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MOVING AHEAD with

Evrysdi® (risdiplam)



1,800+

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

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

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

What is Evrysdi?

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

Important Safety Information

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

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

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

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



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

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

Important Safety Information (continued)

Tell your healthcare provider about all the medicines you take.

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

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

The most common side effects of Evrysdi include:

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

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

You may report side effects to the FDA at 1-800-FDA-1088 or www.fda.gov/medwatch. You may also report side effects to Genentech at 1-888-835-2555.

Please see accompanying brief summary for additional Important Safety Information.

If you cannot afford your Evrysdi medication, visit MySMASupport.com for financial assistance information.

Talk with your doctor about Evrysdi
or visit www.Evrysdi.com/Go to learn more



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

What is EVRYSDI?

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

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

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

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

How should I take EVRYSDI?

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

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

Taking EVRYSDI

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

Reusable Oral Syringes

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

What are the possible side effects of EVRYSDI?

The most common side effects of EVRYSDI include:

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

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

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

How should I store EVRYSDI?

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

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

General information about the safe and effective use of EVRYSDI.

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

What are the ingredients in EVRYSDI?

Active ingredient: risdiplam

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

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EVRYSDI® (risdiplam)

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



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

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The Path Forward

Our first Quest issue of 2024 is another exploration of the expansive offerings of today's MDA and our legacy of innovation and investment. The journey through this issue begins with an introduction to the visionary founder of the ReelAbilities film festival (page 13). Showcasing films created by and about individuals living with disabilities, this festival not only entertains but also contributes significantly to reshaping societal perceptions and challenging stereotypes.



Donald S. Wood, PhD

In a poignant feature that speaks to the resilience of the neuromuscular disease community, some living in our community look back to their childhoods to share what they believe everyone should know about growing up with a neuromuscular disease (page 36). Further enriching our exploration of personal narratives, Quest takes on the complexities surrounding the decision to marry while living with a neuromuscular disease (page 28). Beyond the emotional considerations, our community is faced with potential impacts on health benefits and financial implications.

Finally, we are excited to share a glimpse into the drug development pipeline, where you'll learn about some of the promising research going on in the neuromuscular and genetic medicine spaces (page 42). We also explore the nuanced landscape of limb-girdle muscular dystrophy type 2I/R9 (LGMD2I/R9) with a comprehensive profile of this condition (page 22).

As we usher in 2024, I want to wish you joy and prosperity in a new year that carries the promise of continued progress for the people and families MDA serves. From developing treatments to expanding inclusion to building community bonds across generations, we have never been more optimistic about the impact of our mission-inspired programs and services. Together, we will continue to drive progress across all facets of life with neuromuscular disease. I could not be more proud or more certain of our path forward in 2024. Thank you for being an informed and inspired member of the MDA community.

Sincerely,

Handwritten signature of Donald S. Wood

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

Do you have Becker Muscular Dystrophy? Are you interested in participating in a clinical trial? Have you considered the GRAND CANYON Trial?

The GRAND CANYON Trial

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

The Investigational Therapy

EDG-5506 is an investigational therapy in the form of a daily oral pill. EDG-5506 is designed to prevent contraction-induced muscle injury that occurs with daily activity in Becker. EDG-5506 is designed to limit this damage and help prevent the functional decline that accompanies disease progression in Becker.



Can I join the GRAND CANYON trial?

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

- Genetic diagnosis of Becker Muscular Dystrophy
- Male, ages 18-50
- Ambulatory with the ability to complete physical function activities (i.e., North Star Ambulatory Assessment, 100-meter timed test*)
- Able to meet other criteria as specified

Travel and other resources will be coordinated and provided for participants

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

Sites across the United States, United Kingdom, and the Netherlands will enroll for the GRAND CANYON trial in 2023. Sites in up to 10 additional countries are expected to open enrollment for GRAND CANYON in 2024. For more information, please go to clinicaltrials.gov (NCT05291091) or contact studies@edgewisetx.com



 **Edgewise**
THERAPEUTICS



Fulfilling Our Mission



Welcome to the first Quest Magazine issue of the year. As we embark on another exciting year of creating content for the Quest Blog, Quest Podcast, and this magazine, Quest Media’s mission is at the forefront of my mind — we exist to empower and help foster progress for the disability community.

Every piece of content we create has to pass some important tests: Does it provide valuable information about living with a neuromuscular disease or resources you can use in your life? Does it empower you to go forth and create change for our community or shine a light on progress that is happening? Does it help individuals and families understand what MDA can do for them? Does it tell a story you can see yourself in?

If we can’t answer “Yes” to at least one of those questions, we move on to the next idea.

As cheesy as it sounds, we truly believe that your success is our success. Quest Media’s mission states that we are here to help you live your best life and make the world better for individuals living with disabilities.

Thank you for looking to us to help inform and empower you.

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

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DID YOU KNOW?

QUEST IS MUCH MORE THAN A MAGAZINE! If you’re reading this page, we know you love Quest Magazine. But did you know you can find more informative, empowering content on the Quest Blog (MDAQuest.org/blog) and Quest Podcast (MDAQuest.org/podcast)?

- 2023 Quest Media content by the numbers:
- + 12 Quest Podcast episodes
 - + 159 Quest Blog posts
 - + 400 Quest Magazine articles
 - + 571 total pieces of content — you can read or listen to Quest Media every day of the year!

Find all our content at MDAQuest.org.



PHYSICAL AND OCCUPATIONAL THERAPIES HELP IN DIFFERENT WAYS.

Physical therapy (PT) and occupational therapy (OT) both help people with neuromuscular diseases maintain mobility and accomplish everyday tasks. But there are some important differences in their approaches. Depending on your goals, you might incorporate one or both into your care plan.

Learn more about how PT and OT fit in neuromuscular disease care in a Quest Magazine online exclusive at MDAQuest.org/pt-and-ot.



Getting to Know Ira Walker, MDA's Newest National Ambassador

Ira Walker is a cherished member of the MDA family, with a longstanding connection to our community. As a kid and young adult, MDA Summer Camp was the highlight of his summers and the week he looked forward to most each year. A devoted baseball enthusiast since childhood, Ira, hailing from St. Louis, has remained a devoted fan of the Cardinals. In his leisure time, you'll find Ira immersing himself in the arts, whether it's enjoying a musical, attending a symphony performance, or experiencing a great concert.

"I'm ecstatic to be a valued partner to this amazing organization and offer an energetic voice that exclaims and brings into focus the great initiatives that are active in the fight to bring a cure to those living with neuromuscular disease."

— Ira Walker, 2024 MDA National Ambassador

Read about MDA National Ambassadors Ira Walker and Leah Zelaya at mda.org/national-ambassador



IAFF & MDA: A 70 Year Partnership

Since 1954, members of the International Association of Fire Fighters have collected critical funds in the community as part of the MDA's Fill the Boot program. We are proud to have the IAFF as our largest national partner and are thankful for seven decades of commitment and tradition!



Celebrating the 20th Annual Berks Black-N-Blue Ball

Presented by The Jeremy Carroll Foundation, this long-standing event is a community favorite and raised over \$340,000 in 2023! A special honoree award was presented to the Berks County Harley Owners Group for their continued support of MDA.



Acosta hits \$100M raised through Aisles of Smiles

This 38-year partnership goes beyond corporate generosity - It's a shared journey deeply woven into the fabric of Acosta's culture. We celebrate this truly historic milestone and thank our Acosta leaders nationwide who give their time to make an impact in their local communities.





Experience Is the Best Teacher

Stephen Moore, PhD, studies neuromuscular diseases ... and lives with one

BY REBECCA HUME

Neuroscientist Stephen Moore, PhD, has always been fascinated by the brain and body. He built his career around studying neurological and neuromuscular diseases. When he was diagnosed with a rare neuromuscular disease himself, his professional pursuit became a personal passion.

Growing up, Dr. Moore's parents taught him to strive to do his best and fostered his desire to make a positive impact on the world. He studied psychology at the University of Alabama (UA) Honors College in Tuscaloosa, where he took an upper-level pathophysiology course about the brain's role in certain disorders, including neuromuscular diseases. Intrigued by the subject, Dr. Moore found his calling in neuroscience, and upon graduating in 2010, he enrolled in a doctoral program.

While taking classes in Tuscaloosa, he accepted a lab position with a research team at the University of Alabama at Birmingham (UAB) Heersink School



Stephen Moore, PhD, in the research lab.

of Medicine, about 60 miles away. It was during this busy time that Dr. Moore developed a painful rash on his upper body and began experiencing fatigue, muscle pain, and weakness. In 2012, Dr. Moore was diagnosed with dermatomyositis and took a medical leave of absence for one semester to manage his symptoms.

Resources and research

Dermatomyositis is an inflammatory disease that affects muscles and skin and is often treated with intravenous immunoglobulin (IVIG) treatment, an infusion of antibodies meant to help the immune system work better. After beginning IVIG and learning to navigate his diagnosis, Dr. Moore returned to research with an increased drive to focus on neuromuscular and autoimmune diseases.



SIMPLE SEARCH

All Quest Media's content on dermatomyositis — and other neuromuscular diseases — is at your fingertips with the Diseases A-Z library. Visit MDAQuest.org/diseases-a-z.

Dermatomyositis can cause progressive muscle loss, debilitating fatigue, and a rash on the hands and body. Wearing gloves while doing lab work made the rash on Dr. Moore's hands worse.

Dr. Moore met with the Office for Disability Services to ask for accommodations. They gave him additional time to take hand-written tests, allowing him to give his hands a break from cramping and pain. Research requires standing for long periods of time, which proved challenging with Dr. Moore's muscle fatigue. The lab provided a chair for Dr. Moore to use for breaks and access to the surgical suites after hours so that he could complete his research at his own pace.

"There are systems set up to help accommodate your needs," Dr. Moore says. "There are always going to be some barriers, but the biggest thing I've learned is to communicate my needs and ask for help."

Dr. Moore transferred to the doctoral program at UAB, where he could work and study on the same campus. In addition, UAB offers a program to retain and provide extra training to underrepresented people in the neurosciences. Living with a disability qualified Dr. Moore to join the program, which gave him more educational support and access to career coaches.

Upon finishing his doctoral program, UAB hired Dr. Moore as a full-time researcher. He is currently completing research on amyotrophic lateral sclerosis (ALS) in an MDA grant-funded lab and conducting research on inflammatory myopathies, a class of diseases that includes dermatomyositis, polymyositis, and inclusion-body myositis (IBM).

"My position is an example of using the resources and connections that I have made along the way," Dr. Moore says. "There is so much power in the connections we make."

The power of connection and community

For Dr. Moore, making connections in the disability community has proven to be immensely powerful. While earning his doctorate, Dr. Moore started an MDA Muscle Walk team, and UAB published an online story about his disease, research, and efforts



Dr. Moore (right) started an MDA Muscle Walk team.

to raise funds and awareness. Readers living with dermatomyositis who saw that story reached out to Dr. Moore. He responded by creating a Facebook group, Strength Through Knowledge, to connect individuals and families around the country and began organizing local dinners for families, caregivers, parents, and individuals to share their experiences and tools for success.

"Building that community is so important,"

Dr. Moore says. "Every single person has their own obstacle to overcome, but having another person to talk to and hear your stories reminds you that you aren't alone and provides hope."

Dr. Moore and his wife, Shaylee, share his story and their love for the Alabama outdoors through their blog, Pelican Point Expeditions ([ExplorePelicanPoint.com](https://www.explorepelicanpoint.com)).

Committed to serving his community in every way possible, Dr. Moore successfully advocated for legislation to designate May as Myositis Awareness Month in Alabama and has presented his research at the annual conference for the Society of Neuroscience in Washington, DC. In his ongoing search to find new treatments, Dr. Moore hosts patient-focused drug development panels with the FDA and helps recruit participants for a phase 3 clinical trial for a new dermatomyositis drug.

"There are so many things to be grateful and thankful for," Dr. Moore says. "And I am just trying to do the most that I can to leave a great impact." [Q](#)

Rebecca Hume is Senior Specialist and Writer for Quest Media.



VYVGART[®]
(efgartigimod alfa-fcab)

Injection for Intravenous Use
400 mg/20 mL vial

Picture your life in motion

GENERALIZED MYASTHENIA GRAVIS

*doesn't get to steal
this moment*

What is VYVGART[®] (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:

Improved daily abilities

68% (44 of 65) of participants on VYVGART achieved significant improvement in their ability to perform daily activities*

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>Important Information about VYVGART® (efgartigimod alfa-fcab); Rx only.</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 www.vyvgart.com/pi 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>What are the common side effects of VYVGART?</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>What is VYVGART?</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>• 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 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>What are the effects of VYVGART on other drugs?</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>What is the most important information I should know about VYVGART?</p> <p>VYVGART may cause serious side effects, including:</p> <ul style="list-style-type: none"> • Infection. 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 	<p>Immunization</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>What information should I know about VYVGART and pregnancy and breastfeeding?</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>Can VYVGART be used in children?</p> <p>The safety and efficacy in children (pediatric patients) have not been established.</p>



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Representation on the Big Screen

A Q&A with Isaac Zablocki, co-founder of the ReelAbilities Film Festival

BY MAGGIE CALLAHAN



Since 2007, the ReelAbilities Film Festival has been the world's largest festival dedicated to films by, about, and for people with disabilities. Founded in New York City by Isaac Zablocki and Anita Altman, the interactive festival has expanded internationally, with events held in more than 20 cities worldwide. Quest Media spoke with co-founder and director Isaac Zablocki to learn more about the festival, its ambitious accessibility efforts, and the future of disability representation in the entertainment industry.

An audience enjoys a ReelAbilities Film Festival screening in Manhattan.

Can you tell us what makes the ReelAbilities Film Festival unique?

Our film selections are really what makes the festival unique, not just because they are films about disability, but because of the types of films about disability we choose. We take a cinematic and artistic approach and prioritize showing great films that aren't being shown anywhere else. We make sure that all our films are



entertaining and of the highest quality, with the best storytelling.

We also believe film is a form of education, so we follow every film with a curated conversation to engage with the audience. I am a big believer in the communal experience of film. Having the community be a part of that conversation and meet the people who made the film humanizes the experience and makes it more relatable.

You're a graduate of Columbia University's film school, worked at Miramax Films, and have experience in many different aspects of film, including producing and directing. What inspired you to unite films and disability inclusion?

Film and disability have always been a big part of who I am. I have a variety of learning disabilities and am neurodivergent, although I didn't always admit that and possibly even rejected it. As a student, focusing in school was a problem for me, but when they put a movie on in class, suddenly I could focus. My entire life, I have been deeply impacted by film; it has always been a place I found comfort.

When I started ReelAbilities, I was really creating an extension of who I am. I felt that my job was to start a meaningful film festival that ensures everyone is seen and gives a space for voices not being heard.



Isaac Zablocki, pictured introducing a film last year, founded ReelAbilities in 2007.

REELABILITIES FESTIVALS 2024

ReelAbilities is proud to host festivals this year in the following cities:

- + Cleveland and Akron, OH
- + Columbus, OH
- + Denver
- + Houston, TX
- + Los Angeles, CA
- + Mexico City
- + New Jersey
- + New York
- + Northern Virginia
- + Pittsburgh, PA
- + Toronto, Canada

Find upcoming festival dates and stream movies at ReelAbilities.org.

And there were no voices being heard less, in my opinion, than the disability community's.

Since the start of ReelAbilities, how has disability inclusion and representation changed in the industry? And what work still needs to be done?

It's changed dramatically. We've had some more mainstream movies, such as "Crip Camp" and "Coda," that opened a lot of doors. With the emphasis on diversity, equity, and inclusion, this concept of authentic portrayals and authentic storytelling, which we've promoted for years, is more mainstream. But the playing field is still far from level.

To increase inclusion, Hollywood needs to see that working with a person with a disability doesn't mean they need to make any compromises in terms of quality and production. You can have inclusion, accessibility, authenticity, and quality. You won't have to hire a less good actor. You won't have to hire a less good camera person. You can have it all. I've seen it all.

But the hardest part is actually not about changing the industry; it's about changing the audience. Hollywood's all about making money, so they're doing what is profitable. Unfortunately, I think a lot of people still have prejudices about seeing a film that includes disability, so they need to get comfortable with it. I think it's really about breaking down stereotypes and stigmas that

exist, and telling these stories through film is a way to do that. We need to see more people with disabilities on the screen. We need to see more disability inclusion everywhere around us.

You recently launched the ReelAbilities Industry Summit for professionals. Can you tell us more about that?

For the last couple of years, during the festival we've hosted a summit on accessibility in the film world. This year, we expanded to explore accessibility in the performing arts world. The summit is for industry professionals to discuss best practices to advance accessibility, inclusion, and representation in all aspects of the film and performing arts industries.

The Mayor's Office of Media and Entertainment in New York City became a huge partner in the summit this past year. The industry has gotten very involved. Because Hollywood is much more interested in disability inclusion these days, we constantly have professionals ask us to connect them to talent or performers. It's definitely an area that is expanding and where these Summit partnerships are coming to life.

What measures do you take to make the festival accessible?

First, we show all of our films with open captions, and we create captions for films that don't come with them. We involve the films' directors and creative staff in the captioning and audio description process because it's part of the artistic process.

We also want to be geographically accessible. The first year, we were in 15 locations in New York City, and we've increased to about 40 locations in New York City. Then, when we had to be virtual for the first time during the pandemic, we learned that's even more accessible. Now, we have as much of the virtual component as possible even though we're back in person and love the in-person experience.

In-person, we make all of our spaces as accessible as possible. We go one step beyond universal design and implement what I call individual design. Universal design is there in spaces where everybody is together, but we also save space for the individual's needs.

As a philosophy, it's about listening and paying attention to what each person is asking for. For example, we could offer fidget spinners or headsets.

Sometimes, accessibility needs contradict one another. For example, one person may think having the lights on low is problematic, but another thinks it's helpful. Some people need captioning, and for others, it's distracting. We can't adjust the film to everyone's preference, but we are working to implement more adjustments on our streaming site. We're always learning and finding new ways to be more accessible.

What advice would you give to aspiring actors, screenwriters, and filmmakers living with disabilities?

The advice that I give to all young filmmakers is to show gratitude and be patient. It's a very frustrating industry to work in, especially when you have a disability. Kindness and gratitude go a long way. Also, authenticity is important — not being afraid to share your story and say your disability out loud when you're going for the job. Luckily enough — and this wasn't the case even ten years ago — we live in a world that isn't going to kick you out for having a disability, make fun of you, or bully you. It'll even be part of your calling card. People will recognize that you're telling your story a little bit differently.

When will you feel like your work here is done?

That's something I think about constantly. Because even from the beginning, the goal was to make ReelAbilities obsolete. We're seeing so much more inclusion, but it's still an uphill battle. It will be great if someday we no longer have to fight for disability rights and inclusion, but there will always be a need for a place for underrepresented film communities to come together.

One of the most beautiful things that's come out of ReelAbilities is the community that's been built. Seeing all these people come together once a year to share their experiences and make friends — it's a wonderful community. [👤](#)

Maggie Callahan is a writer and editor for Quest Media.

Progress Now

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

New approvals

FDA Approves Vamorolone for DMD

In October 2023, the US Food and Drug Administration (FDA) approved vamorolone (AGAMREE®), a unique steroidal anti-inflammatory drug, to treat individuals ages 2 and older living with Duchenne muscular dystrophy (DMD). This novel therapy, supported in part by MDA Venture Philanthropy, has efficacy similar to traditional corticosteroids with reduced negative impacts or side effects. Traditional corticosteroids, which aim to reduce inflammation, are a common therapy for DMD, but they can have many side effects, including weight gain, short stature, high blood pressure, decreased bone density (which can lead to fractures), and others. AGAMREE works similarly to

+RECRUITING CLINICAL TRIALS

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

that results in many of the common side effects. AGAMREE also may help preserve heart function in DMD patients.

The FDA's approval was based on data from the pivotal phase 2b VISION-DMD study,



Agamree works similarly to a corticosteroid by activating or inhibiting certain pathways within a cell.

Image: iStock.com/busracavus

a corticosteroid by activating or inhibiting certain pathways within a cell. For example, it inhibits the pathway linked to inflammation, and it helps stabilize the membranes around cells without causing transactivation, which is the increase of gene expression triggered by corticosteroids

supplemented with safety information from three open-label studies. In these trials, AGAMREE was administered at doses ranging from 2 to 6 mg/kg/day for up to 48 months. Compared with current standard-of-care corticosteroids, this novel corticosteroid treatment showed similar efficacy but fewer adverse events, notably related to bone health, growth trajectory, and behavior. AGAMREE is administered as an oral suspension.

The drug is expected to be available in early 2024.

For more information on the phase 2b VISION-DMD study, visit ClinicalTrials.gov, and enter NCT03439670 in the “Other terms” search box.

CLINICAL TRIAL TERMS TO KNOW

Double-blind:

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

Multicenter:

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.

FDA Approves gMG Treatment

In October 2023, the US Food and Drug Administration (FDA) approved zilucoplan (ZILBRYSQ®) for the treatment of generalized myasthenia gravis (gMG) in adult patients who are anti-acetylcholine receptor (AChR) antibody-positive (ab+). ZILBRYSQ is the first once-daily subcutaneous (under the skin) treatment for gMG, and it is the first FDA-approved gMG therapy that can be self-administered.

This is the second gMG therapy approved by the FDA in 2023. In July, the FDA approved rozanolixizumab-noli (RYSTIGGO®) to treat adults for anti-AChR and anti-MuSK ab+gMG.

MG is a rare, chronic autoimmune disease in which antibodies interfere with the communication between nerve and muscle cells, leading to muscle weakness and fatigue. In the most common subtype of gMG, antibodies target the AChR protein and trigger the activation of the complement system, which, instead of helping to eliminate disease-causing agents, attacks this important protein.

Zilucoplan, the active agent in ZILBRYSQ, is a small, artificial molecule that works to block the activation of the complement cascade. It is specifically designed to target a complement



Zilucoplan, the active agent in ZILBRYSQ®, is a small, artificial molecule that works to block the activation of the complement cascade.

protein called C5, which may reduce the damaging immune reaction in gMG.

The FDA approval of ZILBRYSQ is supported by safety and efficacy data from the RAISE study, a phase 3 multicenter, randomized, double-blind, placebo-controlled study. Participants received either daily subcutaneous injections of ZILBRYSQ or a placebo for 12 weeks. The study demonstrated that ZILBRYSQ delivered rapid, consistent, and statistically significant benefits in a broad population of adult patients with mild-to-severe anti-AChR ab+gMG. The most common adverse reactions were injection site reactions, upper respiratory tract infections, and diarrhea.

For more information about the RAISE study, visit [ClinicalTrials.gov](https://clinicaltrials.gov) and enter NCT04115293 in the “Other terms” search box.

Becker muscular dystrophy (BMD)

Phase 2 study accepting more participants

Edgewise Pharmaceuticals is expanding its phase 2 clinical trial testing the investigational oral medication EDG-5506 in people with BMD. The expansion trial, named GRAND CANYON, will enroll 120 additional adults aged 18-50 years old at up to 50 sites in 10 countries. The first cohort of the study, named CANYON, included more participants than originally planned, with a total of 39 adults and 24 adolescents.

The expansion trial, named GRAND CANYON, will enroll 120 additional adults aged 18-50 years old at up to 50 sites in 10 countries.

EDG-5506, taken orally, is designed to protect susceptible muscle fibers from the damage that occurs as a result of everyday activities in people living with BMD.

The decision to expand the trial was based on the positive results of the phase 1 clinical trial ARCH, which showed that most men with BMD who were treated with EDG-5506 had stable or improved motor function and fewer markers of muscle damage after receiving the therapy.

GRAND CANYON is a multicenter, randomized, double-blind, placebo-controlled study to evaluate the safety and efficacy of EDG-5506 in adults with BMD. The primary endpoint of GRAND CANYON is the North Star Ambulatory Assessment

(NSAA). In addition, other functional measures, biomarkers of muscle damage, and safety will be assessed. The treatment period will be 18 months.

To be eligible, individuals must be able to walk at least 100 meters with or without an assistive device in less than 200 seconds.

The trial is expected to be pivotal, meaning it should produce the important data needed for approval by the Food and Drug Administration (FDA). This would be the first approved therapy for individuals with BMD.

To learn more about the GRAND CANYON study, visit ClinicalTrials.gov and enter NCT05291091 in the “Other terms” search box.

Becker muscular dystrophy (BMD) and Duchenne muscular dystrophy (DMD)

Children Needed for Observational Study

Researchers at the University of Delaware are seeking boys living with BMD or DMD to participate in a study to determine the effects of BMD and DMD on the function of blood vessels in the arms, legs, neck, and heart. The study aims to see if changes in blood vessel function can be used to predict heart problems or disease in males with BMD or DMD.

This study uses online surveys and a one-time in-person data collection and laboratory testing session to look at blood vessel and heart function. The in-person testing may include taking a blood sample and various heart function assessments, including an echocardiogram.

Participation in the study will require two study visits; the first visit may occur online, and the second visit will be in person. Follow-up visits at 12 and 24 months are optional.

To be eligible, individuals must have a diagnosis of BMD or DMD by a pediatric neurologist, be 7-21 years old, and meet additional criteria.



This study aims to see if changes in blood vessel function can be used to predict heart problems or disease in males with BMD or DMD.

Travel support and compensation for time and effort are available for study participants.

To learn more and inquire about participation, contact study coordinator Krista Szymanski at WitmanResearchLab@udel.edu.



Image: iStock.com/Boba

Myasthenia gravis (MG)

Study Seeks Pediatric Participants

Researchers at Janssen Research & Development LLC are seeking pediatric patients ages 2 to 17 who are experiencing symptoms of generalized myasthenia gravis (gMG) despite receiving stable, standard-of-care therapy to participate in a phase 2/3 clinical study. The study, called Vibrance MG, will evaluate the safety and efficacy of an investigational medication, nipocalimab, and determine how long it stays in and acts on the body. The drug is expected to help the body get rid of autoantibodies that cause tissue damage in people with MG.

This is an open-label study, which means that all participants will receive the investigational medication. The study will consist of a screening period of up to four weeks followed by a 24-week active study treatment phase. Participation will require approximately 15 clinic visits during the active study treatment phase, with a total duration of approximately 36 weeks. After completion of the active study treatment phase, all participants will have the option to enroll in an optional long-term extension (LTE) phase. Participants who do not enroll in the LTE phase will be required to complete a safety follow-up visit eight weeks after their last

treatment of the study medication.

The investigational medication will be given by intravenous infusion. The effects of the investigational medication will be evaluated using a number of tests and procedures, including physical exams, measurement of vital signs, urine samples, blood draws, and questionnaires to assess how effective the study medication may be in participants.

Support for study-related travel costs is available for participants.

Nipocalimab is expected to help the body get rid of autoantibodies that cause tissue damage in people with MG.

For more information and a full listing of inclusion and exclusion criteria, visit [ClinicalTrials.gov](https://clinicaltrials.gov) and enter NCT05265273 in the “Other terms” search box. To inquire about participation, email the study coordinator at participate-in-this-study@its.jnj.com.

Myotonic dystrophy (DM)

FDA Allows DM1 Study to Start

The US Food and Drug Administration (FDA) lifted a clinical hold it placed on PGN-EDODM1, a drug being developed by PepGen for myotonic dystrophy type 1 (DM1).

A clinical hold is an order issued by the FDA to delay a proposed clinical study or to suspend an ongoing investigation. With the hold lifted, PepGen may begin its phase 1 study, called FREEDOM-DM1, to evaluate PGN-EDODM1 for the treatment of DM1 in the

United States. (The study was already cleared to start in Canada.)

DM1 occurs when a gene on chromosome 19 called DMPK contains a section

where the DNA building blocks are repeated multiple times. This affects cell processes and results in abnormal production of DMPK protein, which is involved in muscle contraction and relaxation. PGN-EDODM1 contains a small molecule called an antisense oligonucleotide that binds to the repeats. This is expected to restore the production of DMPK protein and allow muscles to function normally again.

FREEDOM-DM1 is a randomized, double-blind, placebo-controlled, single-ascending dose study designed to test the safety and efficacy of PGN-EDODM1 at different doses in adults. Participants will be randomly assigned to either a placebo or a single dose of PGN-EDODM1 at 5 mg/kg, 10 mg/kg, or 20 mg/kg.

Earlier preclinical work showed that PGN-EDODM1 was well-tolerated by rodents and non-human primates when given at clinically relevant doses. In mice given a single dose of PGN-EDODM1, there was sustained easing of myotonia, which occurs when your muscles aren't able to relax after they contract.

PGN-EDODM1 contains a small molecule called an antisense oligonucleotide that binds to the abnormal DNA repeats that cause DM1.

To learn more about PepGen, visit [PepGen.com](https://www.pepgen.com).



Image: iStock.com/Boba

Participants will be asked to provide a blood sample during an at-home visit by a mobile phlebotomist.

Spinal muscular atrophy (SMA)

At-Home Research Study Enrolling

Sanguine Biosciences is seeking people living with SMA or carrying SMA-causing mutations to participate in a preclinical study. The goal of this research is to better understand SMA to support the development of new diagnostic and treatment options.

This study is observational and does not test a new intervention or drug. Participants will be asked to provide a blood sample during a one-time at-home visit by a mobile phlebotomist.

To be eligible, individuals must:

- Be diagnosed with SMA or be an SMA-mutation carrier
- Be age 18-85 years old
- Live in the United States

Compensation for time and effort is available for study participants.

To learn more or sign up, visit Patients.SanguineBio.com/sma or call 818-696-4281 to speak with a research coordinator.



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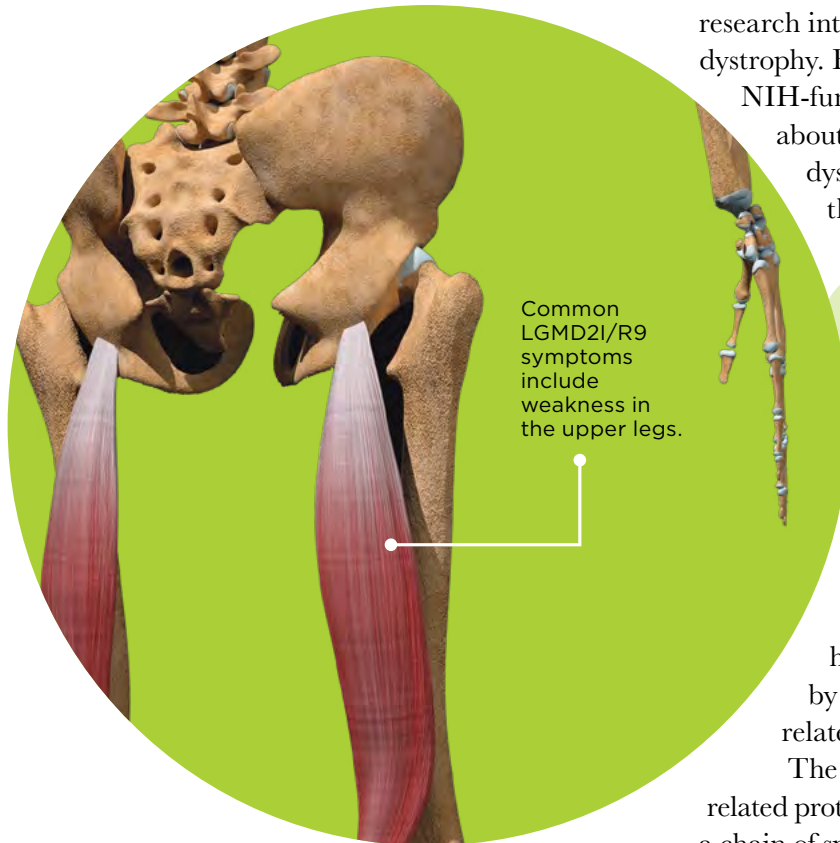




Updates on LGMD2I/R9

A Q&A with Katherine Mathews, MD

BY MYRNA TRAYLOR



Common LGMD2I/R9 symptoms include weakness in the upper legs.

Limb-girdle muscular dystrophies (LGMD) include a diverse group of disorders with many subtypes. LGMD usually causes weakness in muscles around the hips and shoulders. The subtype LGMD2I/R9 (also called LGMD2I or LGMDR9) is the subject of some promising research.

To better understand LGMD2I/R9, we spoke with Katherine Mathews, MD, professor of pediatrics and neurology at the University of Iowa Carver College of Medicine. Dr. Mathews began her career as a pediatric neurologist and pursued a fellowship in genetics, focusing her early work on facioscapulo-humeral muscular dystrophy (FSHD), for which she received funding from MDA. She later expanded her

research interests to include other types of muscular dystrophy. For nearly 20 years, she has directed an NIH-funded natural history study to learn more about LGMD2I/R9 and related muscular dystrophies (dystroglycanopathies) as part of the University of Iowa's Wellstone Muscular Dystrophy Specialized Research Center.

What is LGMD2I/R9?

LGMD2I/R9 is a type of limb-girdle muscular dystrophy that is inherited in an autosomal recessive pattern. Autosomal recessive means both copies of a gene inherited from both parents have a variation (mutation) that leads to disease. People with only one abnormal copy of the gene (from either parent) are healthy (carriers). LGMD2I/R9 is caused by two abnormal copies of the fukutin-related protein (*FKRP*) gene.

The *FKRP* gene is the “recipe” for fukutin-related protein, one of several enzymes required to put a chain of special sugars (called matriglycan) onto the protein alpha-dystroglycan. These sugar molecules help to bind the muscle membrane to a fibrous layer (extracellular matrix, or ECM) just outside the cell, providing stability to the muscle cell membrane. If fukutin-related protein isn't working correctly, this connection to the extracellular matrix is weakened, and the muscle membrane is prone to breakdown. Ultimately, the muscle cell can die and be replaced by fat and scar tissue.

When is it typically diagnosed?

Like all forms of LGMD, there's a wide age range in the onset and rate of progression. It can be diagnosed in young childhood or as late as middle adulthood. The diagnosis is suggested by weakness and often calf hypertrophy (enlargement). The muscle enzyme

test, which measures creatine kinase (CK) levels in the blood, typically shows marked elevation. The diagnosis is confirmed by genetic testing.

What are the early signs and symptoms of LGMD2I/R9?

The most common symptom is muscle weakness or fatigue involving the hips, upper legs, or shoulders, typically first noticed during everyday activities like running or climbing stairs. Children with LGMD2I/R9 may present with intermittent severe weakness associated with a viral illness. They typically recover normal strength after they recover from the viral illness, but persisting laboratory abnormalities are a clue that they have muscular dystrophy.

People with a milder form of the disease may notice after vigorous exercise that their urine turns brown or tea-colored due to the breakdown of muscle cells, which releases iron-containing myoglobin that is excreted in the urine. This can be the first evidence of LGMD2I/R9. Another way people are diagnosed is through abnormal “liver enzyme” tests (such as AST and ALT) identified through blood work. Although they are called “liver tests,” these enzymes are also released from diseased muscle, so they are elevated in people with LGMD2I/R9.

What is the progression of the disease, and how is it managed?

LGMD2I/R9 results in progressive muscle weakness that worsens over time. The rate of progression depends on the person’s genetics, the type of *FKRP* gene mutation, and probably genetic background. Some *FKRP* gene mutations are known to be associated with less rapid progression.

An important part of management is monitoring the heart, as the heart muscle is also affected by abnormal fukutin-related protein activity. Heart monitoring is done by doing an echocardiogram (ECG), cardiac magnetic resonance imaging (MRI), or a combination of these tests every one to three years, depending on the individual situation, as guided by one’s neuromuscular doctor or cardiologist.

The muscles used for breathing can also be affected, so that’s another thing that needs to be monitored, using breathing tests (pulmonary function testing) or

sleep studies. Some people need assistance with generating an effective cough or need breathing support at night.

Some people will need a wheelchair as early as their teens, while others won’t need a wheelchair until late adulthood.



Katherine Mathews, MD

Are there any treatments approved or being studied for LGMD2I/R9?

There are medicines to treat heart dysfunction, but there is not yet an approved medication specifically for LGMD2I/R9.

There are several clinical trials either in progress or in preparation. They revolve around two major approaches to therapy. One possible approach is gene replacement. The healthy gene can be put into a delivery system (currently through modified viruses). The modified virus carrying the *FKRP* gene is injected into the bloodstream. The new *FKRP* gene can turn on in the muscle. There are still questions about this approach. We don’t know the best “dose” of *FKRP* gene, as too much could be harmful and too little might not be effective. The infusion of the large number of virus particles used for gene delivery also has risks, including dysfunction of the heart, liver, clotting system, and kidneys.

Another approach being studied to treat LGMD2I/R9 is to try to make the existing fukutin-related protein work harder. We know that everybody who has LGMD2I/R9 has some fukutin-related protein activity because a complete absence is incompatible with life. Therefore, introducing a lot of one of the building blocks for the matriglycan sugar chain into the body might increase the amount of matriglycan on alpha-dystroglycan by driving fukutin-related protein to do more of what it normally does. This is predicted to increase its matriglycan binding to the ECM, making the muscle membrane less prone to damage and thus slowing the disease progression. [Q](#)

Myrna Traylor is a writer for Quest Media.



How We Support You

There may be more ways to engage with MDA than you realize

BY JASON HENNINGER

Throughout our more than 70-year history, engaging with individuals and families affected by neuromuscular disease and building a sense of community have been at the heart of MDA's mission.

As MDA has continued to grow and evolve along with our community, the ways we support our families have grown. Here are some of the ways you can engage with and receive support from MDA.

MDA Resource Center

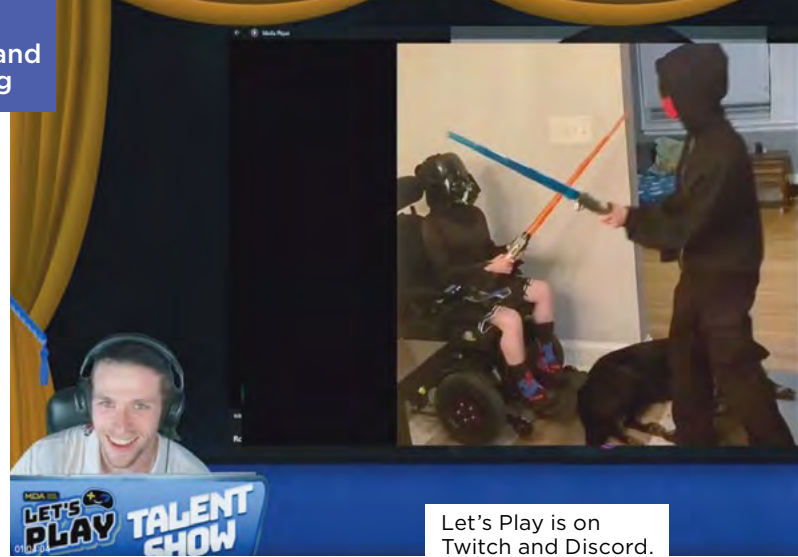
This is the starting place for anyone who has a new diagnosis, is new to MDA, or is looking for support at any point in their journey. Resource Center Specialists will walk you through how to become an MDA member and describe our programs and resources. They can also help you find information on your disease, get started with an MDA Care Center, learn about research and clinical trial opportunities, and direct you to resources and services in your community.

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

MDA Advocacy

The Advocacy program strives to ensure that legislators and others in decision-making positions craft public policies that empower the neuromuscular disease and disability communities.

“We always need more grassroots advocacy volunteers to help us achieve our public policy goals,” says Mark Fisher, MDA’s Director of Advocacy Engagement. “Our volunteers can engage in meaningful activities, including emailing or calling lawmakers, meeting with members of Congress, attending training, writing blog posts, and much more.”



Learn about our latest initiatives and sign up to be a grassroots advocate at mda.org/advocacy.

MDA Ambassador Program

“Ambassadors serve as spokespeople, helping to communicate the MDA mission,” says Alicia Dobosz, MDA’s Vice President of Community Engagement.

Both kids and adults serve as MDA Ambassadors, which involves raising awareness about neuromuscular diseases by sharing their compelling stories at events, online, and through Quest Media and other media outlets. Ambassadors help others understand the impact of neuromuscular diseases while gaining valuable public speaking experience and building connections in the MDA community.

Learn about the current MDA Ambassadors and how to apply for the program at mda.org/ambassador.

MDA Care Center Network

MDA supports a national network of Care Centers that offer specialized, multidisciplinary care for neuromuscular diseases and are at the forefront of research on these diseases. MDA Care Center Network healthcare providers are skilled in the diagnosis and medical management of neuromuscular diseases and keep up-to-date on the latest studies and clinical trials.

The MDA Care Center Network is located in more than 150 of the top health institutions throughout the United States. Find a Care Center near you at mda.org/CareCenters.

MDA Community Education

MDA designs educational programs and materials that empower the neuromuscular disease community

with knowledge and resources. They range from disease-specific in-person seminars to online workshops and webinars covering daily living, social-emotional well-being, and caregiving to printable fact sheets and guides.

“Our Community Education team’s goal is to help individuals and families in the community overcome barriers to access, navigate life’s transitions, and make informed decisions about care,” Alicia says.

Find the full lineup of Community Education offerings at mda.org/community-ed.

MDA Connect

This service offers 30-minute one-on-one video chats with MDA Specialists to introduce individuals with neuromuscular diseases, as well as their families and caregivers, to the broad network of support opportunities MDA offers. These sessions are great for those who are new to MDA, as well as people looking for more resources and support as their disease progresses.

To learn how to schedule a one-on-one session, visit mda.org/connect.

MDA Gene Therapy Support Network

Several gene therapies have been approved to treat neuromuscular diseases, and many more are being studied. (Turn to page 42 to read about the drug development pipeline.) As more gene therapies become available, MDA recognizes the importance of educating our community about the advances and opportunities in the world of gene therapy.

The MDA Gene Therapy Support Network connects people with educational resources and offers video calls with MDA Gene Therapy Support Specialists, who can answer questions about the latest gene therapy news. Visit mda.org/GeneTherapySupport.

MDA Let’s Play

This online community brings 2,500 MDA community members of all ages together in a fun, supportive environment. You can join MDA Let’s Play on Twitch and Discord to play and watch livestreams from other gamers. Organized events include game nights, movie nights, and trivia.

It’s a safe and friendly way to play, learn, and make friends. Learn more at mda.org/lets-play.

ALL ABOUT VOLUNTEERING



MDA builds community among people affected by neuromuscular diseases and those who support them through a variety of programs and events, such as Muscle Walks, golf tournaments, and galas, not to mention MDA Summer Camp, called “the best week of the year.” None of this would be possible without the thousands of generous volunteers who give their time to MDA.

“There are so many ways for folks to get involved with MDA,” says Wendi Dressen, MDA’s Senior Director of Volunteer Programs. “This can include volunteering for a remote or in-person event, supporting a fundraiser, participating in a Muscle Walk, running with Team Momentum, writing a letter to your congressperson, or posting MDA information on your social media.”

Every act of service and every hour of volunteered time helps MDA serve individuals and families living with neuromuscular disease and change lives.

Learn more at mda.org/volunteer.

MDA Summer Camp

Kids and young adults with neuromuscular diseases can attend MDA’s week-long overnight camps around the United States — at no cost to their families. Every year, hundreds of volunteers provide support as camp counselors and medical staff to ensure the campers have an unforgettable summer making friends and trying new things.

MDA Summer Camp offers a fun and safe outdoor experience with activities such as horseback riding, swimming, adaptive sports, arts and crafts, camp dances, and more. They also give parents respite while trained volunteers provide campers with physical and emotional support.

“Summer Camp is a place of complete inclusion, empowerment, exploration, and growth,” Alicia says. For many campers, it’s a life-changing experience. One former camper says: “Camp really gave me a safe place to learn and explore myself. In return, I became more confident in who I am as an adult.”

Learn more about becoming a camper or volunteer at mda.org/SummerCamp.

Jason Henninger is a writer for Quest Media.

Becker Education & Engagement Day 2023

BEED Sponsors



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It's time to
get real
about **Becker**
muscular
dystrophy

A new chapter for the Becker muscular dystrophy community

began on December 2nd, when over 150 people whose lives are impacted by Becker gathered in person at four different locations across the US for the inaugural Becker Education and Engagement Day, or BEED 2023. Held simultaneously in California, Colorado, Florida, and Pennsylvania, BEED 2023 was designed to foster a sense of community for people living with Becker and their families, by providing opportunities to hear from experts in Becker care and research, and most importantly, to meet others affected by the rare neuromuscular disorder.

“ We lump diseases together sometimes, but Becker has unique needs compared to Duchenne. Regarding BEED, I want to shy away from the word “day” and rephrase it to “Becker Education and Engagement Today and Forever. ”

– Sue Apkon, MD
MD Children's Colorado

A highly collaborative group of patient advocates, clinical care providers, researchers, and biotech companies organized

and supported the unique event that provided a mix of in-person talks and national presentations from leading care experts and researchers in Becker. The day was a resounding success, with one attendee saying, “I expected it to be interesting to meet others with Becker, but it was more than that, it was an incredible experience.”

Attendees at BEED 2023 heard from leading experts on care considerations for Becker. In a presentation about the current standard of care for Becker, the importance of a multi-disciplinary approach to monitoring disease progression and treating the effects of the disease was shared. An overview of how Becker affects heart and lung health and a review of interventions recommended to preserve cardiac function were discussed. The importance of appropriate exercise and exercise recommendations for people living with Becker was outlined. Attendees noted the importance of hearing about how these topics were unique for

ADVERTISEMENT

individuals living with Becker. One individual shared, "I work with a physical therapist who is not too familiar with my disease, so I wanted to hear from experts to confirm the treatment I am getting is appropriate."

" So often I have felt alone with this disease and for the first time I have an overwhelming sense of relief that I have others to relate to and who will all support each other. The impact of this event is greater than you know. "

- Individual living with Becker,
BEED attendee

In one of many highlights of the day, the need for a Becker Resource Toolkit was discussed.

The presentation was packed full of ideas and information about what local, state, and federal resources might be available to address financial and quality of life challenges that often come with a Becker diagnosis. Those impacted by Becker were encouraged to get involved in advocacy at the state and federal level on issues related to therapy development and access to appropriate healthcare and services. Later in the day, an overview of the research landscape for Becker highlighted the importance of research, including natural history studies, in establishing Becker clinical care standards, as well as the development of potential disease-modifying therapies. This session concluded with a live Q and A panel about the ongoing studies enrolling individuals with Becker, both natural history studies and studies with investigational treatments.

Most importantly, BEED 2023 provided an opportunity for those impacted by Becker to meet face-to-face for discussions about common experiences and challenges. Time was built into the agenda at each location to allow for open conversations led by Becker community members in groups made up of affected individuals of all ages. "For the first time in my life, I truly felt like I was not alone living with BMD," said one attendee. In a post-event survey, the need for the BEED event was

unanimous, with comments including: "BEED is what the community needed! The first Becker-only session of its kind" and "Finally some TRUE TRUE HOPE for the orphaned Becker community... thank you BEED!!"

" BEED exceeded my wildest expectations. It truly was an example of a pre-competitive collaboration where multi-disciplinary stakeholders united around serving and lifting up the Becker community, which has been underserved for so long. This was truly one of my most rewarding projects in my career in advocacy. "

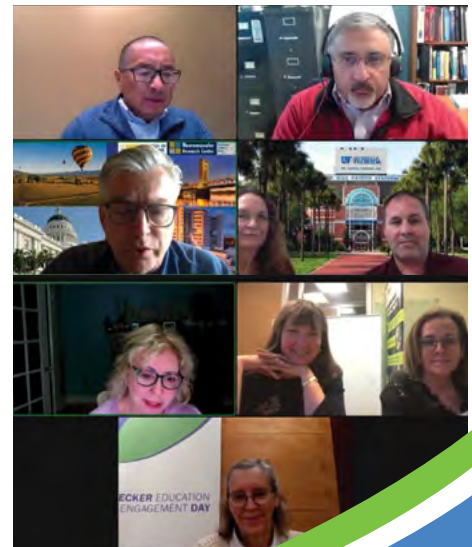
- Abby Bronson
Vice President of Advocacy and External Innovation
at Edgewise Therapeutics



MDA Chief Medical Advisor Barry Byrne kicked off the day at the University of Florida site of BEED 2023

BECKER EDUCATION & ENGAGEMENT DAY • 2023

Zoom panelists for Care Considerations & Clinical Trial Landscape sessions



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For people with disabilities, getting hitched may mean losing vital benefits

After dating for six years, Gabriella Garbero's boyfriend, Juan Johnson, popped the question on New Year's Eve in 2021. She enthusiastically accepted his proposal.

Three years later, the couple still has not tied the knot. Gabriella, an attorney and disability advocate, has spinal muscular atrophy (SMA) and relies on aides and nurses to provide care in her home. Gabriella's and Juan's combined income and assets exceed Medicaid's allowable limits. If they were to marry, Gabriella would lose the Medicaid benefits that help keep her alive. She estimates her in-home care would cost between \$100,000 to \$200,000 per year if she were paying for these services herself.

Gabriella's Medicaid eligibility is tied to qualifying for Supplemental Security Income (SSI). This needs-based government program provides a modest monthly income to people with disabilities. For 2024, the maximum monthly payment is \$943 for an individual.



GE TY

BY BARBARA TWARDOWSKI
AND JIM TWAWRDOWSKI, RN



“We might be engaged for 10 or 20 years and never get married, although I hope not. I hope something changes before then.”
 — Gabriella Garbero

Gabriella Garbero and Juan Johnson



SSI also stipulates that a recipient’s countable assets (a bank account, financial investments, or property) must not exceed \$2,000. For a married couple, SSI counts their combined income and assets regardless of the disability status of the spouse. They may not exceed a total of \$3,000.

Forced to choose between having the daily care Gabriella needs or getting married, the couple has postponed their wedding indefinitely. “We might be



LOVE & MARRIAGE

Listen to a Quest Podcast conversation with couples about the joys and challenges of marriage with a disability at MDAQuest.org/podcast/love-marriage.

engaged for 10 or 20 years and never get married, although I hope not. I hope something changes before then,” Gabriella says.

Marriage inequality

Culturally, Americans have seen inequalities in marriage rights before. We have seen the laws change to allow interracial marriages and, more recently, same-sex marriages.

Marriage equality for the disability community is somewhat different. While disabled people are not prohibited from marrying, the rules to qualify for government benefits penalize those who do marry.

More than 7.6 million Americans who receive SSI benefits are impacted by rules regarding marital status.

Even couples that are not legally married risk facing penalties. If the Social Security Administration, which administers SSI, determines that two people are presenting themselves as married to the community, their SSI benefits are calculated as though they are a legally married couple. Something as simple as how you introduce your significant other can have devastating consequences.

The high cost of marriage

Before Ashleigh and James Ocasio married, they spent a long time discussing the potential ramifications of their union.

Ashleigh has limb-girdle muscular dystrophy (LGMD).

“I need help with everything: getting dressed, doing my hair, brushing my teeth, toileting, and showering. Sometimes, even assistance with eating,” Ashleigh says. Her home healthcare services provided by four professional caregivers cost more than \$1,000 per week. Medicaid and her state’s waiver program pay for this, as well as additional medical expenses.

To keep Ashleigh’s benefits, the couple needed to earn less. James took a lower-paying job and became Ashleigh’s primary caregiver. Due to a shortage of home health workers in their area, Ashleigh now has only one outside caregiver.

“Except for one person who comes in once a week to help, the caregiving is all down to my husband now,” Ashleigh says. This situation takes a toll on their marriage. “Sometimes it’s hard to separate caregiving from the romantic spousal relationship. So, we’ve had to work through that. We go to therapy to help us with managing that relationship, and we’re only a year in.”

More than 7.6 million Americans who receive SSI benefits are impacted by rules regarding marital status.

AN ISSUE OF ECONOMIC INDEPENDENCE

The Supplemental Security Income (SSI) program was established 50 years ago, and the asset limits haven’t been updated since the 1980s. This doesn’t just create barriers to marriage for people with disabilities — it also affects their ability to work, get promoted, and save for the future.

The SSI Penalty Elimination Act, introduced in the US Senate in 2023, would go a long way toward providing economic independence for the disability community.

You can help by contacting your Congress members and telling them how the SSI Penalty Elimination Act would impact you or your loved ones. To act now, scan the QR code and fill out MDA’s easy-to-use online form.



Image: iStock.com/BrianAJackson



Ashleigh and James Ocasio

“If we got legally married or just continued dating while living together ... either way, it was going to count against us.”
— Ashleigh Ocasio

Photo: Stephanie Schneider/Touch of Love Photography

To marry or not?

The rules surrounding eligibility for Medicaid, SSI, and other benefit programs are complex.

Medicaid is a joint federal-state program, and the eligibility and benefits vary from one state to another. Some states use the same eligibility rules and application process for SSI and Medicaid, while other states have different eligibility rules and/or require a separate application for Medicaid. To learn about the rules in your state, contact your state Medicaid agency.

SSI eligibility, payments, and asset limits are determined at the federal level by the Social Security Administration. Currently, when two people who rely on SSI get married, their monthly income benefit

is reduced by 25%. An individual receives \$943 each month, but two people receiving SSI who are married do not receive double that, which would be \$1,886. Instead, their monthly benefit is \$1,415. That’s almost \$500 per month less.

In addition, the allowable assets threshold is \$2,000 for an individual on SSI and \$3,000 for a married couple, even if only one member of the couple qualifies for SSI. This is yet another 25% reduction.

Ashleigh and James, who is deaf, both receive SSI and Medicaid benefits. Since getting married in 2022, they have been living paycheck to paycheck on a tight budget. They do not have a retirement fund or a savings account for emergency expenses.

In her work as a Family and Clinical Support Specialist for MDA, Ashleigh frequently talks to clients who had no idea that getting married would alter their benefits until after they did so. She also talks to people who are contemplating never getting married or divorcing because they need their benefits.

“I don’t regret getting married, especially because of where I live,” says Ashleigh. Iowa, her home state, recognizes common-law marriage. “It didn’t matter if we got legally married or just continued dating while living together because either way, it was going to count against us. At least I get to have the semblance of a normal marriage.”

Marriage equality movement

Many people with disabilities, including those with neuromuscular diseases, rely on SSI, and in most cases, they are eligible for Medicaid. The marriage penalties baked into Medicaid and SSI benefits have a profound effect on the disability community, according to Ayesha Lewis, an attorney with the Disability Rights Education and Defense Fund — and it’s not just economic.



“They treat people as if they’re not able to live the kind of full lives that we know people with disabilities are capable of and live every day.”

— Ayesha Lewis

“These barriers to marriage are patronizing,” Ayesha says. “They treat people as if they’re not able to live the kind of full lives that we know people with disabilities are capable of and live every day. I’ve spoken to a lot of folks across the country, and they see this as an affront to their dignity, humanity, and equality.”

Fortunately, lawmakers are beginning to see that point of view. In 2023, Senators Bill Cassidy (R-LA) and Sherrod Brown (D-OH) introduced the SSI Savings Penalty Elimination Act, bipartisan legislation that changes the existing



A commitment ceremony protesting marriage penalties was held in Washington, DC.

SSI asset limits. If passed, this bill would increase asset limits to \$10,000 for individuals and \$20,000 for couples, with adjustments for inflation each year. (Visit mda.org/advocacy and click on “Take Action” to learn more about MDA’s work on SSI benefits.)

Awareness of the marriage equality issue continues to grow. In September 2023, numerous media outlets covered a mass commitment ceremony held on the National Mall in Washington, DC, where couples with disabilities recited non-legally binding vows under banners declaring, “End the disabled marriage penalties.” Many attendees said they would get married if SSI’s policies changed.

“The engagement around this issue is heartwarming,” Ayesha says. “We value equality. We value love. We want people to be able to live the lives they want regardless of their disability status.” [📺](https://www.youtube.com/watch?v=...)

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

+MAKE YOUR VOICE HEARD

Join MDA’s Grassroots Advocacy Network to add your voice to any of MDA’s initiatives that promote access to care, champion disability rights, and accelerate drug development. Sign up at mda.org/advocacy.



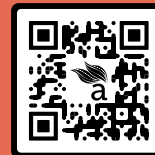
Tough on Duchenne. So it's easier to be him.

Give him the strength to be a kid.

AGAMREE Is a Novel Corticosteroid

- Developed to uncouple anti-inflammatory effects and certain corticosteroid-mediated adverse effects^{1,2}
- Demonstrated statistically significant improvements in motor function^{3,4}
- Established safety and tolerability profile in clinical studies^{3,4}

LEARN MORE
AT AGAMREE.COM



AGAMREE is FDA approved for the treatment of Duchenne muscular dystrophy (DMD) in patients 2 years of age and older³

SELECT IMPORTANT SAFETY INFORMATION

Warnings & Precautions

- **Alterations in Endocrine Function:** Monitor patients receiving AGAMREE for Cushing's syndrome, hyperglycemia, and adrenal insufficiency after AGAMREE withdrawal. In addition, patients with hypopituitarism, primary adrenal insufficiency or congenital adrenal hyperplasia, altered thyroid function, or pheochromocytoma may be at increased risk for adverse endocrine events. Acute adrenal insufficiency can occur if AGAMREE is withdrawn abruptly, and could be fatal.
- **Immunosuppression and Increased Risk of Infection:** Use of corticosteroids, including AGAMREE, increases the risk of new infection, exacerbation of existing infections, dissemination, and reactivation or exacerbation of latent infection and may mask some signs of infection; these infections can be severe, and at times fatal.
- **Alterations in Cardiovascular/Renal Function:** Monitor for elevated blood pressure and monitor sodium and potassium levels in patients chronically treated with AGAMREE.
- **Gastrointestinal Perforation:** Use of corticosteroids increases the risk of gastrointestinal perforation in patients with certain gastrointestinal disorders, such as active or latent peptic ulcers, diverticulitis, fresh intestinal anastomoses, and non-specific ulcerative colitis. Signs and symptoms may be masked.
- **Behavioral and Mood Disturbances:** Potentially severe psychiatric adverse reactions may occur with systemic corticosteroids, including AGAMREE, and may include hypomanic or manic symptoms (eg, euphoria, insomnia, mood swings) during treatment and depressive episodes after discontinuation of treatment.
- **Effects on Bones:** Prolonged use of corticosteroids, such as AGAMREE, can lead to osteoporosis, which can predispose patients to vertebral and long bone fractures. Monitor bone mineral density in patients on long-term treatment with AGAMREE.
- **Ophthalmic Effects:** The use of corticosteroids, such as AGAMREE, may increase the risk of cataracts, ocular infections, and glaucoma. Monitor intraocular pressure if treatment with AGAMREE is continued for more than 6 weeks.
- **Vaccination:** Do not administer live-attenuated or live vaccines to patients receiving AGAMREE. Administer live-attenuated or live vaccines at least 4 to 6 weeks prior to starting AGAMREE.

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

References

1. Liu X, et al. *Proc Natl Acad Sci USA*. 2020;117(39):24285-24293. 2. Heier CR, et al. *EMBO Mol Med*. 2013;5(10):1569-1585. 3. AGAMREE (vamorolone) Oral Suspension [prescribing information]. Catalyst Pharmaceuticals, Inc.; 2023. 4. Guglieri M, et al. *JAMA Neurol*. 2022;79(10):1005-1014.



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AGAMREE® (vamorolone) oral suspension

BRIEF SUMMARY – See Full Prescribing Information at AGAMREEhcp.com

Initial U.S. Approval: 2023

INDICATIONS AND USAGE

AGAMREE is indicated for the treatment of Duchenne muscular dystrophy (DMD) in patients 2 years of age and older.

DOSAGE AND ADMINISTRATION Dosing Information

The recommended dosage of AGAMREE is 6 mg/kg taken orally once daily preferably with a meal, up to a maximum daily dosage of 300 mg for patients weighing more than 50 kg.

Some patients may respond to a dose of 2 mg/kg daily. Doses may be titrated down to 2 mg/kg/day as needed, based on individual tolerability.

Discontinuation

Dosage of AGAMREE must be decreased gradually if the drug has been administered for more than one week.

CONTRAINDICATIONS

AGAMREE is contraindicated in patients with known hypersensitivity to vamorolone or to any of the inactive ingredients of AGAMREE. Instances of hypersensitivity, including anaphylaxis, have occurred in patients receiving corticosteroid therapy.

WARNINGS AND PRECAUTIONS

Alterations in Endocrine Function

Corticosteroids, such as AGAMREE, can cause serious and life-threatening alterations in endocrine function, especially with chronic use. Monitor patients receiving AGAMREE for Cushing's syndrome, hyperglycemia, and adrenal insufficiency after AGAMREE withdrawal. In addition, patients with hypopituitarism, primary adrenal insufficiency or congenital adrenal hyperplasia, altered thyroid function, or pheochromocytoma may be at increased risk for adverse endocrine events.

Acute adrenal insufficiency can occur if AGAMREE is withdrawn abruptly, and could be fatal. The risk of adrenal insufficiency is reduced by gradually tapering the dose when withdrawing treatment. For patients already taking corticosteroids during times of stress, the dosage may need to be increased.

Immunosuppression and Increased Risk of Infection

Corticosteroids, including AGAMREE, suppress the immune system and increase the risk of infection with any pathogen, including viral, bacterial, fungal, protozoan, or helminthic pathogens. Corticosteroids can reduce resistance to new infections, exacerbate existing infections, increase the risk of disseminated infections, increase the risk of reactivation or exacerbation of latent infections, and mask some signs of infection. Corticosteroid-associated infections can be severe, and at times fatal infections can occur. The rate of infectious complications increases with increasing corticosteroid dosages.

Corticosteroids cause a dose-dependent increase in lymphocyte and neutrophil counts. Monitor for the development of infection and consider AGAMREE withdrawal or dosage reduction as needed.

Varicella Zoster and Measles Viral Infections

Varicella and measles can have a serious or even fatal course in non-immune patients taking corticosteroids. In corticosteroid-treated patients who have not had these diseases or are non-immune, particular care should be taken to avoid exposure to varicella and measles.

- If an AGAMREE-treated patient is exposed to varicella, prophylaxis with varicella zoster immunoglobulin may be indicated. If varicella develops, treatment with antiviral agents may be considered.
- If an AGAMREE-treated patient is exposed to measles, prophylaxis with immunoglobulin may be indicated.

Hepatitis B Virus Reactivation

Hepatitis B virus reactivation can occur in patients who are hepatitis B carriers treated with immunosuppressive dosages of corticosteroids. Reactivation can also occur in corticosteroid-treated patients who appear to have resolved hepatitis B infection.

Screen patients for hepatitis B infection before initiating immunosuppressive (e.g., prolonged) treatment with AGAMREE. For patients who show evidence of hepatitis B infection, recommend consultation with physicians with expertise in managing hepatitis B regarding monitoring and consideration for hepatitis B antiviral therapy.

Fungal Infections

Corticosteroids may exacerbate systemic fungal infections; therefore, avoid AGAMREE use in the presence of such infections. For patients on chronic AGAMREE therapy who develop systemic fungal infections, AGAMREE withdrawal or dosage reduction is recommended.

Amebiasis

Corticosteroids may activate latent amebiasis. Therefore, it is recommended that latent amebiasis or active amebiasis be ruled out before initiating AGAMREE in any patient who has spent time in the tropics or any patient with unexplained diarrhea.

Strongyloides Infestation

Corticosteroids should be used with great care in patients with known or suspected Strongyloides (threadworm) infestation. In such patients, corticosteroid-induced immunosuppression may lead to Strongyloides hyperinfection and dissemination with widespread larval migration, often accompanied by severe enterocolitis and potentially fatal gram-negative septicemia. For patients on corticosteroids who develop known or suspected Strongyloides (threadworm) infestation, withdrawal of corticosteroids or reduction of the dose of corticosteroids is recommended.

Alterations in Cardiovascular/Renal Function

Corticosteroids, including AGAMREE, can cause elevation of blood pressure, salt and water retention, and increased excretion of potassium and calcium.

Monitor blood pressure and assess for signs and symptoms of volume overload. Monitor serum potassium levels.

AGAMREE should be used with caution in patients with congestive heart failure, hypertension, or renal insufficiency. Literature reports suggest an association between use of corticosteroids and left free wall rupture after a recent myocardial infarction; therefore, therapy with AGAMREE should be used with great caution in these patients.

Gastrointestinal Perforation

There is an increased risk of gastrointestinal perforation with the use of corticosteroids in patients with certain gastrointestinal disorders, such as active or latent peptic ulcers, diverticulitis, fresh intestinal anastomoses, and non-specific ulcerative colitis. Signs of gastrointestinal perforation, such as peritoneal irritation, may be masked in patients receiving corticosteroids.

Avoid AGAMREE if there is a probability of impending perforation, abscess, or other pyogenic infections; diverticulitis; fresh intestinal anastomoses; or active or latent peptic ulcer.

Behavioral and Mood Disturbances

Potentially severe psychiatric adverse reactions may occur with systemic corticosteroids, including AGAMREE. Symptoms typically emerge within a few days or weeks of starting treatment and may be dose-related. These reactions may improve after either dose reduction or withdrawal, although pharmacologic treatment may be necessary.

Inform patients or caregivers of the potential for behavioral and mood changes and encourage them to seek medical attention if psychiatric symptoms develop, especially if depressed mood or suicidal ideation is suspected.

Effects on Bones

Decreased Bone Mineral Density

Corticosteroids, such as AGAMREE, decrease bone formation and increase bone resorption both through their effect on calcium regulation (i.e., decreasing absorption and increasing excretion) and inhibition of osteoblast function. This, together with a decrease in the protein matrix of the bone secondary to an increase in protein catabolism and reduced sex hormone production, may lead to inhibition of bone growth in pediatric patients and the development of bone loss at any age. Bone loss can predispose patients to vertebral and long bone fractures.

Consider a patient's risk of osteoporosis before initiating corticosteroid therapy. Monitor bone mineral density in patients on long-term treatment with AGAMREE.

Avascular Necrosis

Corticosteroids may cause avascular necrosis.

Ophthalmic Effects

The use of corticosteroids, such as AGAMREE, may produce posterior subcapsular cataracts. Corticosteroids may also cause glaucoma with possible damage to the optic nerves, and may increase the risk of secondary ocular infections caused by bacteria, fungi, or viruses. Corticosteroids are not recommended for patients with active ocular herpes simplex. Intraocular pressure may become elevated in some patients taking corticosteroids. If treatment with AGAMREE is continued for more than 6 weeks, monitor intraocular pressure.

Immunizations

Administer all immunizations according to immunization guidelines prior to starting AGAMREE. Administer live-attenuated or live vaccines at least 4 to 6 weeks prior to starting AGAMREE. Patients on AGAMREE may receive concurrent vaccinations, except for live-attenuated or live vaccines.

Effects on Growth and Development

Long-term use of corticosteroids, including AGAMREE, can have negative effects on growth and development in children.

Myopathy

Patients receiving corticosteroids and concomitant therapy with neuromuscular blocking agents (e.g., pancuronium) or patients with disorders of

neuromuscular transmission (e.g., myasthenia gravis) may be at increased risk of developing acute myopathy. This acute myopathy is generalized, may involve ocular and respiratory muscles, and may result in quadriplegia. Clinical improvement or recovery after stopping corticosteroids may require weeks to years.

Kaposi's Sarcoma

Kaposi's sarcoma has been reported to occur in patients receiving corticosteroid therapy, most often for chronic conditions. Discontinuation of treatment may result in clinical improvement of Kaposi's sarcoma.

Thromboembolic Events

Observational studies have shown an increased risk of thromboembolism (including venous thromboembolism) particularly with higher cumulative doses of corticosteroids. It is unclear if risk differs by daily dose or duration of use. Use AGAMREE with caution in patients who have or may be predisposed to thromboembolic disorders.

Anaphylaxis

Rare instances of anaphylaxis have occurred in patients receiving corticosteroid therapy.

ADVERSE REACTIONS

The following serious adverse reactions are discussed in more detail in other sections:

- Alterations in Endocrine Function
- Immunosuppression and Increased Risk of Infection
- Alterations in Cardiovascular/Renal Function
- Gastrointestinal Perforation
- Behavioral and Mood Disturbances
- Effects on Bones
- Ophthalmic Effects
- Immunizations
- Effects on Growth and Development
- Myopathy
- Kaposi's Sarcoma
- Thromboembolic Events
- Anaphylaxis

Clinical Trials Experience

Because clinical trials are conducted under widely varying conditions, adverse reaction rates observed in the clinical trials of a drug cannot be directly compared to rates in the clinical trials of another drug and may not reflect the rates observed in practice.

Common Adverse Reactions in Clinical Studies

Table 1 lists the adverse reactions that occurred in $\geq 5\%$ of the patients treated with AGAMREE 6 mg/kg/day (N=28) or AGAMREE 2 mg/kg/day (N=30) and that occurred more frequently than in the patients who received placebo (N=29) in Study 1, which was 24 weeks and included patients with DMD between the ages of 4 and 7 years.

Table 1: Adverse Reactions in Patients with DMD that Occurred in $\geq 5\%$ of Patients Treated with AGAMREE and More Frequently than in Patients Who Received Placebo During 24 Weeks (Study 1)

Adverse Reaction	AGAMREE 2mg/kg/d (N=30) %	AGAMREE 6mg/kg/d (N=28) %	Placebo (n=29) %
Cushingoid features	7	29	0
Psychiatric disorders ¹	7	21	14
Vomiting	17	14	7
Weight increased	0	11	3
Vitamin D deficiency	7	11	0
Cough	10	7	3
Headache	7	7	3
Diarrhea	3	7	3
Increased appetite	3	7	3
Rhinitis	3	7	3

¹Includes the following adverse reactions that occurred more frequently in the AGAMREE group than in placebo: abnormal behavior, aggression, agitation, anxiety, irritability, mood altered, sleep disorder, and stereotypy.

In a separate open-label safety study of pediatric patients aged 2 to less than 4 years (n=16) and pediatric patients aged 7 to less than 18 years (n=16) with DMD, adverse reactions were similar to those seen in the Study 1 pediatric patients.

DRUG INTERACTIONS

Effect of Other Drugs on Vamorolone

Co-administration of AGAMREE with itraconazole, a strong CYP3A4 inhibitor, increases vamorolone exposure. Reduce the dosage of AGAMREE in patients when strong CYP3A4 inhibitors are used concomitantly. No dosage adjustments are required when AGAMREE is concomitantly administered with moderate or weak CYP3A4 inhibitors.

USE IN SPECIFIC POPULATIONS

Pregnancy

Risk Summary

AGAMREE is indicated for use for the treatment of DMD, which is a disease of young male patients. However, corticosteroids in general should be used during pregnancy only if the potential benefit justifies the potential risk to the fetus. Infants born to mothers who have received substantial doses of corticosteroids during pregnancy should be carefully observed for signs of hypoadrenalism. There are no data on the use of AGAMREE during pregnancy.

Animal reproduction studies have not been conducted with AGAMREE.

Lactation

Risk Summary

There are no data on the presence of vamorolone in human milk or the effects on milk production.

AGAMREE is indicated for use for the treatment of DMD, which is a disease of young male patients. However, systemically administered corticosteroids appear in human milk and could suppress growth, interfere with endogenous corticosteroid production, or cause other untoward effects. The developmental and health benefits of breastfeeding should be considered along with the mother's clinical need and any potential adverse effects on the breastfed infant.

Pediatric Use

The safety and effectiveness of AGAMREE for the treatment of DMD have been established in patients 2 years of age and older. Use of AGAMREE in pediatric patients is supported by a multicenter, randomized, double-blind, placebo- and active-controlled study in 121 males 4 to less than 7 years of age. Use of AGAMREE in patients 2 years to less than 4 years of age and 7 to less than 18 years of age is supported by findings of efficacy and safety in patients 4 to less than 7 years of age with DMD, and by pharmacokinetic and safety data from patients 2 to 4 years of age and 7 to less than 18 years of age.

The safety and effectiveness in pediatric patients below the age of 2 years have not been established.

Geriatric Use

DMD is largely a disease of children and young adults; therefore, there is no geriatric experience with AGAMREE.

Hepatic Impairment

Moderate hepatic impairment increases vamorolone exposure. Reduce the AGAMREE dosage in patients with mild to moderate hepatic impairment. There is no clinical experience with AGAMREE in patients with severe hepatic impairment, and a dosing recommendation cannot be provided for patients with severe hepatic impairment.

CLINICAL PHARMACOLOGY

Mechanism of Action

Vamorolone is a corticosteroid that acts through the glucocorticoid receptor to exert anti-inflammatory and immunosuppressive effects. The precise mechanism by which vamorolone exerts its effect in patients with DMD is unknown.

Pharmacodynamics

Vamorolone produced a dose-dependent decrease in morning cortisol levels in the clinical studies. Treatment with corticosteroids is associated with a suppression of endogenous cortisol concentrations and an impairment of the hypothalamus-pituitary-adrenal (HPA) axis function. A dose-dependent increase in leukocyte counts and lymphocyte counts was observed in clinical studies with vamorolone.

Cardiac Electrophysiology

Vamorolone does not cause a mean increase in the QTc interval >20 milliseconds (ms) at 16 times the approved recommended dose.

See full Prescribing Information available at AGAMREEhcp.com.



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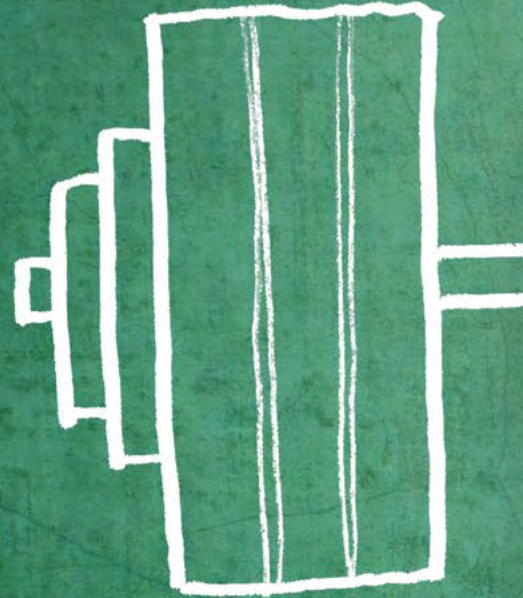


RAI

From the time Alexa Dectis was diagnosed with spinal muscular atrophy (SMA) at 18 months old, her parents spoke openly and honestly about her condition and how it would affect her body. They never tried to hide the fact that SMA is progressive and would weaken her muscles over time. And every time her mother performed a task such as cough assist, she carefully explained to Alexa what she was doing and why.

Now 30 years old, Alexa is thriving and living on her own in Los Angeles – 3,000 miles from her parents in Orefield, Pennsylvania. An entertainment lawyer, she credits her parents’ candid conversations about her condition as a factor driving her independence and success.

“Nothing about my diagnosis was ever kept from me,” says Alexa, who has been listed in the Forbes “30 under 30” list. “My parents’ openness made all the difference in the world. Their willingness to have conversations about the severity of my physical limitations motivated me to make the most of every opportunity that I possibly could.”



Parenting demands the same qualities we want to instill in our kids — honesty, resilience, and courage

BY KAREN DOSS BOWMAN



SING

resilient

KIDS



with
**Neuromuscular
Diseases**





Alexa Dectis is a lawyer in Los Angeles.

“My parents’ openness made all the difference in the world. Their willingness to have conversations about the severity of my physical limitations motivated me to make the most of every opportunity that I possibly could.” — Alexa Dectis

fulfilling lives with those limitations. As Alexa’s experience shows, talking openly with children about their disease is the first step to normalizing the experience and reducing anxiety.

Parents play a key role in building a foundation to encourage their children to dream big. Here are some tips for boosting their confidence to go out and pursue those dreams.

Provide opportunities to make friends

Make sure children with neuromuscular conditions have opportunities to engage in various social settings such as school, church, and community organizations. That will help them form friendships and develop critical interpersonal skills.

After Brock Dahlke was diagnosed with Duchenne muscular dystrophy (DMD) at 6 years old, his parents always welcomed his cousins and friends into their

Generally, parents strive to raise their children to live as independently and productively as they can. That can be especially daunting for parents whose children have physical limitations. But kids with neuromuscular diseases can and do grow up to lead

A MODEL OF RESILIENCE

Resilience, the ability to work through and bounce back from challenges, is a skill children can develop over time as they build problem-solving skills and confidence. The best way to teach that skill is to model it. Here are some strategies:

- + **Talk openly about emotions.** Be the safe place where a child can ask questions about their disease and share their feelings about how it affects their lives. It’s OK to voice your frustrations and fears, too, but also express confidence in their ability to adapt and thrive.
- + **Demonstrate coping skills.** Consider what your child is learning from the way you handle challenges. Can you take a deep breath and

remain calm? Are you willing to ask for help when you need it?

+ **Embrace mistakes.** When kids are afraid to fail, they avoid trying new things. Give your child the message that mistakes help them learn. You can help by talking about a mistake you made and how you recovered from it.

+ **Make time for self-care.** It’s harder to be resilient when you’re running on empty. Follow self-care practices such as meditation, exercise, social activity, and rest. Don’t neglect your mental health ([mda.org/MentalHealth](https://www.mda.org/MentalHealth)). Not only does taking time for yourself help you recharge, but it gives your child some time to practice being independent.

INSPIRING INDEPENDENCE

These MDA programs help kids, teens, and young adults with neuromuscular diseases make friends, build independence, and learn skills.

Let's Play provides a welcoming environment for everyone in the MDA community to enjoy gaming and activities while socializing with others who can truly understand their unique situations. Join events like Saturday Game Night or check out daily streamed content. Learn more at mda.org/lets-play.



Mentorship Programs help teens and young adults living with neuromuscular diseases discover their strengths and interests and explore career paths. There is no cost to participate in these five-week virtual programs for ages 14-21. Learn how to apply at mda.org/mentorships.

MDA Summer Camp is a week-long overnight camp especially for kids with neuromuscular diseases. In a safe and supportive environment, kids build self-reliance and confidence while trying new activities and developing friendships that last a lifetime. Learn more at mda.org/SummerCamp.



Peer Connections brings people in the MDA community together, whether they're in the same neighborhood or across the country. Participants can specify that they are open to being connected to others based on criteria including diagnosis, age, and interests. To get started on a match, reach out to the MDA Resource Center at **833-ASK-MDA1** or ResourceCenter@mdaUSA.org.

home in Shakopee, Minnesota. This created a community of support to help meet Brock's physical and social needs. When Brock was a teenager, his parents even occasionally gave his friends the keys to the family's wheelchair-accessible van. That allowed Brock to spend time with his friends without his parents around, going to movies, sporting events, restaurants, and other adventures.

"My parents wanted me to go out and do things with my friends, but it was a huge step for them to allow my friends to drive me around; not a lot of parents would do that," says Brock, now 32 years old. "Being able to spend time with my friends and go out like other teenagers helped me have a more normal social life."



LOVE YOURSELF

Listen to a conversation with Megan DeJarnett, a speaker and inclusion advocate living with spinal muscular atrophy (SMA), about why she wrote her first children's book, "No Such Thing as Normal," at [MDAQuest.org/podcast/no-such-thing](https://mdaquest.org/podcast/no-such-thing).



Brock Dahlke appreciated having freedom as a teen.

Promote advocacy skills

Encourage and empower children to advocate for themselves. Give them opportunities to ask questions at each medical appointment, and include them in school meetings for their individualized education plan (IEP) or 504 plan. This will teach them to speak up about their needs and wants. But don't get frustrated or give up if this lesson takes a long time.

"The advocacy piece is a long road, and it takes practice and feedback," says Areti Vassilopoulos, PhD, pediatric health psychologist and assistant professor of clinical pediatrics and child psychology at Yale School of Medicine.

Give kids opportunities to ask questions at each medical appointment, and include them in school meetings for their IEP or 504 plan.



Ann Motl learned to advocate by watching her mother.

Ann Motl, a 33-year-old lawyer living with Charcot-Marie-Tooth disease (CMT), learned self-advocacy techniques by observing her mother's persistent requests for appropriate and reasonable accommodations at school.

When Ann was in high school, for example, one of her classes was on the second floor — accessible via an unreliable elevator that had to be operated by a school official. Her teacher resisted moving to a first-floor classroom even though Ann was often late or missed class because of elevator problems. Her mother pushed back.

"My mother persuaded school officials that this was ridiculous, and the teacher finally moved the class downstairs," recalls Ann, who grew up on a dairy farm in Staples, Minnesota. "Situations like this were a learning process for both my mom and me. I was able to see her advocacy style and learn strategies for taking care of myself once I was in college and living on my own."



Areti Vassilopoulos, PhD



Adriana Ferri

Encourage self-reliance

Like their peers, children with neuromuscular diseases desire greater independence as they mature. At the same time, the progressive nature of their conditions increases their need for caregiving support. This can lead to frustration and low self-esteem.

Parents can help kids satisfy their desire for independence by encouraging them to participate in decisions related to their care while giving them opportunities to express their needs to caregivers, teachers, healthcare providers, and others.

"It is essential to grow and cherish children's independence while normalizing seeking and



Sydney Horak is a student at Texas A&M.

there to help you, and you need to explain what you need from them. That was important for preparing me to go to college and eventually live on my own.”

Nurture problem-solving skills

Helping children with neuromuscular diseases to identify their interests and set goals can build confidence. While physical limitations may require these kids to do things differently than others, parents can nurture their problem-solving skills to adjust to challenges.


“It is important to encourage children, teens, and young adults with neuromuscular diseases that it is okay to have dreams and goals, and we can work together to find ways to accomplish them,”

Adriana says.

Sydney Horak, a junior honors student at Texas A&M University, says her parents never limited her choices about what she could or could not do as she was growing up with SMA.

“Where there was a will, there was a way,” says Sydney, an accounting major

who aspires to become an entrepreneur after graduating.

“My parents wanted me to be able to do everything I wanted to do, even if a little alteration was necessary. I believe this is the primary reason I am so determined and motivated to accomplish my goals. It’s also why I created my own saying: ‘Never let your limitations be your limit.’” 

While physical limitations may require them to do things differently than others, parents can nurture problem-solving skills to adjust to challenges.

accepting help from others,” says Adriana Ferri, a child life specialist at Texas Children’s Hospital. “Parents should offer praise for doing things independently while also encouraging children to be brave and aware enough to seek support when needed.”

For Ann, attending MDA Summer Camp for 11 years starting at age 7 offered the chance to learn and practice self-reliance.

“Summer Camp was a really great experience for learning to have someone else take care of you,” she says. “During those weeks, you work with an assistant who’s



INDEPENDENCE STARTS EARLY

The day her daughter left for college wasn’t the first time Linda VanVliet practiced letting her go. Read how Linda helped her daughter prepare to be independent at [MDAQuest.org/prepare-for-adult-life](https://mdaquest.org/prepare-for-adult-life).

Karen Doss Bowman is a freelance writer and editor living with a slow-progressing type of amyotrophic lateral sclerosis (ALS) in Bridgewater, Virginia.

What Is **THE PIP** of Promise?

You've probably heard of the drug development pipeline when people talk about research for neuromuscular disease therapies. But what is it, and why is it called a pipeline?

The world of pharmaceutical research and development is complex. Any company that wants to sell a drug in the United States must go through a series of steps to develop a drug that is safe and effective.

The pipeline represents this process and all the drug candidates that are being researched or tested in one of these **STAGES**:

STAGE 1
Discovery and development
 Early laboratory testing to turn an idea into a potential drug

STAGE 2
Preclinical research
 Laboratory and animal testing to establish basic safety of the drug

STAGE 3
Clinical trials
 Studying how the drug works in people

STAGE 4
FDA review
 The US Food and Drug Administration (FDA) examines data from each stage and decides whether to approve the drug.

Right now, thousands of potentially beneficial therapies for neuromuscular diseases are being researched and tested

BY AMY BERNSTEIN

ELINE

An active pipeline

It's called a pipeline, not a straw, because the biopharmaceutical industry is busy. There are thousands of potentially beneficial therapies in development at any given time.

This is an especially promising time for neuromuscular diseases, with about 165 potentially disease-altering drugs currently in the clinical trial stage. Turn to page 44 to see a chart detailing a portion of these promising drugs in clinical trials. Turn to page 46 to learn about finding and getting into clinical trials.

Important drug approvals

In the past decade, the drug development pipeline has led to some exciting treatment advances for MDA's community. As of January 2024, the FDA has

PHASES of a Clinical Trial

Clinical trials go through three phases before a drug may be approved. Each phase serves a different purpose.

Phase 1

Purpose: *Safety and dosing*
Approximately 70% of drugs move to the next phase.

Phase 2

Purpose: *Efficacy and side effects*
Approximately 33% of drugs move to the next phase.

Phase 3

Purpose: *Efficacy and monitoring adverse reactions*
Approximately 25% of drugs move to FDA review.

approved more than 20 new drugs to treat neuromuscular diseases. This is largely due to the remarkable progress made in the field of genetic medicine.

MDA is proud to play a significant role in this progress. To date, MDA-sponsored research has directly contributed to the development of eight FDA-approved therapies, including Relyvrio[®] and Rilutek[®] for amyotrophic lateral sclerosis (ALS); Myozyme[®] for Pompe disease; Kevevis[®] for periodic paralysis; Spinraza[®] for spinal muscular atrophy (SMA); and Exondys 51[®], Emflaza[®], Vyondys 53[®], and Agamree[®] for Duchenne muscular dystrophy (DMD). MDA also funded the first gene therapy trial for any form of muscular dystrophy in 1999 and the first vector-based gene therapy trial for DMD in 2006. Many of the gene therapies in clinical trials for neuromuscular diseases in the United States are based on strategies developed with MDA funding.

Progress across diseases

Families coping with ultra-rare diseases can feel frustrated as they see therapies developed for more prevalent neuromuscular diseases, like ALS and DMD. But there is a lot of positive news on that front, too.

In addition to advances in genetic medicine, the last decade has seen advances in clinical trial design and drug manufacturing, making it more feasible to develop drugs for specific rare diseases or classes of rare diseases with similar characteristics. Some biotech companies have formed to pursue rare disease research, and MDA created the Kickstart program to develop gene therapies for ultra-rare disorders.

In addition, scientific advances often translate across diseases. "The methods and platforms used to develop drugs for diseases like DMD and SMA have high promise to be applied to the super rare diseases," Alan H. Beggs, PhD, director of the Manton Center for Orphan Disease Research at Boston Children's Hospital, told Quest Media in 2021.

Right now, an unprecedented flow of potential neuromuscular disease therapies is moving from discovery and development to preclinical research to clinical trials, giving the MDA community hope that we'll see more drugs approved to treat neuromuscular diseases in the coming years. That is what MDA calls the "pipeline of promise." [Q](#)

Amy Bernstein is a writer and editor for Quest Media.

THERAPIES IN THE PIP

ANTIBODY TREATMENTS

Disease	Drug	Phase	Organization	Description
ALS	AL001	2	Alector/GlaxoSmithKline	Anti-sortilin (SORT1)
ALS	AP-101	2	AL-S Pharma AG/Neurimmune	Anti-SOD1
ALS	ANX005 (IV)	2	Annexon Biosciences	Anti-C1q
ALS	Actemra (tocilizumab)	2	Genentech	Anti-IL6 receptor
DMD	Pamrevlumab	3	Fibrogen	Anti-CTGF
DMD	Canakinumab (ILARIS)	1/2	Children's National Research Institute	Anti-IL1 β
FSHD	AOC 1020	1/2	Avidity Biosciences	Anti-DUX4 (siRNA-Ab conjugate)
FSHD	RO7204239	2	Hoffmann-La Roche	Anti-myostatin
MG	ALXN1830	1	AstraZeneca	FcRn blocker

CELL THERAPY

Disease	Drug	Phase	Organization	Description
ALS	CK0803	1	Cellenkos	Tregulatory cells
ALS	HYNRCS-Allo	1	Corestem	Stem cells
ALS	NeuroNata-R	3	Corestem	Stem cells
ALS	MSCs	2	Mayo Clinic	Stem cells
ALS	NSI-566	2	Palisades Bio	SC-derived neural stem cells
ALS	RAPA-501	1	RapaTherapeutics	Tregulatory cells

GENE THERAPY

Disease	Drug	Phase	Organization	Description
ALS	APB-102	1/2	Apic Bio/UniQure	<i>SOD1</i> gene
ALS	Engensis	2	Helixmith	<i>HGF</i> gene
CMT	VM202	1/2	Helixmith	<i>HGF</i> gene
DMD	RGX202	1/2	Regenxbio	Micro-dystrophin
DMD	PF-06939926	3	Pfizer	Mini-dystrophin
DMD	rAAVrh74.MCK.GALGT2	1/2	Nationwide Children's Hospital	<i>GALGT2</i> gene
FA	LX2006	1/2	LEXEO Therapeutics/Adverum Biotechnologies	<i>FXN</i> gene
LGMD	ATA-200	1/2	Atamy Therapeutics	<i>FKRP</i> gene
LGMD	SRP-9003	1/2	Sarepta	β -sarcoglycan gene

RNA-TARGETED THERAPIES

Disease	Drug	Phase	Organization	Description
ALS	AROSOD-1	1	Arrowhead Pharmaceuticals	RNAi-SOD1
ALS	BIIB105/ION541	1/2	Biogen/Ionis	ASO-ATXN2
ALS	ION363	3	Ionis	ASO-FUS
ALS	QRL-201	1	QurAlis	ASO-STMN2
ALS	RAG-17	1	Ractigen Therapeutics	siRNA-SOD1
DMD	AOC-1044	1/2	Avidity Biosciences	Exon 44 skipping
DMD	DS-5141	1/2	Daiichi Sankyo	Exon 45 skipping
DMD	DYNE-251	1/2	Dyne Therapeutics	Exon 51 skipping

SMALL MOLECULE DRUGS

Disease	Drug	Phase	Organization	Description
ALS	Masitinib	3	AB Science	Tyrosine kinase inhibitor
ALS	CORT113176 (Dazucorilant)	2	Corcept Therapeutics	Glucocorticoid modulator
ALS	SPG-302	1	Spinogenix	Synapse regeneration
BMD	Vamorolone	2	ReveraGen BioPharma	MR antagonist
CMT	IFB-088	1/2	InFlectis Bioscience	UPR activator
CMT	FLX-787	2	Flex Pharma	TRP ion channel agonist
CMD	Omigapil	1	Santhera Pharmaceuticals	Anti-apoptotic agent
DM	Tideglusib	2/3	AMO Pharma Limited	GSK-3 inhibitor
DM	Mexiletine	3	Lupin	Sodium channel blocker
DMD	Tamoxifen	3	University Hospital, Basel	SERM modulator
DMD & BMD	EDG-5506	2	Edgewise Therapeutics	Muscle stabilizer
DMD & BMD	Givinostat	1/2	Italfarmaco	HDAC inhibitor

Disease	Drug	Phase	Organization	Description
MG	gefurumab (ALXN1720)	3	AstraZeneca	C5 complement inhibitor
MG	Rituximab	2	Genentech/Roche	Anti-CD20
MG	TAK-079	2	Takeda	Anti-CD38
MG	HBM9161 (Batoclimab)	3	Harbour BioMed	Anti-FcRn
MG	inebilizumab	3	Horizon Therapeutics/Viela	Anti-aquaporin-4 (AQP4)
MG	Nipocalimab (M281)	3	Janssen/Johnson & Johnson	Anti-FcRn
MG	satralizumab	3	Roche	Anti-AQP4
SMA	RO7204239/GYM329	3	Roche-Genentech	Anti-myostatin
SMA	SRK-015 (Apitegromab)	3	Scholar Rock	Anti-promyostatin

Disease	Drug	Phase	Organization	Description
CMT	CLZ-2002	1	Cellatoz Therapeutics	MSC-derived schwann cells
CMT	EN001	1	ENCell	WJ-MSCs
DMD	DT-DECO1	1	Dystrogen	Chimeric cells (dystrophin)
DMD	CAP-1002	3	Capricor	Cardiosphere-derived cells
MG	Descartes-8	1/2	Cartesian Therapeutics	rCAR-T
MG	MuSK-CAART	1	Cabaletta Bio	CART-19

Disease	Drug	Phase	Organization	Description
LGMD	SRP-6004	1	Sarepta	Dysferlin gene
LGMD	LION-101	1/2	AskBio (Bayer)	<i>FKRP</i> gene
Myopathies	AT132	1/2	Astellas Gene Therapies	<i>MTM1</i> gene (for XLMTM)
Pompe	ACTUS-101(AAV2/8LSPhGAA)	1/2	Actus Therapeutics/AskBio	<i>GAA</i> gene
Pompe	AT845	1/2	Astellas Gene Therapies/Audentes	<i>GAA</i> gene
Pompe	SPK-3006	1/2	Spark Therapeutics/Roche	<i>GAA</i> gene
Pompe	rAAV9-DES-hGAA	1	Lacerta Therapeutics	<i>GAA</i> gene
SMA	EXG001-307	1/2	Exegenes/Hangzhou Jiayin Biotech Ltd	<i>SMN1</i> gene
SMA	GC101	1/2	GeneCradle Therapeutics	<i>SMN1</i> gene (for SMA2)

Disease	Drug	Phase	Organization	Description
DMD	Vesleteplirs(en exon 51)	2	Sarepta	Exon 51 skipping
DMD	SQY51	1/2	SQY Therapeutics	Exon 51 skipping
DMD	WVE-N531	1/2	Wave	Exon 53 skipping
DMD	ATL1102	2	Antisense Therapeutics	ASO-inhibitor of CD49d
DMD	PGN-EDO51	1	PepGen	Exon 51 skipping
DM	AOC 1001	1/2	Avidity Biosciences	siRNA-TfR1
DM	DYNE-101	1/2	Dyne	ASO-DMPK
SMA	BIIB115	1/2	Biogen/Ionis	ASO-STMN2

Disease	Drug	Phase	Organization	Description
FA	MIN-102 (leriglitzone)	2	Minoryx Therapeutics	PPAR gamma agonist
FA	Elamipretide	1/2	Children's Hospital of Philadelphia	Reduces oxidative stress
FSHD	losmapimod	3	Fulcrum Therapeutics	p38 MAPK inhibitor
LGMD	Ribitol(BBP-418)	3	ML Bio Solutions/BridgeBio	Precursor to alpha-dystroglycan
MG	Mysaterix	2	CuraVac	Antigen receptor
MG	amifampridine	3	Catalyst	K channel blocker
Myopathies	ARM210	1	ARMGO Pharma	RyR1
Myopathies	RENO01 (Mavodelpar)	2/3	Reneo Pharmaceuticals	PMM
Pompe	MZE001	1	Maze Therapeutics/Sanofi	Substrate reduction/stabilizer
SBMA	NIDO-361	1	Nido Biosciences	Androgen receptor BF3
SBMA	AJ201	2	AnnJi Pharmaceutical/Avenue Therapeutics	Nrf1 and Nrf2 activation
SMA	reldesemtiv	2	Cytokinetics	Troponin activator

+DON'T SEE YOUR DISEASE HERE?

This chart shows a fraction of the clinical trials in progress. Learn how to find more clinical trials on page 46.

HOW TO FIND CLINICAL TRIALS

Many people in the MDA community are interested in participating in clinical trials. Here is how you can find and learn about clinical trials for neuromuscular diseases.

Find a trial

MDA's resources for finding clinical trials include the Clinical Trial Finder tool (mda.org/clinical-trials), Clinical Trial Updates list (mda.org/clinical-trial-updates), and Progress Now in Quest Magazine ([page 16](#)).

Your MDA Care Center team is an excellent resource for finding clinical trials that might be right for you or your child. They keep up with the latest research, and many Care Centers serve as clinical trial sites.

Dig deeper

Your next stop is the National Library of Medicine's ClinicalTrials.gov, an online database of clinical research studies taking place in the United States and more than 200 countries.

This website is a wealth of information if you know how to find it. Review the "How to Search for Clinical Studies" page (ClinicalTrials.gov/find-studies/how-to-search) to get started.

Once you know how to search, you can zero in on the studies that are relevant to you. Listings include important information, such as:

- > Whether a study is actively enrolling
- > Eligibility requirements
- > What the study is measuring
- > How the study is designed
- > How long it is expected to last
- > Who to contact about the study

Many trials have a study coordinator who can answer questions and help address barriers to participation for eligible individuals. (Read "9 Questions to Ask Before Joining a Clinical Trial" at MDAQuest.org/9-questions.)

Consider compassionate use

In cases where an individual with a life-threatening condition may benefit from an investigational drug but cannot participate in a clinical trial, a physician may ask the drug developer and health authorities for permission to administer the drug to that individual outside of the clinical trial. This is called "expanded access" or "compassionate use."

Learn more about these options through the Reagen-Udall Foundation Expanded Access Navigator at navigator.reaganudall.org.



TYPES OF THERAPIES IN THE PIPELINE

Researchers are exploring many ways to treat neuromuscular diseases by addressing specific symptoms or altering body mechanisms. Some of the most promising candidates fall into one of the categories below or combine them to target multiple pathways of the disease.

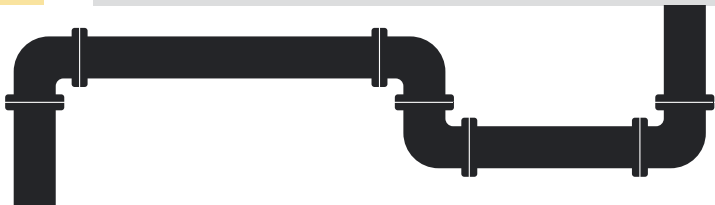
Antibody treatments A natural component of our immune system, antibodies are proteins that can identify and selectively bind and neutralize a targeted protein. For therapies, antibodies have been designed in the lab to target potential cellular contributors to diseases.

Cell therapy These therapies integrate new, healthy cells into the body to replace diseased or damaged cells, use interactive factors to modulate the function of existing cells, or use immune cells to remove dysfunctional cells.

Gene therapy These therapies use techniques to modify a person's genes to change the course of a disease. This might involve introducing a healthy copy of a gene to replace a non-working or missing gene, inactivating a malfunctioning gene, or introducing a new or modified gene into the body.

RNA-targeted therapies RNA, similar to DNA, is present in all living cells and serves many functions, including acting as a messenger carrying instructions from DNA for controlling the synthesis of proteins. By targeting RNA, these therapies seek to reduce, restore, or modify protein expression. Exon skipping is an example of a therapy that modifies messenger RNA (mRNA) to deliver new instructions to cells. Other types of RNA being used in therapies include small interfering RNAs (siRNAs) and microRNAs (miRNAs). Antisense oligonucleotides (ASOs) are a synthetic form of RNA or DNA.

Small molecule drugs Small molecules are organic compounds with low molecular weight, which allows them to pass through cell membranes easily. Small molecule drugs are designed to engage biological targets inside cells and modify disease pathways by activating or inhibiting certain functions.





Advocacy Wins!



Image: iStock.com/guvendemir

MDA Advocacy has been hard at work making a difference in the lives of the neuromuscular disease community.

Better travel experiences

In October 2023, MDA announced a partnership with the Transportation Security Administration (TSA) to develop training to prepare TSA officers to interact with travelers living with neuromuscular disease and other disabilities affecting their mobility. Leveraging the experiences of MDA advocates, the training will educate officers on prioritizing traveler safety, comfort, and dignity. The training program will include watching an MDA video showing the common indignities of air travel for people with disabilities who depend on power wheelchairs and other mobility devices. In addition, MDA will support the TSA Cares program designed to assist travelers with disabilities, medical conditions, and others who may need additional assistance with screening. Learn more about TSA Cares at tsa.gov/travel/passenger-support.

Expanded newborn screenings

MDA is excited to announce that in January all 50 states started screening newborns for spinal muscular atrophy (SMA). This accomplishment was made possible by countless MDA advocates and fellow organizations working tirelessly to ensure all babies are screened for SMA.

In addition, in 2023, West Virginia, Utah, Louisiana, Iowa, Alabama, and Hawaii began screening for Pompe disease, bringing the total to 43 states. MDA also saw progress in screening for Duchenne muscular dystrophy (DMD) and expects New York, Ohio, and Minnesota to begin screening for DMD this year.

From improving air travel to increasing access to newborn screening, MDA's grassroots advocates were a force in 2023. To make 2024 another year of big wins, MDA Advocacy needs your help. Join our grassroots advocacy network at mda.org/advocacy.

New: MDA Community Groups

Looking for a supportive place to gather resources and exchange valuable information with others in the neuromuscular disease community?

Then the new MDA Community Groups are for you.

Currently, Community Groups are held in English and are open for parents of newly diagnosed babies and children, newly diagnosed adults, individuals with ALS, and individuals eligible for gene therapies. Groups may be added based on community needs.

Find more information at mda.org/care/community-groups.



Image: iStock.com/Topdesigner



Matthew, a mentee of the program for the last three years, works on his STEM project.

Meaningful Mentorship

In 2023, MDA's Mentorship Program expanded to offer sessions on general career exploration in addition to science, technology, engineering, and math (STEM). More than 40 teens and young adults participated in the program, which aims to increase the number of people living with neuromuscular diseases in the workforce. Almost 30 mentors volunteered, representing career fields such as medicine, computer programming, education, criminal justice, and engineering. These five-week virtual mentoring sessions are open to individuals with neuromuscular diseases ages 14-21.

To learn more about MDA's Mentorship Program, visit mda.org/mentorships.

Transitioning to Adulthood

Among the many milestones of growing up is transitioning from pediatric to adult healthcare. The printable MDA Transition Guide: Pediatric to Adult Care can make this shift less confusing and challenging. Easy to navigate, the guide includes a helpful checklist, timeline, and glossary of important terms.



To download the Transition Guide, visit mda.org/education and click on "General Neuromuscular Disease Resources."

Announcing MDA Scholarships

MDA is excited to announce a new scholarship program, which will award educational scholarships to college students living with neuromuscular diseases.



These merit-based scholarships are part of a broader effort to support and empower the young adult population. Scholarships will be awarded based on leadership and community advocacy.

To learn more about the program, timeline, and application process, visit mda.org/scholarship.

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MDA Engage Symposium Highlight

On Nov. 11, more than 120 people gathered at Stanford University in California for a day of learning and networking. More than 20 clinicians from Stanford; the University of California, San Francisco;



Clinicians from Stanford, UCSF, and UC Irvine presented at the MDA Engage Symposium in Palo Alto, California.

and the University of California, Davis, met with MDA community members to provide important updates, education, and support for people living with neuromuscular disease and their loved ones. The symposium also brought together members of the biotechnology and pharmaceutical industries and other patient advocacy organizations.

For more information on MDA Engage Symposiums and other Community Education programs, please visit mda.org/community-ed.



The Silver Lining

Connecting with others with my diagnosis is a path to lifelong friendships

BY MELISSA GROVE

For 25 years after I was diagnosed, I never met another person with limb-girdle muscular dystrophy (LGMD). I dealt with everything on my own, which was lonely and hard, but since the doctors told me there was nothing I could do about the gradual progress of my condition, I decided to build a wonderful life.

After graduate school, I backpacked around Europe for three months, and then I began what ended up being a 35-year career caring for people with HIV/AIDS. Besides running a \$2.2 million healthcare nonprofit, I was also a therapist, helping people cope with their issues. I made wonderful friends. I met an amazing man when I was 32, and we have had a spectacular 21-year marriage. He is full of love and care, doing more and more for me as my physical condition

declines. But despite all the support and love in my life, I felt like something was missing.

Making connections

In 2012, my MDA Care Center neurologist told me about the Wellstone Center Dystroglycanopathy Patient and Family Conference in Iowa because it focused on my LGMD subtype, LGMD2I/R9 (also called LGMD2I or LGMDR9). I was eager to go and hear about what was happening with research after a lifetime without treatment.

Within five minutes of walking into the conference hall, I knew I had found my people. “I’m freezing,” “I’m always cold,” “I should have brought a sweater.” These were phrases I had said hundreds of times in my life. Maybe being cold wasn’t a Melissa thing; maybe it had something to do with LGMD. I heard my life reflected in the stories of everyone there. I felt validated, heard, understood, and seen.

The science talks were over my head, but I was happy to be there among my tribe. No one had to explain themselves or feel embarrassed. We all understood. I met Kristen, who is 10 years younger than me and had just been diagnosed. We were both first-timers at the conference, but I wanted to help her because I remembered how it felt to be newly diagnosed. We exchanged phone numbers and email addresses.

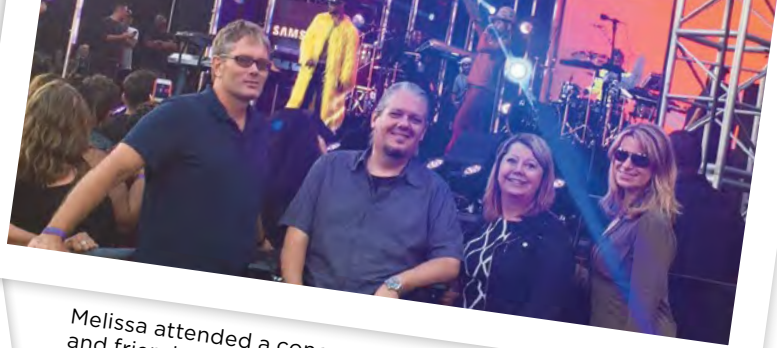
Each year, I rejoined my newfound friends at the conference: in addition to Kristen, Brian, who wrote tax software for Fortune 500 companies; Andrea, an occupational therapist from Ohio; and Lacey, who founded a Facebook group for people with LGMD2I/R9. Lacey inspired me because she used her experience and energy to spread information and support all of us through the Facebook group. These were not just people with my disability but fantastic humans I loved spending time with.

Melissa Grove





Melissa (right) celebrated her birthday in Las Vegas with friends Andrea (left) and Kristen (center).



Melissa attended a concert with her husband Dan (left) and friends Brian (center) and Kristen (right).

I'm not alone

C.S. Lewis said, "Friendship is born at the moment when one person says to another, 'What! You too? I thought I was the only one.'" There have been many of these moments at the conference, and this is a true silver lining to living with a rare disease: the opportunity to not feel alone on this journey.

To grow my friendship with Kristen, I invited her to come to Dallas. We had a blast! This was a real friendship and something that came to mean so much to me. We talk about relationships, work problems, fears and anxieties, and the myriad of things that come with a rare disease.

Kristen and I decided to visit Brian, who had been finding it difficult to travel to the conference. His prime California location made a wine trip appealing, so we rented a wheelchair-accessible van and picked him up. That was a great day! We later met up in Palm Springs, and he introduced me to Christine. She is a friend in the making. There is no end to the wonderful people I keep meeting, thanks to my disability.

When my husband and I turned 50, we celebrated in Las Vegas, and Kristen and Andrea were up there

in the wraparound suite at the Cosmopolitan with me. Andrea and I ganged up on Kristen and insisted she rent a scooter because we knew navigating Vegas would be impossible without it. She reluctantly agreed and ended up overcoming her resistance to using a mobility device. I can only imagine what people thought seeing the three of us tearing down Las Vegas Boulevard in front of the Bellagio fountains — there was no anguish, just plain fun. This memory is a silver lining.

Making more memories

Now, when I travel anywhere, I ask the Facebook group if any of them live at my destination, and I try to see them. When I was in Southern California, Tara, whom I had met at the conference, invited me to her pregnant daughter's birthday dinner. Now, I'm a part of her family, too.

Maria, who lives in Stockholm, Sweden, reached out to me through the Facebook group, and we organized a Zoom chat. Many Zoom chats later, she visited me in Dallas for a week. We talked about our challenges and how we solve them, and the cultural differences between the US and Sweden. We were instant friends, and I will visit her in Sweden. My circle of friends keeps growing, with each person adding another layer of support, information, and camaraderie.

Our friendships aren't just based on travel. Every day, we use Facebook Messenger to share something funny, exchange advice, crowdsource problems, and share our daily lives. When I broke my femur a few years ago, I had hundreds of people on the LGMD2I/R9 Facebook group to support me, which meant a lot.

Connection is the silver lining of living with a rare disease, and it is how we all can find meaning and joy in our journey. [C](#)

Melissa Grove, 56, is a psychotherapist and public speaker living in Dallas.

+FIND A FRIEND

MDA's Peer Connections program helps folks in the MDA community — including individuals living with neuromuscular diseases, parents, spouses, and siblings — connect with each other. To request a connection, contact the MDA Resource Center at [833-ASK-MDA1](tel:833-ASK-MDA1) or ResourceCenter@mdaUSA.org.



An Unforgettable Reunion

Former MDA Summer Campers get together 14 years later

BY MAGGIE CALLAHAN



Kelly Cooper (second row, third from the right) reunited with her MDA Summer Camp friends in Maryland in 2022.

Kelly Cooper, 34, who lives with limb-girdle muscular dystrophy (LGMD), hadn't seen many of her MDA Summer Camp friends since she graduated from the program in 2008, and she wanted to change that. So, in July 2022, Kelly hosted a reunion in Phoenix, Maryland, gathering 13 Summer Camp friends and many counselors.

"Reconnecting with all of my friends — it was a feeling so great I cannot put it into words," recalls Kelly, who attended Summer Camp in Leonardtown, Maryland, for eight years. "Time may have passed since we last saw each other in person, but the strength of our friendships never faded. Getting together for a reunion allowed us to rekindle those long-lost connections, laugh, and create memories for the future."

After the 2022 gathering, another former camper hosted a reunion in 2023. Kelly is planning for summer 2024 and hopes this becomes an annual tradition.

"The friendships I made at Summer Camp have had a profound and lasting impression on me," Kelly says. "Before attending MDA Summer Camp, I did not know anyone else in a wheelchair or with muscular dystrophy. Summer Camp allowed me to develop a support network of people diagnosed with muscular dystrophy who all experience the same struggles that I experience. My Summer Camp friends have all become my best friends, my greatest supporters, and the people I turn to when I am struggling."

+TRUE FRIENDS

Ready to make your Summer Camp connections? Sign up to attend or volunteer at mda.org/SummerCamp.

Maggie Callahan is a writer and editor for Quest Media.

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

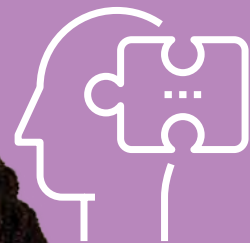
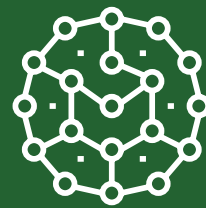
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