STAY INFORMED. LIVE EMPOWERED.

# Quest

MDAQUEST.ORG ISSUE 3 • 2024

## Change the Rules

Advocates and voters with disabilities influence legislation

**GENE THERAPY**
What to know

**GOLDEN YEARS**
Health tips for seniors

Scan this code for your free Quest subscription!

MDA® Muscular Dystrophy Association


## 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.** There is a pregnancy registry for women who take EVRYSDI during pregnancy. The purpose of this registry is to collect information about the health of the pregnant woman and her baby. If you are pregnant or become pregnant while receiving EVRYSDI, tell your healthcare provider right away. Talk to your healthcare provider about registering with the EVRYSDI pregnancy Registry. Your healthcare provider can enroll you in this registry or you can enroll by calling 1-833- 760-1098 or visiting https://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
- **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

Genentech
A Member of the Roche Group

[ADVERTISEMENT]

# **Evrysdi**—the first and only **oral, non-invasive** treatment for spinal muscular atrophy (SMA)

- **Proven results** in adults, children, and infants with SMA, including infants not yet showing symptoms
- Designed to help produce SMN protein **throughout the body**
- Can be taken **at home, at work, or when on the go**‡
- **No needles, sedation or hospital stays** required

**Talk to your doctor about Evrysdi**
Scan the QR code to learn more or visit **EvrysdiResults.com**

‡If refrigeration is not available, Evrysdi can be kept at room temperature up to 104°F for a combined total of 5 days. Please refer to the Instructions for Use for additional information about storage and administration.
SMN=survival motor neuron.

**Important Safety Information (continued)**

- 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.

**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. The purpose of this registry is to collect information about the health of the pregnant woman and her baby. If you are pregnant or become pregnant while receiving EVRYSDI, tell your healthcare provider right away. Talk to your healthcare provider about registering with the EVRYSDI Pregnancy Registry. Your healthcare provider can enroll you in this registry or you can enroll 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.

Genentech
*A Member of the Roche Group*

EVRYSDI® (risdiplam)

Distributed by:

**Genentech, Inc.**
A Member of the Roche Group
1 DNA Way
South San Francisco, CA
94080-4990

EVRYSDI is a registered trademark of Genentech, Inc.

M-US-00007143(v6.0)



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

This Patient Information has been approved by the U.S. Food and Drug Administration.

Approved: 3/2023


# Contents

ISSUE 3 **2024**

MDA-207

## FEATURES




## DEPARTMENTS









Cover image: drnadig/Getty

# Your Voice, Your Vote


Donald S. Wood, PhD

MDA's commitment to advocacy is a cornerstone of our mission, and the power of our united voice has never been more evident than in the passage of the most significant advancement in accessible air travel in four decades. In a monumental step forward for people with disabilities, President Joe Biden signed the FAA Reauthorization Act in May, which includes provisions put forward by MDA to ensure safer, more dignified travel for people with disabilities. I'd be remiss if I did not point out that one of the most powerful voices on this issue has been our own Mindy Henderson, Quest Media's Editor-in-Chief and Vice President of Disability Outreach and Empowerment at MDA.

As important as it is to raise your voice through advocacy, it is equally crucial to exercise your right to vote. Whether you're electing your town council or the president of the United States, you have the right to support the candidates of your choice and issues that are important to you through your ballot. To do so, it's important to know your rights and have a plan to vote.

In this issue of Quest Magazine, we are happy to share a variety of information that will empower you in every stage of life and inform you about how to prepare for current or future gene therapy, as well as continue to explore the power of the ballot and the imperative of making your vote count. In these pages, you will also find information about MDA's Access the Vote campaign (**mda.org/vote**), which provides resources to help you prepare to cast your ballot. If you follow MDA on social media, you've seen the ongoing campaign, and we hope you'll engage in learning about your voting rights.

I hope you'll enjoy this informative issue of Quest. I think it's one of our best yet!

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

## +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 Quest Magazine. Submit your photo at **SurveyMonkey.com/r/QuestPhoto** or scan this QR code.

MDA Muscular Dystrophy Association

For more than 70 years, MDA has led the way in accelerating research, advancing care, and advocating for the support of people living with muscular dystrophy, ALS, and related neuromuscular diseases and their families. MDA's mission is to empower the people we serve to live longer, more independent lives.

**EDITORIAL**
**Mindy Henderson**
Quest Media Editor-in-Chief
For editorial inquiries:
**quest@mdaUSA.org**

**ADVERTISING**
**Elisa N. Beerbohm**
Director, Marketing and Ad Sales
For print and digital advertising opportunities:
**EBeerbohm@mdaUSA.org**
**MDAQuest.org/advertise**

**SUBSCRIPTIONS**
Sign up for free or change your Quest subscription status at **MDAQuest.org/subscribe**.

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

**Quest Volume 31, Issue 3**
Published by
Muscular Dystrophy Association
1016 W Jackson Blvd #1073
Chicago, IL 60607
**800-572-1717**
email: **quest@mdaUSA.org**
Available on the internet at **MDAQuest.org**
ISSN 1087-1578
Postage paid at Bolingbrook, IL
Nonprofit postal permit number 1446.

Postmaster: Treat as Standard A Mail Only



FOLLOW MDA ON

PUBLICATION MANAGEMENT: UNLOCK

## Doug Clough is a Champion in the ALS Community and Beyond

Doug Clough's commitment to serving as a National MDA/ALS Ambassador has ignited inspiration within the ALS community and beyond. At the heart of Doug's journey lies determination to spread awareness and support for those touched by ALS. One of the cornerstones of Doug's advocacy efforts lies in his partnership with Dutch Bros. Doug's ties to Dutch Bros run deep, having frequented a local stand before his diagnosis. Doug has taken the time to meet with hundreds of employees, expressing his gratitude for their unwavering support of the ALS community.

Dutch Bros and its customers came together May 17, for the 18th annual Drink One for Dane to support the Muscular Dystrophy Association and its mission to end ALS. We're stoked to share the Dutch Bros Foundation donated $2.5 million to MDA.


### Mansfield Cares Golf Classic Raises $1.2M

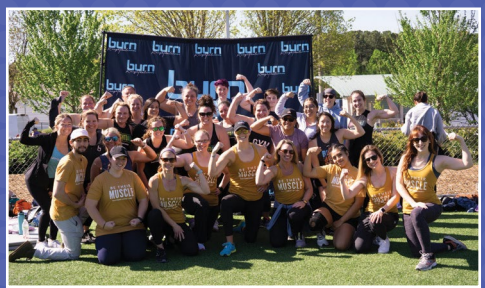
The 38th Annual Mansfield Cares Golf Classic is one of the largest events of its kind. Over 500 golfers, including energy partners and celebrities, come together for this fun event – raising over $11 million for MDA to date. Thank you to Mansfield Energy, and all sponsors and attendees for making the 2024 tournament a success!

### CITGO Lake Charles Refinery Raises over $578,000

The CITGO Lake Charles Refinery raised over $578,000 this year for MDA. Their 39th annual Golf Classic brought together sponsors and golfers to enjoy a day on the course with auctions and great food. They also launched an event called Beer, Bourbon, & BBQ, an exclusive networking dinner to benefit MDA.

### Burn Boot Camp and MDA: A Powerful Partnership

Burn Boot Camp's Be Their Muscle campaign just concluded its eighth consecutive year, bringing the total raised for MDA to over $3M. Each year, fitness enthusiasts and trainers rally together nationwide for special workout events and challenges, community activities, and personal fundraising efforts.



# You Can Influence the World

What are your dreams? If you knew you could do anything you wanted and that you were guaranteed NOT to fail, what would you do?

I am a dreamer, and I have always wanted big things. I've learned a lot about what is possible, and what I have learned is this: If I failed, it was either because I gave up before I should have or because the world was not set up for me to succeed.

The good news? Both of those things are in our span of influence. We have it within ourselves to be creative, to problem-solve, and to stick with it until we accomplish what we want, no matter how long it takes.

We also have the ability to influence how the world sees us and what the world says we can do. One way we do that is far more important than I sometimes believed: voting. Our feature story in this issue is about how advocacy has already changed the world and the power the disability community has at the polls. The upcoming election in November is one of the best ways to make your wishes known. The individuals you choose to vote for make big decisions about your rights in your everyday life. That is not a choice I take lightly anymore.

Dream big, my friends. Dream, and don't quit until it's done. Part of that means voting for the world that will help you get there.

Mindy Henderson
*Vice President, Disability Outreach & Empowerment*
*Editor-in-Chief, Quest Media*

## ICONS TO WATCH FOR THROUGHOUT QUEST

**QUEST MAGAZINE ONLINE**

**QUEST BLOG**

**QUEST PODCAST**

**DISEASES A-Z CONTENT LIBRARY**

## DID YOU KNOW?

**QUEST PODCAST SURPASSED 10K DOWNLOADS!** Thank you to our listeners for helping us reach this milestone. If you're not listening to the Quest Podcast yet, you're missing out.

**Top 3 Quest Podcasts:**

1. Behind the Scenes: A Look at the Science and Research for New Treatments (Episode 39)
2. Unlocking Access and Inspiring Action with Sophie Morgan (Episode 40)
3. Courage Kindled: A Hero's Unbreakable Spirit (Episode 41)

Find these essential episodes and more at **MDAQuest.org/podcast**.

**YOU CAN FIND CLINICAL TRIALS IN MINUTES.**

MDA's Clinical Trials Finder tool is designed to help you locate clinical trial opportunities across the country. Just answer a few questions to pinpoint trials specific to your disease type, location, age, and other criteria, and you can see a list of trials you may be eligible for. To use the tool, go to **mda.org/clinical-trials** and click on the "Find Trials" button.

ADVERTISEMENT

# Do you have Becker Muscular Dystrophy?

# Have you considered the GRAND CANYON Trial?

## The GRAND CANYON Trial

Edgewise Therapeutics is seeking individuals living with Becker for the pivotal cohort of the GRAND CANYON trial of sevasemten (EDG-5506), an investigational treatment for Becker. The GRAND CANYON trial aims to evaluate safety and effects on function and biomarkers of muscle damage in adult males with Becker. Participation is for approximately 19 months and will require up to 7 site visits over the duration of the trial.

## The Investigational Therapy

Sevasemten is an investigational therapy in the form of a daily oral pill. Sevasemten is designed to prevent contraction-induced muscle injury that occurs with daily activity in Becker. Sevasemten is designed to limit this damage and help prevent the functional decline that accompanies disease progression in Becker.

## Can I join the Grand Canyon trial?

Approximately 120 adults living with Becker are expected to be enrolled in the trial. To participate you must fit the following criteria:

- Genetic diagnosis of Becker Muscular Dystrophy
- Male, ages 18-50
- Ambulatory with the 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 participants

*Select assistive devices such as orthotics or a cane can be used during the 100-meter timed test

Sites across the United States began enrolling for the GRAND CANYON trial in 2023. Sites in up to 10 additional countries are expected to open enrollment for GRAND CANYON in 2024. For more information, please go to clinicaltrials.gov (NCT05291091) or scan the QR code to access the GRAND CANYON study website.

Edgewise THERAPEUTICS

# Appraising and Adapting

*Throughout his career, A.J. Bardzilowski has met challenges and found solutions*

BY REBECCA HUME

Real estate appraiser A.J. Bardzilowski has been adapting to the changing real estate market for more than 30 years. When the 58-year-old California State Certified Real Estate Appraiser was diagnosed with sporadic inclusion body myositis (sIBM) in his late 40s and facioscapulohumeral muscular dystrophy (FSHD) last year — a rare combination of conditions — he utilized resources and creative thinking to continue excelling in a career he loves.

A.J. Bardzilowski lives in California with his wife, Marcey.

## Accessibility and appraisals

In his early 40s, A.J. was an avid tennis player when he noticed that he was slowing down on the court and falling frequently. After seeking answers from numerous doctors, he connected with an MDA Care Center neurologist and received a diagnosis of sIBM. As muscle weakness progressed in his legs over the next 12 years, his doctor also became concerned about signs of muscle loss in A.J.'s upper arms and shoulders. A.J. was poised to participate in his second clinical trial for sIBM when a test revealed that he had a dual diagnosis of FSHD. With symptoms occurring in tandem, A.J. has difficulty with walking and navigating stairs, experiences frequent muscle fatigue and exhaustion, and has a hard time typing or writing.

Due to the physical nature of his profession, these symptoms present challenges in A.J.'s day-to-day work. His role requires him to inspect and appraise homes and buildings, often with stairs. As an independent contractor, A.J. didn't have a human resources department to contact for accommodations or assistance; he had to get creative. He found a solution by hiring a friend, Chrissy, who was interested in training to be an appraiser. As his assistant, Chrissy visits inspection sites with A.J. and helps him access all areas of the property.

"Figuring out how to adapt is a valuable tool," A.J. says. "Identifying what you need and how you can accommodate that need or adapt in order to continue

doing what you want to do means getting creative and also leaning on the resources available."

Left: A.J . enjoys time with his three children. Right: New braces improved his mobility.

### Finding support

To overcome other barriers in his line of work, A.J. identifies his needs as they arise and finds solutions, utilizing resources and the network that he has built over the years. "I have met so many nice people along the way who have been extremely helpful," A.J. says. "You still have to advocate for yourself. Sometimes, you have to ask more questions to find out what is available to you, but the resources and support are there."

A.J. connected with the California Department of Rehabilitation (DOR), his state vocational rehabilitation office, and obtained Dragon Easy Speak software and a headset that allows him to complete his documentation hands-free in his home office. He equipped his home with a small ramp to enter and exit, extra handrails on stairs and in his bathroom, and strategically placed long-pole grippers to make reaching easier. But probably the most impactful action that A.J. has taken to maintain mobility and ensure his career independence was obtaining Ottobock C-braces.

"My orthotist told me about these braces. They are a hybrid between a static knee brace and an exoskeleton that does everything for you," he explains. "The C-brace has a microprocessor on the outside of each knee that learns your gait as you walk and pumps fluid to a motor that acts as your knee."

It took A.J. more than four years of appealing insurance denials and working with the DOR to obtain the braces, but he refused to give up. Now, after nine weeks of physical therapy working with the braces, A.J. is learning a new way to walk. He recently took a trip to New York City, where he wore his braces for four days while exploring and enjoying the big city.

**A.J. connected with the California Department of Rehabilitation (DOR), his state vocational rehabilitation office, and obtained Dragon Easy Speak software and a headset.**

Passionate about traveling with his family, A.J. is branching out into a new endeavor: sharing travel advice and reviews for fellow travelers with disabilities on YouTube. "Because of my disability and my issues in my career, I am trying to keep making adjustments and to do something productive that would help others living with disabilities," A.J. says.

He advises others, especially those with adult-onset disabilities, to keep an open mind about embracing change, asking for help, finding creative ways to do things differently, and embarking on new ventures. "If you stay in your career or if you choose to change paths, advocate for yourself. We are at a point in our society where we can — and should — make working with a disability work for us." Q

Rebecca Hume is a Senior Specialist and Writer for Quest Media.

### BETTER WITH HELP

Andrew Zaleski, a journalist living with myotonic dystrophy, writes about how he and others in the community have overcome insecurities about asking for help in daily life at **MDAQuest.org/asking-for-help**.

# GENERALIZED MYASTHENIA GRAVIS

*doesn't get to steal this walk*

## IMPORTANT SAFETY INFORMATION

Do not use VYVGART if you have a serious allergy to efgartigimod alfa or any of the other ingredients in VYVGART. Do not use VYVGART HYTRULO if you have a serious allergy to efgartigimod alfa, hyaluronidase, or any of the other ingredients in VYVGART HYTRULO. VYVGART and VYVGART HYTRULO can cause serious allergic reactions and a decrease in blood pressure leading to fainting.

**VYVGART and VYVGART HYTRULO may cause serious side effects, including:**

- **Infection.** VYVGART and 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. Signs or symptoms of an infection may include 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.
- **Allergic Reactions (hypersensitivity reactions).** VYVGART and VYVGART HYTRULO can cause allergic reactions such as rashes, swelling under the skin, and shortness of breath. Hives were also observed in patients treated with VYVGART HYTRULO. Serious allergic reactions, such as trouble breathing and decrease in blood pressure leading to fainting have been reported with efgartigimod alfa-fcab.
- **Infusion-Related Reactions.** VYVGART and VYVGART HYTRULO can cause infusion-related reactions. The most frequent symptoms and signs reported with efgartigimod alfa-fcab were high blood pressure, chills, shivering, and chest, abdominal, and back pain.

Tell your doctor if you have signs or symptoms of an infection, allergic reaction, or infusion-related reaction. These can happen while you are receiving your VYVGART or VYVGART HYTRULO treatment or afterward. Your doctor may need to pause or stop your treatment. Contact your doctor immediately if you have signs or symptoms of a serious allergic reaction.

**Before taking VYVGART or**

# Discover VYVGART Hytrulo for subcutaneous injection

VYVGART® Hytrulo
(efgartigimod alfa and hyaluronidase-qvfc)
Subcutaneous Injection
180 mg/mL and 2000 U/mL vial

**VYVGART Hytrulo is a 30- to 90-second injection given at an infusion center, doctor's office, or at home.*†**

VYVGART®
(efgartigimod alfa-fcab)
Injection for Intravenous Use
400 mg/20 mL vial

**VYVGART is still available as an IV infusion and is given over 1 hour at an infusion center, neurologist's office, or at home.*†**

VYVGART Hytrulo and VYVGART for IV infusion are given in treatment cycles with an individualized break between cycles (if additional cycles are needed). A treatment cycle consists of 1 treatment each week for 4 weeks (4 treatments per cycle).

*For at least 30 minutes after your subcutaneous injection or 1 hour after your IV infusion, a healthcare professional will monitor you for reactions.
†In some cases, VYVGART Hytrulo for subcutaneous injection or VYVGART for IV infusion may also be given at home by a trained nurse.

## Scan the QR code or visit VYVGART.com to learn more

Questions? Call **1-833-VYVGART (1-833-898-4278)**

IV=intravenous. Visit VYVGART.com/glossary for a glossary of terms.

**VYVGART HYTRULO, tell your doctor if you:**

- take any medicines, including prescription and non-prescription medicines, supplements, or herbal medicines,
- have received or are scheduled to receive a vaccine (immunization), or
- have any allergies or medical conditions, including if you are pregnant or planning to become pregnant, or are breastfeeding.

**What are the common side effects of VYVGART and VYVGART HYTRULO?**

The most common side effects in efgartigimod-alfa-fcab-treated patients were respiratory tract infection, headache, and urinary tract infection. Additional common side effects with 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 and 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® (efgartigimod alfa-fcab) for intravenous (IV) infusion and what is VYVGART® HYTRULO (efgartigimod alfa and hyaluronidase-qvfc) for subcutaneous injection?**

VYVGART and VYVGART HYTRULO are both prescription medicines, each 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 and VYVGART HYTRULO at www.VYVGART.com and talk to your doctor.**

**Please see the Consumer Brief Summary on the following page.**



argenx

**Important Information about both VYVGART® (efgartigimod alfa-fcab) for intravenous (IV) infusion and VYVGART® HYTRULO (efgartigimod alfa and hyaluronidase-qvfc) for subcutaneous injection; Rx only.**

The risk information provided here is not comprehensive. To learn more, talk about VYVGART and VYVGART HYTRULO with your healthcare provider. The US Food and Drug Administration (FDA)-approved product labeling can be found for VYVGART by visiting www.VYVGART.com/PI and for VYVGART HYTRULO by visiting www.VYVGARTHYTRULO.com/PI or calling 1-833-VYVGART (1-833-898-4278).

**What is VYVGART and what is VYVGART HYTRULO?**

VYVGART and VYVGART HYTRULO are both prescription medicines, each 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). It is not known if VYVGART or VYVGART HYTRULO are safe and effective in children under 18 years of age.

It is not known if VYVGART or VYVGART HYTRULO are safe and effective in children under 18 years of age.

**Who should not use VYVGART and VYVGART HYTRULO?**

Do not use VYVGART if you have a serious allergy to efgartigimod alfa or any of the other ingredients in VYVGART. Do not use VYVGART HYTRULO if you have a serious allergy to efgartigimod alfa, hyaluronidase, or any of the other ingredients in VYVGART HYTRULO. VYVGART and VYVGART HYTRULO can cause serious allergic reactions and a decrease in blood pressure leading to fainting.

**What is the most important information I should know about VYVGART and VYVGART HYTRULO?**

VYVGART and VYVGART HYTRULO may cause serious side effects, including:

- **Infection.** VYVGART and 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. Signs or symptoms of an infection may include 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.
- **Allergic Reactions (hypersensitivity reactions).** VYVGART and VYVGART HYTRULO can cause allergic reactions such as rashes, swelling under the skin, and shortness of breath. Hives were also observed in patients treated with VYVGART HYTRULO. Serious allergic reactions, such as trouble breathing and decrease in blood pressure leading to fainting have been reported with efgartigimod alfa-fcab.
- **Infusion-Related Reactions.** VYVGART and VYVGART HYTRULO can cause infusion-related reactions. The most frequent symptoms and signs reported with efgartigimod alfa-fcab were high blood pressure, chills, shivering, and chest, abdominal, and back pain.

Tell your doctor if you have signs or symptoms of an infection, allergic reaction, or infusion-related reaction. These can happen while you are receiving your VYVGART or VYVGART HYTRULO treatment or afterward. Your doctor may need to pause or stop your treatment. Contact your doctor immediately if you have signs or symptoms of a serious allergic reaction.

**Before taking VYVGART or VYVGART HYTRULO, tell your doctor if you:**

- have any of the conditions or symptoms listed in the section **"What is the most important information I should know about VYVGART and VYVGART HYTRULO?"**, any allergies or any medical condition
- have received or are scheduled to receive an immunization (vaccine). It is not recommended to receive a "live vaccine" if you are being treated with VYVGART or VYVGART HYTRULO.
- are pregnant or plan to become pregnant. It is not known if VYVGART or VYVGART HYTRULO may harm your unborn baby.

  - **Pregnancy Registry:** There is a pregnancy registry for pregnant women who take VYVGART or VYVGART HYTRULO. The purpose of this registry is to collect information about the health of you and your baby if you take VYVGART or VYVGART HYTRULO during pregnancy. To learn more, call 1-855-272-6524 or visit https://www.VYVGARTpregnancy.com. You may also talk to your healthcare provider about how you can take part in this registry.
- are breastfeeding or plan to breastfeed. It is not known if VYVGART or VYVGART HYTRULO passes into your breast milk.

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

**What are the ingredients in VYVGART?**

**Active ingredient:** efgartigimod alfa-fcab

Each 20 mL single-dose vial contains 400 mg of efgartigimod alfa-fcab at a concentration of 20 mg/mL. In addition, each mL of solution contains L-arginine hydrochloride (31.6 mg), polysorbate 80 (0.2 mg), sodium chloride (5.8 mg), sodium phosphate dibasic anhydrous (2.4 mg), sodium phosphate monobasic monohydrate (1.1 mg) and water for injection, USP, at a pH of 6.7.

**What are the ingredients in VYVGART HYTRULO?**

**Active ingredients:** efgartigimod alfa and hyaluronidase (human recombinant)

Each 5.6 mL single-dose vial contains 1,008 mg of efgartigimod alfa and 11,200 units of hyaluronidase (human recombinant). Each mL of solution contains 180 mg of efgartigimod alfa, 2,000 units of hyaluronidase (human recombinant) and histidine (1.4 mg), L-histidine hydrochloride monohydrate (2.2 mg), methionine (1.5 mg), polysorbate 20 (0.4 mg), sodium chloride (5.8 mg), sucrose (20.5 mg), and water for injection, USP, at a pH of 6.0.

argenx

VYVGART is manufactured by: argenx BV Industriepark 7 9052 Zwijnaarde, Belgium U.S. License No. 2217
VYVGART HYTRULO is manufactured by: argenx BV Industriepark 7 9052 Zwijnaarde, Belgium U.S. License No. 2217
Halozyme Therapeutics, Inc., 12390 El Camino Real, San Diego, CA 92130, U.S. License No. 2187
VYVGART and VYVGART HYTRULO are distributed by: argenx US, Inc. 33 Arch Street Boston, MA 02110

# Representing the Community

*Behind the scenes of the MDA Ambassador Program*

BY MEGAN KRAMER-SALVITTI

The MDA Ambassador Program is a foundational program at MDA, beginning with its first National Ambassador, Michael Danna, in 1952. Since the program's inception, 48 National Ambassadors and countless General Ambassadors have shared their stories and authentic perspectives to raise awareness and advance the MDA mission. Here, Quest Media speaks with Scott Wiebe, MDA's Director of Community Programs, and Laura McClellan, MDA's Manager of Community Programs, about how the Ambassador Program has evolved and how the community can get involved.


MDA Ambassadors Ira Walker and Jose Flores

**Q What is the Ambassador Program, and how has it changed?**

**Scott:** The MDA Ambassador Program allows individuals living with neuromuscular disease to share their stories, help raise awareness, and connect with the MDA community. Ambassadors are crucial to advancing MDA's mission to empower those living with neuromuscular diseases, and we have deep appreciation for their engagement and support.

Formerly, the program included local, state, and national Ambassadors. Today, it consists of National Ambassadors — the most publicly visible representatives of the neuromuscular community — and General Ambassadors. This shift supports the reality that Ambassadors are

**Since the program's inception, 48 National Ambassadors and countless General Ambassadors have shared their stories and authentic perspectives.**

### +APPLY TO BE AN AMBASSADOR

If you are interested in advancing MDA's mission, serving as an Ambassador provides an opportunity to make an impact. Apply at **mda.org/ambassadors**.

MDA National Ambassador Leah Zelaya (center) with her mother Bevi (left) and father Jaime (right), also an MDA Ambassador


MDA Ambassador Luke

no longer only local; rather, they have an opportunity to share their stories and make an impact on a larger scale while still being able to connect locally with MDA supporters and events.

The MDA National Ambassador program has evolved, too. Historically, the National Ambassador was always a child. Now, MDA has two National Ambassadors — one adult and one child — to better represent the breadth of the community.

The process for National Ambassadors differs in that they are selected by MDA from the group of General Ambassadors.

Over the years, MDA's Ambassador Program has had different names and has evolved considerably. However, the core intent of the program remains the same: Engaging individuals and families living with neuromuscular diseases to raise awareness about the lived experience of the MDA community through their own authentic viewpoints.

### What do MDA Ambassadors do?

**Laura:** There are many ways to be involved, and each individual's engagement is tailored to their strengths and interests. Some Ambassadors are fantastic public speakers, while others write impactful thank-you notes, sharing why research investment or programs like Summer Camp are important to them. Some love spending time with our fire fighter partners during Fill the Boot drives, while others share their insights on educational panels or write guest blogs for Quest (**MDAQuest.org/tag/ambassadors**).

**Scott:** Ambassadors' support of MDA's fundraising efforts is incredible. Sharing personal stories and experiences connects donors and partners to MDA's mission, which strengthens our fundraising initiatives, and that in turn allows MDA's mission to thrive, funding groundbreaking research, advancing MDA's advocacy initiatives, and much more.

### Why is the Ambassador Program important?

**Scott:** The Ambassador program is central to our mission to empower the people we serve.

Everyone processes living with a neuromuscular disease differently and has a unique journey. Many

Ambassadors have said that engaging with this program and sharing their story is empowering. By helping others better understand what living with a neuromuscular disease is like, allowing others who have neuromuscular diseases to feel more supported, and giving back to the community, being an Ambassador is a meaningful experience for many people. Ambassadors are also beacons for others within the community to look to; seeing stories like their own lessens the likelihood in the future that someone feels alone and misunderstood.

**What is the application process?**

**Laura:** All members of the neuromuscular community are invited to apply to represent MDA as General Ambassadors. The application is for a year-long, renewable term.

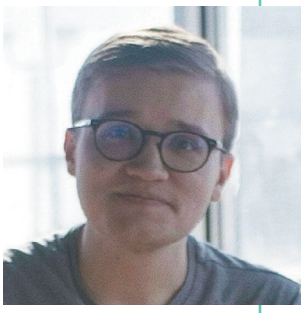
A member of MDA's team meets virtually with each applicant to learn more about their unique journey and discuss how they'd like to engage with the program. Ambassadors can be any age and live with any neuromuscular diagnosis and are selected based on their passion for raising awareness and their commitment to MDA's mission. The process for National Ambassadors differs in that they are selected by MDA from the group of General Ambassadors.

MDA Ambassador Breylyn

## WHAT MDA AMBASSADORS HAVE TO SAY

"I believe it is so important to raise your voice for things you are passionate about. This creates awareness and stimulates others to get involved. It creates community. … If we all just put in a little, we will make the world a better place."
— Leah Zelaya, National Ambassador

"It's my hope that through positive and consistent public appearances, I encourage those early in their neuromuscular journey and clearly illustrate that being an adult with a neuromuscular condition can truly be a life of meaningful significance, happiness, and love."
— Ira Walker, National Ambassador



"The message I hope to spread is that we aren't 'inspiring,' tokenized individuals — we're people. The stories we have are broad, diverse, and powerful. We are dancers, teachers, singers, and doctors. We're someone's kid, someone's parent. Spreading awareness — once a scary phrase for me — is, I believe, my main purpose on this earth."
— Jess Westman, General Ambassador

We are fortunate to have an incredibly talented, diverse, and committed group of Ambassadors.

I am consistently amazed by their accomplishments, authenticity, and the impact they have on the community. Q

Megan Kramer-Salvitti is an editor and writer for Quest Media.

# Progress Now

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

**New approvals**

## Generic Deflazacort Approved for DMD


Image: Rawf8/Getty

The US Food and Drug Administration (FDA) approved the first generic version of deflazacort (Emflaza) oral suspension for the treatment of people with Duchenne muscular dystrophy (DMD) ages 5 and older.

Generics are therapeutically equivalent to brand-name medications and usually sold at lower prices. This approval may make deflazacort more easily available to people with DMD in the United States.

DMD is caused by mutations in the DMD gene, which provides instructions for making the dystrophin protein that muscles need to function properly. Deflazacort, a type of corticosteroid, can reduce inflammation and muscle damage and preserve muscle strength and function in people with DMD.

The generic version of deflazacort is approved for a slightly older population, while brand-name Emflaza is approved for patients as young as 2. Emflaza is available as tablets or as an oral suspension. A generic version of deflazacort oral tablets was approved by the FDA in February for ages 5 and older.

### CLINICAL TRIAL TERMS TO KNOW

**Double-blind:** Neither researchers nor participants know which participants are taking the drug or placebo.

**Multiarm:** 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.

**Becker muscular dystrophy (BMD)**

## Phase 2 Study Seeks Adults

Image: demaerre/Getty

Researchers at the University of Pittsburgh are seeking adults living with BMD to participate in a phase 2 study to evaluate the safety and effectiveness of vamorolone (AGAMREE®) in treating BMD. Studies conducted on boys with Duchenne muscular dystrophy (DMD) have shown that AGAMREE can improve muscle strength and function with lower side effects than traditional

glucocorticoid treatment. This study aims to determine the potential of AGAMREE as a treatment for people with BMD.

The double-blind, placebo-controlled study involves an optional virtual prescreening visit and seven in-person visits over 33 weeks. Eligible participants will be divided into two study groups: one receiving AGAMREE and one receiving the placebo.

The drug or placebo will be administered orally (by mouth). The effects of AGAMREE will be evaluated using multiple tests, including physical exams, laboratory testing, motor function tests, and electrocardiograms.

To be eligible, participants must have a confirmed genetic diagnosis of BMD and be between ages 18 and 65, among other criteria.

**For more information, visit ClinicalTrials.gov and enter NCT05166109 in the "Other terms" search box, or contact study coordinator Gabi Niizawa at niizawaga@upmc.edu or 412-383-9775.**

## Becker muscular dystrophy (BMD)


Image: BanksPhotos/Getty

Results from hand grip strength tests were unchanged, suggesting that the drug could preserve function.

# Phase 1 Results Show Stable Muscle Function

Edgewise Therapeutics released top-line results from its phase 1 ARCH trial of EDG-5506 (sevasemten). This open-label, single-center study involved 12 adults with BMD who were able to walk. Sevasemten was well-tolerated and continued to stabilize muscle function after two years in the men with BMD participating in the trial.

BMD is caused by mutations in the dystrophin (DMD) gene that result in low or defective production of the dystrophin protein. This leads to progressive muscle damage and loss of function. Sevasemten is an orally administered small-molecule drug designed to block the activity of myosin, a protein involved in muscle contractions. The treatment is expected to make muscle contractions gentler, protecting the muscles from damage.

At one year of treatment with sevasemten, researchers saw significantly decreased levels of key biomarkers of muscle damage, including creatine kinase, fast skeletal muscle troponin I (TNNI2), and myoglobin. After two years of treatment, participants' scores on the North Star Ambulatory Assessment improved slightly. Results from hand grip strength tests and the 100-meter timed test of maximal performance were unchanged, suggesting that the drug could preserve function and halt disease progression in BMD.

Edgewise is advancing sevasemten in the phase 2 CANYON trial.

**To learn more about Edgewise Therapeutics' ARCH trial, visit ClinicalTrials.gov and enter NCT05160415 in the "Other terms" search box.**

Image: Dell/Getty

**Duchenne muscular dystrophy (DMD)**

# Participants Needed for Observational Studies

Researchers at the Center for Health + Technology at the University of Rochester are seeking individuals with DMD and their adult caregivers to participate in two observational REDCap studies: the DMD Health Index (DMD-HI) for individuals with DMD and the DMD Caregiver Reported Health Index (DMDCR-HI) for caregivers.

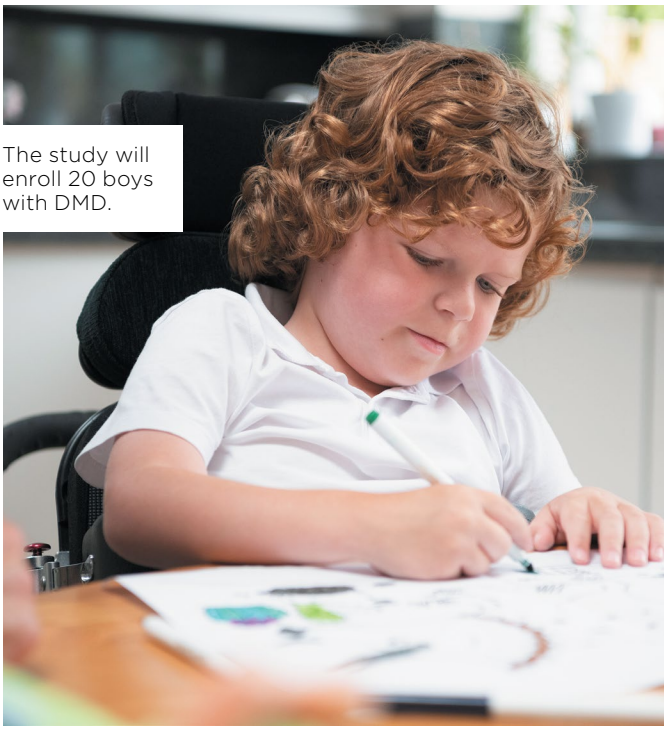
Individuals enrolled in the DMD-HI study will complete a demographics questionnaire, as well as the DMD-HI and other surveys at various intervals.

Caregivers enrolled in the DMDCR-HI study will complete a demographics questionnaire, the DMDCR-HI, and other surveys at various intervals.

The study findings will help determine how the DMD disease burden changes over time and identify demographic and clinical factors associated with disease progression. Both studies will last for 24 months.

To be eligible for the DMD-HI study, individuals must have a DMD diagnosis and be 11 years old or older. To be eligible for the DMDCR-HI study, individuals must be at least 18 years old and be caring for an individual with DMD who is under 21 years old.

**To learn more, contact study coordinator Jennifer Weinstein at jennifer.weinstein@chet.rochester.edu.**



The study will enroll 20 boys with DMD.

Image: JohnnyGreig/Getty

# Phase 2 Clinical Trial Recruits Boys

Researchers at NS Pharma are seeking boys ages 4 to 14 with DMD who are amenable to exon 44 skipping to participate in a phase 2 clinical trial to evaluate the safety and effectiveness of the investigational therapy NS-089/NCNP-02-201 (brogidirsen). Exon skipping is a therapeutic strategy that works in DMD by causing cells to "skip" over faulty or misaligned sections of genetic code (exons) to produce a shortened but still functional dystrophin protein.

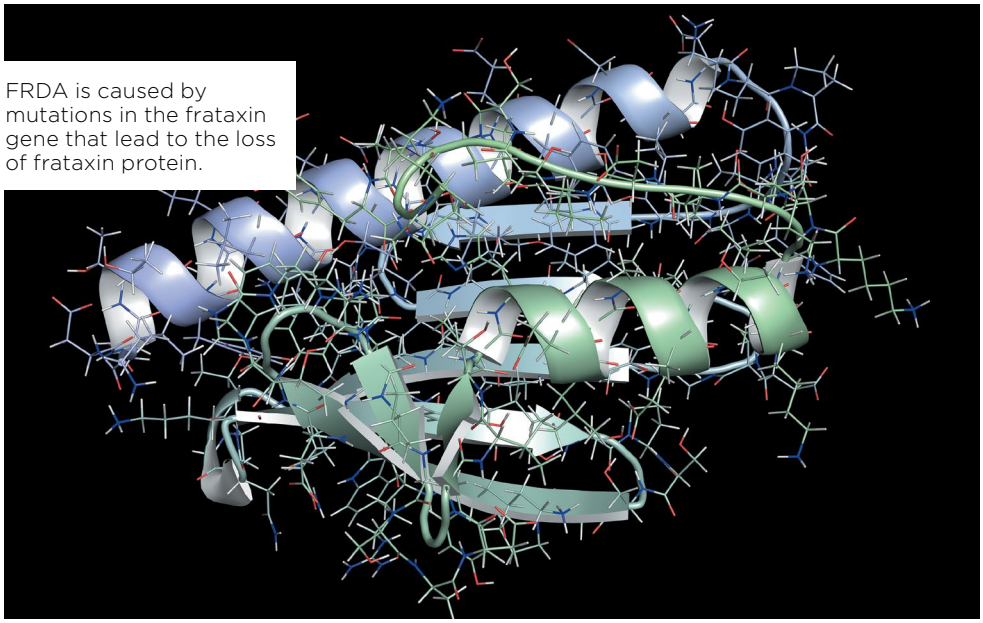
The study will enroll 20 boys with DMD, and all participants will receive brogidirsen treatment intravenously (in the vein). The study will comprise two parts: the first part will include six boys who will be treated with increasing doses of the

drug for 12 weeks, and the second part will include 14 additional boys who will receive the optimal dose (determined by the first part) for 24 weeks. Participants will also have weekly doctor visits.

The effects of brogidirsen will be evaluated with various tests and procedures at each visit, including but not limited to blood draws, urine collections, and physical exams. Motor function tests will be performed every 12 weeks, and muscle biopsies will be performed at the start and end of part two.

To be eligible, individuals must be able to walk independently without assistive devices and be on a stable dose of glucocorticoid for at least three months prior to entering the study, among other criteria. Travel support will be available for eligible participants.

**For more information, visit ClinicalTrials.gov and enter NCT05996003 in the "Other terms" search box, or contact study coordinator Gabi Niizawa at TrialInfo@NSPharma.com.**



FRDA is caused by mutations in the frataxin gene that lead to the loss of frataxin protein.

Image: Wirestock/Getty

**Friedreich ataxia (FRDA)**

# FRDA-related Heart Disease Therapy on Fast Track

The US Food and Drug Administration (FDA) granted Fast Track designation to Lexeo Therapeutics' LX2006, a gene therapy candidate for cardiomyopathy (heart muscle disease) in people with FRDA. The Fast Track program is designed to speed the development and expedite the review of drugs to treat serious conditions and fill an unmet medical need.

According to Lexeo Therapeutics, cardiomyopathy is the leading cause of death in people with FRDA, and there are currently no approved treatment options. In preclinical studies, LX2006 reversed the cardiac abnormalities in FRDA disease models and improved cardiac function and survival while demonstrating a favorable safety profile. Researchers at Lexeo Therapeutics are currently evaluating the safety and efficacy of LX2006 in people in an open-label, multicenter phase 1/2 clinical trial called SUNRISE-FA.

**LX2006 is designed to deliver a functional copy of the *FXN* gene to heart cells to enable them to produce more frataxin protein.**

FRDA is caused by mutations in the frataxin (*FXN*) gene that lead to the loss of frataxin, a protein that the nervous system and heart need to produce energy and work properly. LX2006 is designed to deliver a functional copy of the *FXN* gene to heart cells to enable them to produce more frataxin protein.

All study participants will receive one dose of the investigational treatment intravenously (in the vein). This study will last approximately five-and-a-half years, including a screening period, a 52-week study assessment period, and a four-year follow-up period. Participants will be required to attend 15 office visits during the first year of the study, five study center visits, and three remote visits during the follow-up period.

To be eligible, individuals must be between 18 and 50 years old, have a confirmed genetic diagnosis of FRDA with onset before age 25, have evidence of cardiomyopathy, and meet other inclusion criteria.

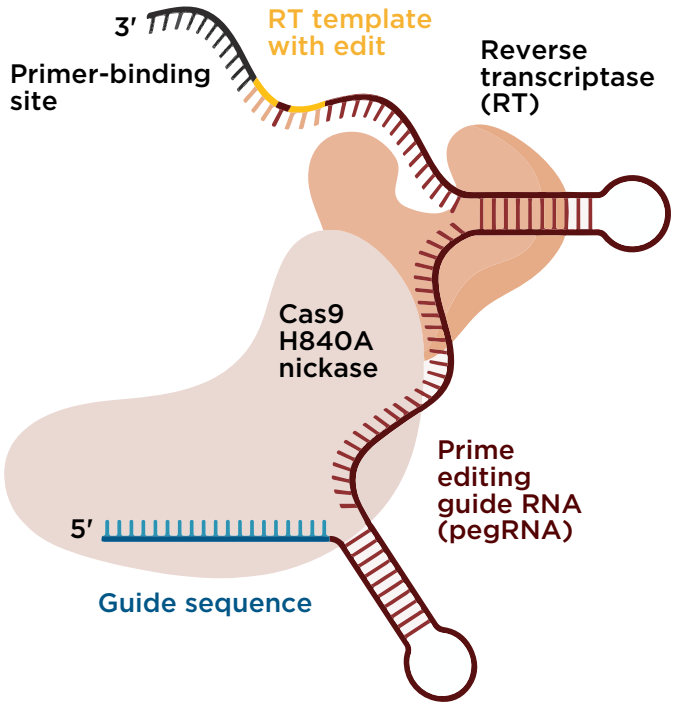
**To learn more about the SUNRISE-FA trial, contact the Lexeo team at ClinicalTrials@lexeotx.com or visit ClinicalTrials.gov and enter NCT05445323 in the "Other terms" search box.**

**Friedreich ataxia (FRDA)**

# Grant Supports Gene Editing Research

MDA and the Friedreich's Ataxia Research Alliance (FARA) announced a $300,000 collaborative grant awarded to Jonathan Watts, PhD, Professor of RNA therapeutics; Erik Sontheimer, PhD, the Pillar Chair in Biomedical Research and Professor of RNA therapeutics; Scot Wolfe, PhD, Professor of molecular, cell, and cancer biology; Wen Xue, PhD, Associate Professor of RNA therapeutics; and a team of investigators at UMass Chan Medical School. This funding will allow further research into using novel genetic technologies to treat FRDA; it does not involve a clinical trial or study on people.

**COMPONENTS OF PRIME EDITING**




The researchers are studying using prime editing (PE), a next-generation CRISPR gene editing tool that can precisely target the removal of GAA repeat expansions in the *FXN* gene. In FRDA, GAA repeat expansions disrupt the production of frataxin protein, leading to progressive damage to the nervous system. PE aims to directly address the cause of the disease.

The research team will compare several PE approaches for their ability to remove the GAA repeats in FRDA cells, with the goal of identifying the optimal tool that can provide high editing efficiency and a reduced rate of off-target modifications to the genome. The investigators have also devised a system called split PE, in which two halves of the PE machinery are delivered as separate molecules. This approach allows them to rapidly test combinations of editing enzymes with desirable properties.

The grant from MDA and FARA will support the diversification of research opportunities, increasing the chance of finding a successful method.

**To learn more about MDA's research grants, visit mda.org/gaag.**


Image: Andy/Getty

**+RECRUITING CLINICAL TRIALS**
Find a list of trials actively recruiting individuals to help advance research and treatment development at **mda.org/clinical-trial-updates**.

**Myasthenia gravis (MG)**

# Participants Needed for Genome Study

Researchers at George Washington University are seeking individuals living with muscle-specific tyrosine kinase-associated myasthenia gravis (MuSK MG) to participate in a genome-wide association study (GWAS). A GWAS compares genetics from many different people to find markers associated with a particular risk of disease.

This GWAS aims to collect a one-time saliva sample from 1,000 people with MuSK MG in the United States. Researchers will use the samples to identify genetic variations associated with MuSK MG, better understand the cause of MuSK MG, and identify biomarkers and targets of therapy.

This study does not involve an intervention. To be eligible, individuals must be US residents and provide a lab report confirming a MuSK MG diagnosis, among other criteria.

**For more information, visit musk1000.smhs.gwu.edu or contact the study team at musk1000@mfa.gwu.edu or 202-677-6205.**

# Understanding Myositis

*Q&A with Tahseen Mozaffar, MD*

BY MYRNA TRAYLOR

Myositis, a chronic inflammation of the skeletal muscles (muscles that connect to bones), can cause symptoms similar to those experienced by people with muscular dystrophy, including muscle weakness and fatigue. To learn more about myositis, Quest Media spoke with Tahseen Mozaffar, MD, a neuromuscular neurologist at the University of California, Irvine, and director of the UC Irvine-MDA ALS and Neuromuscular Center, who has been studying this group of diseases for 30 years.

Tahseen Mozaffar, MD

**Muscular dystrophies usually manifest over years, whereas myositis can manifest within weeks or months.**

Image: Prostock-Studio/Getty

### What is myositis?

Myositis is not a muscular dystrophy. Muscular dystrophies are usually inherited. Myositis is an acquired disease that causes inflammation in the muscles. It could be related to patient susceptibility to autoimmune conditions, or it could be in response to environmental factors. It primarily affects the skeletal muscles, causing significant disability from muscle weakness, but there also can be involvement of the respiratory muscles — the diaphragm or ribcage muscles — which can make it difficult to breathe in enough oxygen, blow out enough carbon dioxide, or clear airway secretions, increasing risk for pneumonia. In very rare instances, myositis can cause inflammation in the heart, where it becomes myocarditis, which can make it harder for the heart to pump blood.

There are five forms of primary myositis. The four more common types of myositis are dermatomyositis, immune-mediated necrotizing myopathy, anti-synthetase syndrome, and inclusion-body myositis (IBM). The fifth type, polymyositis, is considered extremely rare and likely overdiagnosed.

### How is myositis diagnosed?

Muscular dystrophies usually manifest over years, whereas myositis can manifest within weeks or months. It's a much shorter period of onset compared to muscular dystrophy, but both will present with muscle weakness. If a myositis patient comes to

a physician's attention later in their disease course, patients may be misdiagnosed with muscular dystrophy.

A myositis diagnosis will involve a patient history, blood tests for muscle enzymes, muscle and skin biopsies, and perhaps MRI scans, nerve conduction, and electromyogram tests.

### How is myositis treated?

Although there is no cure for myositis, there are several treatments that can help, including corticosteroids or other immunosuppressants; intravenous immunoglobulin; or in some cases, monoclonal antibodies, such as rituximab.

Also, some drugs for myasthenia gravis (MG), such as efgartigimod, are being tested for myositis. One example is a class of drugs called CD19 inhibitors, which are being used in multiple sclerosis, as well as MG. There is significant overlap with the underlying immune process among myasthenia gravis, multiple sclerosis, and myositis, so some of these drugs can be cross-utilized.

### Are there other treatment options on the horizon?

There are a number of clinical trials currently enrolling using some of the same drugs that have been approved for lupus and rheumatoid arthritis, which also involve an immune response.

We're also doing cutting-edge research using CAR T-cell therapy, which is employed in cancer treatments. In CAR T-cell therapy, T lymphocytes (immune cells) are extracted from a patient's own blood and modified to make them target-specific antigens that target antibody producing B and plasma cells (antigens such as CD19 or B-cell maturation antigen (BCMA). The T lymphocytes are reintroduced to the patient's system, where they destroy these immune cells, reset the muscle cells, and reduce the inflammatory response. Q

Myrna Traylor is a writer for Quest Media.

## TYPES OF PRIMARY MYOSITIS

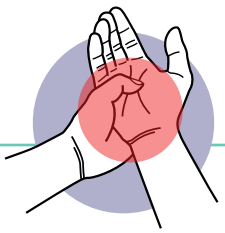


Image: Leremy Gan/Getty

+ **Inclusion-body myositis (IBM)** is an autoimmune disorder that occurs when inflammatory cells infiltrate the spaces between muscle fibers. Although it is generally painless, this form of myositis causes progressive muscle weakness in the arms, hands, and legs, as well as in the neck and esophagus. Disease onset usually begins in the 50s.

+ **Dermatomyositis** is an autoimmune disorder that occurs when inflammatory cells attack the small blood vessels that supply muscles and skin. This causes muscle weakness and a discolored rash. These two symptoms may show up at the same time, or months or years apart. Muscle weakness usually occurs in the shoulders, upper arms, hips, and thighs. Dermatomyositis usually occurs in children 5 to 15 years old or adults 40 to 60 years old. People assigned female at birth are more likely to have the disease.

+ **Immune-mediated necrotizing myopathy (IMNM)** occurs when the immune system attacks muscle fibers, leading to cell death (necrosis). It mainly affects adults, causing weakness in the hips and thighs. It may be misdiagnosed as limb-girdle muscular dystrophy (LGMD). Like the other myositis types, the exact cause is not known; however, IMNM can be a side effect of the use of the group of cholesterol-lowering drugs known as statins. People who receive prescriptions for statins should ask their physicians for muscle enzyme tests before and during treatment to check for the possible development of IMNM.

+ **Anti-synthetase syndrome (ASS)** is an autoimmune disease affecting multiple systems of the body. In addition to causing muscle weakness in the shoulders and hips, it may cause symptoms on the skin that can be mistaken for dermatomyositis. However, many people with ASS also have severe arthritis and interstitial lung disease, which causes fibrosis (scarring) of the lungs. Onset can start in the late teens, but people over 50 are more likely to have it.

+ **Polymyositis** is believed to be an autoimmune disorder in which immune cells attack muscle fibers, causing inflammation and weakness in the muscles closest to the trunk of the body. It is twice as likely to occur in those assigned female at birth but rarely occurs in people under age 18. Polymyositis is considered to be very uncommon and probably accounts for <2% of all myositis.

# Real-life Hacks

*MDA Ambassadors share their favorite tips and tricks for everyday problem-solving*

BY AMY BERNSTEIN

Did you know an MDA Ambassador can be any age and live anywhere in the United States with any neuromuscular diagnosis? (Turn to page 13 to learn more about MDA's Ambassador Program.) This means our current MDA Ambassadors are a pretty varied bunch. But there's one thing they have in common: They're great problem-solvers.

That's why we asked MDA Ambassadors to share some of their favorite life hacks — clever methods of making a common challenge a little (or a lot) easier.

*"My favorite life hack is using a* rolling backpack *to put my breathing ventilator in so I can use it and be mobile. This also helps my assistants easily transport the machine instead of carrying it on their shoulders."*
*— Stephanie Chicas, congenital muscular dystrophy (CMD), Virginia*

*"My favorite life hack is* using fondue forks *or wooden skewers to self-feed foods that would usually require a fork. Regular forks are too heavy, and plastic forks are not sharp enough."*
*— Darci Garcia, amyotrophic lateral sclerosis (ALS), Texas*

*"One of my favorite life hacks is adding* decorative knobs *to my dresser. I choose knobs that are larger than the traditional knobs and make the drawers easier to open — and cuter!"*
*— Allyson Pack-Adair, mitochondrial myopathy (MM), Arizona*

*"My favorite life hack is for* selfie-taking. *Here's how I get a great pic every time, with little to no help: First, I set my phone to take a 4K video. Next, I frame up the shot using a case stand or propping my phone against something. Then, I take a video of myself. When I am done, I can scroll through the video and take screenshots of the best moments."*
*— Fred Graves, limb-girdle muscular dystrophy (LGMD), Connecticut*

*"My favorite life hack involves* having multiples *of my daily go-to items, like sunglasses and car keys. I keep them in different places around my apartment, on my wheelchair, or in my vehicle, so I never have to worry about forgetting them and having to backtrack. When you have a disability, time (and energy) is already compromised, so taking what little steps I can to be proactive and save time and effort is helpful."*
*— Tana Zwart, facioscapulohumeral muscular dystrophy (FSHD), South Dakota*

*"My favorite life hack is choosing products that I can control with* my phone *— from my Nest thermostat to my Victrola record player — which makes life much easier and most accessible!"*
*— Ira Walker, spinal muscular atrophy (SMA), Florida*

*"My favorite life hack is sleeping in* satin pajamas. *Sounds silly, but they have changed my life! Turning over and moving in bed can be a challenge, but silky pajamas make it so much easier to just slide with less effort."*
*— Amy Shinneman, Bethlem myopathy, Indiana*

*"One life hack that helps me a lot is* using gloves with grips *on the palms and fingers to pick up items. I use Stix-On safety gloves when I'm in the kitchen to pick up bottles or cans, carry small bags, or even pick up something off the floor. These gloves have been a lifesaver!"*
*— Mashauna Black, LGMD, North Carolina*

*"One of my favorite life hacks is to wear any kind of* shoe that laces up the front. *I can open the laces nice and wide and slip my foot in easily without needing to have lots of flexibility in my ankles. Then I can pull the laces as tight as I want — pulling the laces tight on my shoes once I have them on almost works like a compression sock and keeps the puffiness in my foot to a minimum. Since I am a full-time wheelchair user and don't have to worry about tripping over my feet, I also buy my shoes a half size to a full size bigger than I need to make them that much easier to get on."*
*— Mindy Henderson, SMA, Texas*

Amy Bernstein is an editor and writer for Quest Media.

## EXPERT IDEAS FOR EASIER INDEPENDENT LIVING

Occupational therapists (OTs) are professionals who assess challenges and limitations in areas of daily living and propose solutions and modifications. Contrary to a common misconception, many of those recommendations are inexpensive and easy to manage at home.

OT Brooke Aarvig provided these hacks for dressing and undressing:

- Learn to dress in a seated position while shifting your weight on your bottom from side to side. This can be less tiring and safer than standing.
- If one side of your body is weaker than the other, always dress your weak side first and your strong side second. Do the opposite when undressing — undress your strong side first and your weak side second.
- Use a reacher grabber tool to reach your feet and help pull on socks, slippers, or sandals.

Brooke offers many more ideas in "Easy, Expert-Level Life Hacks from an Occupational Therapist" at **MDAQuest.org/expert-life-hacks**.

# Research 101: Becker Muscular Dystrophy

The remarkable progress to develop treatments for neuromuscular disorders during the past decade would not have been possible without the participation of patient partners in the research process. This process starts long before a drug, device, or procedure comes before the US FDA or other bodies for marketing approval.

Clinical research is one of the most important ways that scientists can learn and improve our understanding of diseases. Some research studies aim to understand disease progression while others study how disease progression may be modified by a therapeutic drug or device. Results from clinical research can provide much-needed information to optimize clinical care practices. **Currently, multiple studies are enrolling individuals living with Becker muscular dystrophy to evaluate disease progression or to assess the effect of an investigational drug**.

Participation in clinical research helps speed up advancements in care, quality of life, and drug development. There are many ways those interested can get involved.

## Registries

A registry collects clinical and other data for those with a particular disease or drug exposure. They are important in the planning and logistics of rare disease studies and drug development. Registries can either be:

- **Patient-reported:** a patient or caregiver is responsible for entering their data, or
- **Physician-reported:** a care provider or clinical trial investigator is responsible for entering data about patients.

## Natural History Studies

Natural history refers to information about disease onset, how it progresses, and other essential information about living with a specific disease. Natural history studies allow researchers to track the progression of symptoms and how a disease affects patients over time, and information from these studies is important throughout drug product development Information from these studies can help with:

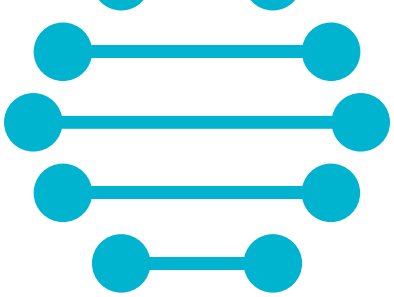
- Discovery and clinical trial design
- Post-approval therapy surveillance
- Understanding how interventions are working
- Regulatory considerations
- Treatment access

## Quality of Life Studies

Quality of life studies look at the impact a condition is having on patient and caregiver quality of life, which can include its physical effects, functional impact, mental wellness, and social health.

The aim of these studies is often to discover new ways to improve patient and caregiver comfort and quality of day-to-day living. Information from these studies can lead to improvements in:

- Innovations in mobility equipment
- Home modifications and automation systems
- Vehicle adaptations
- Public Transportation





## Interventional Studies or Clinical Trials

Clinical trials test experimental therapies, new combinations of therapies, or new approaches to care. They are typically done in phases that range from initial studies in individuals that may not have the disease to phase 4 trials in patients that assess the performance of approved drugs in the real-world setting.

- **Phase 1 trials** test an intervention for safety and side effects and enroll a small number of humans, often healthy volunteers. Trials with investigational drugs are first reviewed by regulators to make sure participants are watched closely for side effects.
- **Phase 2 and 3** include patients living with the condition who have volunteered to enroll in a clinical trial.
  - **A phase 2 clinical trial** gathers data to understand if an investigational treatment is effective on people who have the condition, many times at different dose levels, and collects additional safety information.
  - **A phase 3 (or pivotal) trial** gathers data to find out whether the investigational treatment offers a benefit in larger groups of people living with the condition and typically compares the investigational treatment to other commonly used treatments and/or the standard of care.

Both phases 2 and 3 (phase 3 trials are sometimes called pivotal studies) may include a randomization period, meaning some participants may receive the investigational drug and some may receive a placebo (an inactive form of the investigational drug).

Oftentimes, the sponsor of these trials will cover expenses such as travel and lodging for participants

## Open Label Extension Studies

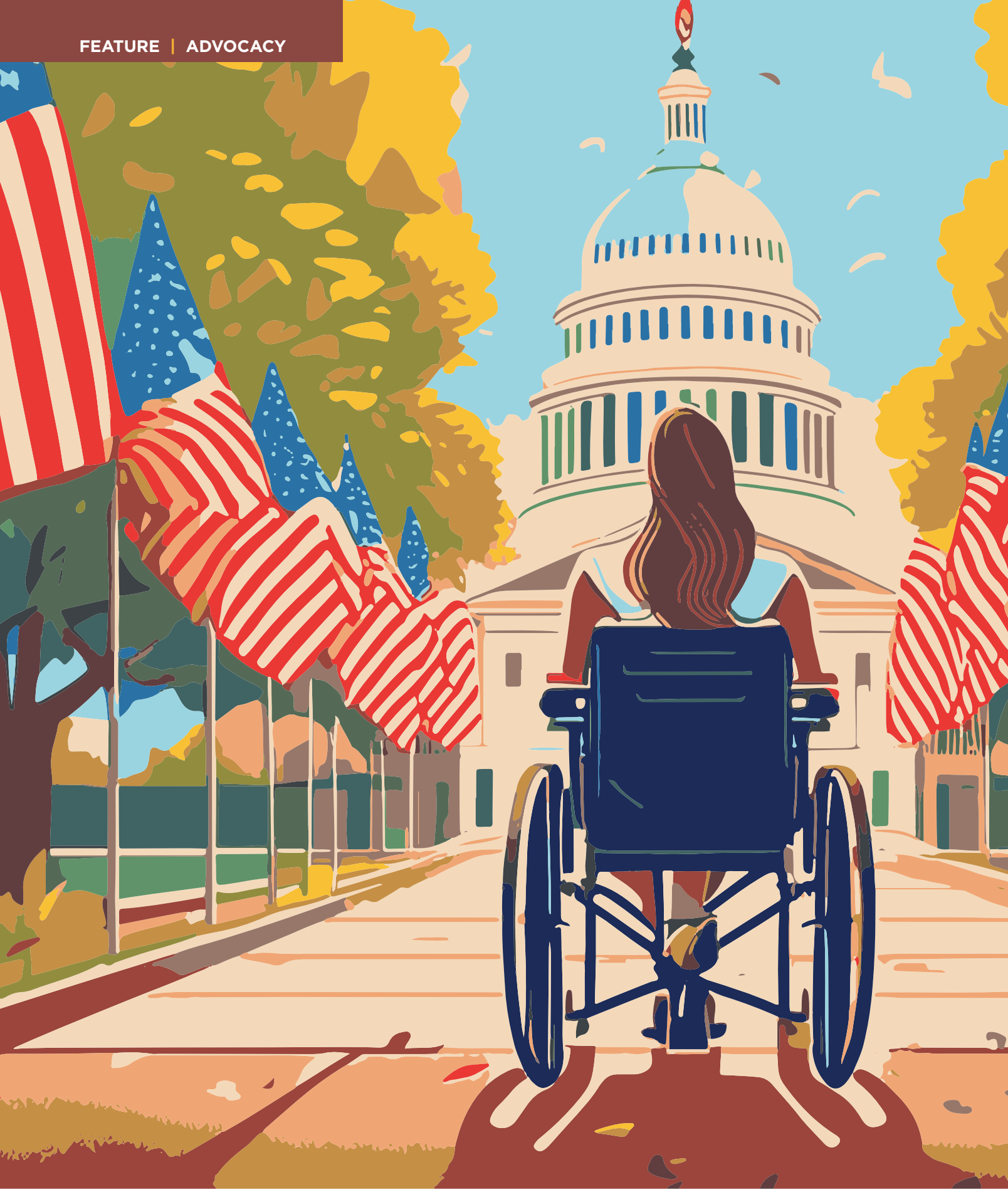
Open label extension (OLE) studies happen after the randomization phase of a trial is complete to continue to understand longer-term treatment. Eligible trial participants go on to receive the active form of the investigational treatment, typically until a decision is made by regulators about if the drug is eligible for approval.

# Learn More

For many people living with neuromuscular disorders, participation in clinical research provides more than the hope for treatment. It provides a chance to contribute to scientific understanding and advancements that will improve lives now and in the future. To find out more about Becker trials currently enrolling, go to mda.org/research/clinical-trials or clinicaltrials.gov and search for “Becker”.

## Becker Education and Engagement Day

On September 28th, there will be a Becker Education and Engagement Day will be held in six locations across the United States: Seattle, Sacramento, Dallas, St. Louis, Orlando, and Pittsburgh. This in-person event will provide education about many facets of living with Becker muscular dystrophy, including information about research and clinical trial opportunities. For more information or to register for the Becker Education and Engagement Day, go to beckereducationandengagement.com.



*Advocates and voters with disabilities influence legislation for the better*

BY STEVE WRIGHT

# Change the Rules

When you think about great legislation for people with disabilities, you probably think of the Americans with Disabilities Act of 1990 (ADA). This important law prohibits discrimination against individuals with disabilities in many areas of public life, including jobs, schools, businesses, and transportation. It is designed to ensure that people with disabilities have the same rights and opportunities as everyone else.

*"While passing the ADA was a major achievement for the disability community, much work remains to be done. I can't emphasize enough how important it is to make your voice heard."*

***— Deborah McFadden***

"The disability community came together to make this landmark law, showing the collective power we have," says Deborah McFadden, who was instrumental in writing the ADA and getting it passed in her role as US Commissioner of Disabilities under President George H. W. Bush. Deborah now operates Abilities Count (**Abilities Count.com**) to assist families in navigating disability benefits.

"While passing the ADA was a major achievement for the disability community, much work remains to be done," she says. "I can't emphasize enough how important it is to make your voice heard."

Thanks to the dedication of countless advocates like Deborah, in the decades since the ADA was passed, we have seen some important progress in public policy and disability awareness. From lobbying efforts by MDA and other organizations to calls, emails, and letters to representatives by grassroots advocates, making our voices heard has been a powerful tool for change.

### Success stories

It's encouraging to look at how the disability community has positively influenced public policy regarding disability benefits in the last few years.

Deborah McFadden

In 2022, Congress passed the ABLE Age Adjustment Act. Starting in 2026, the age of onset of disability to be eligible for an ABLE account will change from 26 to 46. These tax-advantaged savings accounts allow individuals with disabilities to save and pay for disability-related expenses without endangering their benefits. The policy change is estimated to make an additional 6 million people eligible for ABLE accounts.

In addition, the 2022 Inflation Reduction Act requires Medicare to cap the out-of-pocket costs of medications at $2,000 starting in 2025. Currently, people with neuromuscular diseases on Medicare can see drug costs in the many thousands of dollars. Advocates from the neuromuscular and disability communities who contacted their members of Congress were crucial in getting this law passed.

In 2023, MDA teamed up with other disability advocacy organizations to successfully defend the Medicaid program from work requirements and cuts that would have made it harder to qualify for Medicaid services or reduced benefits for people with disabilities.

Another win for the neuromuscular community came in May 2023, when Medicare announced it would cover seat elevation systems for power wheelchairs. Leading up to the announcement, the MDA Advocacy team submitted formal comments to the Centers for Medicare and Medicaid Services (CMS) supporting this additional coverage. In addition, MDA's grassroots advocates sent in their own personal comments encouraging the CMS to make this change.

"Our advocates are spectacular at telling their stories to show why changes to laws and rules are important," says Mark Fisher, MDA's Director of Advocacy Engagement. "One thing I love about advocacy at MDA is how our advocates fight so hard for

change, whether it's a big piece of legislation or a small tweak within an agency. I'm so proud of what our advocates have accomplished."

### Advocacy in flight

Improving air travel for people with disabilities is an area MDA has been engaged in for many years.

For example, in 2022, the US Department of Transportation (DOT) held a listening session on air travel with wheelchair users. People with neuromuscular diseases were among the broad base of advocates who detailed unacceptable conditions — everything from injuries caused by improper seat transfers to damaged mobility devices. In response, the DOT released the Airline Passengers with Disabilities Bill of Rights.

In late 2023, after MDA advocates shared their personal air travel experiences with Transportation Security Administration (TSA) officials, the TSA invited MDA to provide officer training on interacting with travelers who use mobility devices.

This year, Congress passed the long-awaited Federal Aviation Administration (FAA) reauthorization. This major piece of legislation includes reforms that will make air travel more accessible for people with disabilities, including:

- Airport and airline personnel will receive stronger, hands-on training on assisting passengers with disabilities and stowing wheelchairs.
- Airlines will be held accountable for reporting damage to wheelchairs.
- The FAA will continue to study the option of allowing passengers to stay in their wheelchairs during flight.
- People with disabilities will have a seat at the table when the government makes future decisions about air travel.

This law is the result of advocates sharing their air travel stories and telling lawmakers how the provisions in the bill will improve their flying experience. MDA advocates made personal connections with their representatives to provide key information during the reauthorization process.

"I would say that accessible air travel is one area where MDA has helped create immense progress," Mark says.

## ONE PERSON CAN MAKE A DIFFERENCE

Two MDA advocates on the power of sharing your story:

"There were eight kids in my family, and five of us have limb-girdle muscular dystrophy [LGMD], and I have two nieces with LGMD. I was more involved in advocacy after I was in the first gene therapy trial for muscular dystrophy back in 1999. It opened up doors for me to tell my story and to try to get the government to help people living with neuromuscular diseases.

I believe one person can make a difference in telling their story and connecting to the political people in Washington, DC, because it's the personal stories that politicians need to hear.

I hope that being involved in advocacy will make a difference for my family and others."

— Donavon Decker, Kansas

"I had the typical misdiagnosis journey before I learned I have facioscapulohumeral muscular dystrophy [FSHD].

One of the many things I love about MDA Advocacy is they send emails encouraging us to advocate. These emails are filled with all the information you need to send comments to senators and representatives. With one positive experience reaching out to your elected officials, you get a good handle on how easy it is to do, how worthwhile it is, and how you can make an impact on many individual's lives.

One person advocating can make a big difference. It is like that little snowball rolling down a hill that just keeps getting bigger and bigger and bigger."

— Ranae Beeker, Washington State

### +HOW WILL YOU VOTE?

Did you know the Voting Rights Act of 1965 allows a person with a disability to bring a friend, family member, or other person to help them vote? Before election day arrives, brush up on your voting rights and the policies in your area, such as absentee and early voting, at **mda.org/vote**.

**Current issues**

While the neuromuscular and disability communities celebrate the progress made, they know there is still much to do to ensure that people with disabilities have the same opportunities as everyone else.

"Our community has told us to focus on reforming federal benefit programs, to prioritize SSI benefits that are better tailored to the century we live in," says Paul Melmeyer, MDA's Vice President of Public Policy and Advocacy. "Without changes, people risk SSI or Medicaid benefits if they pursue a career and their pay makes them ineligible."

The Supplemental Social Security Income (SSI) Penalty Elimination Act, introduced in the Senate in 2023, addresses this issue. If approved, it would increase SSI asset limits from $2,000 for individuals and $3,000 for couples to $10,000 for individuals and $20,000 for couples — and these limits would be adjusted annually for inflation. This would make it easier for individuals with disabilities to hold jobs and save for unexpected expenses without jeopardizing their benefits.

Congress is also considering legislation related to Home and Community-Based Services (HCBS). The HCBS Access Act would increase access to in-home caregiving for people who need assistance with activities of daily living, such as bathing and dressing. The HCBS Relief Act supplies temporary additional funding for HCBS services with the goal of increasing the caregiver workforce.

Other important issues and pending legislation include:

- Making sure telehealth continues to be accessible for those who use it for essential medical care.
- Increasing access to genetic medicine and counseling.
- Improving access to specialized multidisciplinary care for the amyotrophic lateral sclerosis (ALS) community via the ALS Better Care Act.

If approved, it would increase SSI asset limits from **$2,000** for individuals and **$3,000** for couples to **$10,000** for individuals and **$20,000** for couples — and these limits would be adjusted annually for inflation.

**Access to voting**

From the presidential election in November to the thousands of statewide, district, and local races taking place throughout the year, MDA encourages its members to make their voices heard at the ballot box.

Shaun Hill

Shaun Hill, MDA's Manager of Public Policy and Advocacy, is leading the Access the Vote campaign (**mda.org/vote**) to provide resources and guidance for navigating the electoral process. She urges MDA members to use their right to vote to champion legislation impacting the disability community.

"Access the Vote is all about education, engagement, and empowerment," Shaun says. "The initiative aims


to help people understand our government, the issues that affect our community, and how these things intersect with our daily lives."

Along with teaching individuals how to be active, informed voters, Access the Vote addresses the barriers people with disabilities still face to exercising their right to vote — from lack of transportation to inaccessible voting machines. It is a vital resource for information on disability voting rights and practical tools for preparing to vote.

Deborah recounts: "I was talking to five people who had just turned 18 that I helped with SSI and vocational rehabilitation, and I asked them if they were registered to vote. Not one was. I told them voting is the cornerstone of Democracy. If there is one thing I can say to parents of children with disabilities, it is to vote and encourage your children to register when they turn 18."

MDA wants to ensure that everyone in the neuromuscular community has the knowledge and opportunity to engage in the electoral process at all levels of government.

### Making our voices heard

Voting is a right of all Americans, but too few are exercising that right. According to the US Census Bureau, 67% of eligible voters cast ballots in the 2020 presidential election, but 62% of people with disabilities voted. An analysis by the Election Assistance Commission found that if people with disabilities had voted at the same rate as people without disabilities, there would have been about 1.75 million more voters.

"One in four people have a disability. It's the largest minority voting bloc in America. It is even bigger when you consider the family, friends, and caregivers," Deborah says. "Our voices have been heard. But if we were even more active — as voters and as advocates pressing for inclusion — we would have a huge voice that could positively impact many things." Q

Steve Wright is an award-winning writer and advocate based in Miami. He lectures throughout the US and abroad on creating a better built environment for people with disabilities.

*"Access the Vote is all about education, engagement, and empowerment. The initiative aims to help people understand our government, the issues that affect our community, and how these things intersect with our daily lives."*
***— Shaun Hill***

# ARE YOU READY FOR GEN

# E THERAPY?

Image: grivina/Getty

*Many factors go into deciding to get gene therapy. Here's what you need to know.*

BY LARRY LUXNER

When Marie Bedirlian's son Douglas, 6, was diagnosed with Duchenne muscular dystrophy (DMD) as an infant, she thought she knew what to expect. Douglas's older brother, Brayden, 14, also lives with DMD. At a young age, Brayden couldn't jump and had trouble running. Later, he started tiring quickly while walking, and at 11, he started using a wheelchair full-time.

But in 2023, the US Food and Drug Administration (FDA) approved a gene therapy for DMD in young boys that may delay or halt the progression of the disease. Marie, of Bellflower, California, seized the opportunity for her younger son. On October 24, 2023, Douglas became the second person to receive the newly approved delandistrogene moxeparvovec-rokl (Elevidys®) at the MDA Care Center at Children's Hospital of Los Angeles.

"My hope was for him to walk, jump, run, and play like any other child his age could do," Marie says. "And this gene therapy has made that possible, for now."

In fact, gene therapy is emerging as a promising area of treatment for neuromuscular diseases. However, access to gene therapies depends on many factors, including age, proximity to an infusion center, and insurance approval. As we enter this new era of gene therapy, here's what individuals with neuromuscular diseases and their families need to know.

### Why gene therapy?

Most neuromuscular diseases are caused by genetic mutations. Gene therapy aims to treat a genetic disease at its source by replacing a mutated gene with a working gene, repairing a flawed gene, or altering how a gene is controlled.

So far, gene therapy results have varied, but when it works, the effects can be dramatic. Some children who were identified with spinal muscular atrophy (SMA) through newborn screening and given the gene therapy drug onasemnogene abeparvovec (Zolgensma®) as infants have not developed symptoms of the disease years later. The hope is that they never will.

Marie Bederlian's sons Douglas (front left) and Brayden (front right) have Duchenne muscular dystrophy.

**Most neuromuscular diseases are caused by genetic mutations. Gene therapy aims to treat a genetic disease at its source by replacing or repairing a flawed gene, or altering how a gene is controlled.**

Zolgensma was approved in 2019 for children under 2 years old with SMA. Elevidys was approved in 2023 for boys 4 and 5 years old with DMD, and the approval was expanded in June 2024 to ages 4 and older. These are currently the only two gene therapies approved for neuromuscular diseases. However, many more are being tested to treat diseases such as amyotrophic lateral sclerosis (ALS), Charcot-Marie-Tooth disease (CMT), Friedreich ataxia (FA), limb-girdle muscular dystrophy (LGMD), and Pompe disease.

MDA has made key contributions to the gene therapy field that helped lead to these advances, including funding the first muscular dystrophy gene therapy clinical trial in 1998 and the first DMD gene therapy trial in 2006. Many of the gene therapies in clinical trials for neuromuscular diseases in the United States are based on strategies developed with MDA funding.

Researchers expect more approvals in the next few years, and many people will want to know how they can access gene therapies as they become available.

### Access to gene therapy

Several factors are involved in who can get gene therapy:

- **Diagnosis.** Gene therapies are designed to replace or block the function of specific genes. Genetic testing to confirm a genetic diagnosis — or, in some cases, a certain type of gene mutation — is required to qualify for a gene therapy.
- **Age.** New drugs are tested and approved for specific ages. As with Elevidys, some therapies are initially approved for a narrow age range, but as more data emerge to suggest they are safe and effective in a wider population, that approval may be expanded.

Image: grivina/Getty

- **Preexisting antibodies.** Adeno-associated virus (AAV) vectors are the leading platform for delivering gene therapy to cells in the body. Zolgensma and Elevidys both use AAV vectors. AAVs are small, naturally occurring viruses that do not cause sickness in humans and can carry genetic material. However, anyone who has been exposed to an AAV before may have developed some immunity to it. Before receiving a gene therapy that uses an AAV vector, individuals must be tested for antibodies to the virus.
- **Cost.** The current market price for Zolgensma is $2.1 million, and for Elevidys, $3.2 million. The extensive research involved in developing a new gene therapy and the cost of manufacturing it contribute to the price. Health insurance companies require preauthorization before approving coverage for these expensive drugs. "Each insurer is different, and while some cover gene therapy, some don't," says Nora Capocci, MDA's Executive Vice President for Healthcare Services. Because gene therapy is so new, many insurers don't have policies in place for them and are looking at each case individually. "There have been instances where people have to appeal a decision two or three times," Nora says.
- **Clinical readiness.** Not all healthcare facilities have an infusion center with the proper equipment to store gene therapy drugs or staff trained in administering them and monitoring patients afterward.

"Families looking to pursue treatment may find that their local hospital or clinic may or may not be prepared to administer gene therapy. That doesn't mean they won't be able to get it," says Crystal Proud, MD, a neurologist at the MDA Care Center at Children's Hospital of the King's Daughters in Norfolk, Virginia. Her facility has dosed more than 40 children with Elevidys or Zolgensma.

**BEHIND THE SCENES**

Listen to a conversation on the science and research behind new treatments for neuromuscular diseases at **MDAQuest.org/podcast/new-treatments**.

Laszlo Mechtler, MD

"We're going to need more collaboration between healthcare centers. Some are very experienced in administering these therapies," Dr. Proud says.

MDA created the Gene Therapy Support Network to help individuals and families navigate the new landscape and challenges to access. Learn more about this service in "A Partner on the Gene Therapy Journey" on this page.

### Risks and rewards

Nora stresses that while new gene therapies offer incredible promise, "there are no guarantees that gene therapy will improve function or stop progression in any individual."

"In clinical trials of gene therapies, patients have responded differently in terms of benefits and side effects," Nora says. "Potential risks, side effects, and expectations of benefit must be thoroughly reviewed with your physician before receiving any gene therapy."

Gene therapy is generally considered safe, but Laszlo Mechtler, MD, director of neurology at the Dent Institute in Buffalo, New York — also an MDA Care Center — notes that all gene therapies have the potential to cause adverse side effects. In clinical trials, serious but rare side effects have included severe immune responses and damage to internal organs, such as the heart and liver. There have been several gene therapy-related deaths in neuromuscular disease trials.

Researchers are continuing to look for ways to make gene therapy safer. For example, in some recent DMD gene therapy trials by Sarepta, Solid Biosciences, and Pfizer, five participants developed immune-mediated myositis, a condition in which the immune system attacks the muscles.

"What was remarkable was how all these pharmaceutical companies got together and pooled their resources to try to figure out what was causing this very serious reaction," says Omer Abdul Hamid, MD, a pediatric neurologist and Director of the MDA Care Center at Nemours Children's Hospital in Orlando, Florida. "They found that all five boys had large deletions that included exons 8 and 9." Based on this finding, they revised the eligibility criteria to exclude individuals with those genetic characteristics from that type of gene therapy.


Omer Abdul Hamid, MD

## A PARTNER ON THE GENE THERAPY JOURNEY

MDA created the Gene Therapy Support Network to help individuals and families navigate the new landscape and challenges of accessing gene therapies. It aims to partner with patients and their care teams throughout decision-making, gene therapy treatment, and post-gene therapy care.

The Gene Therapy Support Network can connect you to educational materials and resources, as well as to MDA Care Centers and the new Gene Therapy Community Group. Gene Therapy Support specialists are available to answer questions and provide one-on-one support by phone, email, or a scheduled video call.

The Gene Therapy Support Network also serves as a resource for healthcare providers, sharing insights and best practices from clinicians across the MDA Care Center Network who have successfully helped their patients access gene therapy.

To contact the Gene Therapy Support Network, call **833-ASK-MDA1** or email **ResourceCenter@mdaUSA.org**. Learn more and schedule a video call at **mda.org/GeneTherapySupport**.

## +FREE GENE THERAPY WORKSHOP

Take MDA's online Access Workshop: Access to Gene Therapy. This self-paced course explains gene therapy treatments, the FDA drug approval process, insurance coverage options, and more. Find it at **mda.org/AccessWorkshops**.

Dr. Laszlo attempts to put the risks and potential rewards of gene therapy in perspective: "You're taking a risk, but you're taking a risk with a progressive disease that has no treatment. When you take chemotherapy for cancer, you're also taking a risk. This is something the family must consider in their decision-making."

Image: grivina/Getty

### What to expect

At the MDA Care Center at Nemours Children's Hospital, Dr. Hamid has dosed children with Elevidys and Zolgensma. Both drugs are given as an intravenous (IV) infusion in a single dose.

Patients usually spend a day at the clinic to receive the infusion and be monitored afterward for adverse reactions. "The IV infusion goes over an hour and a half to two hours. On dosing day, families get there early," Dr. Hamid says. "In a sense, it's a very anticlimactic day, although it's a very important and memorable day for our families."

Following the infusion, the individual will need blood tests and follow-up visits for months or years to monitor the potential long-term effects of the gene therapy, and they will continue to manage their underlying neuromuscular disease.

Dr. Hamid stresses that gene therapy is a commitment and says that before seeking gene therapy for their child, parents should ask themselves: "Are they ready for the commitment it takes to undergo the frequent lab monitoring, as well as the risks of liver, heart, and other organ injuries? They will need to talk to their provider to see if their child is a good candidate."

The final consideration is, perhaps, the biggest mystery of all when it comes to gene therapy: How long will it last? Based on animal studies, the answer seems to be at least several years — although the exact duration is unknown and may differ from person to person.

"Factors that determine how long gene therapy will last in the body include the type of cells being treated, age of the patient at dosing, dose level, disease progression, and patient-specific responses," Nora says.

Gene therapies delivered via adeno-associated virus (AAV), such as Elevidys and Zolgensma, are limited to a single treatment because of current technology

*"Factors that determine how long gene therapy will last in the body include the type of cells being treated, age of the patient at dosing, dose level, disease progression, and patient-specific responses."*

***— Nora Capocci***

and human biology. In 2023, MDA co-funded a project with Parent Project Muscular Dystrophy and CureDuchenne to focus on the hurdles of preexisting antibodies and redosing.

There are many challenges to overcome in developing and accessing gene therapies, but the potential to correct a gene mutation at its source, thus preventing or reversing the effects of a disease, motivates researchers to keep going. MDA is committed to funding gene therapy research and bringing scientists together at its annual Gene Therapy Summit to work through remaining obstacles.

Marie has seen encouraging results since her son Douglas received Elevidys in October 2023. "Within three weeks, we noticed a major difference," she says. "Before, he couldn't jump and would get tired easily. To go upstairs, he'd have to hang on and climb up one by one. Now, he gets up in four seconds and is walking normally." Q

---

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

*Living well as a senior with neuromuscular disease*

BY BARBARA AND JIM TWARDOWSKI, RN

# OLDEN YEARS

Aging is a journey, and everyone's path is unique. Older adults with neuromuscular diseases experience many of the same milestones as others — retiring, adapting to changing mobility and energy levels, modifying the home for convenience and safety, learning to accept help and caregiving — but managing a progressive disease while dealing with these common challenges requires extra flexibility and resilience.

Here, experts and members of the neuromuscular disease community weigh in on how aging intersects with neuromuscular disease and share six strategies to make this time of life truly golden.

Image: Halfpoint/Getty

## 1 Find purpose

Post-career blues are common. For Ranae Beeker, 65, it was especially difficult because she wasn't ready to retire when her facioscapulohumeral muscular dystrophy (FSHD) progressed to the point that she could not perform the physical duties of her job. As a registered nurse, she was on her feet, carrying heavy materials, doing paperwork, and filing. Her 40-year career in nursing ended when she applied for disability at 62.

This "forced retirement" left her feeling down. For a year, she wondered how she was going to fill her time. "My brain was used to working really fast and really hard, and all of a sudden, I didn't have anything to do. So, I started volunteering," says Ranae.

She began answering calls for a suicide prevention hotline, and she became an MDA Ambassador and participated in an advocacy trip to Washington, DC. Ranae also serves on three county committees and the disability ministry board of her church. "Volunteering makes me feel like I have a purpose — I can be productive," says Ranae.

## 2 Stay active

"A healthy older person needs to exercise more than they did at a younger age to maintain their muscle mass," says Catherine Loman-Hoerth, MD, PhD, a neurologist and Director of the MDA ALS Care Center at University of California San Francisco Medical Center. "For someone with a neuromuscular disease, even a mild underlying weakness can become more severe as they age due to loss of muscle mass."

Ranae Beeker participated in an advocacy trip in Washington, DC.

If a patient's weakness is severe, however, exercising can damage their muscles. Healthy muscle fibers will deteriorate over time from aging and the neuromuscular disease.

Dr. Loman-Hoerth recommends asking your physician for a neurological physical therapist (PT) referral. This healthcare professional instructs clients on appropriate stretching moves and strengthening exercises that will help maintain strength without damaging muscles.

"I try to make sure my patients are in physical therapy as much as possible because as the disease changes, the recommendations change. A PT will know how much exercise is too much or too little," Dr. Loman-Hoerth says.

The best way to stay active is to find activities you enjoy. Ed Linde, 66, lives with Charcot-Marie-Tooth disease (CMT) and uses a wheelchair.

"As I went through life, I progressed from ankle-foot orthoses (AFOs) to walking with a cane, to a walker, to using a wheelchair part-time. Now, I'm permanently in a wheelchair. So yes, I've faced my challenges, especially getting older," Ed says. He's chosen to continue doing the things he loves by making them work for his needs. Twice a week, he gets his exercise by bowling with adaptive equipment in a local league. Another activity he enjoys is wheelchair dancing with his wife at music venues, bars, and the American Legion hall. They took lessons with an instructor who adapted the moves for them. Ed and his wife also enjoy outings on his three-wheel motorcycle.

## 3 Think ahead

It pays to be prepared for changes that might be coming down the road. Ranae's training as a nurse helped her prepare for aging with FSHD. She listened to other people who have her disease and observed how they adapted. One smart decision she made was choosing a single-level home.

Ed Linde's three-wheel motorcycle allows easy transfer from his wheelchair.

"Living with a disability is not cheap; it's expensive," says Ranae, who consulted financial advisors and contributed to retirement savings plans through her work. Now, she lives modestly and tries to be wise with her money.

By developing friendships with like-minded people, Ranae has found affordable ways to have fun and not feel like she is missing out. "We get together and play games as opposed to going to shows or big vacations. Not that I have anything against vacations, but it's a lot of money I could use for something else," she says.

Ed and his family have lived in the same home for 25 years. Their shower doesn't accommodate a wheelchair, so he takes baths. This year, he bought a bathtub transfer bench when the weakness in his legs made it difficult to stand up. A walk-in tub might be a solution in the future.

Making home modifications can enhance independence and improve safety for aging residents. Before implementing changes, consult with an occupational therapist, who can help identify challenges before they occur and propose solutions.

## 4 Know your coverage

Medicare is available to anyone 65 or older and some people under 65 with disabilities. Medicaid requirements are based on income and other factors and vary from state to state. Medicaid offers some benefits that Medicare doesn't usually cover, like home healthcare and home modifications. It is possible to have both Medicare and Medicaid coverage.

Take the time to thoroughly review your health insurance coverage — some plans provide benefits such as discounts on meal delivery, gym membership, and wellness products. Gina Rossi, LCSW-BACS, DCSW, an adjunct instructor at the Louisiana State University School of Social Work, recommends asking your doctor for a referral for a clinical social worker who is familiar with local resources that may be able to assist you with a variety of needs, from building a ramp to finding respite care.

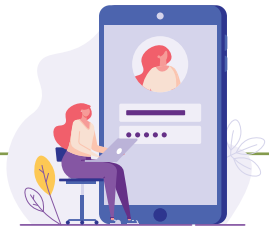
Dr. Loman-Hoerth recommends considering long-term care insurance, which would cover home healthcare, respite care, residential care, and other services. However, unlike health insurance, long-term care insurance is not required to cover preexisting conditions. (Learn more about planning for long-term care needs at **MDAQuest.org/long-term-care-needs**.)

**SPOT SCAMS**
Learn how to protect yourself from online scams that target older adults and people with disabilities at **MDAQuest.org/scams**.

## 5 Accept help

To do her volunteer work, Ranae needs help from the people around her. "If I can't get through a door or whatnot, my saying is, 'May I borrow your muscle?' That usually makes people giggle a little bit. I've never met anybody who didn't want to help me," she says. "I try to be very independent, but I also know every one of us is dependent upon somebody, whether it is to grow your food, provide you with electricity, or fix your car; we are all very much interdependent."

After Ranae retired, one day, she fell in her bathtub and could not get up. She used Alexa to call her son. "It was very scary," says Ranae. After the accident, her son moved into her home. He does the cooking, shopping, and gardening. He assists her with transfers from her wheelchair in the bathroom and into bed. Accepting her son's help was not easy, but she has set aside her embarrassment and pride. "I am blessed he is as kind and caring and understanding as he is," says Ranae.



## RESOURCES FOR SENIORS

You don't have to do the journey alone. Use these resources to find the support you need.

**BENEFITS**
Centers for Medicare and Medicaid Services
**cms.gov**

**MEDICAID**
**medicaid.gov**

**MEDICARE**
**medicare.gov**

**SERVICES**
Area Agencies on Aging
**ElderCare.acl.gov/Public/About/Aging_Network/AAA.aspx**

**CATHOLIC CHARITIES**
**CatholicCharitiesUSA.org**

**CENTERS FOR INDEPENDENT LIVING**
**ilru.org/projects/cil-net/cil-center-and-association-directory**

**DISABILITY INFORMATION AND ACCESS LINE**
**888-677-1199**
**dial.acl.gov**

**MDA RESOURCE CENTER**
**833-ASK-MDA1**
**ResourceCenter@mdausa.org**

**HOME MODIFICATIONS**
Government home repair assistance programs
**usa.gov/home-repair-programs**

**RAMPS**
**ramps.org**

**REBUILDING TOGETHER**
**RebuildingTogether.org**

**EDUCATION**
MDA Access Workshop: Access to Financial Independence
**mda.org/AccessWorkshops**

**MDA GUIDE FOR CAREGIVERS**
**mda.org/sites/default/files/2024/01/MDA_Caregiver_Guide.pdf**

**MDA MENTAL HEALTH HUB**
**mda.org/MentalHealth**

**MAKING CONNECTIONS**
MDA Community Groups
**mda.org/CommunityGroups**

**MDA PEER CONNECTIONS**
**833-ASK-MDA1**
**ResourceCenter@mdausa.org**

Image: Alexandr Makarov/Getty

Paid caregivers can also be valuable members of your caregiving team and help family caregivers avoid fatigue. MDA's Resource Center can provide one-on-one support via phone or email for individuals and families looking for local caregiving resources, support groups, disease information, and more. (Contact the Resource Center at **833-ASK-MDA1** or **ResourceCenter@mdausa.org**.)

### 6 Pay attention to your mental health

"People who have chronic illness are susceptible to depression," says Gina. Be proactive in keeping a healthy mental state by seeking the help of a therapist, finding hobbies you enjoy, and maintaining a social life. (Read more about taking care of your mental health at **MDAQuest.org/mental-health-for-everyone**.)

Ranae joined a CMT support group. Joining a support group provides an opportunity to talk openly about your challenges and realize you are not alone.

"My advice is to keep moving forward and don't let the challenges in life stop you from your passion," Ed says. "I watched my uncle, who had CMT and was in the same position I am at this age, give up. Basically, he would just sit in his room, watch TV, and never go anywhere. I've made the choice to be the opposite — to get out there and try to keep moving. My son has CMT, and I hope my positive attitude encourages him." Q

Gina Rossi recommends working with a social worker.

---

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

# A Meaningful Day at the Ballpark

On June 2, the MDA community gathered in Major League Baseball (MLB) clubs across the country to celebrate Lou Gehrig Day, honoring the legendary New York Yankee known for his remarkable baseball career and fight against amyotrophic lateral sclerosis (ALS). From coast to coast, fans, families, and supporters wore commemorative gear, shared personal stories, and participated in activities designed to raise awareness about ALS.

Eleanor Gehrig, Lou's widow, was among MDA's earliest and most dedicated supporters. Her commitment to fighting the disease that claimed her husband's life helped shape our lasting dedication to the ALS community and formed a legacy that endures to this day.

Thank you to MLB and all who attended for your ongoing support and commitment to end ALS.


Larry Leiberman, Sharon Hesterlee, Scott Weibe, and Sandra Sullivan (center, standing) with friends

Susan Manning and one of her sons at the Boston Red Sox game

The Diaz family at the LA Angels game

# Congrats, MDA College Scholarship Winners!

MDA is proud to complete the inaugural year of the MDA College Scholarship program, an effort intended to support individuals with neuromuscular disease who are pursuing higher education. This program is a part of MDA's broader effort to focus on resources, education, and programs geared toward supporting the young adult community.

MDA awarded $5,000 merit-based scholarships to 10 college-bound students in the neuromuscular community. MDA recognizes that navigating higher education can be complex and various forms of support can be helpful. As such, in addition to financial assistance, MDA Scholars also receive supplemental support that includes leadership development and connection to a network of students.

Congratulations to the 2024 MDA College Scholarship winners:

**To learn more, visit mda.org/scholarship.**

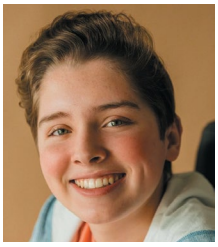
Sydney Bryant, Lewis University

Olivia Calvert, Washington College

Blake Deakin, Arizona State University

Serena Desiderio, University of Arizona

Abby Dreyer, Eastern Connecticut State University



Jonathan Lengel, Fordham University at Lincoln Center

Grace LoPiccolo, Saint Louis University

Alex Mavian, Rensselaer Polytechnic Institute

Grace Nelson, University of Iowa

Jacy Thomas, Indiana University - Indianapolis

# Upcoming MDA Engage Symposiums

MDA Engage Symposiums are not just great opportunities to get up-to-date, disease-specific information — these in-person meetings also allow you to connect with other individuals and families impacted by neuromuscular disease and learn about resources available to the community.

Upcoming Symposium dates include:

+ Oct. 5 – Atlanta, GA
+ Nov. 2 – Stanford, CA
+ Nov. 9 – Dallas, TX
+ Nov. 9 – Irvine, CA

A recent attendee said, "Thank you for putting together such an interesting and informative day. It gives me motivation to look into additional ways to help myself and make living with this disease more tolerable."

Symposiums are half-day, multi-session programs. They are free to attend, but registration is required.

**Find event details and register at mda.org/symposiums.**

Family Getaway participants enjoyed kayaking.

# Ready to Get Away?

Fun and adventure await this fall with MDA Family Getaways. When you take part in a Family Getaway, you and your family can experience a week away from home, enjoying things like outdoor activities, campfires, arts and crafts, and bonding with each other and the community, all in a fully accessible location and at no cost to families.

A 2023 attendee shared, "I loved this, I needed this, I feel like I finally found my planet. Everybody here is like me. Here, we all have the same pain. [People here] walk, run, and jump around like I do."

**Find upcoming getaway locations and dates at mda.org/family-getaways.**

Fishing is a favorite Family Getaway activity.

# MDA Community Groups Are Here for You

MDA recently created Community Groups for members to connect with each other and share information in a safe and supportive environment. Currently, there are specific Community Groups for those newly diagnosed with a neuromuscular disease, parents of newly diagnosed babies and children, individuals with ALS, and individuals eligible for gene therapies. Here is what a few Community Group members have to say about their experiences so far:

"The gene therapy Community Group was the first time I was truly able to see that I am not alone in this experience. This has been so powerful for me."

"Being a part of a Community Group has been so reassuring! Hearing from people who have been through the same things as you, even though they don't have the same kind of disease, and knowing that we all have the same kinds of feelings after getting the diagnosis … makes me feel not so alone in everything we have gone through to this point. It is nice to hear stories and get advice from people whose children have had their diagnosis longer than ours."

"I was having an extremely hard time accepting the fact I must use a wheelchair now. I knew it was for the best but just couldn't get myself to do it. During our very first meeting, I expressed my concerns, and after talking to the group, they got through to me. That weekend, I left my house [using my wheelchair] and went to a store for the first time in two years. I was able to explore the entire store. Finally free!"

**Interested in joining? Register at mda.org/CommunityGroups.**


Image: Svetlana Shamshurina/Getty

# Access the Vote This Election

With the November election fast approaching, voters need the tools and resources to make informed decisions at the polls. The effect of your vote goes far beyond Election Day, and the candidates we vote for shape how the entire federal government works, influence policy, and make decisions that impact our daily lives. MDA is excited to provide you with all the information you need with our Access the Vote campaign, aimed at ensuring everyone can make their voice heard at the ballot box.

**Get started at mda.org/vote.**

# Congress Passes Major Air Travel Reforms

In May, Congress passed the Federal Aviation Administration (FAA) Reauthorization Act. This major piece of legislation includes many reforms that will make air travel more accessible for people living with disabilities, including:

- Mandating regular training for airline and airport personnel who assist passengers with disabilities and load and stow wheelchairs for flights
- Holding airlines accountable for reporting damage to wheelchairs
- Establishing a pathway for wheelchair spots on airplanes
- Continuing to study wheelchair tie-down systems
- Giving the disability community a seat at the table when the government makes future decisions about air travel

This legislation is the most significant advancement in accessible air travel in nearly 40 years.

**Learn more at mda.org/AirTravel.**

# Quest Poll: Voting

## Do you plan to vote in November 2024?

(145 responses collected between April and June)

- Definitely will
- Probably will
- Definitely will not
- Probably will not

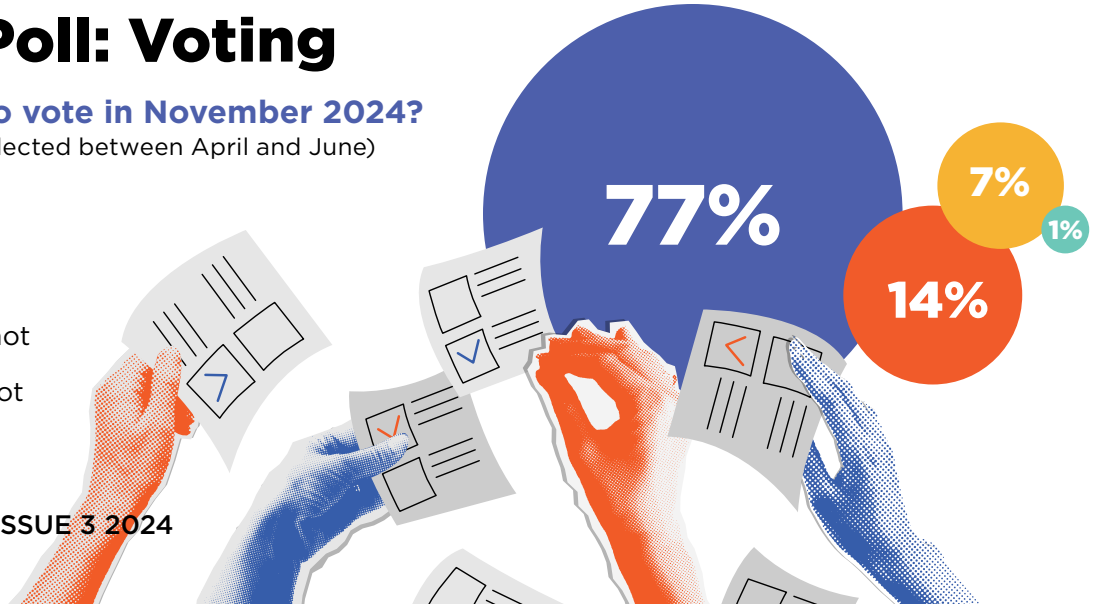



Image: Svetlana Shamshurina/Getty

# New Rules Against Discrimination

Image: cagkansayin/Getty Images

In May, the US Department of Health and Human Services (HHS) finalized a rule to update section 504 of the Rehabilitation Act. This action is a historic milestone for disability rights and marks the first update to the law in 50 years.

The rule supports banning biased treatment decisions and rejecting tools that devalue life based on disability, defines accessibility standards for web-based tools, ensures diagnostic medical equipment is accessible to all patients, and much more. MDA has been highly involved during the public comment process to support the neuromuscular community, and we are excited to see this work come to fruition.

**Learn more about MDA's Advocacy efforts and join us at mda.org/advocacy.**

# Embracing My Differences

*My experiences made me want to help others understand life with a disability*

BY OLIVIA HOLLER

In 2013, at age 13, I was diagnosed with Congenital muscular dystrophy (CMD) and restrictive lung disease. My proximal muscles are weak: legs, hips, neck, and core. I fatigue easily, so walking is difficult, and I cannot walk long distances. I use bilevel positive airway pressure (BiPAP) at night to assist me with breathing.

It wasn't until I went to MDA Summer Camp at 15 that I started embracing my diagnosis and advocating for the community. Before then, I was lost and in complete denial. I wanted nothing to do with my disease. Summer Camp was a transformative experience. I made lifelong friends and gained a support system of people who understood what I was going through. I knew I wasn't alone, and it fueled a passion and love for a community I was now a part of.

Olivia Holler graduated from college with a degree in journalism.

### Discovering my passion

Transitioning into a power wheelchair during my freshman year of high school opened my eyes to barriers and challenges I didn't know existed until I had experienced them. Some people would stare at me, ask my family what happened to me and not ask me, or treat me like a child because I was in a wheelchair. These things frustrated me and fueled me. From that moment on, I wanted to educate others about the disability community.

> *"I knew I wasn't alone, and it fueled a passion and love for a community I was now a part of."*
> *— Olivia Holler*

I used my passion for writing and communicating to connect my passion for bringing awareness about my community. I started a blog to share my experiences of living with a disability. I hoped to influence others with disabilities to start sharing their stories and help those who do not have disabilities gain more understanding of the community and not make assumptions.

The blog made me realize I wanted to go into journalism. If I had the gift of writing, why not use it to empower others to share their stories and amplify

their voices? I attended the University of Missouri-Columbia and received my bachelor's degree in journalism in 2023.

My journey through college also exposed me to unaddressed accessibility barriers. As a member of the Chancellor's Committee for People with Disabilities and Vice President of the Mizzou Disability Coalition from 2020 to 2023, I helped improve campus accessibility. I also kept writing about my lived experiences. One thing I learned from my college journey as a disabled individual is that people don't know what they don't know, and bringing awareness is the way that people will understand.

Olivia made friends and built confidence at MDA Summer Camp.

*"Summer Camp was a transformative experience. I made lifelong friends and gained a support system of people who understood what I was going through."*
***— Olivia Holler***

### A time of transition

After graduating from college and returning home, I struggled to find resources for independent living and transportation, and funding for services. The services I needed had always been given to me. Now, the ball was in my court to find the resources and communities to support me.

Because of this challenging time, I wanted to share what I found with other young adults with disabilities, so I created my disability advocacy brand, Rolling with Liv (**RollingWithLiv.com**). This brand includes a website where I provide resources that I have found helpful, a blog that showcases my story, and social media where I share day-in-the-life posts, bathroom reviews, and more. I also interview friends and people in my support circle on my podcast. With this brand, I hope to bring awareness and education while utilizing the journalism skills I gained in school.

### Ready for the future

It took time to be where I am now, and the journey was not easy, but embracing and accepting my disease has given me the confidence to keep fighting for the future of accessibility and inclusion.

I aspire to make a meaningful impact on the disability community through my writing and communication skills. I want to continue sharing my journey and experiences to raise awareness and create a more inclusive and supportive environment for individuals with disabilities.

Reflecting on my journey and how far I have come, I realized it started with MDA Summer Camp. At camp, I was able to be myself without fear of judgment and self-doubt. If I hadn't gotten to experience camp, I wouldn't be where I am today. Q

**INSPIRING ACTION**
Listen to a conversation with Sophie Morgan, entrepreneur, TV personality, and producer, about media inclusion for people with disabilities at **MDAQuest.org/podcast/sophie-morgan**.

---

Olivia Holler, 23, lives in Lake St. Louis, Missouri, with her family. When she's not writing and podcasting for **RollingWithLiv.com**, she enjoys reading and crafting.

# Voodoo Royalty

*Patricia Perez and her husband's handmade costumes won Halloween*

Patricia Perez and her husband, Dave, don't usually dress up for Halloween together. The retired couple from Arizona has been married for 44 years, but each has a different approach to the holiday.

"My husband is a mild-mannered, kind-hearted introvert—until Halloween rolls around. Once in costume, he morphs into a complete weirdo," says Patricia, who lives with muscular dystrophy and an incomplete spinal cord injury. "I'm always an introvert, and spooky isn't my deal; I don't even watch scary movies."

Patricia normally helps create costumes solely for Dave. However, one year, the couple decided to dress up together. Despite Patricia's usual aversion to all things spooky, Dave convinced her to go with a voodoo theme. Other than their hats, the costumes were made with thrift and dollar store finds. Patricia made her voodoo doll using an old shirt and some neon Sharpies and she applied both her and Dave's makeup. The couple also enlisted the help of a friend to convert Patricia's wheelchair into a throne with a cardboard box, some spray paint, plastic spiders, and lights.

Patricia Perez (left) and her husband in their winning costumes.

The couple debuted their costumes on Halloween night at The Office, a pub in Lake Havasu City, Arizona, where they won first prize for best couple's costume.

Patricia and Dave normally spend their days tinkering around the house or taking road trips to visit friends and family. Patricia's care team at her MDA Care Center recently identified her particular genetic mutation, though it doesn't have a name yet.

"I am told medical science has not seen that particular mutation before," she says. "That seems fitting, as I've always been a little bit different." Q

**+ENTER THE PHOTO CONTEST**

Share a great photo of a meaningful moment for you or a loved one with a neuromuscular disease, and it could be selected to appear on this page in a future issue. Photos must be submitted by Sept. 12, 2024. Enter the photo contest at **SurveyMonkey.com/r/QuestPhoto**.

Abilities EXPO

*Serving the Community Since 1979*


# Access Your World

***Find out how at Abilities Expo.***

## Knock these off your bucket list:

- Test drive the latest product & tech
- Consult experts from the disability community
- Push your limits with adaptive activities
- Network with your Expo family
- Get the answers you need

| | | | |
|---|---|---|---|
| **Ft. Lauderdale** October 13-15, 2023 | **Dallas** Dec. 1-3, 2023 | **Los Angeles** March 15-17, 2024 | **New York Metro** May 3-5, 2024 |
| **Chicago** June 21-23, 2024 | **Houston** August 2-4, 2024 | **Phoenix** September 6-8, 2024 | |

**FREE ADMISSION**

## Abilities.com

## Get registered and join us!

@AbilitiesExpo @AbilitiesExpo @abilities_expo

# 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**.

CDC ATSDR