

STAY INFORMED. LIVE EMPOWERED.

# Quest

MDAQUEST.ORG ISSUE 2 • 2023

# HOME Mods

Small changes  
boost home  
accessibility

## INDEPENDENT LIVING

Getting your  
own place

## GENE THERAPY

What to expect

# MOVING AHEAD with

**Evrysdi® (risdiplam)**

# 1,800+

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

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

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

## What is Evrysdi?

Evrysdi is a prescription medicine used to treat spinal muscular atrophy (SMA) in children and adults.

## Important Safety Information

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

- are pregnant or plan to become pregnant, as Evrysdi may harm your unborn baby. Ask your healthcare provider for advice before taking this medicine
- are a woman who can become pregnant:
  - Before you start your treatment with Evrysdi, your healthcare provider may test you for pregnancy
  - Talk to your healthcare provider about birth control methods that may be right for you. Use birth control while on treatment and for at least 1 month after stopping Evrysdi
  - **Pregnancy Registry.** Talk to your healthcare provider right away if you become pregnant while taking Evrysdi. Ask about registering with the Evrysdi Pregnancy Registry, which was created to collect information about your health and your baby's health. Your healthcare provider can enroll you in this registry by calling 1-833-760-1098 or visiting [www.evrysdipregnancyregistry.com](http://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

**Genentech**

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# Studied in the **most inclusive** clinical study program in SMA<sup>‡§</sup>

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



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

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

## Important Safety Information (continued)

### Tell your healthcare provider about all the medicines you take.

You should receive Evrysdi from the pharmacy as a liquid. If the medicine in the bottle is a powder, **do not use it**. Contact your pharmacist for a replacement.

Avoid getting Evrysdi on your skin or in your eyes. If Evrysdi gets on your skin, wash the area with soap and water. If Evrysdi gets in your eyes, rinse your eyes with water.

### The most common side effects of Evrysdi include:

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

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

You may report side effects to the FDA at 1-800-FDA-1088 or [www.fda.gov/medwatch](http://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](http://MySMASupport.com) for financial assistance information.

Talk with your doctor about Evrysdi  
or visit [www.Evrysdi.com/Go](http://www.Evrysdi.com/Go) to learn more



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

**What is EVRYSDI?**

- EVRYSDI is a prescription medicine used to treat spinal muscular atrophy (SMA) in children and adults.

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

- are pregnant or plan to become pregnant. If you are pregnant, or are planning to become pregnant, ask your healthcare provider for advice before taking this medicine. EVRYSDI may harm your unborn baby.
- are a woman who can become pregnant:
  - Before you start your treatment with EVRYSDI, your healthcare provider may test you for pregnancy. Because EVRYSDI may harm your unborn baby, you and your healthcare provider will decide if taking EVRYSDI is right for you during this time.
  - Talk to your healthcare provider about birth control methods that may be right for you. Use birth control while on treatment and for at least 1 month after stopping EVRYSDI.
  - **Pregnancy Registry.** There is a pregnancy registry for women who take EVRYSDI during pregnancy. If you become pregnant while receiving EVRYSDI, tell your healthcare provider right away. Talk to your healthcare provider about registering with the EVRYSDI Pregnancy Registry. The purpose of this registry is to collect information about your health and your baby's health. Your healthcare provider can enroll you in this registry by calling 1-833-760-1098 or visiting <https://www.evrysdipregnancyregistry.com>.
- are an adult male planning to have children: EVRYSDI may affect a man's ability to have children (fertility). If this is of concern to you, make sure to ask a healthcare provider for advice.
- are breastfeeding or plan to breastfeed. It is not known if EVRYSDI passes into breast milk and may harm your baby. If you plan to breastfeed, discuss with your healthcare provider about the best way to feed your baby while on treatment with EVRYSDI.

**Tell your healthcare provider about all the medicines you take**, including prescription and over-the-counter medicines, vitamins, and herbal supplements. Keep a list of them to show your healthcare provider, including your pharmacist, when you get a new medicine.

**How should I take EVRYSDI?**

**See the detailed Instructions for Use that comes with EVRYSDI for information on how to take or give EVRYSDI oral solution.**

- You should receive EVRYSDI from the pharmacy as a liquid that can be given by mouth or through a feeding tube. The liquid solution is prepared by your pharmacist or other healthcare provider. If the medicine in the bottle is a powder, **do not use it**. Contact your pharmacist for a replacement.
- Avoid getting EVRYSDI on your skin or in your eyes. If EVRYSDI gets on your skin, wash the area with soap and water. If EVRYSDI gets in your eyes, rinse your eyes with water.

**Taking EVRYSDI**

- Your healthcare provider will tell you how long you or your child needs to take EVRYSDI. Do not stop treatment with EVRYSDI unless your healthcare provider tells you to.
- For infants and children, your healthcare provider will determine the daily dose of EVRYSDI needed based on your child's age and weight. For adults, take 5 mg of EVRYSDI daily.
  - Take EVRYSDI exactly as your healthcare provider tells you to take it. Do not change the dose without talking to your healthcare provider.
- Take EVRYSDI 1 time daily after a meal (or after breastfeeding for a child) at approximately the same time each day. Drink water afterwards to make sure EVRYSDI has been completely swallowed.
- Do not mix EVRYSDI with formula or milk.
- If you are unable to swallow and have a nasogastric or gastrostomy tube, EVRYSDI can be given through the tube.
- If you miss a dose of EVRYSDI:
  - If you remember the missed dose within 6 hours of when you normally take EVRYSDI, then take or give the dose. Continue taking EVRYSDI at your usual time the next day.
  - If you remember the missed dose more than 6 hours after you normally take EVRYSDI, skip the missed dose. Take your next dose at your usual time the next day.
- If you do not fully swallow the dose, or you vomit after taking a dose, **do not take** another dose of EVRYSDI to make up for that dose. Wait until the next day to take the next dose at your usual time.

**Reusable Oral Syringes**

- Your pharmacist will provide you with the reusable oral syringe(s) that are needed for taking your medicine and explain how to use them. Wash the syringes per instructions after use. Do not throw them away.
- Use the reusable oral syringe(s) provided by your pharmacist (you should receive 1 or 2 identical oral syringes depending on your prescribed daily dose) to measure your or your child's dose of EVRYSDI, as they are designed to protect the medicine from light. Contact your healthcare provider or pharmacist if your oral syringe(s) are lost or damaged.
- When transferred from the bottle to the oral syringe, take EVRYSDI right away. Do not store the EVRYSDI solution in the syringe. If EVRYSDI is not taken within 5 minutes of when it is drawn up, EVRYSDI should be thrown away from the reusable oral syringe, and a new dose should be prepared.

**What are the possible side effects of EVRYSDI?**

**The most common side effects of EVRYSDI include:**

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

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

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

**How should I store EVRYSDI?**

- Store EVRYSDI in the refrigerator between 36°F to 46°F (2°C to 8°C). Do not freeze.
  - If necessary, EVRYSDI can be kept at room temperature up to 104°F (up to 40°C) for a combined total of 5 days. EVRYSDI can be removed from, and returned to, a refrigerator. The total combined time out of refrigeration should not be more than 5 days.
- Keep EVRYSDI in an upright position in the original amber bottle to protect from light.
- Throw away (discard) any unused portion of EVRYSDI 64 days after it is mixed by the pharmacist (constitution) or if EVRYSDI has been kept at room temperature (below 104°F [40°C]) for more than a total combined time of 5 days. Discard EVRYSDI if it has been kept above 104°F (40°C). Please see the Discard After date written on the bottle label. (See the **Instructions for Use** that comes with EVRYSDI).

**Keep EVRYSDI, all medicines and syringes out of the reach of children.**

**General information about the safe and effective use of EVRYSDI.**

Medicines are sometimes prescribed for purposes other than those listed in a Patient Information leaflet. Do not use EVRYSDI for a condition for which it was not prescribed. Do not give EVRYSDI to other people, even if they have the same symptoms you have. It may harm them. You can ask your pharmacist or healthcare provider for information about EVRYSDI that is written for health professionals.

**What are the ingredients in EVRYSDI?**

**Active ingredient:** risdiplam

**Inactive ingredients:** ascorbic acid, disodium edetate dihydrate, isomalt, mannitol, polyethylene glycol 6000, sodium benzoate, strawberry flavor, sucralose, and tartaric acid.

**Genentech**

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EVRYSDI® (risdiplam)

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For more information, go to [www.EVRYSDI.com](http://www.EVRYSDI.com) or call 1-833-387-9734.

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## Progress Comes to Life

Since our inception 73 years ago, MDA's mission has been to empower the people we serve to live longer, more independent lives. That mission has been a constant source of hope for more than a million people and their families during our seven decades. At MDA, families are at the heart of our mission, and the strength of the MDA community is resolute.

Through MDA's relentless focus on research to improve diagnosis and treatment and create a nationwide network of state-of-the-art clinical facilities to deliver that diagnosis and treatment, more people living with neuromuscular diseases are now feeling the extraordinary impact of MDA's mission in their lives.

This is reflected in the incredible stories we hear from our community, such as a five-year-old girl who was treated for a previously incurable and fatal form of infant spinal muscular atrophy (SMA). Now she is starting elementary school. And a young man under the latest treatment regimen for Duchenne muscular dystrophy (DMD), the most common and rapidly progressive form of childhood muscular dystrophy, has finished college and is planning a career. Stories like these abound.

*“For almost three decades, Quest magazine has been in your homes, bringing our community together through stories, progress, and resources.”*

— Donald S. Wood, PhD

And many can be found in Quest. For almost three decades, Quest magazine has been in your homes, bringing our community together through stories, progress, and resources. More recently, Quest Media has expanded as an adaptive lifestyle platform, a critical resource to our community, providing the information, tools, and resources they need to live out their dreams.

Quest Media brings MDA's legacy and progress into homes, and this platform unites us as we navigate the journeys in this community, together. We move through 2023 with great momentum in continuing to grow our leadership in genetic medicine and rare disease treatment development. New methods are bringing hope of developing therapies for many more neuromuscular diseases, including ultra-rare ones, allowing members of our community to live longer, more independent lives.



Donald S. Wood, PhD

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

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

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Donald S. Wood, PhD

President and CEO

Muscular Dystrophy Association

**SKYCLARYS™**  
(omaveloxolone) 50 mg capsules

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FIRST AND  
ONLY

# NOW APPROVED

prescription medicine for the treatment of **Friedreich's ataxia**  
in adults and children 16 years of age and older



## Got questions about SKYCLARYS?

Talk to your doctor and visit our site for more information. Scan the QR code or visit [SKYCLARYS.com](https://www.skyclarys.com).

[SKYCLARYS.com](https://www.skyclarys.com)

### BRIEF SUMMARY OF PATIENT INFORMATION

**SKYCLARYS™ (skye klar' is) (omaveloxolone) capsules, for oral use**

#### What is SKYCLARYS?

SKYCLARYS is used for the treatment of Friedreich's ataxia in adults and children 16 years of age and older. It is not known if SKYCLARYS is safe and effective for use in children younger than 16 years of age.

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

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

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

Taking SKYCLARYS with other medicines can cause serious side effects.

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

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

#### How should I take SKYCLARYS?

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

#### What should I avoid while taking SKYCLARYS?

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

#### What are the possible side effects of SKYCLARYS?

SKYCLARYS may cause serious side effects, including:

- ▶ **increase in blood liver enzymes.** Some people taking SKYCLARYS have had an increase in the level of liver enzymes in their blood. Your healthcare provider will do liver function tests
  - before you start taking SKYCLARYS
  - every month for the first 3 months after starting your treatment with SKYCLARYS
  - during certain times as needed while taking SKYCLARYS

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

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

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

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

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

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

#### The most common side effects of SKYCLARYS include:

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

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

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

#### General information about the safe and effective use of SKYCLARYS

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

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





## Always Innovating



Welcome to this issue of Quest magazine. As we transition from spring into summer, the team here at MDA's Quest Media has been working hard, interviewing experts to give you the latest on gene therapy and unearthing tools and resources to help you accomplish accessible home modifications on a budget, transition out of your family home to live independently for the first time, and more.

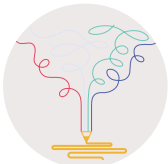
I promised in the previous issue that we have big plans to continue to innovate Quest Media and to give you as much as we can to help you thrive in every way in your everyday life. To that end, I am excited to share that we have established an external advisory board, full of experts on things that matter to this community.

The board is still growing, but we have already confirmed a media executive, the president and CEO of an educational institution that creates opportunities for individuals with disabilities, a judge living with a neuromuscular disease, and an accessibility strategist and expert on inclusive design in built environments.

There's more to come as the board rolls up our sleeves this year. Meanwhile, please enjoy this issue of Quest magazine.

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

## ICONS TO WATCH FOR THROUGHOUT QUEST



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**DID YOU KNOW?**

**There are more than 6,000 ways to get involved and volunteer with MDA each year.**

- Consider these ways to have fun while giving back:
- Spend a week at MDA Summer Camp as a caregiver and friend to a camper. [mda.org/summer-camp](https://mda.org/summer-camp)
  - Support an MDA Muscle Walk, where communities unite along a 1-mile course. [walk.mda.org](https://walk.mda.org)
  - Join an MDA Golf Outing to help MDA raise funds and awareness through sport. [mda.org/golf](https://mda.org/golf)

Learn about these opportunities — and 5,997 more — at [mda.org/volunteer](https://mda.org/volunteer).

**In 2022, MDA had almost 300 phone calls in languages other than English.**

The MDA Resource Center has access to live translation services in more than 100 languages. This allows Resource Center specialists to provide one-on-one support to individuals and families who call looking for disease information, programs, resources, and more, regardless of their preferred language.

To use the translation service, call the Resource Center at **833-ASK-MDA1**. Resource Center specialists are available Monday through Friday, 9 a.m.-5 p.m. CT. You may also email [ResourceCenter@mdaUSA.org](mailto:ResourceCenter@mdaUSA.org).





# Investing in Research Success

*MDA's impact on the drug development process*



Following our mission to transform the lives of those living with neuromuscular disease, MDA has made investing in the most promising and innovative research a priority. Before a new neuromuscular disease treatment makes it to market, it must undergo a rigorous research and development (R&D) process — a process made possible by funding research at different stages of drug development. As the largest source of funding for neuromuscular disease research outside of the federal government, MDA has committed more than \$1 billion in grants and funding since its inception to accelerate the necessary research and development processes.

“MDA finds various ways to de-risk drug development by seeding funds to different stages of the R&D pipeline to relieve bottlenecks in the process,” says Sharon Hesterlee, PhD, Executive Vice President and Chief Research Officer for MDA. By de-risking, she means reducing potential time delays and costs to motivate more companies to develop neuromuscular disease therapies.

The process of developing a new therapy or drug involves four main stages:

- Stage 1: Basic research
- Stage 2: Identifying and testing drug targets
- Stage 3: Translational research
- Stage 4: Clinical trials

“MDA uses funds to de-risk each stage of drug development for all the diseases we cover, and we have different granting mechanisms to address each of the stages,” says Angela Lek, PhD, MDA’s Vice President of Research.

Research that MDA has supported through our grants and programs is directly linked to life-changing therapies across multiple neuromuscular diseases. Turn the page to learn more about the drug development cycle and the pivotal role MDA funding has played.

## FUNDING THE NEXT GENERATION OF SCIENTISTS

Along with investing in the drug development process, MDA has two ways to support promising candidates interested in a career in the neuromuscular disease field: Development Grants and Clinical Fellowships.

### Development Grant Success Story

In 1995, MDA awarded Rachelle H. Crosbie, PhD, the Robert G. Sampson Development Grant to support her postdoctoral training. She investigated autosomal recessive forms of limb-girdle muscular dystrophy (LGMD) and identified a small sarcoglycan-associated protein, sarcospan.

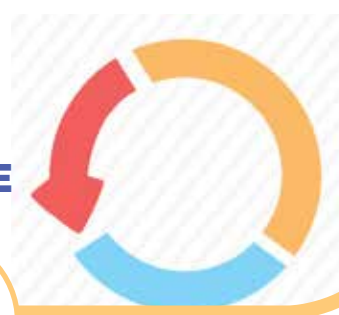
Dr. Crosbie is now a professor and chair of the Department of Integrative Biology and Physiology at UCLA. MDA contributed additional funding to her lab’s work to verify that sarcospan improves skeletal and cardiac aspects of Duchenne muscular dystrophy (DMD) and develop potential drugs to target sarcospan as a treatment for DMD.

“MDA has been instrumental at every step of my career and has provided continual support of our research, from basic science to translation.”

— Rachelle H. Crosbie, PhD

### Clinical Fellowship Success Story

MDA awarded Russell Butterfield, MD, PhD, a Clinical Fellowship in 2009 to complete his neuromuscular fellowship at the University of Utah. His funded project aimed to determine the clinical and molecular characterization of collagen VI-related muscular dystrophies. Dr. Butterfield currently leads a research program focusing on genetic therapies and understanding genetic modifiers of severity in various types of muscular dystrophy, including DMD, facioscapulohumeral muscular dystrophy (FSHD), and collagen VI-related muscular dystrophies. He also directs the MDA Care Center at the University of Utah and Primary Children’s Hospital and leads their efforts to implement novel genetic therapies.



## MDA'S IMPACT ON THE DRUG DEVELOPMENT CYCLE

# 1

### Stage 1: Basic research

During this stage, researchers work to understand the root cause of a disease, such as identifying a disease-causing gene, characterizing how a DNA mutation can affect the function of a gene, or determining which molecular pathways are disrupted. Funding at this phase can set the stage for developing a treatment strategy in the future. MDA supports academic research labs at this stage with Research Grants, Development Grants, and Idea Awards, and by organizing scientific conferences for sharing ideas.

### Success Story

MDA provided Robert Brown, MD, DPhil, a three-year research grant in 1992 to look for genes that increase the risk for amyotrophic lateral sclerosis (ALS). This project led to the discovery of the gene *SOD1*, a familial form of ALS. About 5%-10% of ALS cases are linked to familial or hereditary forms, and *SOD1* is the second most common form of genetically linked ALS, accounting for 10%-20% of those cases. The discoveries from this project have led to other milestones in the field, such as the development of antisense oligonucleotide (ASO) therapeutics to target *SOD1*. Biogen's tofersen (QALSODY), an ASO, was recently granted accelerated approval by the US Food and Drug Administration (FDA) to treat ALS in patients with *SOD1* mutations.



# 2

### Stage 2: Identifying and testing drug targets

Once the cause of a disease is identified, researchers devise a strategy to address the cause using available technologies, such as drug screening or gene therapy. These strategies are tested in cells and animal models of the disease. MDA supports this stage with Research Grants and Infrastructure Grants, which support the tools, techniques, and resources needed for the research.

### Success Story

In 1996, MDA provided a small starter grant to Steve Wilton, PhD, BSc, a primary investigator at the University of Western Australia in Perth, for research into a process to address the effects of certain gene mutations. This led to the process of exon skipping, a therapeutic strategy that has been used to develop four FDA-approved disease-modifying drugs by Sarepta for Duchenne muscular dystrophy (DMD). Since Dr. Wilton's discovery, MDA has funded the development of exon-skipping technology for many years, from a concept first demonstrated in experiments with cells, then in animal models, and finally in clinical trials with DMD patients.

### Success Story

In 2007, MDA provided Adrian Krainer, PhD, a Research Grant to develop and test an ASO approach to treat the underlying cause of motor neuron loss in spinal muscular atrophy (SMA). The most common form of SMA is caused by a mutated or missing gene known as the survival motor neuron gene 1 (*SMN1*). This gene produces the survival motor neuron (SMN) protein, which maintains the health and normal function of motor neurons. In this project, Dr. Krainer developed an ASO to enhance the amount of SMN protein in animals. This therapeutic approach has since been translated into humans. The success of this project attracted Biogen and Ionis Pharmaceuticals to collaborate with Dr. Krainer to develop his approach into a drug named Spinraza, the first treatment for SMA approved by the FDA, in 2016.

# 3

## Stage 3: Translational research

This is the early stage of drug development when processes typically transition out of the academic lab to a biotech or pharmaceutical company. During this stage, MDA grants help start-up companies seeking to translate promising technologies, discoveries, or drugs into clinical trials. The funds can also be used to create tools, techniques, or services to support translational research. MDA funds this stage with Research Infrastructure Grants and the MDA Venture Philanthropy (MVP) program. “Right now, it’s more important than ever to invest in early-stage biotech companies because it’s hard for them to find funding in the current market. This funding makes MDA an important lifeline for actually getting new drugs into clinical testing,” says Sharon Hesterlee, PhD, Executive Vice President and Chief Research Officer for MDA.

### MVP Grant Success Story

Through the MVP program, MDA invested in ReveraGen from the early phase of preclinical development of its drug Vamorolone to its phase 1 clinical trial for individuals with DMD. Vamorolone is a steroid alternative that may have fewer side effects than the steroids commonly prescribed for DMD. In early 2023, the FDA accepted the new drug application for Vamorolone for the treatment of DMD. If the FDA approves Vamorolone, it will become available later this year and is anticipated to provide an alternative option to traditional steroids.

### Infrastructure Grant Success Story

MDA awarded a three-year Research Infrastructure Grant to Michael Shy, MD, in 2002 to help fund the goals of the Inherited Neuropathy Consortium (INC), an integrated group of academic medical centers, patient support organizations, and clinical research resources dedicated to conducting clinical research on Charcot-Marie-Tooth disease (CMT) and improving care for patients. Since its inception, the INC has developed CMT-specific clinical outcome assessments to measure the effects of CMT on adults and children; identified disease biomarkers; identified 23 novel genetic causes; initiated the CMT Variant Browser, which promotes sharing genetic information on CMT for patients and their families, as well as researchers; and successfully trained 14 young investigators to carry on future CMT research.

# 4

## Stage 4: Clinical trials

After receiving FDA approval to test drugs on people, companies work with clinicians to identify eligible individuals to participate in clinical trials. MDA supports this stage through Clinical Trial Grants, Clinical Research Network Grants, and Clinical Trial Travel Grants.

### Clinical Trial Travel Grant Success Story

MDA is helping fund a project to ensure equitable access for individuals to participate in a study investigating the utility of the National Institutes of Health Toolbox Cognition Battery (NIHTB-CB) to measure mental skills in DMD clinical trials. MDA will support travel and travel-related costs, so families and research participants will have access to this study regardless of their socioeconomic status or geographic location.

### Infrastructure Grant Success Story

This grant helped support David Lynch, MD, PhD, and other investigators collaborating on developing clinical measures to assess Friedreich ataxia (FRDA; often referred to as FA) in a natural history study known as the Clinical Research Network in Friedrich’s Ataxia. The outcome helped guide patient selection and data analysis of the clinical trial for the recently approved drug SKYCLARYS, the first medication available to treat FRDA — a major milestone for the FRDA community.

### Clinical Research Grant Success Story

Charcot-Marie-Tooth disease type 2C (CMT2C) is caused by mutations in the gene *TRPV4*, which regulates a channel allowing calcium to enter cells. By researching animal models, Brett McCray, MD, PhD, discovered that mutations in *TRPV4* cause the channel to be overactive, and drugs that block *TRPV4* are highly effective as treatments. In 2022, MDA awarded a grant to support Dr. McCray in exploring the use of available *TRPV4*-blocking drugs as a potential treatment for patients with CMT2C. Dr. McCray intends to lay the framework for clinical trials in CMT2C. If successful, this would be a groundbreaking accomplishment and pave the way for clinical trials for other forms of CMT.





# 'I Belong Here'

*Leticia Tatum's career is powered by her abilities*

BY REBECCA HUME

Leticia Tatum, 42, knows first-hand that many different roads can lead to success. Living with spinal muscular atrophy (SMA), Leticia forged her own path to become Vice President of Human Resources (HR) at Valent Group, a risk consulting firm. She hopes her story will remind others that they are entitled to pursue their own quests for success.

"You're not just allowed to want more, but you are deserving of it," she says.

## Developing confidence

Leticia was diagnosed with SMA when she was almost 2 years old. She is the oldest of four siblings and the only one in her family who lives with a physical disability. Her parents expected all their children to go to college and pursue careers. They instilled in Leticia the idea that she could and would do everything she wanted to do.

Leticia was raised in Birmingham, Alabama. Throughout her early education, her mother fought for Leticia's right

to be in mainstream classrooms instead of automatically assigned to special education, as was the custom for students with disabilities at the time.

Leticia used a wheelchair throughout her childhood and traveled to middle and high schools outside her district to be in accessible buildings. The school district provided an attendant to assist her with transfers in the bathroom. Leticia was active in her school community, joining the choir and school newspaper.

"I never felt I was treated any differently, which is so important," Leticia says. "I had influential teachers who

*"No individual should feel that they have to reveal a disability. Employers should be looking at qualifications."*

— Leticia Tatum



Leticia Tatum

reminded me that there was no difference between me and the student sitting next to me who was able to walk."

## Transitioning to independence

After high school, Leticia enrolled at the University of Alabama at Birmingham. She commuted to campus and used the school's Disability Support Services to navigate her accessibility needs. For example, when a classroom location was a barrier, she requested to move the class to an accessible building. She also received extended time on exams.

Transitioning to becoming an independent adult came naturally because Leticia's parents helped her develop the tools she needed to live independently. "My parents were very hands-off, in a good way," she says. "I could ask for advice, but if I needed something, they directed me to pursue resources myself instead of doing it for me. This put me in a position to advocate for myself and navigate my own path."

This path changed direction a few times as she sought her passion. “I didn’t have all the answers, but I learned that it’s OK to figure it out as you go along,” Leticia says. “I think a lot of individuals in the neuromuscular disease community have to be planners. Pursuing an education and career takes some planning, but you don’t always have to have the answers. And the answers can change, and that is fine.”

### Beginning a career

When Leticia began applying for entry-level positions after college, she didn’t disclose her disability before job interviews for two reasons: “One, my disability is physical, and I can’t hide it, so once I was there, it was evident to employers,” she says. “And two, no individual should feel that they have to reveal a disability. Employers should be looking at qualifications. So, I went to interviews with the intent that knowing I met the qualifications and believing I could do the job, the rest we would figure out.”

Leticia didn’t drive at the time, so she scheduled rides through the Birmingham-Jefferson County Transit Authority (BJCTA), calling so often that the scheduler eventually asked her where she was traveling so frequently. When Leticia shared that she was going to job interviews, the scheduler told her that the transit authority had an administrative assistant opening and promptly scheduled her a ride to apply. The executive director met with her and offered her the job that week.



### MINDSET RESET

Listen to motivational speaker Jose Flores discuss using the power of your mind to overcome anything that life throws your way at [MDAQuest.org/podcast/Jose-Flores](https://MDAQuest.org/podcast/Jose-Flores).



Leticia and her husband, Douglas, enjoy traveling.

Leticia’s boss at BJCTA recognized her potential and upgraded her title to HR Assistant. This small change helped launch her career in HR.

### Advocacy and accommodations

Throughout her career, Leticia has had a variety of accessibility accommodations. Her employers have modified doors for easier opening and installed automatic doors and motion-activated lights. Employees typically make accommodation requests through their company’s HR department, but because she works on the HR team, Leticia meets directly with

In college, Leticia commuted to campus and used the school’s Disability Support Services to meet her accessibility needs.

### +PERSONAL STORIES

Read about people living and thriving with neuromuscular diseases at [MDAQuest.org/personal-stories](https://MDAQuest.org/personal-stories).



*“It is so important to advocate for yourself, as an individual with a disability and an individual working through a career, and to remember, ‘I belong here.’”*

— *Leticia Tatum*



Leticia and Douglas enjoy time at the beach.

the company president to advocate for her needs.

Valent Group offers a hybrid work environment. Leticia lives in Birmingham with her husband, and he assists with transfers and toileting when she works from home. On days in the office, a caregiver accompanies her for half an hour to help prepare her lunch and assist in the restroom.

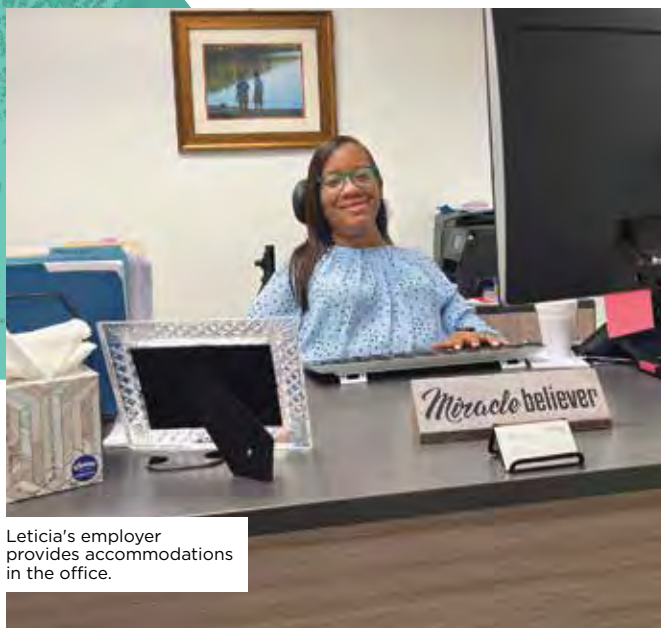
Leticia worked with a vocational rehabilitation counselor to coordinate funding for an accessible van, so she can drive to work.

As her career progressed, Leticia surpassed the income threshold to obtain state-funded caregiver services. Determined to pursue high-level positions and receive the services she needed, she appealed the decision. She argued that if she was denied services, she could not remain a productive member of society, outlining how that would financially impact her ability to meet basic needs and live independently. The power of self-advocacy was evident, as the state granted her appeal.

Her employers have **modified doors** for easier opening and installed **automatic doors** and **motion-activated lights**.

“It is so important to advocate for yourself, as an individual with a disability and an individual working through a career, and to remember, ‘I belong here,’” Leticia says. “I focus on my ability and what I bring to the table. My disability is an important part of me, but it is not at the forefront. It takes a backseat to my strengths. I am not Leticia who has a disability and works in HR. I am Leticia, the HR leader, who happens to have a disability.” [Q](#)

Rebecca Hume is Senior Specialist and Writer for Quest Media.



Leticia's employer provides accommodations in the office.

# Progress

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

# NOW

## New approvals

## FDA Approves Friedreich Ataxia Drug

The US Food and Drug Administration (FDA) has granted approval to Reata Pharmaceuticals' omaveloxolone (SKYCLARYS) for the treatment of Friedreich ataxia (FRDA; often referred to as FA) for ages 16 and older. SKYCLARYS is the first disease-modifying drug approved to treat FRDA. Reata also announced the launch of the Reata Education, Access, and Care Helpline (REACH), an integrated specialty pharmacy and patient services program designed to help eligible patients access prescribed Reata medicines.

The FDA based its approval decision on positive results from the MOXle Part 2 trial and an open-label MOXle Extension trial.

In the MOXle Part 2 trial, 172 participants were randomly assigned to receive either 150 mg of SKYCLARYS or an inactive placebo control over the course of the 48-week study. The effects of the drug were measured using the modified

Friedreich's Ataxia Rating Scale (mFARS), a clinical assessment tool used to measure disease progression. Participants receiving SKYCLARYS

demonstrated significantly lower mFARS scores (less impairment) relative to participants receiving the placebo.

In the follow-up extension period, in which all 136 participants received SKYCLARYS, analysis suggested that long-term treatment with SKYCLARYS leads to lower mFARS scores and functional improvements.

The most common treatment-related adverse reactions were elevated liver enzymes (AST/ALT), headache, nausea, abdominal pain, fatigue, diarrhea, and musculoskeletal pain.

**For additional information about REACH programs, call 1-844-98-REACH or visit [reataREACH.com](http://reataREACH.com).**



Image: iStock.com/RudZhan Nagiev

### **+RECRUITING CLINICAL TRIALS**

Find a list of trials actively recruiting individuals to help advance research and treatment development at [mda.org/clinical-trial-updates](http://mda.org/clinical-trial-updates).

## Spotlight on Rare Diseases

MDA sheds light on neuromuscular diseases in our Spotlight series of Q&As with expert researchers and clinicians. Read all the Spotlight articles at [MDAQuest.org/tag/spotlight](http://MDAQuest.org/tag/spotlight).

## Becker muscular dystrophy (BMD)

# Clinical Trial Seeks Participants



An open-label study in which all participants receive EDG-5506 may be available after the first 12-month period.

Image: iStock.com/SeventyFour

Researchers at Edgewise Therapeutics are seeking adults and adolescents living with BMD to participate in the phase 2 CANYON clinical trial to evaluate EDG-5506 for the treatment of BMD. EDG-5506 is designed to protect susceptible muscle fibers from the damage that occurs as a result of everyday activities in individuals living with BMD.

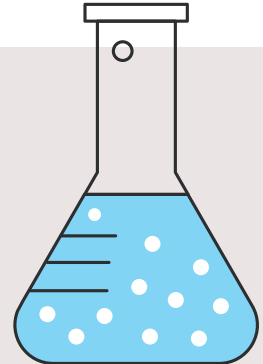
In the double-blind CANYON study, participants will be randomly assigned to take either EDG-5506 or a placebo. The total trial duration for each participant will be approximately 14 months and will include up to 10 on-site clinic visits and five telephone visits.

EDG-5506 and the placebo will be administered as oral tablets given once per day. The safety and efficacy of EDG-5506 will be evaluated using blood tests, safety assessments, and functional assessments.

Travel and other resources are available for eligible participants. To be eligible, individuals must be:

- Adults (ages 18 to 50) who are currently ambulatory (able to walk) and have a documented dystrophin mutation or adolescents (ages 12 to 17) with genetic confirmation of in-frame dystrophin mutation
- Able to complete the 100-meter timed test in less than 150 seconds
- Able to perform the North Star Ambulatory Assessment scale with a score of 10-32 for adults or more than 10 for adolescents

To learn more, visit the trial website at [EdgewiseTX.com/clinical-trials](https://EdgewiseTX.com/clinical-trials), or email the study coordinator at [studies@EdgewiseTX.com](mailto:studies@EdgewiseTX.com).



### CLINICAL TRIAL TERMS TO KNOW

**Double-blind:**

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

**Multiaim:**

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

**Multicenter:**

The trial is completed at more than one site.

**Randomized:**

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

**Open-label:**

Both the researchers and participants know which treatment a participant is receiving.



## Facioscapulohumeral muscular dystrophy (FSHD)

# Experimental Drug Granted Orphan Status

The US Food and Drug Administration (FDA) has granted orphan drug designation to Avidity Biosciences' experimental drug AOC 1020 for the treatment of FSHD. Avidity's phase 1/2 FORTITUDE clinical trial is getting underway to evaluate safety and exploratory efficacy in adults who have type 1 or type 2 FSHD.

The FDA's Office of Orphan Drug Products grants orphan status to support the development of medicines for rare disorders, which are defined as diseases or conditions that affect fewer than 200,000 people in the United States. Orphan drug designation provides certain benefits, including market exclusivity after regulatory approval, exemption of FDA application fees, and tax credits for qualified clinical trials.

AOC 1020 is designed to target the disease-causing gene *DUX4*, with the goal of treating the

underlying biological cause of FSHD. The abnormal expression of DUX4 protein leads to the changes in gene expression in muscle cells that are associated with the progressive loss of muscle function in people with FSHD.

Researchers expect to enroll approximately 72 adults to participate in the FORTITUDE trial, which will test single and multiple doses of AOC 1020, administered intravenously (into the vein), versus a placebo control. Avidity has announced plans to share data from a preliminary assessment of AOC 1020's effect on about half of the FORTITUDE trial participants in the first half of 2024.

Learn more about the drug status and trial enrollment at [AvidityBiosciences.com](https://www.AvidityBiosciences.com), or visit [ClinicalTrials.gov](https://ClinicalTrials.gov) and enter NCT05747924 in the "Other terms" search box.

The FDA defines an orphan drug as one intended for the treatment, prevention, or diagnosis of a condition that affects fewer than 200,000 people in the United States.

image: iStock.com/AlexandrVedmed

## Spinal muscular atrophy (SMA)

# Phase 3 Trial Seeks Participants

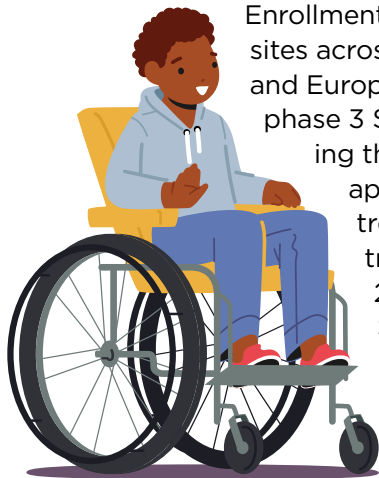


Image: iStock.com/Pavel Sivak

Enrollment is ongoing at 37 sites across the United States and Europe for Scholar Rock's phase 3 SAPPHIRE trial assessing the experimental drug apitegromab for the treatment of SMA. The trial is recruiting up to 204 individuals with SMA types 2 or 3 who are nonambulatory (unable to walk) but can sit independently. During this randomized,

double-blind trial, the drug and placebo will be administered intravenously (into the vein).

Apitegromab is a lab-made antibody designed to inhibit the production of a protein called myostatin. Myostatin is found in skeletal muscles, which are involved in voluntary movement. It's thought that reducing levels of myostatin protein

will increase muscle mass and improve motor function. Apitegromab may work as an add-on to other approved SMA therapies.

To be eligible to participate in the SAPPHIRE trial, individuals must:

- Be 2 to 21 years old at the time of screening
- Have a documented diagnosis of 5q SMA
- Have been diagnosed with later-onset SMA (type 2 or 3) before receiving an approved survival motor neuron protein (SMN) upregulatory therapy (e.g., nusinersen or risdiplam)
- Be receiving one background therapy for SMA (e.g., nusinersen or risdiplam)
- Meet additional criteria

Top-line data from the SAPPHIRE trial are anticipated in 2024. If the trial is successful and the drug becomes FDA-approved, Scholar Rock expects to start a commercial product launch in 2025.

To learn more about SAPPHIRE, visit [ClinicalTrials.gov](https://clinicaltrials.gov) and enter NCT05156320 in the "Other terms" search box.

## Milestones Seen With Zolgensma

Most children with SMA who are treated with the gene therapy Zolgensma retain motor function and, moreover, achieve one or more new motor milestones, according to a new analysis. These outcomes differ from the outcomes of people with SMA who do not receive treatment.

To inform their analysis, a team of scientists analyzed data from 10 studies covering 250 children with SMA who were treated with Zolgensma.

The researchers analyzed the treatment's effectiveness using Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders (CHOP INTEND) scores. This test is a standardized measure of motor function for infants and young children with respiratory and



neuromuscular deficits associated with SMA or other neuromuscular diseases. Sixteen items are scored to assess motor skills, each of which is graded on a scale of 0 to 4, with higher scores indicating better function.

In the natural history of SMA, an untreated child's total CHOP INTEND score rarely reaches 40 points, and it gradually decreases over time. However, the scores were markedly higher for children treated with Zolgensma, both in clinical trials and in follow-ups after six months or longer, the researchers said. Some children treated with Zolgensma gained the ability to stand and even walk without assistance.

The team concluded that the maintenance of scores greater than or equal to 40 points is clinically meaningful in individuals living with SMA, and that the available evidence supports Zolgensma as an effective treatment for SMA, especially for younger individuals with early disease. Studies are needed to assess whether Zolgensma might benefit older individuals with more advanced SMA.

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To view the full study results, visit the Journal of Paediatrics and Child Health online at [OnlineLibrary.wiley.com/doi/full/10.1111/jpc.16340](http://OnlineLibrary.wiley.com/doi/full/10.1111/jpc.16340).

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## Data Confirms Nusinersen's Positive Effects

An analysis of SMARtCARE Registry data shows a positive effect of nusinersen treatment on motor function in ambulant (able to walk) children and adults with SMA over a 38-month period. Researchers observed not only stabilized disease progression but clinically meaningful improvements in walking distance in a subgroup of patients. The study's findings have been published in the Journal of Neuromuscular Diseases.

SMARtCARE is a disease-specific registry with 58 participating centers in Germany, Austria, and Switzerland. Data are collected during routine patient visits. The study's analysis included all patients being treated with nusinersen who were able to walk independently before the start of treatment, with a focus on changes in motor function.

Data from 231 individuals were included in the analysis. During the observation period, changes in walking distance were assessed using the six-minute walk test. The study showed that 31 pediatric walkers (27.2%) and 31 adult walkers (26.5%) experienced a clinically meaningful improvement. In contrast, only five adult walkers (7.7%) showed a decline in walking distance, and two pediatric walkers (1.8%) lost



Increasing evidence of the positive long-term effects of treatment with nusinersen may lead to new treatment recommendations.

the ability to walk unassisted under treatment with nusinersen.

Researchers say the results contribute to the increasing evidence of nusinersen treatment's positive long-term effects in this patient population.

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To read the full study, visit [Content.IOSPress.com](http://Content.IOSPress.com) and search for "Improvements in Walking Distance During Nusinersen Treatment — A Prospective 3-Year SMARtCARE Registry Study."

## Neuromuscular diseases

# Medical Device Study Seeks People With Paralysis

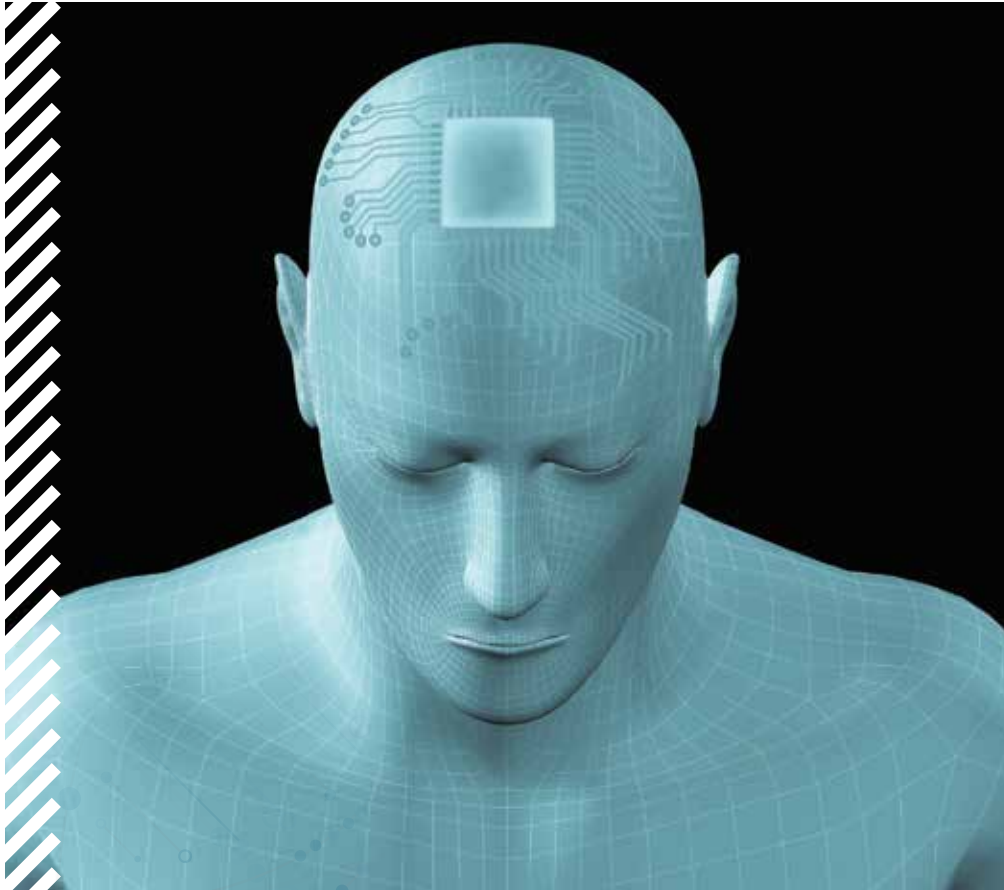


Image: iStock.com/Nixxphotography

A brain-computer interface is a direct communication pathway between the brain's electrical activity and an external device.

Researchers at Synchron are seeking individuals living with paralysis from muscular dystrophy, amyotrophic lateral sclerosis (ALS), spinal muscular atrophy (SMA), stroke, and spinal cord injury to participate in a study to evaluate the safety and feasibility of a motor neuroprosthesis (implanted medical device) to improve neural communication. Researchers will assess how well the neuroprosthesis enables people with severe paralysis to control digital devices using brain-computer interface technology.

The motor neuroprosthesis will be

implanted directly into a vessel in the brain. To analyze safety, researchers will be studying the incidence of device-related serious adverse events within 12 months post-implant. The researchers will assess the device's effects using medical evaluations, questionnaires, scans, and other methods.

This trial is a multicenter, open-label safety feasibility trial. There is no placebo control. The total trial duration for each participant will be approximately 15 months, which will include up to 15 visits. Home visits are a possibility.

To be eligible to participate, individuals must be aged 21 to 75 years, have severe quadriplegia (weakness in all four limbs), be able to give consent, have a caregiver who can assist, and meet additional criteria.

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To learn more about the study or inquire about participation, contact study coordinators Marta Lapinska at [646-689-1776](tel:646-689-1776) or [Marta.Lapinska@MountSinai.org](mailto:Marta.Lapinska@MountSinai.org), or Aidan Rogers at [718-308-9450](tel:718-308-9450) or [Aidan.Rogers@MountSinai.org](mailto:Aidan.Rogers@MountSinai.org).

# Participants Needed to Test Muscle Assessment Tool

Researchers at Ohio State University are seeking individuals living with neuromuscular diseases, including muscular dystrophy, facioscapulo-humeral muscular dystrophy (FSHD), and limb-girdle muscular dystrophy (LGMD), to participate in the phase 2 EIM Bedside clinical trial. This trial will evaluate the effectiveness of using data from a noninvasive type of medical imaging called electrical impedance myography (EIM) to assist in muscle assessment in neuromuscular disorders. The trial aims to test an EIM software prototype and determine its capability to accurately classify disease type and determine disease severity.

Participants must complete a one-time visit of approximately 45-60 minutes, which will include a manual muscle test. EIM data also will be collected for analysis.

To be eligible to participate, individuals must be aged 5 to 90 and have a definite clinical neuromuscular diagnosis associated with weakness or muscle dysfunction.

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To learn more about the study, contact study coordinator Kaneshia Hives at **614-685-5661** or [Kaneshia.Hives@osumc.edu](mailto:Kaneshia.Hives@osumc.edu).



## LISTEN IN

With more innovations and advances in treatment, it's important to know all your neuromuscular care options. Listen to a conversation with a neurologist about this changing landscape at [MDAQuest.org/podcast/neuromuscular-care](https://MDAQuest.org/podcast/neuromuscular-care).



Image: iStock.com/Arlette Lopez

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# GENERALIZED MYASTHENIA GRAVIS

*doesn't get to steal  
this moment*

## **What is VYVGART<sup>®</sup> (efgartigimod alfa-fcab)?**

VYVGART is a prescription medicine used to treat a condition called generalized myasthenia gravis, which causes muscles to tire and weaken easily throughout the body, in adults who are positive for antibodies directed toward a protein called acetylcholine receptor (anti-AChR antibody positive).

### **IMPORTANT SAFETY INFORMATION**

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

VYVGART may cause serious side effects, including:

- **Infection.** VYVGART may increase the risk of infection. In a clinical study, the most common infections were urinary tract and respiratory tract infections. More patients on VYVGART vs placebo had below normal levels for white blood cell counts, lymphocyte counts, and neutrophil counts. The majority of infections and blood side effects were mild to moderate in severity. Your health care provider should check you for infections before starting treatment, during treatment, and after treatment with VYVGART. Tell your health care provider if you have any history of infections. Tell your health care provider right away if you have signs or symptoms of an infection during

treatment with VYVGART such as fever, chills, frequent and/or painful urination, cough, pain and blockage of nasal passages/sinus, wheezing, shortness of breath, fatigue, sore throat, excess phlegm, nasal discharge, back pain, and/or chest pain.

- **Undesirable immune reactions (hypersensitivity reactions).**

VYVGART can cause the immune system to have undesirable reactions such as rashes, swelling under the skin, and shortness of breath. In clinical studies, the reactions were mild or moderate and occurred within 1 hour to 3 weeks of administration, and the reactions did not lead to VYVGART discontinuation. Your health

# VYVGART is a first-of-its-kind, FDA-approved treatment for adults with anti-AChR antibody positive generalized myasthenia gravis (gMG)

AChR=acetylcholine receptor  
Visit [VYVGART.com/glossary](https://www.vyvgart.com/glossary) for a glossary of terms.

When added to their current gMG treatment, VYVGART helped clinical trial participants with anti-AChR antibody positive gMG achieve:

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## Reduced muscle weakness

**63%** (41 of 65) of participants on VYVGART achieved a significant reduction in muscle weakness†

\*Improvement maintained for 4 or more weeks was measured by a decrease of 2 or more points on the Myasthenia Gravis Activities of Daily Living (MG-ADL) scale, with the first reduction occurring no later than 1 week after the last infusion of treatment cycle 1. The MG-ADL scale assesses the impact of gMG on daily functions by measuring 8 signs or symptoms that are commonly affected in gMG. Each item is measured on a 4-point scale, where a score of 0 represents normal function and a score of 3 represents the loss of ability to perform that function. Total scores range from 0 to 24 points, with a higher score showing more severe gMG.

†Improvement maintained for 4 or more weeks was measured by a decrease of 3 or more points on the Quantitative Myasthenia Gravis (QMG) scale, with the first reduction occurring no later than 1 week after the last infusion of treatment cycle 1. The QMG scale assesses muscle weakness in gMG based on 13 items. Each item is assessed on a 4-point scale, where a score of 0 represents no muscle weakness and a score of 3 represents severe muscle weakness. Total scores range from 0 to 39, with a higher score meaning muscle weakness is more severe.

➔ Talk to your neurologist and visit [VYVGART.com](https://www.vyvgart.com) or call **1-833-VYVGART (1-833-898-4278)**

care provider should monitor you during and after treatment and discontinue VYVGART if needed. Tell your health care provider immediately about any undesirable reactions.

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

- Have a history of infection or you think you have an infection
- Have received or are scheduled to receive a vaccine (immunization). Discuss with your health care provider whether you need to receive age-appropriate immunizations before initiation of a new treatment cycle with VYVGART. The use of vaccines

during VYVGART treatment has not been studied, and the safety with live or live-attenuated vaccines is unknown. Administration of live or live-attenuated vaccines is not recommended during treatment with VYVGART.

- Are pregnant or plan to become pregnant and are breastfeeding or plan to breastfeed.

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

### What are the common side effects of VYVGART?

The most common side effects of VYVGART are respiratory tract

infection, headache, and urinary tract infection.

These are not all the possible side effects of VYVGART. 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.

Please see the full Prescribing Information for VYVGART and talk to your doctor.



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US-VYV-22-00075 V2 06/2022

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



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# Talking About a Diagnosis

*It's a personal decision, but there are good reasons to share*

BY CHERYL ALKON

When her late husband, Kevin, was diagnosed with amyotrophic lateral sclerosis (ALS) in 1995 at the age of 30, Jodi O'Donnell-Ames gathered their closest family and friends to tell their news.

“We shared that we would need help and hoped they would be our village — and that’s exactly what happened, and we were so grateful,” says Jodi, the founder of Hope Loves Company (**HopeLovesCompany.org**), a nonprofit that supports children and adolescents who have or had a loved one with ALS.

“With a complicated disease, being able to express your feelings, feel supported, and find helpful resources is extremely important,” she says.

ALS is one of the more common and severe neuromuscular diseases diagnosed in adulthood. Other neuromuscular diseases typically diagnosed in adulthood include some types of myotonic dystrophy (DM), Emery-Dreifuss muscular dystrophy (EDMD), facioscapulohumeral muscular dystrophy (FSHD), and limb-girdle muscular dystrophy (LGMD). How symptoms affect daily life and the rate of progression can vary greatly between diseases, and even from person to person.

As each disease journey is personal, so is the decision to tell family and friends about a diagnosis, what to disclose, and how to present it.



Jodi O'Donnell-Ames

“I don’t believe there is a one-size-fits-all process,” says Rebecca Axline, LCSW-S, CSM, APHSW-C, a clinical social worker at the Houston Methodist Neurological Institute, who has worked with ALS patients for 17 years. “Just as you have made individual

*“We shared that we would need help and hoped they would be our village — and that’s exactly what happened, and we were so grateful.”*  
— Jodi O'Donnell-Ames

## +ASK AN MDA SPECIALIST

The MDA Connect program provides live support from MDA Specialists. Learn more on page 48 or visit [mda.org/care/connect](http://mda.org/care/connect).



Sabrina Johnson



and family choices in the past, this is your choice to make.”

However, communicating about your disease is important. “I would encourage individuals to share with someone because connection, acceptance, and support is a universal need,” Rebecca says.

Some people are ready to share right after their diagnosis, while others need some time to process the information first.

### Choosing to tell

Consider the following questions:

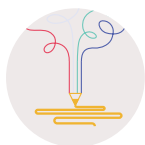
- **Who will you tell?** Everyone you know, or just those closest to you, such as your children, parents, or siblings?
- **Why should you share?** Some people want to withhold a diagnosis to protect loved ones from sadness, or because they don’t want to be treated differently. But Rebecca maintains that honesty strengthens relationships. In addition, sharing can help you locate resources and get the support you and your family need.
- **What do you want to say?** Depending on your audience, you might share your specific diagnosis or a more general term, such as a “muscle disease.” Consider whether you want to explain how the disease will affect you as it progresses, and be up-front about how you want to be treated and what help you and your family might need.
- **When will you share your news?** Some people are ready to share right after their diagnosis, while others need some time to process the information first.
- **Where will you have the conversation?** Whether you talk in person, on video, or on the phone, a private setting is best.

### Handling questions

Sabrina Johnson, a board-certified patient advocate ([SabrinaJohnsonAdvocate.com](http://SabrinaJohnsonAdvocate.com)), understands that the questions, concerns, and comments that follow these conversations, though they come out of love, can be overwhelming. Her father, known as Ton, died from ALS in 2019.

To avoid being overwhelmed, Sabrina recommends designating a point person to help spread the news and answer questions. That way, you don’t need to repeat everything. It also gives you space to process your feelings and enjoy time with your loved ones.

Managing emotional energy is important, Rebecca adds. She acknowledges that some people don’t know how



### HANDLING GRIEF

Strong emotions come with losing functions in progressive diseases. Read personal stories and tips at [MDAQuest.org/grief](http://MDAQuest.org/grief).



## SUPPORT FOR LIFE'S TRANSITIONS

MDA Next Steps Seminars offer information and resources on key topics related to life transitions, including receiving a diagnosis, transitioning to adulthood, and palliative and hospice care.

These virtual seminars are delivered over multiple days and include opportunities for participants to meet in small groups for

facilitated discussions and support. The goal of the Next Steps Seminars is to empower individuals and families with the information they need to navigate the next steps in their journey with confidence.

There is no cost, but registration is required. Find upcoming Next Steps Seminars at [mda.org/seminars](https://mda.org/seminars).

Image: iStock.com/Sofitukka

to react and say unhelpful things, like offering advice or launching into their own family story of a disease. “I encourage individuals to build ‘scripts’ to get out of awkward moments with these well-meaning people,” she says.

It’s OK to stick to the facts when family and friends ask for updates. “People do not need to know your feelings, stats during exams, or concerns,” Sabrina says. If someone asks how a medical appointment went, a facts-only response could be: “The appointment was helpful. The doctor checked my breathing, arm movement, and overall comfort. My next appointment is in three months.”

### Talking to children

How you tell a child about a diagnosis depends on their age, maturity level, and personality. Be honest, but don’t overwhelm them with too much information at once. Make sure the conversation feels safe, secure, and supportive.

Jodi and Kevin’s daughter was 2 when Kevin was diagnosed. “When children are curious and comfortable enough to ask questions, give them answers to the best

of your ability, or have them speak to a counselor who can,” Jodi advises. “You or a counselor won’t have all the answers, but you can welcome open conversations, and that allows children to feel heard and included.”

Sabrina, whose oldest son was 2 when her father was diagnosed, suggests letting a child’s teachers know about a relative’s diagnosis. “Together, the parents and school staff can make adaptations to ensure the child is cared for,” she says.



Rebecca Axline, LCSW-S, CSM, APHSW-C

### Starting the conversation

Many of us struggle to start difficult conversations, but don’t let that become an excuse to put it off. One strategy to help the conversation feel less daunting is to set a time limit for the discussion. “For example, you could say, ‘We are going to talk about this for 30 minutes, and then we will have lunch,’” Rebecca says. This helps keep the conversation on track and gives you both a chance to pause to process emotions.

When in doubt, remember that each person you tell about your diagnosis helps grow your support system. [Q](#)

*“I don’t believe there is a one-size-fits-all process. Just as you have made individual and family choices in the past, this is your choice to make.”*

— *Rebecca Axline, LCSW-S, CSM, APHSW-C*

Cheryl Alkon is a freelance writer based in Massachusetts.

# PLAYING LIKE BROTHERS

ADVERTISEMENT

## BECAUSE THEY CAN

### EMFLAZA® has been shown to preserve muscle strength and function

In a clinical trial of 196 boys aged 5 to 15 with Duchenne muscular dystrophy, the effectiveness and safety of EMFLAZA was compared with placebo (sugar pills) and prednisone. EMFLAZA improved muscle strength at 12 weeks compared with placebo (0.15 change in strength score vs -0.10 change in strength score)

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



Mikey, Age 9

Reid, Age 7

Actual EMFLAZA patients.

### STUDY INFORMATION

Real-world outcomes of long-term prednisone and deflazacort use in patients with Duchenne muscular dystrophy: experience at a single, large care center.

**Objective:** To assess outcomes among patients with DMD receiving deflazacort or prednisone in real-world practice.

**Methods:** Clinical data for 435 boys with DMD from Cincinnati Children's Hospital Medical Center were studied retrospectively using time-to-event and regression analyses.

**Results:** Median ages at loss of ambulation were 15.6 and 13.5 years among deflazacort- and prednisone-initiated patients, respectively. Deflazacort was also associated with a lower risk of scoliosis, improved ambulatory function, greater % lean body mass, shorter stature, and lower weight, after adjusting for age and steroid duration. No differences were observed in whole body bone mineral density or left ventricular ejection fraction.

Marden JR, Freimark J, Yao Z, Signorovitch J, Tian C, Wong BL. Real-world outcomes of long-term prednisone and deflazacort use in patients with Duchenne muscular dystrophy: experience at a single, large care center. J Comp Eff Res. 2020;9(3):177-189. doi:10.2217/ cer-2019-0170.

### Delayed onset of scoliosis (curved spine)

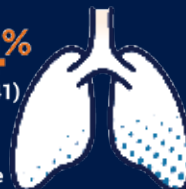
**LOWERED RISK OF SCOLIOSIS** compared to prednisone



Over 11.9 years, 17.9% of patients developed scoliosis taking prednisone vs 7.9% taking deflazacort.

### Preserved lung function

**9.24%** (CI: 3.06 - 15.41) higher forced vital capacity %-predicted vs prednisone



\*Forced vital capacity is a type of test that measures the amount of air your son can inhale and exhale.

**Emflaza®**  
(deflazacort)

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



## Summary of Information for EMFLAZA®

### What is EMFLAZA® (deflazacort) used for?

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

### When should I not take EMFLAZA?

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

### What warnings should I know about EMFLAZA?

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

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

### What should I tell my health care provider?

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

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

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

### What are the side effects of EMFLAZA?

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

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


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US-EMF-0292 11/22



smooth flooring

lighting

large bathroom



*Small changes can make a big difference  
when it comes to home accessibility*

BY ELIZABETH MILLARD

## Budget-friendly

# HOME Mods

**L**orraine Woodward, 62, has remodeled four accessible homes and has learned a thing or two along the way. As a mother who lives with limb-girdle muscular dystrophy (LGMD), Lorraine used to spend hours researching vacation homes for accessible features, so she could enjoy time with her family. In 2021, she decided to take a different approach: Remodeling short-term rentals and founding Becoming rentABLE ([Becoming RentABLE.com](https://www.becomingrentable.com)), an online accessible short-term

rental resource that lists her own properties and many others. What she discovered in the process is that accessibility is sometimes achievable through small changes and thoughtful modifications.

“When people think about renovations that focus on accessibility, they tend to envision large-scale and expensive projects, but you can achieve quite a bit even on a modest budget,” Lorraine says.

With that in mind, here are key home modifications and strategies to consider.

**Barrier-free entry**



Image: iStock.com/Justin Krug

**Consider a Certified Aging-in-Place Specialist (CAPS) as your contractor**

This specialization may sound like it’s only for older adults, but they have extensive knowledge for people of all ages with mobility limitations, according to Anthony Persiani,

a registered home improvement contractor and CAPS in Pennsylvania who often directs accessibility renovations.

“These professionals understand the unique needs of this population and, from a contractor’s standpoint, know what modifications can be completed in a home,” he says. “The

CAPS professional contractor will be able to walk through your home to make

appropriate recommendations for modifications that would be the most beneficial to your needs.”

Products in bold are included in **Helpful Home Products** on page 32.

**Start with the entry**

Figuring out what needs to be done can feel overwhelming, so a good place to begin is outside, making sure you can get into and out of your home easily. Resurfacing a driveway or paving a walkway are possibilities, but Lorraine recommends first seeing if you can clear obstacles to create a smooth path to the house.

“Make sure the pathway to get into the house is free of rocks, shrubs, and other plantings,” she says. “Also, keep in mind that your best entry might not be the front door. A garage door or back door could be easier to modify.”

If one of these doors allows entry with a small ramp — or none — it may cost less to modify. She adds that a rubber **threshold ramp** can make a typical home entrance more accessible.

**Add handles and grab bars**

The fix that Woodward appreciates the most in her properties as well as her own home, is using **lever-style door handles**

instead of traditional knobs. Her grip strength and hand movement are limited, and the levers are easier to grab and even allow her to open a door with her elbow instead.



**Lever door handles**

Image: iStock.com/Visit Reemvanitch



**WHAT’S A SMART HOME?**

Read how people with disabilities are using home automation technology at **MDAQuest.org/smart-homes-provide-accessibility**.



## + FINDING RESOURCES

The MDA Resource Center can help guide you to national and local resources for improving accessibility. Call **833-ASK-MDA1** or email [ResourceCenter@mdaUSA.org](mailto:ResourceCenter@mdaUSA.org).

### Relocate outlets

Bringing in an electrician to change outlets and light switches can be another boon for access. Outlets are usually located low to the floor, and light switches may be too high to reach from a wheelchair. In kitchens, outlets and switches are often out of reach on the other side of countertops.

“In our properties, we tend to elevate floor outlets and lower switches so everything is reachable,” Lorraine says. “In the kitchen, we put outlets and switches on the sides of cabinets. For a bedroom, focus on putting switches and outlets within reach of the bed.” Smart home technology can be helpful for turning lights on and off, but it’s best not to rely on the technology completely.

Another mobility aid is **grab bars** placed in strategic locations around the house, not just in the bathroom. Anthony recommends putting grab bars anywhere they might help — for example, next to the bed to make it easier to get in and out of bed.

### Conveniently placed switches



Image: iStock.com/IP Galanternik D.U.

## HOME MODIFICATION FUNDING

Although there are many affordable home modifications, the costs can add up if you’re making an entire home accessible, or if you’re undertaking large-scale renovations. Medicaid home and community-based services waivers are a common way to receive financial assistance for home modifications. These services are offered through state-level programs that may provide funds to make home modifications affordable for people with disabilities or elderly individuals.

You can find the resources available in your area by reaching out to your local Chamber of Commerce, Area Agencies on Aging, or Center for Independent Living (CIL). (Find a CIL at [acl.gov/programs/centers-independent-living/list-cils-and-spils](https://acl.gov/programs/centers-independent-living/list-cils-and-spils).) Your local CIL is the best starting point to assess if you qualify for waiver services, according to Rebecca Hume, a former Service Coordinator Supervisor for the Pennsylvania Medicaid waiver program who is now Senior Specialist and Writer for Quest Media. Rebecca also offers these tips:

- > Waiver programs are more likely to approve a home modification if you can show it will improve safety and accessibility in the home. Commonly approved modifications include stair lifts and ceiling lifts, for example.
- > Most waiver programs require a home assessment by an occupational therapist or home modification specialist to determine which changes will best meet an individual’s needs. Then, a contractor should determine if these modifications are structurally feasible.
- > Most waiver programs allow modifications to rented properties if participants can confirm that they plan to stay in that residence for the foreseeable future and the landlord is willing to sign permission for the modifications to be completed.
- > Medicaid is considered a payor of last resort, so if there are other funding options available, you should pursue those first. For example, veterans can qualify for federal grants that can pay for modifications and should reach out to the US Department of Veterans Affairs (VA) first. Some communities have private organizations or nonprofits that offer funding for home modifications.

## HELPFUL HOME PRODUCTS

Some products mentioned in this article can help make your home more accessible without breaking the bank. Here are suggestions on where to find them:

> Threshold ramps help mobility devices roll through doorways and over short rises. [DiscountRamps.com](http://DiscountRamps.com)



< Lever door handles are easier to open than doorknobs. [DirectDoorHardware.com](http://DirectDoorHardware.com)

> Grab bars can be placed anywhere they help with mobility, not just in bathrooms. [GrabBarSpecialists.com](http://GrabBarSpecialists.com)



< Rubber utility mats help prevent slipping in areas that get wet, such as bathrooms and laundry rooms. [Lowe.com](http://Lowe.com)

> Electric toilet lifts help users with lower body weakness to stand up. [DignityLifts.com](http://DignityLifts.com)

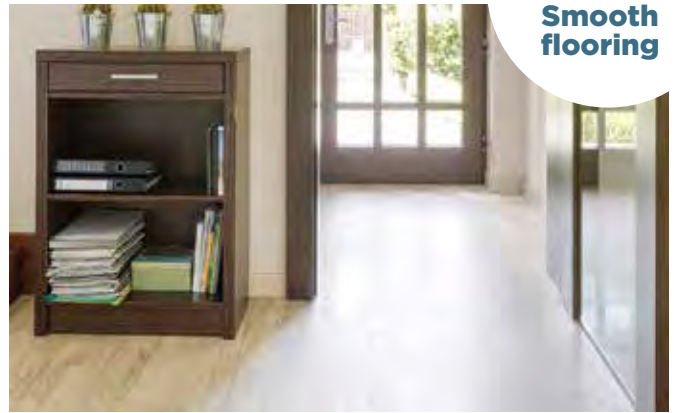


< Tilting vanity mirrors can be positioned to see yourself while seated. [Lowe.com](http://Lowe.com)

> A floor shower caddy is easy to reach while seated in the shower. [ToiletTreeProducts.com](http://ToiletTreeProducts.com)



< A freestanding toilet paper holder can be positioned to allow you to reach it easily. [HomeDepot.com](http://HomeDepot.com)



Smooth flooring

Image: iStock.com/KatarzynaBialasiewicz

### Focus on flooring

For people who use manual wheelchairs, walkers, or canes, thick carpets can be difficult to navigate and may increase injury risk. Throw rugs also can cause trips and falls. Lorraine suggests either replacing thick carpeting with a low-pile option or hardwood-style flooring. For example, there's a breadth of laminate flooring options that look like wood but are more affordable and durable.

In the bathroom, Lorraine opts for **rubber utility mats**, rather than fluffy bathmats, to prevent slipping. These mats are useful for other areas that might get wet, too — for example, in front of the kitchen sink or in a laundry room.

### Expand the toe kick

Kitchen cabinets usually have a “toe kick” at the bottom to make it easy to stand close to a sink or counter. But for those in a wheelchair, the standard toe kick area isn't enough. Expanding it may involve lifting the cabinets a few inches and making the toe kick deeper.



A tall, deep toe kick allows a wheelchair user to get close to the counter.

“Deepening that area to 8 inches or more allows you to get your knees closer to the cabinet,” Lorraine says. Consider picking a few cabinets for a contractor to modify in this way to make the kitchen more accessible overall.



### QUEST PODCAST

Listen to a discussion about how one community member adapted his home for his changing mobility level at [MDAQuest.org/podcast/accessible-home](http://MDAQuest.org/podcast/accessible-home).

## Maximize bathroom space

The bathroom tends to be where Lorraine spends most of her renovation budget on each project, and that makes sense. Being able to shower, use the toilet, and do other hygiene tasks can be challenging in an inaccessible bathroom. Fortunately, products geared toward bathroom accessibility are becoming more affordable.

“When I needed to get an **electric toilet lift** 30 years ago, the price was out of this world,” Lorraine says. “Now, you can get one for \$800 in some cases. Those are game changers, so it’s worth spending the money on a good one.”

Other low-cost modifications she loves:

- A **tilting vanity mirror** that pivots forward, so she can see herself while sitting
- A **floor shower caddy** that fits in the corner to keep all her products in easy reach
- Extra grab bars on walls in the main bathroom area, not just by the shower or toilet, so she can hold onto them while getting dressed



Image: iStock.com/Onzeg

- A **freestanding toilet paper holder** positioned close to the toilet because she doesn’t have full arm extension  
“Many times, seemingly simple and small changes like these can do so much when it comes to accessibility,” Lorraine says. [🔗](#)

Elizabeth Millard is a freelance writer in Northern Minnesota.

## ADVERTISEMENT

A new oral investigational treatment option may help you manage your generalized myasthenia gravis symptoms.

If you or a loved one has been diagnosed with generalized myasthenia gravis (gMG), the ExpanD Study may be an option. This clinical research study aims to evaluate the effects of an oral investigational medication in managing symptoms in adults with gMG.

### TO BE ELIGIBLE FOR THIS STUDY, YOU MUST:

- Be at least 18 years of age
- Have a confirmed diagnosis of gMG
- Be on a stable dose of gMG medication  
(only applies to those currently on gMG treatment)

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**ALEXION**<sup>®</sup>  
AstraZeneca Rare Disease

# A Place of



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*More Americans with disabilities are living independently. Community resources and self-advocacy help make it happen.*

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BY KAREN DOSS BOWMAN

# One's Own



**M**ore Americans with physical disabilities are living on their own, working or volunteering, starting families, and leading full lives. This makes sense, as medical treatments and care advance, because those with neuromuscular diseases are uniquely prepared for these experiences. Throughout their lives, most have been taught to face adversity, advocate for themselves, manage personal care attendants, and access resources vital to their care. These skills offer an advantage when it comes to adaptability and problem-solving.

Image: iStock.com/Elena Iakovleva, Jeremy

*“Because of my condition, I rely on others for everything. But when I became an adult and didn’t have a place of my own, I didn’t feel complete.” —Gil Vasquez*



Gilbert “Gil” Vasquez of San Antonio, embodies that spirit. The 43-year-old, who was born with spinal muscular atrophy (SMA) and needs 24-hour care, has proven time and again his capacity to set goals and achieve success. For example, after earning a Master of Business Administration degree in 2005, he worked for a decade in the banking industry. In his 30s, he decided to make a career change and enrolled in law school, graduating in December 2020.

These academic and professional accomplishments — and the life skills he’s learned along the way — gave Gil the confidence to move forward last year with his lifelong dream of living independently. Last December, he bought a house and moved out of his parents’ home. Now, he lives with his partner, who shares expenses and provides backup care when necessary.

“When you live in a situation where assistance is needed and always available, it’s easy to stay in your comfort zone,” says Gil, an attorney in private practice and adjunct professor at St. Mary’s University School of Law, his alma mater. “Because of my condition, I rely on others for everything. But when I became an adult and didn’t have a place of my own, I didn’t feel complete. So being able to live independently, even with the challenges, is worth it.”

Sarah Dunks, Accessibility Assessment and Aging-in-Place Specialist with Options for Independent Living, a Center for Independent Living in Wisconsin, regularly works with adults like Gil.

“People with disabilities gain an even greater sense of self-worth and quality of life knowing they can live independently,” she says.

“That independence can make them an even stronger person. The experience could be a stepping stone to going out into the community, getting a job, and contributing to society in that way. It’s just this trickle effect that can happen.”



Sarah Dunks

## GETTING BY WITH HELP FROM FRIENDS



When it comes to facing the unknown, it’s helpful to connect with others in similar situations. The MDA Peer Connections Program helps folks in the MDA community build connections with people of all ages — from parents of children to individuals in their 90s. The program is open to anyone in the neuromuscular

disease community — individuals living with neuromuscular diseases, caregivers, parents, spouses, and siblings.

Ready to make connections? Contact MDA’s Resource Center at **833-ASK-MDA1** or **ResourceCenter@mdaUSA.org**. After gathering information about you and your interests, MDA’s care specialists will connect you with others living with relatable and shareable experiences.

## Leaving the nest

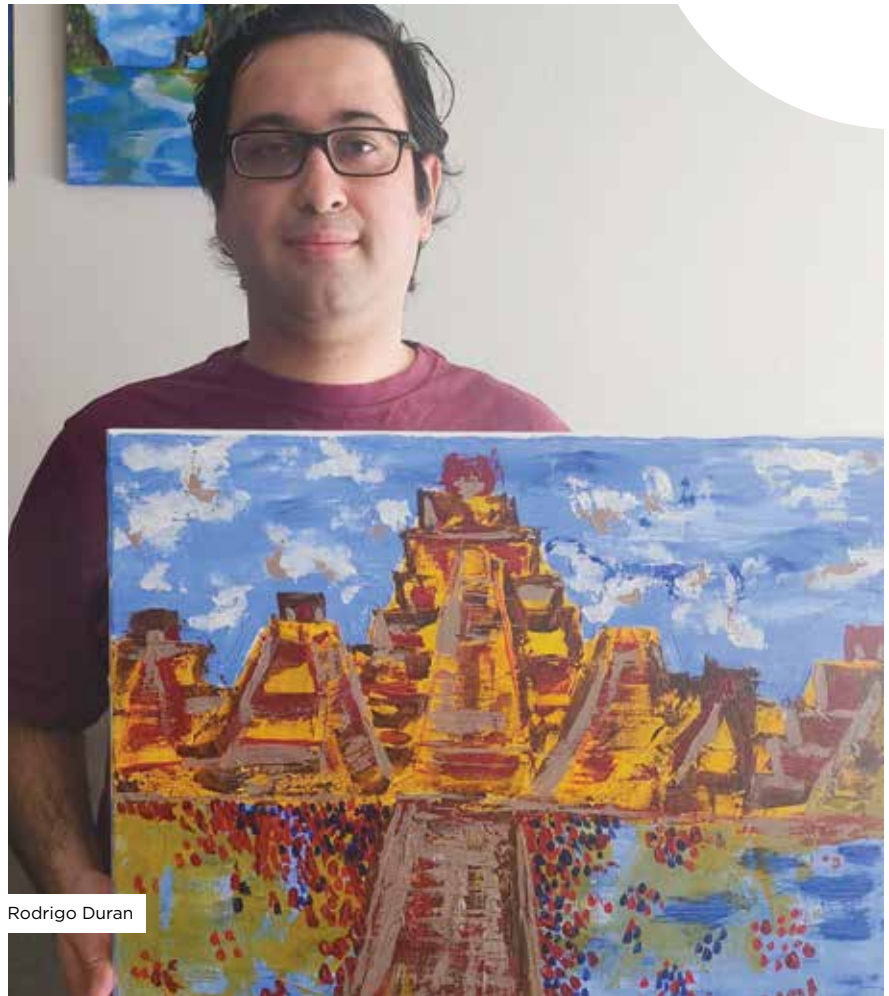
Parents naturally worry about their children — even if those children are adults — when they leave the nest. When Rodrigo Duran, of McKinney, Texas, decided to move into an apartment nearly four years ago, he had a heart-to-heart talk with his mother to allay her concerns. The 31-year-old artist, who lives with congenital muscular dystrophy (CMD) and epilepsy, assured her that he had carefully chosen two trustworthy roommates — one of whom is also his partner. They understand Rodrigo's disabilities, are prepared to help him, and know how to reach his family in case of an emergency.

“I asked my mom to give me a chance to learn for myself what I can and cannot do,” says Rodrigo, who walks with a cane and has difficulty standing up from a sitting position. “It gave her peace of mind knowing that my roommates know how to help me if something goes wrong.”

## Options for independence

For Beckley Lorenz, a 26-year-old living with Duchenne muscular dystrophy (DMD), moving into a licensed residential community for people with intellectual and developmental disabilities gave him the opportunity to live independently while receiving the care and services he needs. The Maryland man — who fought a successful legal battle to live in the community of his choice after the Maryland Department of Health initially denied him — moved last June into a house with two other residents. A “house mom” lives onsite and helps with cooking, cleaning, and transportation. Nursing staff visit every few weeks to check on him.

Beckley's new home is only 20 minutes from his parents' house and the community where he was raised. This



Rodrigo Duran

*“I asked my mom to give me a chance to learn for myself what I can and cannot do. It gave her peace of mind knowing that my roommates know how to help me if something goes wrong.” —Rodrigo Duran*

arrangement allows him to be independent and still be surrounded by a loving and familiar support system.

“Through this legal battle, I've learned that anything is possible when you believe in yourself and surround yourself with people that love and support you,” says Beckley, who loves hanging out with friends, going to the movies, and playing Scrabble.

His mother, Michele, reinforces that message. “That's kind of the attitude we've always had because you never know what is going to be the next obstacle. And now, he can make his own decisions and advocate for himself,” she says.



## A FIGHT FOR INDEPENDENCE

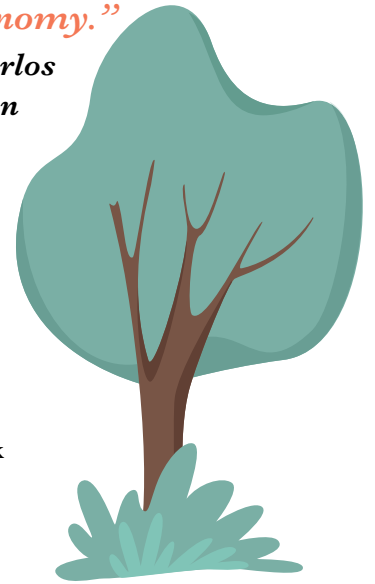
Read how a young man with DMD won his right to live independently at [MDAQuest.org/Beckley-Lorenz](https://mdaquest.org/Beckley-Lorenz).



Carlos Colon

*“You have to change your mindset and take steps to facilitate your new way of living. If you want to live independently, you have to develop autonomy.”*

—Carlos Colon



### Practical tips for adulting

Moving into your own home can be an exciting adventure. But the responsibilities of adulting can be overwhelming for anyone. Adding the unique challenges of living with a neuromuscular disease requires careful planning. These five tips can ensure a smooth transition.



#### 1. Find a place configured to your needs.

Consider your accessibility needs and look for homes or apartments that have these accommodations in place or can be modified. For example, does the dwelling have a ramp or lift? Are the interior and exterior doors wide enough for a wheelchair to pass through? If you're renting, what types of modifications will the landlord allow you to make? You'll want a responsive landlord with good communication skills.

**2. Plan for practical needs.** How will you manage daily living tasks, such as getting groceries, cooking, and housecleaning? Can you drive yourself to employment, social outings, or medical appointments, or will you need to call on family, friends, or a transportation service? Be sure these supports are in place wherever you decide to live.

**3. Create a budget.** Whether your income is earned through a job or disability benefits such as Social Security or Medicaid, make a budget to ensure that you can meet your expenses each month. When Carlos Colon, of Puerto Rico, moved into an apartment with his girlfriend, he had to think more carefully about money to navigate the new responsibilities of paying for rent and utilities, buying groceries, and other expenses.



“When you’re living with your parents, you don’t have to think about those things as much,” says Carlos, 29, who is studying for a doctoral degree and lives with Charcot-Marie-Tooth disease (CMT). “You have to establish priorities. Maybe you cook your own meals instead of going out to eat as much, for example. You have to change your mindset and take steps to facilitate your new way of living. If you want to live independently, you have to develop autonomy.”

**4. Build a support network.** One of the biggest challenges of moving away from your family is hiring and managing caregivers. About a week after moving into his new home, Gil’s daytime caregiver quit. He has struggled to find a replacement and is relying on his roommate for care during those hours. “You can have everything perfectly planned out, and then all of a sudden, life throws a curve ball at you,” Gil says. “Nobody can do everything on their own. You always need help around you from people you can trust.”

Keep a list of family and friends who can be on call to provide backup care when needed. You’ll also want to stay close to people who can provide emotional support when times get tough and join you in celebrating life’s victories.

**5. Seek resources.** Contacting your local Center for Independent Living ([acl.gov/programs/centers-independent-living/list-cils-and-spils](http://acl.gov/programs/centers-independent-living/list-cils-and-spils)) is a good starting point. This nationwide network of organizations was established to support community living and independence for people with disabilities. They can help you think through the pros and cons of your choices, explain rental agreements, or point you to other helpful resources.

“Independent living centers have many staff and board members who are people with disabilities, so it’s not just that we have helpful information — we have experienced the same situations as many of our clients,” says Sarah. “We are able to offer connections and mentoring.” 

### **+MONEY MATTERS**

Managing your own money is an important part of independent living. Build your financial know-how with MDA’s educational workshop Access to Financial Independence. Find it at [mda.org/AccessWorkshops](http://mda.org/AccessWorkshops).

Karen Doss Bowman is a freelance writer and editor living with progressive muscular atrophy, a subset of ALS, in Bridgewater, Virginia.

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AstraZeneca Rare Disease

# Gene Therapy Gains Steam

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*A decade ago, there were no gene therapies. Now, we're poised for a wave of drug approvals.*

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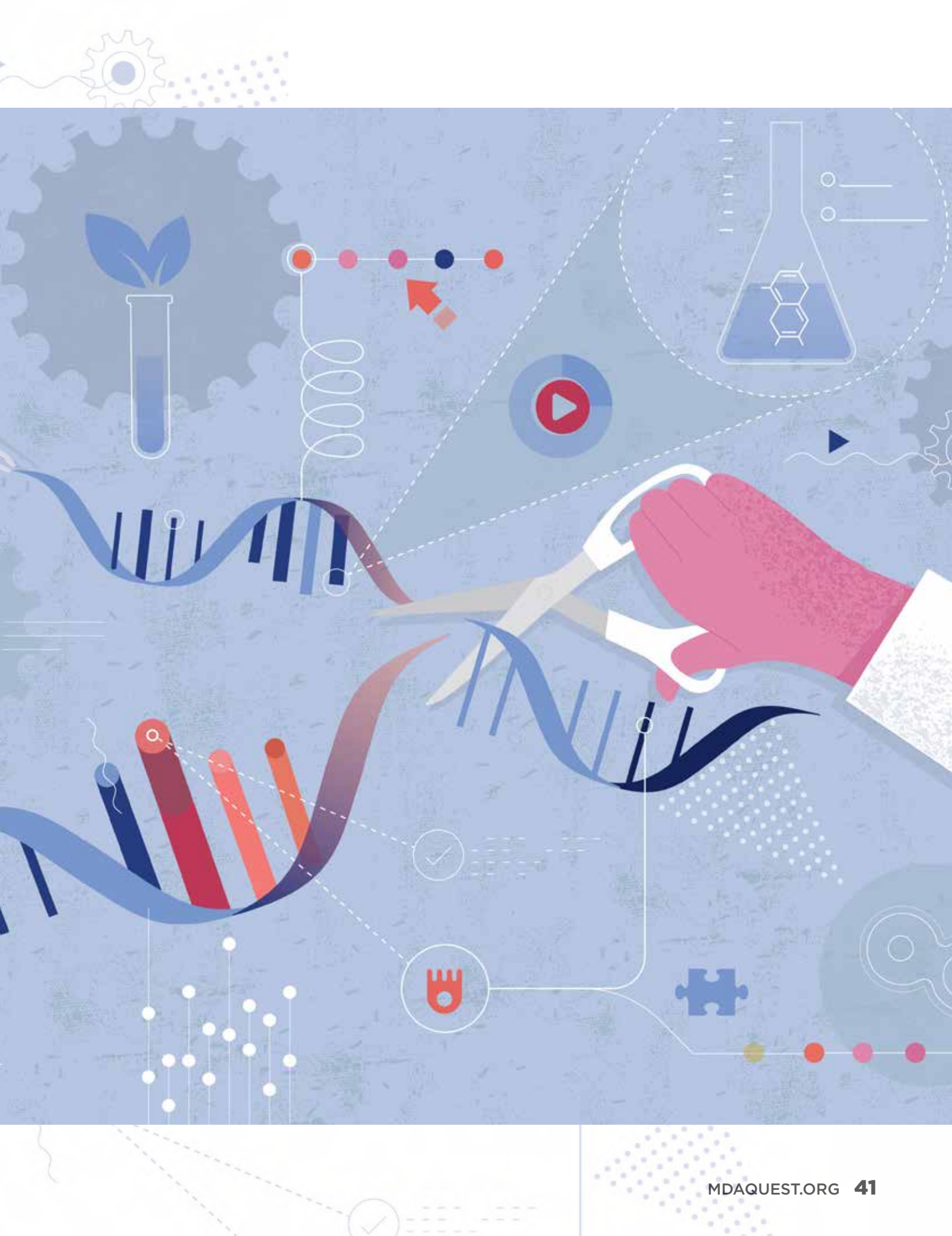
BY LARRY LUXNER

If current trends are any indication, 2023 may see the approval of more new gene therapies for more diseases than any year in history.

Consider this: In the past six years, the US Food and Drug Administration (FDA) gave its blessing to 12 such therapies — including five in 2022 — compared to zero not even a decade ago. Among this new class of treatments is Zolgensma, a \$2.1 million one-time infusion approved in 2019 to treat spinal muscular atrophy (SMA).



Image: iStock.com/DrAfter123





Two-year-old Lennox received a gene therapy treatment for SMA at 19 days old.

Photo by Opal & Onyx

Today, Lennox acts like any other 2-year-old. She runs, jumps, climbs, and does gymnastics.

That **gene therapy** has already proven nothing short of a miracle for Lennox Joy Schill of Brighton, Colorado, who was diagnosed with SMA type 1 a year after her state added SMA to its newborn screening program. At only 19 days old, Lennox received her Zolgensma infusion at Children’s Hospital Colorado. Her parents were able to choose between two disease-modifying therapies — Zolgensma and nusinersen (Spinraza) — for their infant, who was not yet showing any signs of SMA.

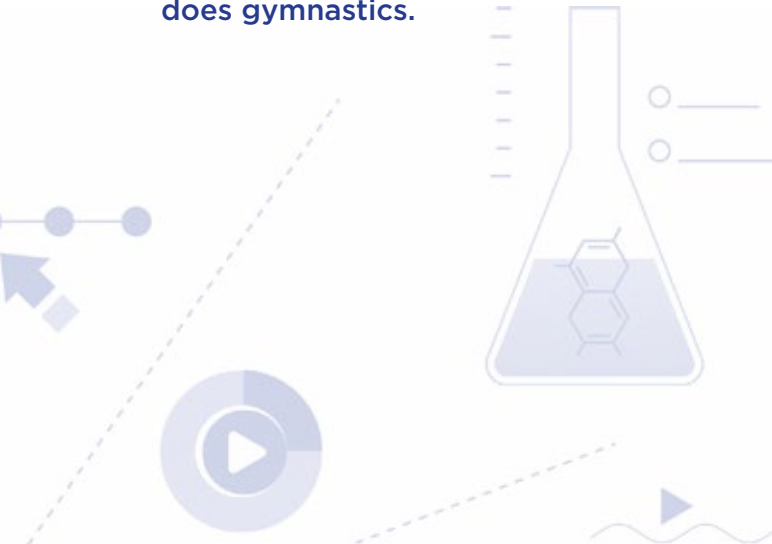
“While there were more possibilities of acute effects up-front with Zolgensma, we chose to go with that since it’s a once-in-a-lifetime thing, and Spinraza is every four months for the rest of her life,” says Lennox’s mother, Erin. “We thought that would be best for our child.”

Today, Lennox acts like any other 2-year-old. She runs, jumps, climbs, and does gymnastics.

**New frontier of progress**

The FDA approved the first gene therapy to treat a **genetic disease** in 2017. Called Luxturna, it was a game-changer for people with biallelic RPE65 mutation-associated retinal dystrophy, which causes vision loss. Since then, 11 more gene therapies have been approved, and each success has yielded knowledge that helps researchers develop new therapies for other diseases. Currently, about 2,000 gene and cell therapies are in clinical trials for diseases from cancer to sickle cell disease to neuromuscular diseases.

At his opening address to the clinicians, researchers, and pharmaceutical experts gathered for the 2023 MDA Clinical



**+FIND RESEARCH AND CLINICAL TRIALS**

Read the Quest blog ([MDAQuest.org/blog](https://mdaquest.org/blog)) for breaking news on treatments and research. Use MDA’s Clinical Trials Finder Tool to search for available neuromuscular disease clinical trials at [mda.org/clinical-trials](https://mda.org/clinical-trials).

& Scientific Conference in March, Donald S. Wood, PhD, MDA's President and CEO, called this a "dynamic time" for gene therapy.

"The frontier of progress in neuromuscular diseases has moved from the research labs more and more toward clinics," Dr. Wood said. "However, we are in an era when it's not enough to come up with a new idea. You need to take that idea and execute it in a way that can help patients."

The same year that Luxturna was approved, researchers at the University of California-Berkeley used a gene editing tool called **CRISPR-Cas9** to treat Duchenne muscular dystrophy (DMD) in mice with a single injection. This marked the first time any genetic disease had been successfully treated in an adult **animal model** using gene editing. While promising, translating advances in a lab to humans in a clinic can take a long time. Currently, there are no gene editing-based treatments on the market.

However, efforts continue. Earlier this year, German scientists announced they had used CRISPR-Cas9 to fix the **genetic mutation** that most commonly causes type 2A limb-girdle muscular dystrophy (LGMD) in muscle stem cells. The idea is that muscle stem cells could be extracted from an individual with LGMD, repaired, and transplanted back into the person. The researchers are testing the implantation of repaired cells in an animal model to see if it helps mice generate healthy muscle.

As of press time, the FDA is considering whether to approve delandistrogene moxeparvovec (SRP-9001), a gene therapy developed by Sarepta Therapeutics to treat DMD. If approved, SRP-9001 will become the world's first gene therapy for DMD, as well as the most expensive drug in history, with a retail price likely exceeding \$4 million. At the MDA Conference, Barry J. Byrne, MD, PhD, and John W. Day, MD, PhD, facilitated a critical discussion among MDA Care Center Directors about ensuring all clinicians and their teams are ready to support our DMD community through what will be a brand-new process of administering the therapy and conducting lab work and follow-up visits.

"I'm very excited about the prospect of gene therapy," says Mindy Cameron, a patient advocate and government affairs consultant for Santhera Pharmaceuticals, as well as the mother of a son with DMD. "I've always been a big believer in the concepts behind gene therapy. Yes, we do have to overcome some issues, but I think that first approval's going to open the gates, and we're going to get better and better treatments."

### Big benefits and challenges

As with most treatments, gene therapy comes with potential benefits and risks.



## WORDS TO KNOW

Here is a guide to some common gene therapy terms used in this article.

**Adeno-associated virus (AAV):** A harmless virus that can be engineered to deliver genetic material to target cells. AAV vectors are the leading platform for delivering gene therapies in the human body.

**Animal model:** A non-human animal used in research because it can mimic aspects of a biological process or disease found in humans. Typically, animal models are used to test the safety and efficacy of new therapies before human clinical trials are started.

**CRISPR-Cas9:** A tool used to "edit" pieces of a cell's DNA. It uses a specially designed RNA molecule to guide an enzyme called Cas9 to a specific sequence of DNA. Cas9 then cuts the strands of DNA. The cut ends can be joined, or a new piece of DNA may be inserted in the gap.

**Gene therapy:** Modifying the content of a person's genetic code or changing gene expression with the goal of treating or curing a disease. Approaches to gene therapy include gene replacement, gene silencing, and gene editing.

**Genetic disease:** A disorder caused by changes in one or more genes. These genetic mutations, or variants, can be inherited from parents or occur spontaneously.

**Genetic mutation:** A copy of a gene with a difference in its DNA sequence. Genetic mutations, or variants, can be pathogenic (disease-causing) or benign (not causing disease).

Read the Quest article "These 7 Things Can Help You Understand Gene Targeting Therapy" at [MDAQuest.org/targeting-genes](https://mdaquest.org/targeting-genes).

Among the advantages: it has the potential to treat a broad spectrum of inherited diseases, only one dose may be required, and long-term disease benefits or even cures are possible.

“Gene therapy has the potential to be one-and-done, but we don’t know how long the benefit will last,” says Sharon Hesterlee, PhD, MDA’s Chief Research Officer.

The disadvantages of gene therapy include its complexity, high manufacturing costs, potential for irreversible side effects, and special expertise required for administration. A gene therapy is not a drug that you’ll be able to pick up at the local pharmacy.

Dr. Byrne directs the Powell Gene Therapy Center at the University of Florida. He’s been involved in research on gene therapies for several diseases, including DMD, Friedrich ataxia, and Pompe disease.

“When we started Luxturna studies, we thought there was a certain degree of vision loss that would never be recovered, but people who had lost their vision actually recovered it,” he says. He has seen similarly surprising results in ongoing DMD gene therapy trials being conducted by Sarepta, Pfizer, and Solid Biosciences.

“We’re seeing boys gain skills that they would otherwise have never developed,” he says. “Even with something as simple as running, you have to get both feet off the ground. Boys with Duchenne can’t do that, even before they decline. And now we see that many of the kids who’ve been treated with gene therapy can run.”

### Ensuring access

Creating a gene therapy that works is only the first step. Once a new therapy goes through the long process of being developed and approved, the next challenge is to ensure that the people who need it have access to the drug — and to the professionals who must administer it. “Even at the best-run clinics, the number of patients with Duchenne they can treat with gene therapy will be about two a week — and that’s if there are no significant complications,” Dr. Wood said at the MDA Clinical & Scientific Conference.

Experts point out that, with approximately 30 million American living with rare diseases, the healthcare system isn’t ready to meet the needs of all the people who could benefit from genetic treatments. It’s not uncommon for people trying to get access to the currently approved gene therapies to run into barriers ranging from insurance approvals to hospital staffing.

The pharmaceutical industry will have to adapt, too, according to Peter Marks, MD, PhD, Director of the FDA’s Center for Biologics Evaluation and Research, who delivered a keynote address on “the promise of gene therapy” at the MDA Clinical & Scientific Conference.

“The pharma industry is used to ‘pay as you go’ — you get your prescription refilled every 30 days. But when you give someone a therapy once and they don’t need it again ... then once you’ve taken care of the patients with that disease, now, on a yearly basis, your market will be smaller,” he said.

This is one of the factors that lead to higher costs for drugs that treat rare diseases, and drug cost is another significant barrier to access.

MDA’s research grants and programs aim to address some barriers at the source by reducing potential time delays and costs to motivate more companies to develop neuromuscular disease therapies. (Read “Investing in Research Success” on page 7.)

In addition, public-private partnerships are emerging to help develop new drugs and enable access for small patient populations.

The Foundation for the National Institutes of Health (NIH), along with 35 public, private, and nonprofit partners, launched the Bespoke Gene Therapy Consortium (BGTC) in 2021. This venture involves the NIH’s National Center for Advancing Translational Science (NCATS) and is led by Philip J. Brooks, PhD, Acting Director of the NCATS Division of Rare Diseases Research Innovation.

Among other things, the consortium aims to make **adeno-associated virus** technology available for a broader range of diseases, streamline preclinical and product testing, and bring gene therapies to all individuals who need it sooner.

“The consortium is looking for better and more efficient ways to get gene therapy trials up and running, particularly for diseases that are of no commercial interest because they’re so rare,” Dr. Brooks says, noting that only 600 of the 7,000 known rare diseases have approved therapies. This means that at the current rate of development, it would take about 2,000 years to find treatments for all of them.

“This illustrates the point that we have to do something different if we want to treat, or even open, clinical trials for all these diseases,” Dr. Brooks says.

MDA also has launched its own gene therapy program, the Kickstart Program, focused on ultra-rare neuromuscular diseases.

The world got a peek at many of the new ways of doing things — and the leading minds behind them — at this year’s MDA Clinical & Scientific Conference. It’s apparent that the wave of gene therapies is coming. And the rare disease community is ready for them. [Q](#)

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

# Quest Marketplace

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# MDA Conference Makes an Impact



Donald Wood, PhD (left), and Peter Marks, MD, PhD (right), kicked off the 2023 MDA Clinical & Scientific Conference.

In March, more than 2,000 people gathered in person and virtually for MDA's 2023 Clinical & Scientific Conference.

Held at the Hilton Anatole in Dallas, the three-day event brought together healthcare providers, researchers, investors, pharmaceutical giants, advocacy teams, patients, and caregivers, with nearly 1,500 in-person and 555 virtual attendees. This is the biggest, most comprehensive conference of its kind in the world, allowing professionals in the neuromuscular disease space to present their research, form collaborations, and learn about the latest developments and therapies, ultimately supporting the development of better care and treatments for our community.

This year's educational sessions were broken into tracks that offered in-depth education on the latest in ultra-rare diseases, technology, gene therapy, and care throughout the patient journey. Sessions also covered timely topics, such as building an infrastructure to safely and efficiently administer new gene therapies and practical matters like digestive health.

### A good start

To kick off the conference on March 20, Donald S. Wood, PhD, President and CEO of MDA, welcomed attendees and introduced keynote speaker Peter Marks, MD, PhD, Director of the

Center for Biologics Evaluation and Research (CBER) at the US Food and Drug Administration (FDA).

Dr. Marks explained how the FDA is implementing several strategies to help researchers and pharmaceutical companies develop gene therapies for rare diseases and speed the review process. One example is a pilot project to apply methods used during Operation Warp Speed — the federal initiative that sped up the development of COVID-19 vaccines — to develop promising therapies.

“We're very committed to advancing the time and availability of gene therapies for all sorts of disorders,” he told the audience.

### Legacy Award

Stanley Appel, MD, Director of the Ann Kimball and John W. Johnson Center for Cellular Therapeutics at Houston Methodist, presented MDA's Legacy Award for Achievement in Clinical Research. Dr. Appel, a world-renowned researcher and clinician in the amyotrophic lateral sclerosis (ALS) field, presented the award to Merit E.



Merit E. Cudkowicz, MD, MSc, received MDA's Legacy Award.



Cudkowicz, MD, MSc, to recognize her innovative work researching ALS treatments.

Dr. Cudkowicz started the Healey Center for ALS at Massachusetts General Hospital, where she and her team designed the first-of-its-kind Platform Trial for ALS. This clinical trial tests multiple drugs simultaneously on the same platform, reducing the number of trial participants taking a placebo and the time it takes to develop treatments.

“We are at a transformational time in ALS, as well as all the illnesses ... in the neuromuscular world,” Dr. Cudkowicz said after accepting the award. “It is so nice to be able to tell [our patients] that we do have options for them today that are on the market as well as many trial options for them.”

Further highlighting this pivotal time for ALS research, Matthew Harms, MD, Associate Professor of Neurology at Columbia University, oversaw an ALS education track. Its topics ranged from describing major collaborative efforts to new information about cellular changes and gene therapy efforts in ALS.

### MDA families

While the MDA Clinical & Scientific Conference is geared toward the scientific community, MDA knows individuals living with neuromuscular diseases and their families must stay informed on

research and care. MDA welcomed members of the MDA community to participate in the virtual conference at no cost; some even made the trip to Dallas to attend in person.

In addition, MDA families were invited to speak in some sessions,

helping the attending scientists understand the impact of their work.

For example, in a session on Practical Management of Gene Therapy, Kody and Sydney Graves, of Villa Ridge, Missouri, shared their daughter Kenzie’s treatment journey. Kenzie was diagnosed with spinal muscular atrophy (SMA) through newborn screening in 2019. She received Zolgensma, a one-dose gene replacement therapy, when she was 1 month old. At 2 months, she began physical

and occupational therapy, and she started taking risdiplam (Evrysdi) at 1 year old. Kenzie, now 3, is able to walk and continues her therapy and Evrysdi regimen.

“At the end of the day, it was that newborn screening and [early] identification that triggered all the successes to come and the fact that she’s walking, talking, and very active today,” said Sydney, Kenzie’s mom.

Next, Brent Furbee, of Columbia, Tennessee, spoke about his experience getting his son, Emerson, into a gene therapy clinical trial for Duchenne muscular dystrophy (DMD) in March 2021.

The process involved a series of steps, each of which “came with its own challenges and blessings,” Brent explained. The most emotional and nerve-racking phase was the two-hour testing and evaluation to determine if Emerson was accepted into the trial. When he and his wife, Sabina, learned their son was accepted, “it was like a weight was lifted off our shoulders,” he said.

Then, there was another waiting period before Emerson received the infusion, during which he needed to stay healthy. “We had to make sure he didn’t get COVID ..., make sure he didn’t fall down and break a leg or hurt himself,” Brent said. “We felt like we needed to wrap him in bubble wrap, but we wanted him to live his life.”

Since receiving the infusion, the family has traveled regularly to St. Louis for Emerson’s post-therapy follow-ups.

Emerson, now an active 6-year-old, is able to walk and keep up with his peers. “When we first received this diagnosis, we expected for him to start declining by the time he was 6,” Brent said. “We had a reasonable expectation that this [trial] was going to benefit him, and what we have seen is nothing short of remarkable.”

Brent ended by thanking and encouraging the scientists in the room. “Continue to provide that hope that there is something in the pipeline,” he said, emphasizing that the hope is vital to families like his.

**Dr. Cudkowicz started the Healey Center for ALS at Massachusetts General Hospital, where she and her team designed the first-of-its-kind Platform Trial for ALS.**

### +COMMUNITY EDUCATION

Throughout the year, MDA offers high-quality educational programs and materials focused on helping MDA community members overcome barriers to access, navigate life’s transitions, and make informed decisions about care. Visit [mda.org/community-ed](https://mda.org/community-ed).



## Introducing MDA Connect for Live Support

Sometimes you just need to talk with someone. We created the MDA Connect Program, a collaboration between the MDA Resource Center and the Family & Clinical Support Specialist team, so you can talk with a real person about resources and information that will help you navigate life with a neuromuscular disease.

With MDA Connect, members of the neuromuscular disease community — including parents and caregivers — can schedule 30-minute, one-on-one video calls with MDA Support Specialists. Our specialists can help locate resources, navigate care within MDA’s Care Center Network, and share information on MDA programs and opportunities for engagement.

“MDA Connect is an exciting opportunity we can now offer our community — to simply connect. Thanks to technology, MDA can come to members anywhere,” says Nora Capocci, Vice President of Healthcare Services at MDA. “Virtual appointments offer another way to learn more about MDA programs and services and request resources. This program complements the MDA Resource Center as another means of connection — allowing us to be ‘virtually’ together.”

MDA community member Crystal, who lives with mitochondrial myopathy, used MDA Connect to get information about her local MDA Care

Centers. It was easy for her to schedule a meeting with an MDA Support Specialist. “It was just like scheduling a haircut or any other online appointment,” Crystal says, adding how valuable it was to have a face-to-face interaction.

“Putting a face to a name is invaluable,” Crystal says. “It makes a difference to have someone see your expressions and emotions, especially if you are discussing a heavy topic, such as a progression in your diagnosis.”

MDA Connect video appointments are available in English and Spanish for United States and Puerto Rico residents. All appointments are scheduled and accessed online.

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

Appointments can be scheduled Monday-Friday, 9 a.m.-5 p.m. CT. For support in other languages, please contact the MDA Resource Center at [833-ASK-MDA1](tel:833-ASK-MDA1) or [ResourceCenter@mdaUSA.org](mailto:ResourceCenter@mdaUSA.org) to schedule a phone call using a translation service.

Please note that MDA Specialists do not provide medical advice or counseling services, but they can help connect you to those services.

## Advocacy Gains

MDA’s advocacy team has been hard at work on critical initiatives, such as:

### Improving air travel

MDA’s advocacy program and grassroots volunteers have a unique opportunity to spur meaningful reforms this year. Congress must reauthorize the Federal Aviation Act to prevent

disruptions in the air travel system, and MDA is working diligently to ensure reforms are included in this “must-pass” legislation. These reforms include:

- Improving training for all flight crew members, baggage handlers, and ramp agents who assist passengers with disabilities or handle

wheelchairs and mobility devices. This training includes how to transfer people in and out of wheelchairs and prevent wheelchair damage.

- Requiring regular maintenance and inspection of onboard wheelchairs
- Incorporating the Air Carrier Access Amendments Act (H.R. 1267 / S. 545). This bill would strengthen nondiscrimination provisions, allow passengers with disabilities to file private legal action against the airline, improve aircraft design, and increase access to better seating accommodations.
- Strengthening enforcement of fines and penalties for wheelchair damage and injuries
- Continuing the effort to allow people to stay in their wheelchairs during flight



### **Adding Duchenne muscular dystrophy (DMD) to the Newborn Screening Program**

Despite hurdles, MDA continues to lead the effort to add DMD to the Recommended Uniform Screening Panel, which would bring the country one step closer to screening all newborns for this disease.

### **Ending discrimination in accessing treatments**

MDA is advocating for Congress to pass the Protecting Healthcare for All Patients Act of 2023, which would end the use of “quality-adjusted life years” to deny coverage of treatments for people with disabilities who participate in federally funded health programs.

### **Urging Medicare to cover seat elevating systems**

Medicare has released a proposal that would finally cover wheelchair seat elevating systems for those who use Group 3 wheelchairs (typically used by someone living with ALS) and is considering covering seat elevating systems for Group 2

wheelchairs (typically used by someone living with inclusion body myositis and myasthenia gravis). These systems are vital for those living with a disability. MDA and its advocates sent comments to the Centers for Medicare & Medicaid Services, urging it to finalize this proposal as soon as possible. Regardless of what CMS decides, we will continue to work to ensure that everyone has access to these important devices.

### **Awarding Advocacy Collaboration Grants**

Helping fulfill MDA’s mission of empowering the people we serve, MDA awarded more than \$100,000 in grants to five organizations to fund innovative and impactful advocacy projects across the country.

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**Learn more about MDA advocacy projects and become a grassroots member at [mda.org/advocacy](https://mda.org/advocacy).**

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## **In-person Community Education Is Back!**

MDA is excited to announce the return of in-person education with four upcoming Engage Symposiums. These full-day, multi-session programs provide up-to-date information from experts in the field, empowering learners with actionable information to support their care and

life goals. Attendees can also connect with others impacted by neuromuscular diseases and explore exhibitor booths to learn about different resources.

“There will be something for everyone at each symposium,” says

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**Register and find event updates and agendas at [mda.org/symposiums](https://mda.org/symposiums), or send questions to [MDAEngage@mdaUSA.org](mailto:MDAEngage@mdaUSA.org).**

Marissa Lozano, MEd, MDA’s Director of Community Education. “We will have disease-specific and general quality-of-life tracks, focusing on topics such as navigating insurance and accessible travel.”

The symposiums are free and open to the neuromuscular disease community, but registration is required. Mark your calendar for these upcoming events:

### **June 3, 2023**

Westin Princeton at Forrestal Village, Princeton, NJ

**Learning tracks:** General neuromuscular, amyotrophic lateral sclerosis (ALS), spinal muscular atrophy (SMA)

### **July 22, 2023**

University of Florida Research and Academic Center at Lake Nona, Orlando, FL

**Learning tracks:** General neuromuscular, Duchenne muscular dystrophy (DMD)/Becker muscular dystrophy (BMD), Pompe disease

### **Sept. 23, 2023**

McNamara Alumni Center at the University of Minnesota, Minneapolis, MN

**Learning tracks:** General neuromuscular, Charcot-Marie-Tooth (CMT), DMD/BMD

### **Nov. 11, 2023**

Paul and Mildred Berg Hall at Stanford University, Stanford, CA

**Learning tracks:** General neuromuscular, myasthenia gravis (MG), and myotonic dystrophy (DM)

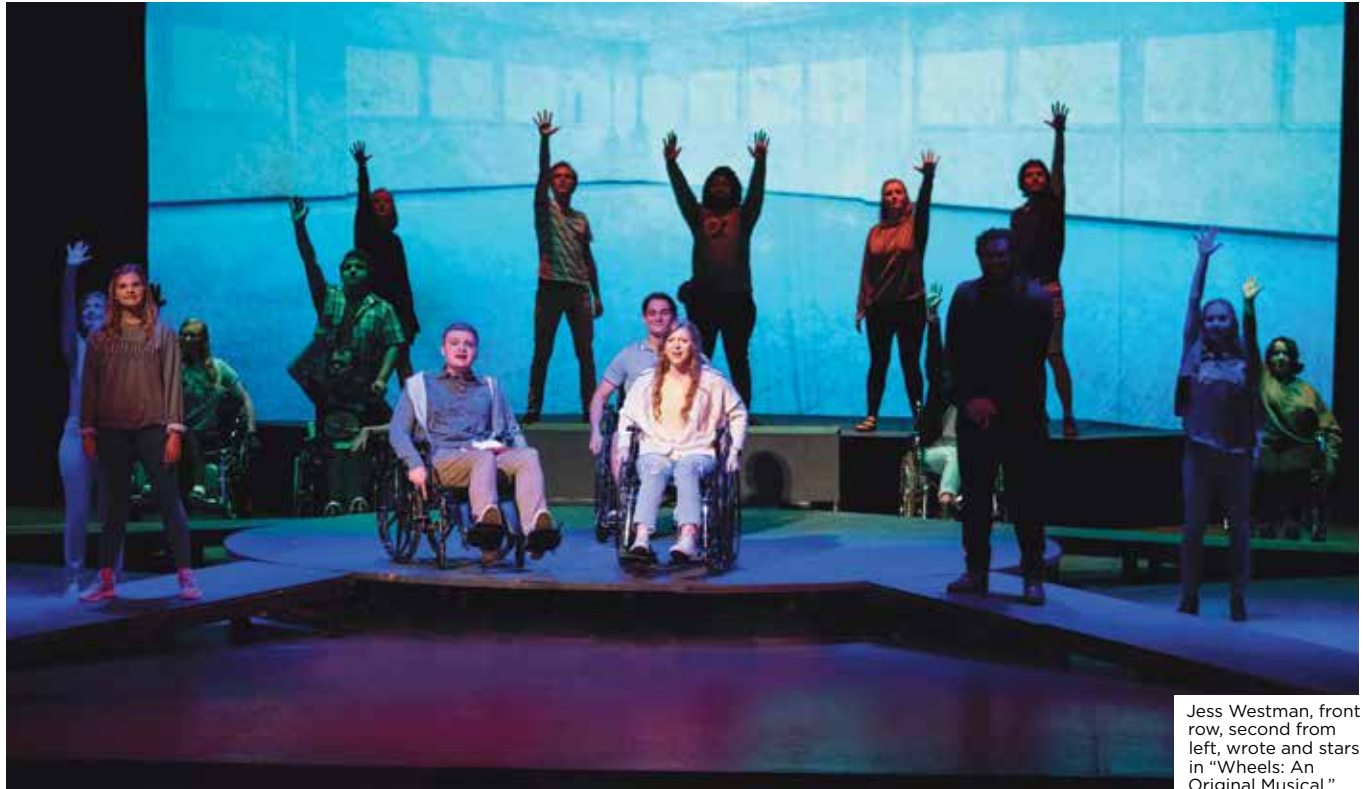




# There's More to Life

*Why I wrote a musical about muscular dystrophy*

BY JESS WESTMAN



Jess Westman, front row, second from left, wrote and stars in "Wheels: An Original Musical."

**G**enetic testing, heart and lung monitoring, muscle biopsy, and electromyography are all methods for diagnosing muscular dystrophy, but there's only one method for breaking the news.

"Shocked, sad, and scared," my mother recalled feeling when my brother and I were diagnosed with Becker muscular dystrophy (BMD). She and my dad pondered the unthinkable: "How can we make sure our boys will live happy, healthy, and productive lives?" Too many parents ask themselves that question, and ironically, the answer is pain. My disease isn't a preference, but it's taught me there's more to life.

## The game of pretend

I was starting preschool when my parents first put me onstage. Having recently been diagnosed with BMD, all athletics were thrown into the backseat, and my brother and I began wholesome childhoods of entertaining people. Beginning my acting career as a 5-year-old mobster in

a community production of "Bugsy Malone," I'd eventually act in high school and university musicals like "Seussical," "Footloose," "Shrek: The Musical," "Anything Goes," and "Into the Woods."

Other students, over the years, suggested I pace myself, but every moment, onstage and off, was a brand-new, make-them-laugh opportunity, free from my disease. Theater was everything; I couldn't get enough. And then I turned 19.

I still enjoyed character acting, but it was taking a physical toll. My "doing too much" was adding up. At first, I was exiting the stage a

How long could I continue coping theatrically? And just like that, **making people laugh** meant less, and **making them aware** meant everything.

little winded, but by my final year of college, I was collapsing backstage in a wolf costume.

I hated what was happening to my body, but I had no control. And then, a friend asked me why I was limping. I recall staring at them in horror. Nobody was supposed to notice. I had trained myself to correct the limp — to walk it off. But they still noticed.

I found myself at a crossroads. What do you do when you know the truth, but facing it is the scariest thing on earth? How long could I continue coping theatrically? And just like that, making people laugh meant less, and making them aware meant everything.

### Making it make sense

Newly filled with a need to make my disability make sense, I decided to write a musical about someone like me, called “Wheels.” I had written other plays, songs, and musicals, but I hoped that creating something honest would have a greater impact.

What I pitched to my peers as “a showcase of everyday life” grew into the story of a troubled man named Elijah, his life with muscular dystrophy, and the complex relationship between him and his caretaker, Amelia. I thought of it as a theatrical window into the normalcy of what society might consider abnormal, juxtaposed with the wheels of life — wheels that keep spinning, whether we like it or not.

The initial, tri-collegiate iteration of “Wheels: An Original Musical” premiered in Abilene, Texas, in June 2021 and was met with local acclaim. Following the production, I contacted MDA, and their PR director connected me with Tony Award-winning Broadway director and dramaturg Sheryl Kaller. I moved to New York City two months later.

When I arrived, I was expecting an indescribable, life-changing city. But it wasn't. To me, it was a tourist trap laden with physical obstacles. However, I needed to be here. If I wanted to continue dramaturgical meetings with Sheryl and truly make “Wheels” a Broadway show, I'd have to deal with the stairs and walking.

This experience taught me that I won't succeed in the entertainment industry if all I do is sulk about how hard my life is. So, anytime I'd think, “Other composers don't have to suffer like this,” I'd start putting that passion into my script and songs. Suddenly, those 58 days of freelance graphic design that would get me to one meeting with a producer or a PR team were worth it. It all became worth it.

I've learned that powerful storytelling isn't about who is telling the story, but why.



Jess played Pinocchio in a 2018 college production of “Shrek: The Musical.”

### Impact is everything

New York City, though still inaccessible, is my home now. Where the stairs have failed me, the people haven't. God hasn't. My friends have become my family, I've discovered rich cultures, and I've grown in ways I couldn't have predicted. My entertainment pursuit has become a means of honestly spreading awareness, and I didn't do it on my own.

Today, “Wheels: An Original Musical” is a Broadway show in the making and a 2022 Eugene O'Neill National Music Theater Conference semifinalist. And, through musical collaborations aside from “Wheels,” I've kept my passion for storytelling alive. I've learned that powerful storytelling isn't about who is telling the story, but why. Why a protagonist like Elijah, why his condition, cynicism, growth — everything? Like him, I don't like my disease, but I own it, and I try to keep learning from it.

My ultimate hope is that the truths I've shared can make a difference for anyone living with a disability they can't control. Our stories aren't just grief, hilarity, candor, and suffering — they're real. And I hope they'll teach you, as they've taught me, that there's more to life. [👉](#)

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Jess Westman, 23, was diagnosed with BMD when he was 1 and created “Wheels: An Original Musical” at 19. He lives in Queens, New York, and blogs at [JessWestman.org](http://JessWestman.org).



# Two Uncles. One Mission.

*Motivated by their nephew, two Oklahoma uncles raise funds for MDA*



Talon Smith (left) and his uncle Kirby Walker, a fire captain, at a Fill the Boot event.

his pro season. The proceeds go to MDA. In 2021, the event raised \$20,000.

“Hosting this fundraiser to help my nephew has been a dream,” Matt says. “I love that I not only get to help someone close to me, but I get to help others like him. My goal is to make it possible for everyone to enjoy a week of Summer Camp. I never want a child to be turned away from this experience.”

Talon, who enjoys attending the fundraisers with his uncles every year, hopes he can pay it forward.

“Seeing my uncles fund-raise shows me that they care about not only me but also others like me,” says Talon. “They don’t have to do these events every year, but they do, and it makes me so happy to have that special connection and understanding of why they do it.” [Q](#)

### +LEARN MORE

Find fundraiser information at [FireFighters.mda.org](#) and [BassforBeckers.com](#).

**T**alon Smith, 14, of Coweta, Oklahoma, is one lucky nephew. Talon, who lives with Becker muscular dystrophy (BMD), has not one but two uncles who go above and beyond each year to raise money for MDA and his favorite MDA program, Summer Camp.

His uncle Kirby Walker, a fire captain with the Broken Arrow Fire Department in Oklahoma, has volunteered at Fill the Boot events for 23 years. When Talon was diagnosed in 2017, the fundraiser became personal to Kirby, and he hopes to help other kids enjoy a week at MDA Summer Camp. “Not only do I have a family member now affected by muscular dystrophy, but I was able to experience the happiness that camp brings to children when I attended the Fire Fighter Day at my local camp,” Kirby says.

Talon’s other uncle, Matt Looney, is a professional fisherman and started the fundraiser Bass for Beckers in 2019. The event sells raffle tickets year-round for a chance to win the boat Matt uses during



Talon (right) and his uncle Matt Looney at a Bass for Beckers event.

### CITGO Petroleum Corporation’s 37 Year Partnership



Team CITGO employees, retirees, and their families and friends, have donated countless hours of their time and talents to MDA.

### Dutch Bros is Stoked to Serve



Dutch Bros employees are always stoked to serve at local golf outings, galas, MDA Muscle Walks, MDA Summer Camps, and Fill the Boot drives.

### MDA Care Center Pediatric Nurse & Camp Volunteer



MDA Summer Camp Medical Team Volunteer, Kathryn Sawyer, RN, CPNP at Children’s Healthcare of Atlanta, has volunteered for 3 summers, and counting.

### Circle K Goes Above and Beyond for MDA



Every year, over 25 employees of Circle K South Atlantic Business Unit volunteer through two fundraising campaigns and an annual golf tournament.



### Why I Volunteer for MDA

“The ability to work with the MDA Volunteer Advisory Committee has been tremendously rewarding. Collaborating with other members of the committee has provided both a learning experience and the application of distinct backgrounds and perspectives to developing a national MDA Volunteer Program. Through our work, we know that there are thousands of individuals across the country willing and able to volunteer for the association and all the families we serve. There is a tremendous opportunity ahead to positively impact our communities.”

— Alan Cohen, MDA Volunteer Advisory Committee Chairperson

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

**Join the  CANYON Trial.**

## **The Canyon Trial**

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



## **The Investigational Therapy**

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

## **Who Can Participate?**

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

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

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

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



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