing help is a common experience for everybody, but it can take on a different texture for people living with neuromuscular diseases. Being embarrassed by or uncomfortable with requiring assistance to navigate social situations can be emotionally difficult. Even the most basic tasks can leave one feeling like they’re a problem.

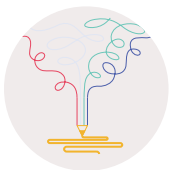
After I was diagnosed with myotonic dystrophy (DM) at age 32, I felt sheepish asking others to open bottles for me when my hands were excessively cramped, a common symptom of DM. I started moving my hands more slowly to prevent cramps and make sure I could unscrew the cap on my own.

“A lot of people will just choose not to do the activity because they don’t want to have to ask for help,” says Brooke Aarvig, OTR, CBIS, an occupational therapist (OT) in Austin, Texas, who works with people who have a variety of neurological and neuromuscular conditions, including muscular dystrophy.

Yet overcoming the uneasiness and guilt sometimes associated with receiving help is an important skill for people living with neuromuscular diseases. And learning how to be appropriately assertive while teaching others — family members, friends, coworkers, and, in some cases, strangers — how to lend a hand is key for navigating daily life.

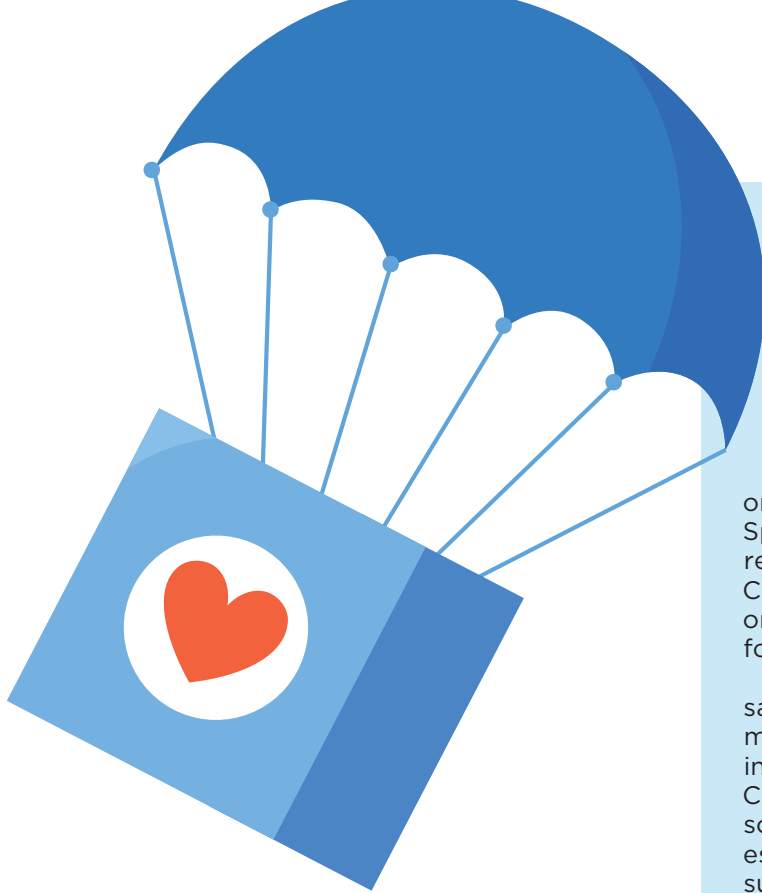
“You just have to get used to it because it will become a big part of your life,” says AJ Likhatskaya, a 17-year-old entering his senior year of high school.

He uses a power wheelchair, like Jess, due to SMA, and he knows some mundane tasks, like pulling open a door, are impossible for him. From an early age, his parents encouraged him to ask for help, and he has benefitted from a supportive group of friends who give him a boost



12 WAYS TO ASK FOR HELP

Not sure how to respond when a friend asks how they can help? Find a dozen ideas at [MDAQuest.org/12-ways-to-ask-for-help](https://www.mdaquest.org/12-ways-to-ask-for-help).



HERE TO HELP: MDA CONNECT

The MDA Connect program makes it easy to ask for help as you navigate your neuromuscular disease journey. Members of the neuromuscular disease community — including parents and caregivers — can schedule 30-minute, one-on-one video calls with MDA Support Specialists. Our specialists can help locate resources, navigate care within MDA's Care Center Network, and share information on MDA programs and opportunities for engagement.

"Putting a face to a name is invaluable," says Crystal, who is living with mitochondrial myopathy. She used MDA Connect to get information about her local MDA Care Centers. "It makes a difference to have someone see your expressions and emotions, especially if you are discussing a heavy topic, such as a progression in your diagnosis."

Simply visit mda.org/connect, and select a specialist, date, and time. You'll be asked a few questions so the specialist can better serve your personal needs during your meeting. After scheduling, you will receive a confirmation email and a meeting link for your scheduled appointment.



"A lot of people will just choose not to do the activity because they don't want to have to ask for help."

*—Brooke Aarvig,
OTR, CBIS*

General Hospital's Think:Kids program.

"Ask very clearly, in words that anyone would understand," she says. "Clarifying a situation by making your need clear is more likely to get someone to behave in the way you want them to."

when he needs it. An effective method he employs is dishing out in-the-moment tips on what his buddies can do to make activities easier for him.

For example, before he had surgery for scoliosis, AJ had a hard time holding up his head. His friends jumped in during those moments.

"I would give them directional cues," he says. "'Move my head more to the left, move it to the right.' I've realized that's the easiest way for people to understand."

Being direct is a great way to communicate immediate needs, according to Alisha Pollastri, PhD, a clinical child psychologist, as well as the director of research and evaluation at Massachusetts

When possible, Dr. Pollastri recommends having a conversation about the help you need upfront. That might mean taking time before hanging out with a friend or visiting a family member to explain what help you might need and when you'll need it. After all, it might be the case that others want to help but are unsure about when or how to step in.

"Often, what gets in the way and makes things awkward is not knowing what to do," says Dr. Pollastri. "It activates those feelings of guilt or embarrassment."

I've found that if I tell my wife or other family members what to expect, it's much easier for me to just be myself. I let them know when I'm in pain or when my hands aren't working properly, and that's when they know to step in to help me do something, even if it's as simple as opening a bottle of water.

Pushing past the awkwardness is something Jess learned to do while attending MDA Summer Camp. She recalls the counselors telling campers that there was nothing burdensome about their needing help to go to the bathroom, eat and drink, or use their medical equipment.

High school student AJ Likhatskaya practiced asking friends for help from an early age.



AJ Likhatskaya finds it best to give clear instructions when asking for help.

“I would give them directional cues. Move my head more to the left, move it to the right.’ I’ve realized that’s the easiest way for people to understand.”

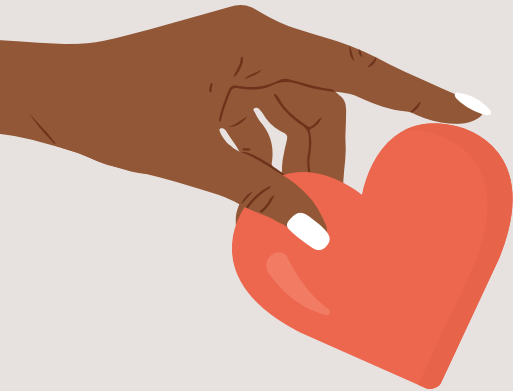
—AJ Likhatskaya

“That was the first time people started talking about the fact that they’re there to help me, and it’s not really a big deal,” Jess says.

It was also a growing experience for her brother, who’s four years older and does not have a neuromuscular condition. He was a counselor at Summer Camp for five years, starting at age 16, and Jess says the experience opened his eyes.

“The years before he went to camp, he didn’t really understand the help I needed,” she says. “But after he

Learning how to be appropriately assertive while teaching others how to lend a hand is key for navigating daily life.



saw a different perspective, he noticed what I needed and was super accommodating.”

Learn by doing

Becoming comfortable asking for help takes practice.

As an OT, Brooke coaches her patients through social exercises. That means, for example, going out somewhere and working on saying hello to people and making eye contact. Taking the time to deliberately insert yourself into social situations can not only help you learn how to ask for help; it can also demonstrate to others how to offer support. Sometimes the activity is as simple as asking a stranger to please open a door.

“I think if you can practice it enough times, it becomes a new norm, and it becomes a little more comfortable,” Brooke says.

This is something Jess accomplished over time, first at MDA Summer Camp, and then as a student at Central Michigan University. She will graduate in December 2023 with a major in public relations and a double minor in event management and disability studies — a line of study that changed her own perspective on living with a disability. Courses that dove into how living in an often-inaccessible society leads those with disabilities to feel burdensome helped her recognize and let go of her internalized feelings of guilt. Jess grew more comfortable and more assertive in asking for help.

“It’s not a big deal to ask. It’s not embarrassing. It’s just stigmatized — but that doesn’t mean it’s true,” she says. “You’re not the problem, and your body is not defective or faulty. Remind yourself of that.” [Q](#)



“Ask very clearly, in words that anyone would understand. Clarifying a situation by making your need clear is more likely to get someone to behave in the way you want them to.”

—Alisha Pollastri, PhD

Andrew Zaleski is a journalist who lives near Washington, DC, and wrote about his experiences with type 1 myotonic dystrophy for GQ magazine in February 2023.

Expect the Unexpected



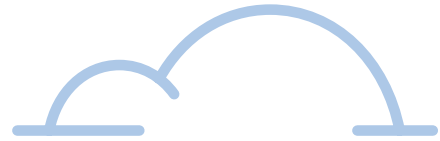
*Set yourself up for success
in a medical emergency*

BY DARLENE DEMETRI

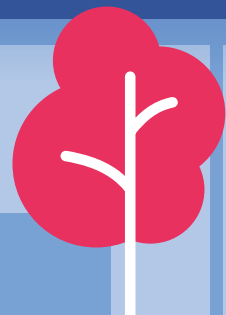
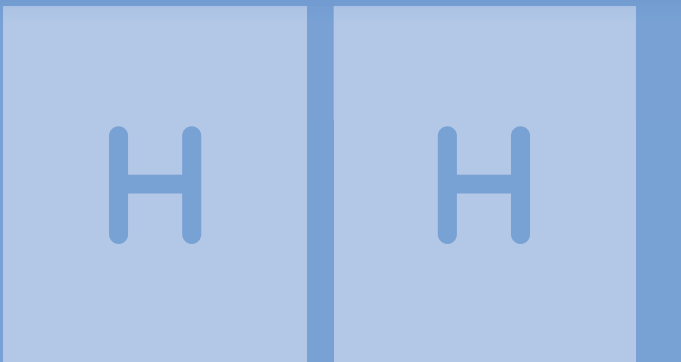
Sometimes, Crystal Killian, who lives with mitochondrial myopathy (MM), suddenly runs out of energy. When she experiences a severe “mito crash,” her nerves and muscles don’t work, her coordination is off, and she’s unable to communicate. Going to the emergency room (ER) in this state puts her in a difficult position because the symptoms look a lot like a stroke, but stroke medications won’t help her — they’ll cause serious complications.

The first responders and ER nurses and physicians who treat Crystal are in a difficult position, too. They generally haven’t had any training on mitochondrial myopathy and may have never encountered a patient with this rare neuromuscular disease. They’re trying to provide the best care they can; they want to help, not harm her.

When you live with a neuromuscular disease, it can be challenging to get appropriate emergency medical care. Taking some smart steps to prepare for a medical



EMERGENCY



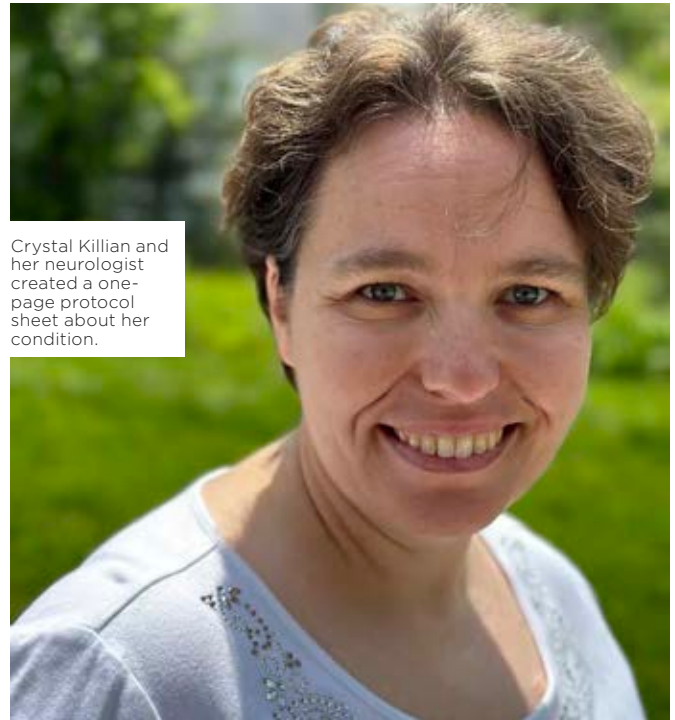
emergency can make the situation better for everyone involved.

Backup plans

Crystal, 41, and her partner, Michael, live in a Philadelphia suburb close to their community hospital. “I always plan for the worst and hope for the best,” Crystal says.

After what she calls her “diagnosis odyssey” in her 20s, Crystal has visited the ER many times. Her medical history is in the hospital’s electronic records, but medical staff members don’t always look at it.

As a backup, Crystal and her neurologist at the MDA/ALS Care Center at Temple University Hospital in Philadelphia, created



a one-page protocol sheet printed on the Care Center’s letterhead that provides first responders and ER staff with this critical information:

- A brief explanation of MM
- How to treat dystonia (muscle contractions that cause repetitive or twisting movements), a common complication of MM
- Special considerations for administering anesthesia
- A list of her essential medications and potential adverse interactions
- Her neurologist’s contact information

Crystal keeps one copy of the protocol sheet on her refrigerator and another in her medication bag. She always carries a copy if Michael is not with her.

She also stores the respiratory settings for her non-invasive ventilation device and her respiratory therapist’s phone number on her smartphone. Having a photo of her ventilator settings is helpful, too.

Life-saving documents

Lisa Wolfe, MD, Professor of Pulmonary and Critical Care Medicine and Neurology at Northwestern Medicine in Chicago, has seen many people with neuromuscular diseases come to ERs with breathing difficulties. She recommends the International Ventilator Users Network’s (IVUN’s) ‘Take Charge, Not



Chances' (ventnews.org/tcnc) as a resource. Ventilator users and caregivers can download this set of checklists and information sheets, fill out their information in advance, and share them with first responders and ER personnel.

"With the completed forms, everything is set up and ready to go in an emergency," Dr. Wolfe says.

She advises keeping hard copies on hand and asking your physician to manually enter the completed forms in your hospital's electronic records system. That way, "not only are they readily available, but it indicates that your physician has signed off on them," she says.

MDA's Emergency Care Resources (mda.org/education) include Emergency Room Alert Cards for amyotrophic lateral sclerosis (ALS), Duchenne muscular dystrophy (DMD), and myasthenia gravis (MG), and a general Emergency Room Alert Summary. These are designed to be printed, filled out, and carried with you. They include vital information, such as special care for fractures and cardiac precautions, that can help emergency care providers treat you safely.

When you need an ambulance

Before an emergency occurs, it's a good idea to reach out to your local fire department or ambulance service to let them know about your situation and how they can get into your house if you can't open the door.

During the day, Crystal feels safe leaving her front door unlocked so first responders can enter her home. Two trusted neighbors know where her spare key, protocol sheet, and rescue meds are located. If her neighbors see an ambulance or first responders attempting to get into her home when the doors are locked, they would know what to do.

When she goes to the ER in an ambulance, Crystal always asks the first responders to bring her manual wheelchair and noninvasive ventilator to the hospital. The manual wheelchair is easier and safer to use when she's given an anti-spasmodic for her cramped muscles. The noninvasive ventilators at the hospital are uncomfortable, so she prefers her own device and mask. The first responders usually comply with these requests, but if they don't, her caregiver or a family member brings them to the hospital as soon as they can.

Navigating the ER

Dr. Wolfe recommends requesting the hospital's neurologist as soon as possible after entering the ER. Not all neurologists are well-versed in neuromuscular diseases, so it's also a good idea to alert your own neurologist or

RESOURCES FOR A MEDICAL EMERGENCY

Emergency Care Resources:

MDA's emergency resources tailored to the neuromuscular disease community include Emergency Room Alert Cards designed to be printed, filled out, and carried with you. Find them at mda.org/education.

Access to Medical Care Workshop:

Part of MDA's Access Workshops series, this self-paced digital workshop covers navigating the healthcare system and educating providers about your neuromuscular disease. Find it at mda.org/AccessWorkshops.

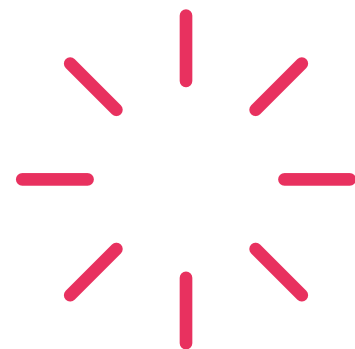
Virtual Learning Programs: MDA's educational webinars cover a variety of topics, including Preparing for Emergencies. Visit mda.org/virtual-learning.



neuromuscular disease specialist. Together, they should give input to the ER team.

James Naprawa, MD, is a pediatric emergency medicine physician at UCSF Benioff Children's Hospital in Oakland. He co-authored a 2018 publication with the American Academy of Pediatrics on ER management of patients with DMD.

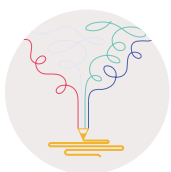
Dr. Naprawa recommends patients and parents take a tactful approach to avoid putting ER personnel on the defensive. Even though a medical emergency is a high-stress situation, try to remain calm and start the





“In a practical sense, there’s too much information out there, and although guidelines exist for neuromuscular disease emergency care, a busy ER may not have time to find the information.”

—James Naprawa, MD



CRITICAL CARE

Learn more ways to prepare before a medical emergency occurs in the Quest article “Be Prepared” at [MDAQuest.org/be-prepared](https://www.mdaquest.org/be-prepared).

conversation from a perspective of wanting to help, such as, “Here’s what my child’s specialist has advised me to do when I bring him to the ER.”

If the patient or caregiver feels the staff is not listening to their concerns, it’s time to use your self-advocacy skills. Providers who are not trained in a neuromuscular diseases may not be aware of special concerns with common interventions.

For example, for a child with DMD having breathing trouble, providing supplemental oxygen without positive-pressure ventilation “can be very dangerous as it can decrease the respiratory drive and cause them to retain carbon dioxide,” Dr. Naprawa says. People with DMD should be given oxygen in combination with non-invasive positive-pressure

ventilation (such as BiPAP) and close monitoring of carbon dioxide levels.

Most ER providers are willing to consult with your or your child’s specialists, and many MDA Care Center specialists have experience with advising ER doctors on their patients’ care. Be sure to have your specialist’s contact information easily available to make the process smoother.

You also have the right to request a transfer to a different hospital. If Crystal’s community hospital doesn’t have a neurologist knowledgeable in muscular dystrophy available when she visits the ER, she requests to be transferred to her neurologist’s direct care. The hospital obliges, but the transfers take some time and tend to occur at odd hours, like late at night or early morning.

Proactive education

For a busy ER or small community hospital, it can be helpful if you take the initiative to get them the information they need to provide the best care.

“In a practical sense, there’s too much information out there, and although guidelines exist for neuromuscular disease emergency care, a busy ER may not have time

to find the information,” Dr. Naprawa says. “Providing them with expert, medically screened information can be a big help.”

If you’ve been to the ER several times for the same problem or have a specific concern about a medical emergency, he recommends asking your neuromuscular specialists if they are willing to contact your nearest ER to have a discussion with the director and offer to educate ER staff.

It’s also a good idea to ask each of your specialists about scenarios when you should go to the ER and when it may not be necessary. “If you can go straight to the specialist, circumventing the ER may be just as safe and less traumatic,” Dr. Naprawa says.

Making the system better

Access to medical care for people with disabilities is required under the Americans with Disabilities Act (ADA), but Dr. Wolfe acknowledges that, especially in the current climate of healthcare staffing shortages, many hospitals may not have the resources to provide specialized care for people with neuromuscular diseases.

Throughout the nation, there’s wide variation in the emergency care available to people with disabilities and complex conditions. Dr. Wolfe believes this has a lot to do with advocacy within the community. “In Chicago, our access living group is very engaged with medical providers in the area,” she says.

Dr. Wolfe recommends contacting local disability rights and disability access groups to discuss needs that aren’t being met by your hospital or medical system. They can reach out to the hospital or medical care offices and help you get access problems resolved on a one-to-one basis.

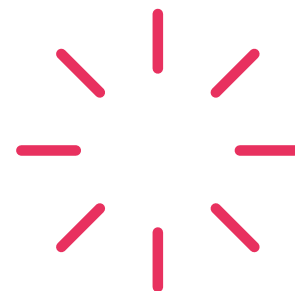
Many medical systems include ombudsmen and disability advocates on their staff. Request both whenever possible. Also, ask if your hospital has a Physical Medicine and Rehabilitation department. This specialty works with people experiencing disability due to medical conditions, and they can be helpful in resolving access issues.

After an ER visit, Crystal completes all hospital surveys and writes letters to the hospital board to alert them to any issues she encountered. When the hospital does its follow-up call after she is discharged, she names specific hospital staff, discussing what did and didn’t go well. Usually, the hospital calls her back to let her know they followed up with the personnel she named.

“Although sadly I’m a repeat customer, it’s a good thing that I’ve gotten to know the staff, nurses, nursing assistants, and doctors, and they’ve gotten to know me, too,” Crystal says. [Q](#)

Darlene Demetri is a Connecticut-based freelance writer living with facioscapulohumeral muscular dystrophy (FSHD).

It’s also a good idea to ask each of your specialists when you should go to the ER and when it may not be necessary.





Got Questions About Gene Therapy?

MDA's new Gene Therapy Support Network is here to offer guidance and resources to help navigate emerging treatment options.

"Gene therapy has created so much excitement and is the future of medicine," says Nora Capocci, MPH, MDA's Vice President of Healthcare Services. "We at MDA want to be supportive to the NMD community, and we're in a good position to be with our access to the clinician community through our Care Center Network and to the family community to support facilitating access to these treatments."

What resources are available through the Gene Therapy Support Network?

- **Gene Therapy Community Education.** Learn about gene therapy through MDA's print-ready gene therapy education materials, available in English and Spanish.
- **MDA's Care Center Network.** Individuals can explore the Care Center Network and reach out to Care Centers to see which are or are planning to offer gene therapies.

- **Access to Gene Therapy Support Specialists.** Connect with a Gene Therapy Support Specialist by phone, email, or video call.

Learn about gene therapy through MDA's print-ready gene therapy education materials.

"The Network is for everyone in the community — people planning to start gene therapy or those who just want to learn about it. It's for everybody," Nora says.

In addition to supporting the community, MDA's Gene Therapy Support Network provides gene therapy resources to clinicians to keep them informed about the latest treatments.

To find all of MDA's gene therapy resources, visit mda.org/gene-therapy-support.

Quest Marketplace

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Introducing the Quest Media Adaptive Lifestyle Website

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Media

MDAQuest.org is MDA's newly launched adaptive website offering the latest in magazine, blog and podcast content.



MDA Hill Group 2023



Advocacy Roundup

The year is more than halfway over, and MDA's advocacy team has been working nonstop on crucial advocacy initiatives. Check out the latest update from Washington, DC.

Medicare covers seat-elevating systems.

It's official: Medicare will cover wheelchair seat elevating systems for many types of power wheelchairs. These systems are vital for those living with a disability. Earlier this year, MDA and our advocates sent comments to the Centers for Medicare & Medicaid Services (CMS), asking the agency to cover these systems. Thank you to all the MDA advocates who spoke up. Your voices made a difference.

MDA and our advocates sent comments to the Centers for Medicare & Medicaid Services.

MDA won the fight against Medicaid work requirements.

MDA successfully fought against adding work requirements to the Medicaid program as part of legislation to raise the debt ceiling. Work requirements would've added unnecessary burdens in accessing care and could've resulted in many in

the neuromuscular community losing coverage. We will continue to fight efforts to make accessing Medicaid more difficult for those who need it.

MDA renominated DMD to the newborn screening program.

In May, MDA co-sponsored the renomination of Duchenne muscular dystrophy (DMD) for inclusion in the Federal Recommended Uniform Screening Panel (RUSP) for Newborn Screening. This renomination addresses concerns the Secretary's Advisory Committee raised during their February quarterly meeting. We anticipate the committee will consider the proposal again later this summer, and we hope the nomination will successfully move to the next stage of the process.

Advocates continue to fight to improve air travel.

In May, MDA's grassroots advocates traveled to Washington, DC, for the first time in three years to meet in person with their lawmakers for MDA on the Hill. Our grassroots volunteers were a force, urging their lawmakers to make air travel more accessible for people living with disabilities. Check out some of the highlights of the visit on page 52.

Ready to be an advocate? Become a member of MDA's grassroots advocacy network at mda.org/advocacy.

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- Get the answers you need



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New York Metro May 3-5, 2024	Chicago June 21-23, 2024	Houston August 2-4, 2024	

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Doug and Karen Clough joined MDA in celebrating Lou Gehrig Day at an Arizona Diamondback game in Phoenix on June 2. Doug lives with amyotrophic lateral sclerosis (ALS). Each year on Lou Gehrig Day, MDA community members gather at ballparks across the country to spread awareness of people living with ALS and their families.

Community Education: Together Again

Community education is back in person. MDA held its first in-person Engage Symposium since 2019 on June 3 in Princeton, New Jersey.

Individuals living with neuromuscular diseases, their caregivers, clinicians, and industry representatives gathered for the full day of education, empowering learners with actionable, up-to-date information to support their life goals.

Matthew Harms, MD, Associate Professor of Neurology at Columbia University and Medical Advisor to MDA, presented the keynote address, followed by sessions on topics including mental health, advocacy, orthopedic care, respiratory care, daily living tips and tricks, research updates, and transitioning to adulthood.

Attendees had the opportunity to connect with one another while also learning about resources and supports available in their



Matthew Harms, MD (center), was the keynote speaker at the in-person Engage Symposium.

community. When asked what was the most important thing they learned at the symposium, one attendee shared, “That there is help and hope.”

Join us for future MDA Engage Symposiums in Minneapolis on Sept. 23, and in Stanford, California, on Nov. 11. To register, visit mda.org/symposiums.



Searching for Answers

How I'm navigating life after college

BY JONATHAN PIACENTINO

Picture this: You've just received a college diploma — a four, maybe six-year endeavor, depending on how many credit hours you could handle. You've defied many expectations, perhaps living longer than your physician's initial prognosis or living on campus because the facilities could accommodate your needs. You take pictures with friends; you flash your diploma with your family. But in the back of your mind, you're thinking, "Now what?"

Sure, college grads all over the world struggle with that question, but with a complex condition — and specifically my diagnosis, Duchenne muscular dystrophy (DMD) — the answers are harder to find.

My personal FAQ

For the past seven years since I graduated from college, I've been asking myself the same questions.

Do I look for a job and try to earn money? That's the first thing that comes to mind. Going on disability is a steady source of income, but if I so much as make a dollar over what is allowed, those benefits are cut.

Where will I live? My parents' home is an accommodating space, and they have been my primary caregivers for my whole life. But I am getting older, and they are getting older, too. I am going to have greater needs, and they won't be able to take care of me forever.

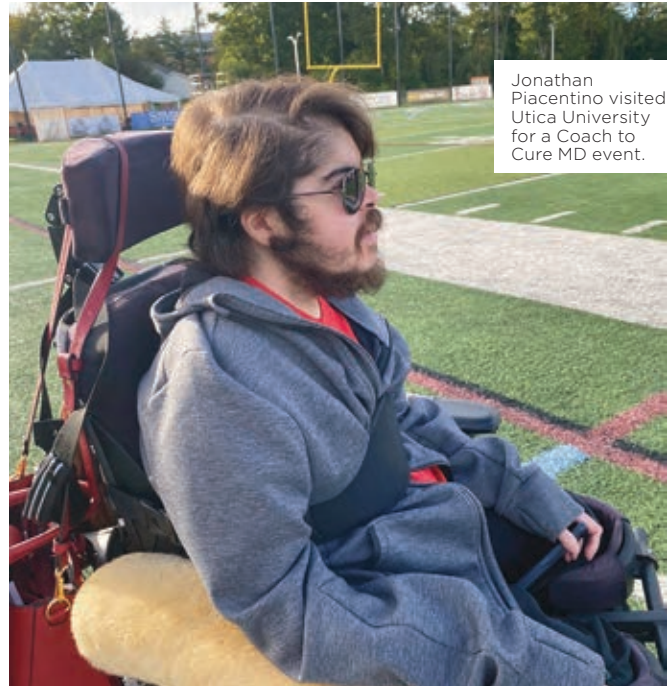
How can I get the help I need? I'm currently waiting on caregiving staff to be cleared for work through a home health agency. I must wait on the agency to go through the necessary paperwork. I must wait on the potential staff to find time to fulfill the prerequisites to work. While I'm working toward a solution, the solution doesn't feel good enough.

There are no clear answers to the questions above, and that is what weighs on me.

Here's one last question — and it might be the most important one: How do I get through this?

Finding connection

Sometimes it feels like there are too many questions weighing me down. But, under the crushing weight, I've found other things to focus on that get me through. I have made connections with new people online.



Jonathan Piacentino visited Utica University for a Coach to Cure MD event.

I like to create things. From painting and putting together model kits to building out fantastic digital worlds filled with all manner of wonderful creatures. And with that creative fire, I've found like-minded individuals who enjoy crafting equally awesome worlds. The social platform Discord has helped me connect with new people and new experiences, and this has helped me make it through.

So even when you feel weighed down, remember you are not alone. Just close your eyes and breathe. If you get tired of holding all that weight, simply rest and take a breath. Then, right yourself and keep going.

Laws of probability state that the longer something happens, the greater the chance that something else will occur. Maybe the next time you push against the weight of all those questions, the weight shifts. But focusing on you can ground you. And the best you can do is be alive. [👉](#)

+ADDRESSING BARRIERS

MDA's Access Workshops provide information and resources to help you overcome barriers in many aspects of living with a neuromuscular disease, from employment to equipment. Visit mda.org/AccessWorkshops.

Jonathan Piacentino, 29, of Penfield, New York, was diagnosed with DMD in 1998. In 2016, he earned his Bachelor of Science degree from Nazareth College, where he majored in Biology and minored in Chemistry and Toxicology.



Back on the Hill

MDA advocates returned to Washington to fight for accessible air travel

In May, MDA advocates returned to Washington, DC, for the first in-person MDA on the Hill event in more than three years. MDA staff and grassroots volunteers met with lawmakers to push for more accessible air travel for people with disabilities.

The timing of MDA on the Hill was crucial because Congress must reauthorize the Federal Aviation Act (FAA) this year and won't be able to adjust it again until 2028. Advocates implored lawmakers to include the following reforms in the reauthorization:

- Making airplanes more accessible
- Ensuring increased safety for air travelers with disabilities
- Preventing and holding airlines accountable for mobility device damages

Former MDA National Ambassador Reagan Imhoff, 18, from Milwaukee, attended MDA on the Hill for the first time this year.

“Not only was it rewarding to share my stories about the inaccessibility of air travel and my experiences with

lawmakers and their staff, but I met so many wonderful advocates during this journey,” says Reagan, who lives with spinal muscular atrophy (SMA).

To compel lawmakers, advocates shared personal stories about being dropped by airline staff, having their wheelchairs destroyed, and being excluded from travel.

“I think it’s important for people to keep in mind that a person’s mobility aid is an extension of their body and allows them to experience the world,” Reagan says. “Categorizing mobility aids as



From left: Amy Shinneman, Leah Zelaya, and Reagan Imhoff



Reagan met with Senator Tammy Baldwin of Wisconsin.

MDA ON THE HILL BY THE NUMBERS

- 31 advocates 11 states represented
- 31 strategic meetings with representatives and senators
- 370+ digital messages sent by advocates from across the country

‘luggage’ is completely stripping away a disabled person’s humanity while flying, as they truly are separated from their body. And shouldn’t everyone have the opportunity to travel and connect with others?” [Q](#)

+ ENTER THE PHOTO CONTEST

What meaningful moments have you captured on camera? Share a great photo of you or a loved one with a neuromuscular disease, and it could be selected to appear on this page in a future issue of Quest. Photos must be submitted by Sept. 10, 2023. Submit your photo at SurveyMonkey.com/r/QuestPhotos.

Do you have Becker Muscular Dystrophy?
Are you interested in participating in a clinical trial?

Join the  CANYON Trial.

The Canyon Trial

Edgewise Therapeutics is seeking individuals living with BMD for the phase 2 trial of EDG-5506, an investigational treatment for BMD. The Canyon trial aims to evaluate the effect of EDG-5506 on safety, tolerability, biomarkers of muscle damage and functional measures in individuals living with BMD. Participation is approximately 14 months and requires up to 10 site visits during the trial.



The Investigational Therapy

EDG-5506 is designed to reduce the skeletal muscle stress and injury that occurs in individuals with BMD. EDG-5506 aims to prevent skeletal muscle breakdown, inflammation, and the functional decline that accompany disease progression in BMD.

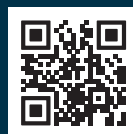
Who Can Participate?

Approximately 48 adults and 18 adolescents living with BMD are expected to be enrolled at sites across the United States, United Kingdom, and the Netherlands.

- Genetic diagnosis of Becker Muscular Dystrophy
- Male, ages 12 - 50 years
- Ability to complete physical function activities (i.e., North Star Ambulatory Assessment, 100-meter timed test)
- Able to meet other criteria as specified

Travel and other resources will be coordinated and provided for eligible participants.

Sites across the United States, United Kingdom, and the Netherlands are enrolling for the CANYON trial. For more information, please go to clinicaltrials.gov (NCT05291091) or contact studies@edgewisetx.com.



 **Edgewise**
THERAPEUTICS

There are so many questions about ALS. You can help find answers.

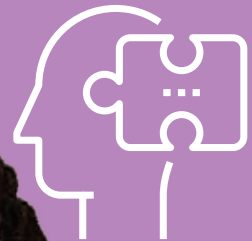
The National ALS Registry is a program that allows people with ALS to fight back and help defeat the disease.

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

Collecting and analyzing data

Striving to better understand the disease

Helping researchers find possible risk factors



Your participation can make a difference

Ask us about the Registry today.
For more information, call **800-232-4636** or visit **cdc.gov/als**.

