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# Quest

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## Your Best *Holiday* Season Yet!

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challenges and how  
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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](http://www.evrysdipregnancyregistry.com)
- are an adult male. Evrysdi may affect a man's ability to have children (fertility). Ask a healthcare provider for advice before taking this medicine
- are breastfeeding or plan to breastfeed. It is not known if Evrysdi passes into breast milk and may harm your baby

**Genentech**  
A Member of the Roche Group

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## ADVERTISEMENT

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

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



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

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

### Important Safety Information (continued)

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

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

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

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

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

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

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

**Please see accompanying brief summary for additional Important Safety Information.**

If you cannot afford your Evrysdi medication, visit [MySMASupport.com](http://MySMASupport.com) for financial assistance information.

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



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

**What is EVRYSDI?**

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

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

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

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

**How should I take EVRYSDI?**

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

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

**Taking EVRYSDI**

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

**Reusable Oral Syringes**

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

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

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

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

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

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

**How should I store EVRYSDI?**

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

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

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

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

**What are the ingredients in EVRYSDI?**

**Active ingredient:** risdiplam

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

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





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MDA-168

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

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**Quest Volume 30, Issue 4**

Published by  
Muscular Dystrophy Association  
1016 W Jackson Blvd #1073  
Chicago, IL 60607  
**800-572-1717**  
email: [quest@mdaUSA.org](mailto:quest@mdaUSA.org)  
Available on the internet  
at [MDAQuest.org](https://MDAQuest.org)  
ISSN 1087-1578  
Postage paid at Bolingbrook, IL  
Nonprofit postal permit number 1446.

Postmaster: Treat as Standard  
A Mail Only

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# 'Tis the Season for Inclusivity

I am thrilled to present the latest issue of Quest magazine, a publication that embodies our commitment to information, inclusivity, and progress. As we delve into the holiday season and celebrate with hearts full of warmth and anticipation, we explore topics reflective of the season and the needs of this community.

Our cover story is an invitation to make this holiday season your most memorable yet (page 30). From accessible holiday parties to practical shopping and financial tips, and from holiday travel insights to maintaining balanced nutrition during these times of celebration, we embark on a journey to ensure that everyone can participate fully in the joys of the season.

In the area of healthcare, we take a deep dive into transitioning from pediatric to adult care, a significant step for those living with neuromuscular diseases (page 34).

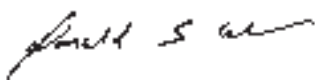
We also explore the field of genetic testing, focusing on its significance and implications (page 40), as well as navigating insurance for these services ([MDAQuest.org](https://MDAQuest.org)), empowering our readers to make informed decisions. And you will find a timely update on COVID-19 and neuromuscular disease to help you stay at the forefront of knowledge and awareness (page 26).

On the business side, we celebrate the strides made by notable companies like Google, QVC, and Slick Chicks (page 13). These trailblazers are setting new standards for disability inclusion and representation, redefining the boundaries of what's possible.

I'd also like to share with you that MDA has been officially approved as an accredited charity by the Better Business Bureau (BBB), meeting each of their 20 requirements. (Look us up on [Give.org](https://Give.org).) The BBB Accredited Charity Seal is a symbol of our adherence to the highest standards of transparency, accountability, and ethical practices.

As we wind down 2023 and move into 2024, let us carry forward the spirit of unity, empowerment, and progress. Together, we illuminate the path toward a more inclusive future where potential knows no bounds.

Wishing you a joyful holiday season!



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



Donald S. Wood, PhD





### Devoted MDA Summer Camp Volunteers: A Lifelong Commitment

Volunteers play a pivotal role in advancing the MDA mission, leaving a profound impact on both the world at large and our ability to offer support and opportunities to the MDA community. Among these volunteers, there are those who have steadfastly devoted decades to enhancing the experiences of children and young adults at MDA Summer Camp. Allow us to introduce you to just three of these remarkable individuals: Marie, Kathy, and John.

#### Marie Clavenna, Michigan

Finishing up her 46th year of volunteering at MDA Summer Camp, Marie says her one word to describe camp is “soul-altering.”

#### John Remillard, Pennsylvania

John’s first-time volunteer role at MDA Summer Camp was 21 years ago. If he was to sum up camp in just one word, his would be “enriching.”

#### Kathy Weibel, Indiana

For over 20 years, Kathy has been volunteering for “the best week of the year” at camp locations in Indiana. Her one word to describe camp is “unconditional.”



### Golf Classic’s Best Year Yet

The 19th Annual Edgar Martinez Golf Classic presented by QFC & Liberty Mutual had its best year yet! An incredible day with 65+ sponsors, 40 volunteers, & appearances from Hall of Fame baseball player, Edgar Martinez, & MDA-funded researcher, Dr. Jeff Chamberlain, University of Washington.

### The Hoskins Roll Up Their Sleeves

Philadelphia Phillies First Baseman Rhys and wife, Jayme, volunteered at MDA Summer Camp in Philadelphia, getting messy with campers and counselors during Messtival. Campers enjoyed TastyKake treats and were gifted custom MDA/New Era Caps.

### Acosta Group Adds Magic to MDA Summer Camp

Long-time sponsor added to the magic of the camp talent show in Florida. Dance routines, musical performances, and skits were brought to life with extra dazzle, thanks to props made possible by Acosta’s generous donation. Thank you for your ongoing commitment to fund camps nationwide.





## It's Goal-setting Time!



**A**s 2023 comes to a close, I am working through my usual personal and professional goal-setting exercises. In my personal life, I always explore five areas and how I can become better in all of them: social life/relationships, physical health, mental health, financial health, and spirituality.

In my professional life, I am thinking about all of you and how I can better serve you. This year, through Quest Media, we introduced a quarterly product guide ([MDAQuest.org/product-guide](https://mdaquest.org/product-guide)) to share goods that our MDA Ambassadors find accessible and useful in their own lives. We implemented two new columns in the magazine that focus on personal success stories from our community (page 8) and transparency from MDA and other organizations (page 13).

And we gave the microphone to our ambassadors through monthly guest blogs ([MDAQuest.org/blog](https://mdaquest.org/blog)), where they share how they are empowered in their own lives.

Now, we are thinking about 2024 and how we can make Quest Media an even more powerful hub of information, tools, and resources for you. One of my favorite things is to hear from YOU. This is the perfect time to let us know how we are doing. Please email us at [quest@mdaUSA.org](mailto:quest@mdaUSA.org) and tell us what we are doing well, what you would like to see more of, and how we can improve.

Sending you and your families love this holiday season, and looking forward to spending 2024 with you.

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

### ICONS TO WATCH FOR THROUGHOUT QUEST



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#### DID YOU KNOW?

**NEW GENE THERAPIES CAN BE EXCITING ... AND OVERWHELMING. MDA OFFERS SUPPORT AND EDUCATION.**

MDA launched the Gene Therapy Support Network to help the neuromuscular disease community navigate this new landscape. The network shares resources with the community and facilitates clinical best practice sharing throughout the MDA Care Center Network.

Contact the MDA Gene Therapy Support Team at **833-275-6321** or email [ResourceCenter@mdaUSA.org](mailto:ResourceCenter@mdaUSA.org). Learn more at [mda.org/GeneTherapySupport](https://mda.org/GeneTherapySupport).

**LEARN SOMETHING NEW ABOUT OUR COMMUNITY IN 60 MINUTES OR LESS.**

Quest Podcast host (and editor-in-chief of this magazine) Mindy Henderson strives to demystify, educate, and inspire in every episode. Don't miss her stimulating conversations with experts and influencers in our community at [MDAQuest.org/podcast](https://mdaquest.org/podcast).

**Top 5** downloaded podcasts in 2023:

1. The Changing Landscape of Neuromuscular Care (Episode 27)
2. Expert Accessible Travel Tips for Your Next Vacation (Episode 29)
3. ALS Podcast with Brooke Eby (Episode 31)
4. There Is No Such Thing as Normal (Episode 28)
5. New Year, New Mindset (Episode 25)



## Fortify is a Phase 3 clinical trial evaluating if an investigational oral therapy (BBP-418) is safe and effective for treating Limb Girdle Muscular Dystrophy type 2I / R9, FKRP-related (LGMD2I/R9)



### About Fortify

Fortify is a 36 month randomized, double-blind, placebo-controlled trial for individuals with genetically confirmed LGMD2I/R9 measuring patient response to treatment by measuring both biomarkers and clinical assessments. For every three study participants, two will receive BBP-418 and one will receive placebo.

### About the Therapy

In patients with LGMD2I, the enzyme FKRP does not work properly. FKRP is responsible for a critical step in a process called “glycosylation”, whereby a crucial string of sugars are added to alpha dystroglycan ( $\alpha$ -DG). Without this string,  $\alpha$ -DG does not work correctly in its role as a “shock absorber” for muscle fibers. BBP-418’s theoretical mechanism of action supplements the FKRP enzyme by adding more of the molecule that FKRP normally reacts with to drive residual activity of FKRP and helping it to stabilize muscle cells and act as a shock absorber. BBP-418 is an investigational therapy and is not yet approved by any health authorities for the treatment of LGMD2I/R9.

### Who Can Participate

You may be eligible to participate in Fortify if you:

- Have a genetically confirmed diagnosis of LGMD2I/R9
- Are 12 to 60 years of age
- Have not used ribose or systemic corticosteroids prescribed for the treatment of LGMD or other investigational therapies for the treatment of LGMD within 90 days of screening

There are other requirements to participate in Fortify. A physician or study team member will help determine if you are eligible to participate and if this study is a good fit for you. Speak with your physician about your ability to participate in Fortify.

### Fortify Locations:

The trial will be conducted at clinical sites in the United States and Europe.

Additional information about our BBP-418 study is available at [www.clinicaltrials.gov](http://www.clinicaltrials.gov) and at [www.mlbsolutions.com](http://www.mlbsolutions.com).

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# Success Is Possible

*Abhiram Srivatsa, DO, turns his disability into a strength*

BY REBECCA HUME

In his first year of residency as a psychiatrist, Abhiram Srivatsa, DO, is using his gift of being both a patient and a physician to show others that their disability does not define them. Diagnosed with Emery-Dreifuss muscular dystrophy (EDMD) at the age of 3, Dr. Srivatsa practices medicine with a unique level of empathy and understanding.

“That duality helps me to get seated at the patient’s eye level and tell them that their diagnosis doesn’t define them, and it doesn’t define their life’s trajectory,” says Dr. Srivatsa, a resident at Gateway Behavioral Health in Savannah, Georgia. “My life experience and where I have taken my goals so far helps me to drive that home.”

Dr. Srivatsa always knew he wanted to be a doctor. He chose psychiatry because he believes it offers career longevity and accessibility, especially with the increased popularity of telehealth. “Regardless of how my EDMD progresses, I have a career and a path to success,” he says.

## Expressing himself

Dr. Srivatsa has difficulty using stairs, walking distances, and standing for long periods of time. His disability is not



Abhiram Srivatsa graduated from Augusta University in Georgia with a bachelor’s degree in cell and molecular biology.

always visible to others, and he has learned to be open about the accommodations he needs.

In elementary and high school, he found his schools were willing to work with him when he explained his needs. For example, his high school assigned the centralized nurse’s office as his homeroom and reassigned his two best friends so he would have people to walk with. He got elevator privileges and permission to replace physical education with an additional music class.

Dr. Srivatsa is passionate about music and beatboxing and started an a cappella group with fellow students in his Sanskrit class. “The Sanskrit Boys” found viral success on YouTube and were invited to perform at the Library of Congress in Washington, DC. This experience, along with

Dr. Srivatsa chose psychiatry because he believes it offers career longevity and accessibility, especially with the increased popularity of telehealth.



Abhiram is flanked by his parents, Bharath Srivatsa, MD (left), and Sumathi Srivatsa, MD (right), at the start of his medical school training.



During the residency application process, Dr. Srivatsa decided that disclosing his disability was an opportunity to control the narrative.

participating in the band, chorus, debate team, and math team, helped Dr. Srivatsa gain confidence in discussing his disability.

“It is important to tell people that this is who I am, this is what I need, and this is how I need it in order to do the best that I can do,” he says.

During medical school at the Philadelphia College of Osteopathic Medicine’s campus in Suwanee, Georgia (PCOM Georgia), medical rotations between different hospitals required special considerations. PCOM Georgia notified each clinical faculty team that Dr. Srivatsa would need the opportunity to sit down, avoid walking for long distances, and take a little extra travel time.

When it came time to graduate, PCOM Georgia initially offered Dr. Srivatsa an alternate route to the stage. “But I wanted to be with everyone that I had experienced challenges and accomplishments with during those four years of medical school,” Dr. Srivatsa says. “I wanted to walk with my peers, not separately from them.” The school built a large ramp for the entire graduating class to walk to the stage together.

After graduating, Dr. Srivatsa met with his residency program director and learned that she had already spoken to his rotation sites and ensured that everything was accessible, with close parking and elevator access. “The people in charge at my job have gone above and beyond to be aware and accepting of my needs. Their support helps me to prove to myself that I can do this,” he says. “I bring a lot to the table; I just need to get to the table.”

### Finding his voice

During the residency application process, Dr. Srivatsa debated about disclosing his disability. He reached out to advocacy groups, disability rights groups, and MDA



At his medical school graduation, he officially became Abhiram Srivatsa, DO.

for guidance and ultimately decided that disclosing his disability was an opportunity to control the narrative.

“It ended up being a really good decision to disclose,” he says. “Every interviewer felt comfortable asking how disability impacts who I am as a physician. And I was able to tell them, ‘It’s not that I am going to be a physician, but I am limited by my disability — it is because of my disability that I am going to be a fantastic physician.’”

Finding his voice and learning how to talk openly about his disability has been empowering. “Your disability does not define you, but it can strengthen you as you pursue your dreams,” Dr. Srivatsa says. “At any stage in your life, don’t sell yourself short because of your diagnosis. There are different avenues to pursue — you just have to shape and mold the world around you to do what you want to do. In this day and age, it is increasingly possible to be successful. And that success is whatever success means to you.” [Q](#)

*“That duality helps me to get seated at the patient’s eye level and tell them that their diagnosis doesn’t define them, and it doesn’t define their life’s trajectory.”*  
— Abhiram Srivatsa, DO



### INCLUSION MATTERS

Listen to a discussion about advocacy and inclusion with Madison Lawson, aka Wheelchair Barbie, a journalist, model, and disability rights activist, at [MDAQuest.org/podcast/inclusion](https://MDAQuest.org/podcast/inclusion).

Rebecca Hume is Senior Specialist and Writer for Quest Media.



## VYVGART® Hytrulo

(efgartigimod alfa and  
hyaluronidase-qvfc)

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

*Picture your life in motion*

# GENERALIZED MYASTHENIA GRAVIS

*isn't canceling this  
weekend getaway*

### IMPORTANT SAFETY INFORMATION

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

VYVGART HYTRULO may cause serious side effects, including:

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

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

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

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

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

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



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

VYVGART Hytrulo usually takes

## 30 to 90 seconds to inject

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

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

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



## Learn about the effectiveness and safety of VYVGART Hytrulo



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

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

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

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

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

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

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

### What is VYVGART HYTRULO?

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

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

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

**Please see brief summary on next page.**

**argenx** 

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US-ESC-22-00021 V1 06/2023

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







# Inclusive Trailblazers

*Recognizing three companies that prioritize accessible and adaptive products*

Companies are getting wise to the fact that 1 in 4 US adults lives with a disability. They are finally realizing that increasing inclusivity makes sense from ethical and business standpoints. Many are answering the call for products and features that more people can use and enjoy.

To celebrate this progress, we are spotlighting three companies leading the way in accessibility and inclusion.

## **QVC: Creating more accessible products**

Over the last few years, the video commerce retailer QVC has made great strides in promoting inclusivity and accessibility. Actor and activist Selma Blair, who lives with multiple sclerosis, joined QVC as their brand ambassador for accessibility earlier this year, and the company developed a dedicated accessible and adaptive product line and launched the monthly on-air show “Accessible Living.”

Now, people living with disabilities; aging in place or in assisted living; recovering from surgery, illness, or injury; or supporting others as a caregiver can find everything from adaptive clothing to housewares to mobility aids.

Inclusivity has long been part of QVC’s philosophy. For more than 30 years, the company has been a leader in size inclusivity, offering all apparel options at price parity. About five years ago, the QVC team started discussing ways to make their offerings even more universal.

“We started talking about how we have the perfect medium and platform to serve the disability community



in a different way,” says Rachel Ungaro, Vice President and General Merchandise Manager at QVC. “With our live broadcast and ability to tell stories — 100% live and unscripted — we felt like it was something we wanted to venture into. We wanted to change the narrative for a broader community.”

The company created a cross-functional internal task force and began conducting research and consulting with experts and vendors. Then, they began offering some of the pieces online.

“We started seeing interest and felt like the best way for us to continue this journey was to offer adaptive, accessible, and universal items from our own brands to the customer,” Rachel says. So, they developed and recently launched Denim & Co. Adaptive, the first accessible collection from a QVC private-label fashion brand. Rachel says shoppers can expect to see new items released monthly.

Apparel options include pants with loops or larger leg openings for easy dressing. Those with dexterity issues can find clothing with magnets, Velcro, or zippers with pull rings. “It’s much easier to grab a loop than it is to grab the actual zipper edge, and we offer it in metal or other kinds of fabrication so it looks like a fashion detail versus a utilitarian detail,” Ungaro says. “We are trying many different things, and we love feedback.”



QVC’s Denim & Co. Adaptive line, pictured here, releases new items monthly.

Image: iStock.com/fkuvshinov



Outside of apparel, QVC sells the EV Rider mobility scooter, hearing aids, home safety items, canes, kitchen tools and gadgets, and compression socks and vests.

### Lessons from the journey

Rachel says the early feedback has been positive. “We might find out the first version of a product didn’t have something the customer was looking for, and then we can add it in the future,” she says.

Authenticity is also in QVC’s DNA, and Rachel says it has guided them throughout the process.

“A brand owner, inventor, or designer is selling their product to the customer live on any number of our platforms from on-air to digital or streaming. They’re authentically telling their story. Authenticity runs through our veins because of the type of business that we do.”

Venturing into the world of adaptable and accessible goods was the next logical step.

“It’s the most important thing I’ve done in my career,” she says. “It just feels right.”



Rachel Ungaro, QVC

### Slick Chicks: Offering a fully adaptive clothing line

Adaptive clothing company Slick Chicks was created to empower the disability community with fashionable and functional apparel.

Founder Helya Mohammadian got the idea for Slick Chicks when her sister was recovering from a C-section. Seeing the struggle involved in daily tasks like putting on underwear, Helya, who studied product development at the Fashion Institute of Technology, wanted to find a solution. So, she launched a Kickstarter campaign to develop adaptable panties. With all of the interest she got from the disability community, she immediately saw that there was a much bigger need.

Since then, the Slick Chicks team has grown to five employees who wear many hats and are passionate about what they do. The success of the panties has led to an expanded product line, including sweatshirts, hoodies, lounge pants, shorts, and bras. The team is also hard at work on a men’s line. Slick Chicks products are available on SlickChicksOnline.com and at Target, QVC, JC Penney, Aerie, Nordstrom, Lane Bryant, Zappos, CVS, Third Love, and Kohl’s.

### Clothes for everyone

Rather than trying to cater to one type of disability, Slick Chicks aims to make their products as universally accessible



Slick Chicks has expanded to offer adaptive loungewear.

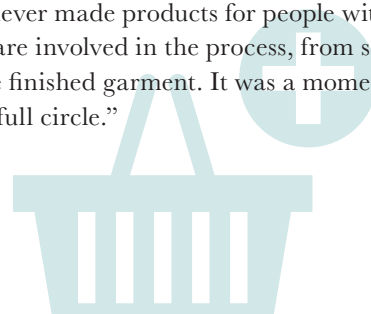
as possible, from the right design details to affordable price points. For example, they aim to make tagless garments, depending on the fabric. As for fasteners, it’s a tougher mission to meet as many needs as possible, but one they are dedicated to. “When we used a hook, not everybody had the dexterity to use it, so we started working with Velcro,” Helya says. “Some people experienced sensory issues and skin allergies with Velcro, so we placed the Velcro where it wouldn’t be touching the skin. Then we changed to magnets, but some people were uncomfortable with the idea of magnets touching their skin. So then we switched to zippers. Now we’re working on a self-fastening fabric.”

Helya loves getting community feedback to inspire these tweaks. “I’m very open to suggestions and feedback from our community. We don’t always hit the mark, but we definitely try to evolve,” she says.

Slick Chick’s inclusive approach extends to its production process. They found a manufacturing partner in Sri Lanka that has similar values and employs about 380 people with disabilities. “Making the garments was a learning experience for both of us,” Helya says. “They employ so many people with disabilities, but they had never made products for people with disabilities. Now, they are involved in the process, from sourcing the fabric to the finished garment. It was a moment that brought us both full circle.”



Helya Mohammadian, Slick Chicks





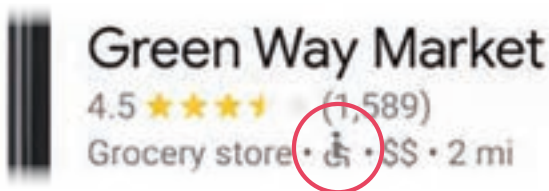
## Fueling momentum

As mainstream retailers are starting to introduce accessible and adaptive products, Helya believes brands like hers can educate them. “It’s a big responsibility because they’re not going to know the space and the disability community the way we do,” she says. “We have to make sure the products are showcased in a way that is thoughtful and respectful to the community and that people can actually find them. Anything that we learn, we share with our retailers because it’s a win-win.”

## Google: Developing products for everyone

When it comes to inclusive software development, technology powerhouse Google is leading the way, guided by its mission “to organize the world’s information and make it universally accessible and useful.”

The company has been busy creating accessible features, apps, and products. Of the company’s extensive innovations, here are some exciting examples:



### Accessibility icon on Google Maps

Google has released a Google Maps feature that identifies places that are wheelchair accessible — and those that aren’t.

When a business has a wheelchair icon next to it, that indicates it has a wheelchair-accessible entrance. If a business doesn’t have one, the icon will have a slash through it. You can also check out the “About” tab for information on accessible seating, parking, or restrooms and a place to add missing data. The information is gathered through crowd-sourcing from local guides and businesses who contribute their information. “We know that planning outings can be a challenge, and we hope this will make it easier,” says Lisie Lillianfeld, Product Manager, Products for All, at Google.

### Project Relate

People with nonstandard speech can train this app to understand them by recording 500 phrases. Once the app has processed these phrases, the user chooses between three modes:

- Listen transcribes what they say in real time.
- Repeat restates what they said in a clear, synthesized voice.

- Assistant connects to Google Assistant to respond to their query or request.

“This is a big step toward making speech recognition that works for more people,” Lisie says. You can learn more about the app at [g.co/ProjectRelate](https://g.co/ProjectRelate).

### Project Activate

This communication app is designed for people who cannot speak or use a phone with their hands. With Project Activate, users assign specific gestures — such as smiling, raising eyebrows, opening their mouth, or looking left, right, or up — to specific actions — such as sending a text message, speaking a phrase, or playing an audio file. The app uses the phone’s camera to detect the gesture to trigger the action and can be downloaded from the Google Play Store.


“We worked with people with ALS and others with profound paralysis for over a year to develop this app,” Lisie says.

### Success for all

For companies beginning their journey to inclusivity, the key to creating a useful product is to know your users. Lisie recommends speaking to many different people to get a more representative sample. “We ask, ‘What’s working?’ and ‘What’s not working?’ to understand their challenges,” she says. “Then we look at the technology and see if there is an opportunity to make an impact.”

For example, when developing Project Relate, the team met with people living with muscular dystrophy and ALS. “We wanted to know, ‘What are the frustrations and the challenges?’ We heard that communication was a huge challenge. Machine learning was just getting to the point that it could start understanding a wider variety of speech. It was that beautiful match of the user need and the technological innovation that made us feel like we could move the needle.”

Lisie adds that prioritizing accessibility often drives more innovation. She feels that as more people begin designing and creating with disability inclusion in mind, it will become the norm, with designers routinely considering visual, auditory, physical, and cognitive accessibility.

“We believe that it’s important for everybody to have access to technology,” she says. “We believe this is an equitable thing to do and the right thing to do.” 



### MORE ONLINE

Read an expanded version of this article at [MDAQuest.org/inclusive-trailblazers](https://MDAQuest.org/inclusive-trailblazers).



Lisie Lillianfeld,  
Google

# Progress Now

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

## New approvals

### FDA Approves Treatment for gMG

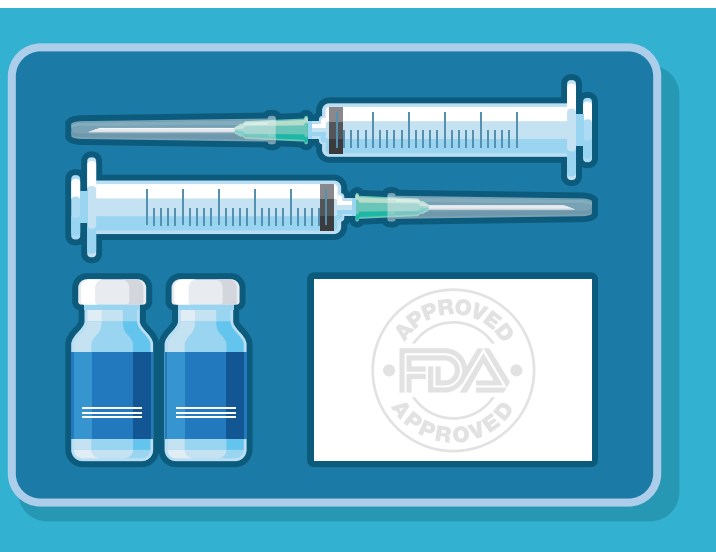


Image: iStock.com/KrizzDaPaul

The US Food and Drug Administration (FDA) approved rozanolixizumab-noli (RYSTIGGO) for the treatment of generalized myasthenia gravis (gMG) in adult patients who are anti-acetylcholine receptor (AChR) or anti-muscle-specific tyrosine kinase (MuSK) antibody-positive (ab+).

RYSTIGGO is the only FDA-approved treatment in adults for anti-AChR and anti-MuSK ab+ gMG, the two most common subtypes of gMG. It will be administered as an injection for subcutaneous (under the skin) infusion.

MG is a rare and chronic autoimmune disease in which immunoglobulin G (IgG) autoantibodies

disrupt communication between nerves and muscles, causing debilitating and potentially life-threatening muscle weakness.

RYSTIGGO works by binding to the neonatal Fc receptor (FcRN) (a receptor that prevents the breakdown of IgG), resulting in the reduction of circulating IgG. RYSTIGGO is designed to block the interaction of FcRN and IgG, accelerating antibody breakdown and reducing pathogenic IgG autoantibody concentration. Although treatment with RYSTIGGO will not cure gMG, it could lead to functional improvements in the daily life of individuals with gMG.

The FDA based its decision to approve RYSTIGGO on the positive results of the phase 3 MycarinG study, which showed treatment with rozanolixizumab-noli resulted in statistically significant improvements in gMG-specific outcomes, including everyday activities such as breathing, talking, swallowing, and rising from a chair.

In the past six years, the FDA has approved four therapies to treat adults living with anti-AChR ab+ gMG. They include eculizumab (Soliris), efgartigimod (Vyvgart), ravulizumab (Ultromiris), and recently approved efgartigimod alfa and hyaluronidase-qvfc (Vyvgart Hytrulo).

**To learn more about the phase 3 MycarinG study, visit [ClinicalTrials.gov](https://clinicaltrials.gov) and enter NCT03971422 in the search box.**

### FDA Approves Pompe Disease Treatment

In September, the US Food and Drug Administration (FDA) approved cipaglucosidase alfa-atga (Pombiliti) plus miglustat (Opfolda) for adults living with late-onset Pompe disease (LOPD). This two-component therapy is indicated for adults weighing at least 40 kg (88.18 lbs.) who are not improving on their current enzyme replacement therapy (ERT). Pombiliti + Opfolda was granted



## CLINICAL TRIAL TERMS TO KNOW

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

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

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

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

breakthrough designation from the FDA in 2019 and has now been approved as the first and only two-component therapy for eligible adults with LOPD. Pompe disease is caused by mutations in a gene that makes the acid alpha-glucosidase (GAA) enzyme, which aids in the breakdown of glycogen, a sugar used for energy. This causes glycogen to build up and damage muscle cells. This therapy combines Pombiliti, a long-term ERT, with Opfolda, an oral enzyme stabilizer. Pombiliti is a unique enzyme with optimized carbohydrate

structures designed to enhance uptake into muscle cells. Once in a cell, Pombiliti can be processed into its most active form to break down glycogen. Opfolda helps stabilize the enzyme and minimize the loss of enzyme activity in the blood.

During treatment, Opfolda is administered orally in a 65 mg capsule approximately one hour before the four-hour intravenous (IV) infusion of Pombiliti. The treatment is administered every other week.

The FDA based its approval on clinical data from the pivotal phase 3 study (PROPEL). The 52-week, double-blind study assessed the efficacy, safety, and tolerability of Pombiliti + Opfolda compared to the current standard of care. Findings from PROPEL showed the therapy led to significant improvements in muscle strength, pulmonary and motor function, patient-reported outcomes, and biomarkers.

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For more information about the phase 3 PROPEL trial, visit [ClinicalTrials.gov](https://ClinicalTrials.gov) and enter NCT03729362 in the search box.



Image: iStock.com/Atefanamer

## Becker muscular dystrophy (BMD)

### Natural History Study in Males

Researchers at the University of Florida are seeking males between the ages of 18-62 years old living with BMD to participate in a natural history study. This study is observational and does not involve an intervention. The objective is to better understand the course of BMD and identify disease biomarkers.

Participants will complete one in-person study visit each year over the course of the three-year study period. During these visits, participants will be evaluated using tests such as magnetic resonance imaging (MRI), magnetic resonance spectroscopy (MRS), and functional tests. In addition, participants may be asked for a blood sample, a urine sample, and a one-time needle biopsy of leg muscles. Following each visit, participants will meet with the investigators by Zoom to review the collected data and analysis.

Travel is covered for participants and a caregiver if necessary.

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To see the full list of inclusion and exclusion criteria, visit [ClinicalTrials.gov](https://ClinicalTrials.gov) and type NCT01484678 in the "Other terms" search box. To learn more about the study or inquire about participation, visit [ImagingNMD.org](https://ImagingNMD.org) or contact study coordinator Claudia Senesac at 352-273-6453 or [CSenesac@php.ufl.edu](mailto:CSenesac@php.ufl.edu).

## Duchenne muscular dystrophy (DMD)

# Clinical Trial of MG Drug for DMD

A collaborative grant from MDA, CureDuchenne, and Parent Project Muscular Dystrophy (PPMD) will fund a clinical trial to test if the FDA-approved drug efgartigimod alfa-fcab (Vyvgart) could help more individuals with DMD become eligible for gene therapy.

Vyvgart is used to treat autoimmune diseases by reducing overall levels of circulating immunoglobulin G (IgG) antibodies. It has been effective in reducing pathogenic anti-acetylcholine receptor antibodies present in myasthenia gravis (MG). The study will investigate whether Vyvgart can reduce anti-adenovirus (anti-AAV) antibodies to levels compatible with the safe and effective delivery of gene therapies in DMD patients.

This clinical trial aims to suppress antibodies in individuals with DMD who are currently not eligible for gene therapy due to pre-existing anti-AAV antibodies from natural infections. The trial will also explore the potential for reducing in DMD patients anti-AAV antibodies acquired from previous participation in gene therapy trials so they can become eligible for redosing.



Image: iStock.com/noppant

This trial is an open-label, single-center, multi-arm, phase 2 study that will take place at Powell Center for Rare Disease Research and Therapy in Gainesville, Florida. Twelve DMD patients will participate in the study—six with elevated anti-AAV antibody levels before gene therapy and six with high levels after receiving gene therapy. The study will also examine the safety and efficacy of Vyvgart in boys with DMD.

Barry Byrne, MD, PhD, MDA's Chief Medical Advisor and Director of the Powell Gene Therapy Center at the University of Florida, will lead the one-year clinical study.



The study will investigate whether Vyvgart can reduce anti-AAV antibodies to levels compatible with the safe and effective delivery of gene therapies in DMD patients.

If you have questions about gene therapy, MDA's Gene Therapy Support Network can provide guidance. Learn more at [mda.org/GeneTherapySupport](https://mda.org/GeneTherapySupport).



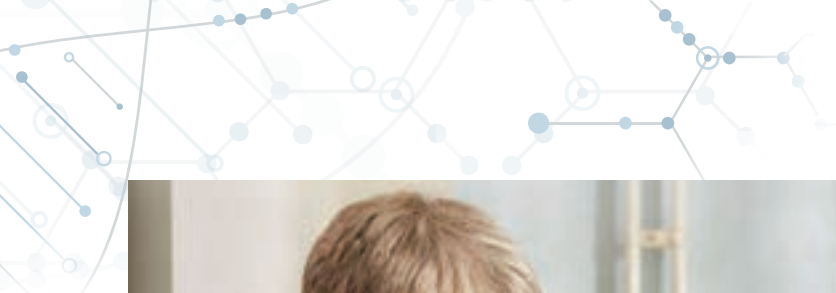


Image: iStock.com/filmstudio



# Heart Disease Drug Study

Researchers at Cumberland Pharmaceuticals Inc. are seeking boys and men living with DMD to participate in a phase 2 clinical trial to evaluate the safety and efficacy of oral ifetroban to treat the heart disease associated with DMD. Oral ifetroban is being evaluated for its ability to protect the heart from scarring and maintain normal heart function in people with DMD.

This is a multicenter, randomized, double-blind, placebo-controlled, 12-month study with an optional open-label extension. This means that participants will be randomly assigned to either receive low-dose or high-dose ifetroban or an inactive placebo control over the course of the study. Each treatment group will include eight participants with early-stage (defined as left ventricular ejection fraction (LVEF) of >45%) DMD heart disease and eight participants with advanced-stage (defined as LVEF 35-45%) DMD heart disease.

## ADVERTISEMENT

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Oral ifetroban is being evaluated for its ability to protect the heart from scarring and maintain normal heart function in people with DMD.

The drug or placebo will be administered once daily as a dime-sized oral capsule. The effect of oral ifetroban will be evaluated at each of three doctor visits during the study period using five tests: cardiac MRI, pulmonary function test, quantitative muscle

strength test, measurement of

daily activity for seven days using a wrist-worn actigraph, and quality-of-life questionnaires. A finger-stick test will be performed at the center and at home to detect ifetroban and its metabolite levels. All participants who complete the 12-month study will have the opportunity to receive ifetroban in an open-label period.

Travel support and a participant stipend will be provided. Criteria to be eligible include:

- Being a male 7 years of age or older
- Having a diagnosis of DMD
- Being on a stable dose of oral corticosteroids for at least eight weeks OR no corticosteroids for at least 30 days (FDA-approved exon-skipping therapies are permitted)
- Having stable cardiac function, including those on stable cardiac medications

To see the full list of inclusion and exclusion criteria, visit [ClinicalTrials.gov](https://ClinicalTrials.gov) and type NCT03340675 in the “Other terms” search box. To learn more about the study or inquire about participation, visit [FightDMDTrial.com](https://FightDMDTrial.com) or contact study coordinator Ingrid Anderson, PhD, at 615-627-4121 or [IAnderson@CumberlandPharma.com](mailto:IAnderson@CumberlandPharma.com).

## Working Caregivers Survey

Researchers at Rice University are seeking working parents of children living with DMD to participate in a survey about their experiences. The goal is to evaluate whether organizational resources could improve the well-being of employed caregivers of children with disabilities.

This study does not involve a new intervention. Enrolled participants will be asked to complete two surveys expected to take a total of 45 minutes. The surveys include questions about home caregiving and work experiences.

To be eligible for the survey, individuals must:

- Be working parents (not self-employed or unemployed) who are caregivers of children with DMD
- Be over 18 years old, with affected children younger than 18 years old

To learn more, email the study coordinator at [fyw1@rice.edu](mailto:fyw1@rice.edu).



Image: iStock.com/Milko



## Spinal-bulbar muscular atrophy

# Oral Drug Study Starts New Phase

Avenue Therapeutics Inc. announced the start of a phase 1b/2a clinical trial of AJ201 for the treatment of SBMA, also known as Kennedy's Disease.

SBMA is estimated to affect 1 in 6,887 males and is caused by a genetic defect on the X chromosome. The defect is an expanded section of DNA, called a trinucleotide repeat, in the *AR* gene, which carries instructions for the androgen receptor (AR) protein.

The 12-week, multicenter, randomized, double-blind, placebo-controlled trial expects to enroll 24 participants. The trial aims to assess the safety and tolerability of AJ201 in people with SBMA. The trial will also explore how the oral drug could

reduce the accumulation of abnormal AR aggregates to decrease neuroinflammation, protect cells from oxidative stress, and improve clinical outcomes.

Data from phase 1 of the clinical trial showed the drug was safe in healthy individuals. Preclinical data in a mouse model showed improvement in motor function and a decline in AR aggregates, among other results. Early results from the 1b/2a study are expected in 2024. Enrollment in the trial is expected to be complete by late 2023 or early 2024.

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For more information, visit [ClinicalTrials.gov](https://ClinicalTrials.gov) and enter NCT05517603 in the "Other terms" search box.

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### +RECRUITING CLINICAL TRIALS

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



## Amyotrophic lateral sclerosis (ALS)

# Tofersen Study in Presymptomatic Adults

Researchers at Biogen are seeking adults with a mutation in the superoxide dismutase 1 (*SOD1*) gene who are not yet experiencing ALS symptoms to participate in a phase 3 clinical trial (ATLAS) to evaluate the safety, effectiveness, and pharmacological properties of the investigational therapy tofersen.

In April 2023, tofersen (Qalsody) received accelerated marketing approval for the treatment of ALS associated with a mutation in the *SOD1* gene. The ATLAS study tests the ability of Qalsody to delay the onset or progression of ALS in people at risk of developing the disease who have not yet shown any symptoms.

This is a multi-part study. The sponsor is currently recruiting for part A, in which participants will not receive Qalsody but will be monitored to establish the natural history of their condition. The study team will evaluate blood samples from monthly home visits for levels of a blood biomarker called neurofilament light chain (NfL). If a participant's NfL level is above a certain threshold, they can be considered for part B of the study, in which they may receive tofersen as part of a placebo-controlled trial period.

Part A may last up to four years and three months and require two to three clinic visits, as well as monthly home visits. Participants who continue to part B will receive Qalsody or

a placebo via lumbar puncture in the spine. The effects of Qalsody will be evaluated using several tests and procedures, which may include blood samples, urine samples, vital signs,

health questionnaires, electrocardiogram, lumbar puncture, electromyography, neurological exam, physical exam, and clinical function assessments.

Travel assistance and reimbursement for study-related expenses may be available. Criteria to be eligible include:

- Being 18 or older
- Having a documented disease-causing mutation in the *SOD1* gene
- Being clinically presymptomatic for ALS

To see the full list of inclusion and exclusion criteria, visit [ClinicalTrials.gov](https://ClinicalTrials.gov) and type NCT04856982 in the "Other terms" search box. To learn more about the study or inquire about participation, visit [ALSatlasStudy.com](https://ALSatlasStudy.com) or contact study coordinator Gigi van den Hoef at 613-227-3170 or [gigi.vandehoef@iqvia.com](mailto:gigi.vandehoef@iqvia.com).



Image: iStock.com/Evgenii Kovalev

### +MDA CLINICAL & SCIENTIFIC CONFERENCE

MDA members can attend the 2024 conference virtually for free March 3-6. Learn more at

[MDAConference.org](https://MDAConference.org).

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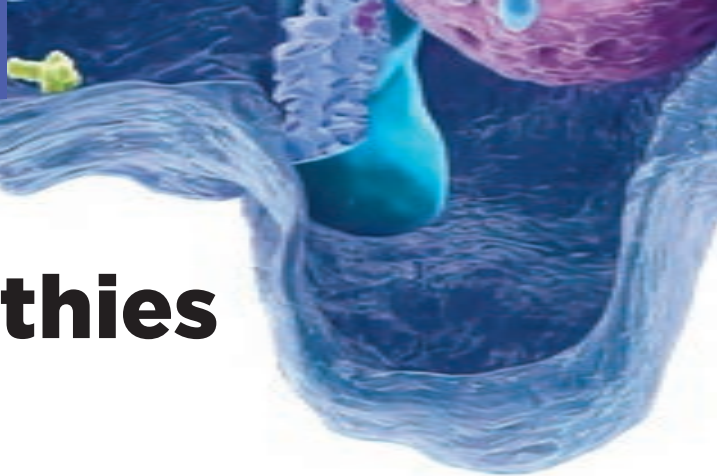


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# Understanding Metabolic Myopathies

A Q&A with Mark Terrelonge, MD

**M**etabolic myopathies are a general category of disorders characterized by muscle cells that don't function properly, leading to muscle weakness.

“Metabolism” is the over-arching term for how cells use chemical processes to convert substances, like carbohydrates, fats, and proteins, into energy. When one or more of those processes breaks down, the cell can't do its job as effectively.

In muscle cells, there are several ways that metabolism can fail. They generally fall into three categories: failure to process carbohydrates, failure to process fats, and a failure in the mitochondria — the cell's “powerhouse” that is responsible for generating energy. These failures occur because a critical enzyme is either missing or too low.

We spoke with Mark Terrelonge, MD, Assistant Professor of Neurology at the University of California at San Francisco Weill Institute for Neuroscience, to better understand metabolic myopathies.

## What are metabolic myopathies?

Whenever things affect the metabolism of the muscle, they usually affect how the muscle can deal with energy. Energy could come from different sources. In metabolic myopathies, there are issues with the machinery responsible for turning those different energy sources into the actual energy.

## What conditions fall under this umbrella?

Different conditions are labeled based on the type of energy source that is inhibited or isn't working. There are disorders of glycogen metabolism (such as Pompe disease or myophosphorylase deficiency, also known as McArdle's Disease), which have to do with how glycogen is stored and turned into energy. There are disorders of lipid metabolism, which have to do with the way that fatty acids are turned into energy. Carnitine palmitoyltransferase deficiency is a relatively common example of lipid metabolism disorder.

Mitochondrial diseases are not necessarily specific to muscle — they normally have multisystem manifestations. So, while many of the enzymes described in the other diseases are muscle-specific, enzymatic changes in the mitochondria can cause muscle disease and issues with the nerves, brain, liver or other body parts. For example, one

mitochondrial disease, Kearns-Sayre syndrome, is specific to muscle cell mitochondria. But other mitochondrial disorders can cause neuropathy, ataxia, or retinitis pigmentosa. (See sidebar for a list of metabolic myopathies.)

## What are the symptoms of metabolic myopathies?

Usually, it shows as muscle weakness.

One of the most frequent presentations is exercise intolerance — when someone has to exert themselves more than they would for normal daily movement, such as walking.

This activity stresses their system and causes energy failure, which can lead to either weakness or, in some cases, issues with having dark urine or other problems. Some people will say that they just feel “generalized fatigue.” Others will feel that their muscles are actually failing. And though it is rare, some people's breathing is affected.



Mark Terrelonge, MD

## What causes these conditions?

Generally, these tend to have a genetic basis, but some may be acquired. Most metabolic diseases have a recessive pattern, meaning a person must inherit two copies of the flawed gene — one from each parent. But some metabolic diseases may be autosomal dominant, which means a person has to inherit a flawed gene from only one parent to have disease symptoms. There are no risk factors for those who don't have a hereditary basis — the disease arises spontaneously.

## When are they typically diagnosed?

Frequently, people are diagnosed when they're children, but many types first manifest in the teenage or adult years. It could be that until that age, some people haven't had a sufficient challenge that has caused the muscle weakness to become apparent. Or for some of the mitochondrial diseases, more mitochondria can become involved over time, so an affected person hits a point where their typical activities become harder.



Infections or changes in eating habits, such as fasting, also can cause the symptoms to present for the first time. For example, somebody may have a viral illness, feeling weak or fatigued, and they think that it's the virus when really the metabolic disease has reached a tipping point in terms of how much stress their body could take. This can happen for those with either the genetically based illness or one they acquired.

**How does a metabolic myopathy affect the people who live with it? What are some of the specific challenges they live with?**

Depending on the specific type of metabolic changes a patient has, a patient may feel fine between bouts of exercise, but when they exercise, it leads to cramps, muscle pain, or dark urine. Other patients may have more fixed muscle weakness, leading to difficulty participating in activities of daily living. Patients with mitochondrial diseases may have multisystem involvement that can cause changes in vision, cognition, gastrointestinal transit, or hearing, among other things.

**Are there dietary or lifestyle modifications that could benefit those living with it?**

Depending on the kind of disorder — which is why it's important to identify a specific type — we have strategies for supplements or dietary modifications in addition to medications.

For example, with Pompe disease, the patient can be given enzymes to help with some of its manifestations, such as respiratory changes.

For somebody with a glycogen storage disease, when they choose to eat carbohydrates in relationship to when they're exercising or exerting themselves can have major impacts on how they manifest weakness. The same thing is true for those with lipid disorders. For others, having frequent small meals with fast-acting carbohydrates might be helpful, while others may need more long-acting carbs; it really depends on the specific disease.

**What is the current care plan for people living with metabolic myopathies? How does care change as the disease progresses?**

Generally, the best care plan is avoidance of the stressors that make people feel worse, such as over-exercising or long stretches of exercising. Avoiding fasting can be very, very important. Unless they have a myopathy that can affect the heart or lungs, people can use behavioral changes to manage the disease and not need additional mobility devices in the long term. For those with conditions that lead to progressive weakness, such as Pompe disease, some of those

**10 COMMON METABOLIC MYOPATHIES**

Here is a list of 10 common metabolic myopathies, which are distinguished by the enzyme deficiency they cause.

- > Acid maltase deficiency (AMD, Pompe disease, glycogenosis type 2, lysosomal storage disease)
- > Carnitine deficiency
- > Carnitine palmitoyl transferase deficiency (CPT deficiency)
- > Debrancher enzyme deficiency (Cori or Forbes disease, glycogenosis type 3)
- > Lactate dehydrogenase deficiency (glycogenosis type 11)
- > Myoadenylate deaminase deficiency
- > Phosphofructokinase deficiency (tarui disease, glycogenosis type 7)
- > Phosphoglycerate kinase deficiency (glycogenosis type 9)
- > Phosphoglycerate mutase deficiency (glycogenosis type 10)
- > Phosphorylase deficiency (McCardle disease, myophosphorylase deficiency, glycogenosis type 5)

To learn more about each of the disorders listed, visit [mda.org/disease/metabolic-myopathies/types](https://mda.org/disease/metabolic-myopathies/types).

patients might need a cane, walker, or wheelchair, depending on their progression. For myopathies with respiratory manifestations, though, patients might need to use noninvasive ventilators or other assistance devices like BiPAP or two-level airway pressure, for example, to help them take deeper breaths.

**Are there any recent advances in treatment or disease management?**

In September, the US Food and Drug Administration (FDA) approved cipaglucosidase alfa-atga (Pombiliti) plus miglustat (Opfolda) for adults living with late-onset Pompe disease (LOPD). For acid maltase deficiency, the standard of care treatment includes the use of alglucosidase alfa, which leads to improved outcomes in patients with infantile and adult-onset forms. A newer enzyme, avalglucosidase alfa, was approved in 2021 and has improved cellular uptake and muscle targeting.

**What treatments are on the horizon?**

There is research on supplements that may help with metabolism when patients are missing specific enzymes. Mitochondrial disease researchers are investigating peptides that may target mitochondria and change the way they work (but this is not the full spectrum of patients with these diseases). Lastly, gene therapy may be on the horizon, but no active trials are underway as of now. [G](#)



# What You Need to Know About COVID-19

*The virus is less frightening, but it's still spreading*

BY LARRY LUXNER

**W**e've been living with COVID-19 for more than three years now, and the pandemic is over, but we're still hearing news reports about surges and variants. It's tempting to tune out those reports, but there are good reasons to pay attention. While we're learning to live with COVID-19, the infectious disease is still a concern, especially for people with respiratory issues or compromised immune systems.

That's why people with neuromuscular diseases — and those who come into regular contact with them — should remain on alert, say experts who have been tracking the spread of COVID-19.

## Trends and protection

In the United States, Omicron has been the dominant variant since December 2021. The Omicron subvariant HV.1 is responsible for about 25% of new COVID-19 infections, according to data issued by the Centers for Disease Control and Prevention (CDC) in October.

In second place is the Omicron subvariant EG.5 (22% of new infections), followed by FL.1.5.1 (12% of new infections) and XBB.1.16.6 (9% of new infections).

Elizabeth McNally, MD, PhD, a cardiologist and Director of Northwestern University's Center for Genetic Medicine in Chicago, calls COVID-19 an "ever-evolving target."

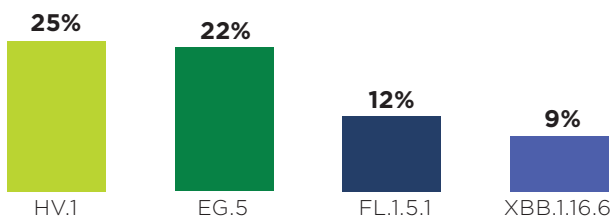


Image: iStock.com/RUSSELLTATEdotCOM

In September, the US Food and Drug Administration (FDA) approved updated COVID-19 vaccines for everyone over 6 months old, and the CDC recommends them. The revamped Pfizer and Moderna shots are aimed at a viral variant called XBB.1.5, a close relative of the other currently circulating subvariants. According to Dr. McNally, both vaccines offer good protection against serious infections.

"Just like the flu vaccine, which is also recommended, the COVID vaccine does not eliminate the chance you can get infected. But it does mean you are much less likely to get really sick from the infection," Dr. McNally says, noting this protection is especially important for the neuromuscular disease community. "A lot depends on your pulmonary condition. People with neuromuscular diseases tend to have weakness in their respiratory muscles. The best approach is to make sure you're up to date on your vaccines."

## DOMINANT SUBVARIANTS IN THE US



Source: Centers for Disease Control and Prevention, October 2023

The new COVID-19 vaccines are expected to prevent about **400,000 hospitalizations** over the next two years.



Image: iStock.com/Boba

### Winter surge

According to the CDC, 81% of Americans (and 95% of those ages 65 and older) have received an initial COVID-19 vaccine. Yet only 17% of Americans

(and 43% of those 65 and up) have received boosters. Since the beginning of the pandemic, the disease has killed more than 1.1 million Americans and led to 6.4 million hospitalizations.

Looking back, the pandemic took an enormous toll on people with neuromuscular diseases, says Nora Capocci, Vice President of Healthcare Services at MDA. “Anyone who has a compromised immune system — along with

their disease — has had their own challenges navigating COVID and the impact it’s had on their health, as well as on their personal lives,” she says. “People are learning to live with it, but there’s still so much we don’t know.”

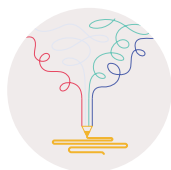
Fortunately, health officials at the state and local levels know a lot more than they did in 2020, and they’ve been preparing for the expected winter surge in respiratory illnesses for months.



Nora Capocci

### VACCINE CONCERNS

MDA’s Chief Medical Advisor weighs in on the risks and benefits of vaccines at [MDAQuest.org/addressing-vaccine-concerns](https://www.mdaquest.org/addressing-vaccine-concerns).



Studies have shown that wearing masks is a safe and effective way to prevent the spread of COVID-19. Many state health departments recommend wearing masks in public places when you can’t maintain six feet of distance from others — not only to protect yourself but also to avoid infecting others.

Social distancing, while difficult, is also an effective way to limit the spread of COVID-19 and other infections, as is frequent hand washing, seeking treatment if you feel sick, and staying away from people who are suspected or confirmed to have a virus.

Besides the updated COVID-19 vaccine and seasonal flu shot, there’s also a newly approved vaccine for respiratory syncytial virus (RSV), an illness that can be especially serious for older adults. The FDA approved the vaccine for ages 60 and older, and the CDC recommends adults in this age group talk with their doctors about getting the vaccine.

“It’s fine to get all these vaccines at the same time,” says Dr. McNally, who urges people with neuromuscular diseases to stay on top of their vaccinations, regardless of their specific disease.

### Using our knowledge

The new COVID-19 vaccines are expected to prevent about 400,000 hospitalizations and 40,000 deaths over the next two years, the American Medical Association (AMA) stated in September.

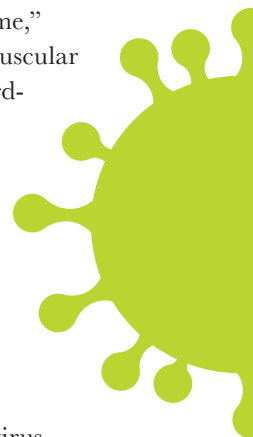
“This is the first respiratory virus season where we have vaccines against the biggest respiratory virus threats, including an updated COVID-19 vaccine, an annual flu vaccine, RSV vaccines for older adults, as well as a long-acting monoclonal antibody for infants that reduces the risk of both hospitalizations and healthcare visits for RSV,” the AMA said. “We will continue to support evidence-based vaccines and treatments to help prevent severe disease and protect public health.”

We might not be able to escape the news about COVID-19 surges, but we are in a better place than we were in 2020 — or even last winter — with more knowledge and more tools to protect ourselves from respiratory illnesses. [Q](#)

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





Elizabeth McNally, MD, PhD







# Putting the Pieces Together for Becker Muscular Dystrophy



Becker muscular dystrophy is a serious health condition, but it is often underappreciated and regarded as less significant than other related neuromuscular disorders. However, Becker has substantial negative impacts on those who live with the condition, and to fully recognize this, a dedicated effort by the neuromuscular community is needed. Let's work together to turn the Becker puzzle pieces into a detailed picture that includes comprehensive disease-specific clinical care, a robust therapeutic research pipeline, patient and family educational resources, and a global community where those affected by Becker can find support and share experiences.



## CARE



For some types of muscular dystrophy, established best practices in clinical care, often called standard of care, have led to significant improvements in overall health and quality of life for patients. An effort by all stakeholders in the neuromuscular community to establish a standard of care for Becker may also bring great results. The range of symptoms experienced by those living with Becker, including muscle weakness, mobility challenges, and cardiac and respiratory issues, could be optimally managed by teams of collaborative healthcare professionals working together at medical centers.

In an effort to improve our understanding of current gaps in care for Becker patients, during the last quarter of 2023, a US survey of people living with Becker was launched that included questions about current care as well as considerations for optimal comprehensive care. Results from the survey will be shared in the coming months with the Becker community.

## COMMUNITY



For patients living with rare diseases, sharing experiences with others who have the same condition and face similar challenges can be an invaluable way to gain knowledge and feel supported. Recent outreach efforts focused on people who live with Becker highlight that many do not feel like they have adequate opportunities to get together in person or online as a distinct community.

Getting together, whether virtually, locally, nationally, or globally, provides unique opportunities to learn more about what's going on in research, where to find healthcare providers, how to access available

## ADVERTISEMENT

### COMMUNITY CONTINUED

resources and how to navigate mobility and fatigue challenges. Additionally, sometimes it is helpful to simply be around others who understand what it means to live with Becker. In early December of 2023, a group of patients, advocates, healthcare providers, and researchers convened the first “Becker Education and Engagement Day” with the goal of starting to build a community to support anyone whose life has been touched by Becker.



## RESEARCH

Clinical research is one of the most important ways that scientists can learn and improve our understanding of diseases. Some research studies aim to understand disease progression while others study how disease progression may be modified by a therapeutic drug or device. Results from clinical research can provide much-needed information to optimize clinical care practices. Currently, multiple studies are enrolling individuals living with Becker to evaluate disease progression or to assess the effect of an investigational drug. To find out more about Becker trials currently enrolling, go to [mda.org/research/clinical-trials](https://mda.org/research/clinical-trials) or [clinicaltrials.gov](https://clinicaltrials.gov) and search for Becker.

## EDUCATION AND RESOURCES

For those who live with Becker, having Becker-specific educational information, resources, and programs is crucial to optimizing the quality of everyday life. With promising new therapies for Becker being tested, education about clinical trials will also be important. Understanding what services and supports may be available at the local, state, and national level, including Medicaid and Medicare programs, Vocational Rehabilitation services, legal services, affordable mental health counseling, financial assistance, and transportation options, can also be critical to achieving optimal quality of life with Becker. Creating a dynamic and robust Becker community which offers these resources will result in more informed individuals who are better able to advocate for themselves and receive the support and care they need. Importantly, these resources will also help move Becker out of the shadows and recognize the community's needs.

## JOIN US

Great effort is needed to build an engaged and active Becker community, but the rewards could lead to improvements in care, quality of life, and progress for the entire community. As more and more initiatives in care, community, research, and resources are implemented, sharing best practices for the community will become easier. We are getting real about Becker muscular dystrophy. We hope you will join us!



## Common Symptoms

of Becker Include:



Arm weakness



Difficulty walking that gets worse over time



Toe walking



Hard getting out of chair



Muscle pain and/or spasms



Frequent falls

## Other Symptoms

of Becker Include:



Cardiomyopathy



Fatigue



Learning Differences



Breathing Problems

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

– adult living with Becker





# Your Best Holiday



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*5 common holiday challenges  
and how to beat them*

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BY QUEST MEDIA STAFF



# Oh Season Yet!

**T**he holiday season isn't just joyful — it can also be challenging. From navigating airports to planning budgets and parties, this time of year can go from festive to frustrating fast. Fortunately, it doesn't have to. With some preparation, you can meet those holiday challenges, manage them, and still have the energy to enjoy yourself.

We spoke with experts about five common holiday-related challenges for people in the neuromuscular disease community. Here are their top tips to make this holiday season your best one ever.

### Challenge 1: Managing energy

Juggling the usual holiday expectations of gift buying, cooking, and entertaining — on top of a neuromuscular disease — can leave you feeling sapped. Pamela Glazener, OTR, ATP, an occupational and assistive technology therapist at Houston Methodist Hospital, offers tips for enjoying the holidays without being wrung out.

“The people I work with usually know that if they are very busy one day, they may not have a good day the next day because of the fatigue factor,” she says. “But when everyone is ‘go, go, go,’ it’s hard to say, ‘I need to stop and take a break.’”

Her first piece of advice: Pace yourself. “If your tank is empty by noon, take a rest so you can recharge,” she says. “If you are looking forward to a very important party, maybe avoid doing anything the day before.”



Pamela Glazener,  
OTR, ATP

When you go to holiday gatherings, Pamela advises bringing along tools that will help you do what you need to do with less effort, such as a portable ramp or a toilet lift.

She also notes that some people travel with a manual wheelchair for convenience, but this can contribute to fatigue. “Do the prep work necessary to be able to take your power chair because you’ll save energy and be more independent,” she says.

Start preparing for trips early so you’re not packing on the same day you’re traveling. “I tell clients to load the car the night before or even a few days before. It’s just less stress and less energy expense,” she says.

### Challenge 2: Travel

Getting to your holiday destination can easily become the most stressful part of the season. Travel blogger Cory Lee ([CurbFreeWithCoryLee.com](http://CurbFreeWithCoryLee.com)), who lives with spinal muscular atrophy (SMA), shared his biggest piece of advice for minimizing challenges during the busiest travel time of the year: Get there early.

This is especially important if you’re flying. “Planning your flight a day earlier than you need to be there will ensure you are prepared for flight delays and cancellations. No one wants to spend the holidays stuck at the airport,” he says.

He also emphasizes communicating with your airline. “Discuss the details of your accessibility needs so they can be documented,” he says. “If you are using a power wheelchair, give them the height, weight, and type of battery. The more the airline knows in advance, the easier your travel day will be.”

The Transportation Security Administration (TSA) also offers a free service called TSA Cares ([tsa.gov/contact-center/form/cares](http://tsa.gov/contact-center/form/cares)). This allows you to alert TSA if you need screening assistance, so you can get through airport security quickly.

Lastly, Cory says don’t over-plan activities once you’re there. Ideally, getting to your destination early will give you some time to rest after your travel. “We need to find a way to slow down enough to truly enjoy this time with our family and friends,” he says.



Cory Lee

### Challenge 3: Accessible gatherings

The joy and togetherness of the holiday season should be reflected in get-togethers that are accessible to everyone.

If you’re attending an event and you’re not sure if it will be accessible, Lauren Herringdine, Senior Director of Signature Events at MDA, advises

reaching out to the organizer to ask questions and let them know what you need.

“You are your own advocate, so don’t be shy about expressing your needs,” she says.

“We all learn from sharing, and the more vocal we can be about our needs, the more we can continue to make a comfortable environment for anybody who might come through the door.”

If you’re planning a holiday party, Lauren offers some advice: “During the planning process, walk through every single piece of the space that you will use to ensure the space is wide enough for wheelchairs and it’s accessible all the way through, from the moment guests arrive to the moment they leave,” she says.

Lauren also emphasizes paying attention to small details when setting up your space for accessibility — for example, make sure food and drink stations are at a height that is accessible for those in wheelchairs and there is ample accessible parking.



Lauren Herringdine



## GIFT-GIVING MADE EASY

Is someone with a neuromuscular disease on your gift list? The MDA Quest Media product guide is your destination for thoughtfully curated, seasonally inspired products recommended by MDA Ambassadors.

MDA Ambassadors are a mix of youths and adults living around the nation with different neuromuscular diseases. The guide includes their favorite products and descriptions of how each product helps them in their daily lives. Whether you're looking for something fun or practical, adapted for a specific need or inclusive for all, you're sure to find a gift that will enhance your loved one's life.

Find the MDA Quest Media product guide at [MDAQuest.org/product-guide](http://MDAQuest.org/product-guide).

### Challenge 4: Eating healthy

It wouldn't be the holidays without delicious treats, but all those tempting foods and drinks can make it difficult to stick to a healthy diet. So, we asked our expert: Is it possible to balance good nutrition with enjoying seasonal goodies?

Tad Campbell, Assistant Professor of Clinical Nutrition at the University of Texas Southwestern Medical Center, believes so, and he offers some tips for approaching holiday feasting sensibly:

- + **Don't skip meals.** It's tempting to skip breakfast, for example, if you're looking forward to a big family lunch. But skipping meals leads to poor food choices and overeating. "Someone with a neuromuscular disease may need to watch out for weight gain, especially if they're not as mobile as they used to be," Tad says.
- + **Eat a healthy snack before an event.** A healthy protein (such as Greek yogurt or a handful of nuts) or complex carbohydrates (such as whole-wheat bread, fruits, or veggies) will keep you from arriving hungry and overfilling your plate.
- + **Drink in moderation.** "Some medications don't interact well with alcohol," Tad says. He recommends asking your healthcare team if it's safe for you to imbibe.



Tad Campbell

When it comes to nonalcoholic drinks, it's good to stay hydrated but if toileting is a concern, keep in mind that consuming a lot of liquid will lead to more bathroom trips.

- + **Watch your portions.** If there's something that you look forward to every year, have some, but don't feel like you must have an entire serving. Maybe share a dessert so you don't go overboard.
- + **Spread out your favorite treats.** Most holiday foods freeze beautifully. Have a bit now and save some for later.

### Challenge 5: Sticking to a budget

It's difficult to enjoy the holidays when you're worried about money. Jonathan Greeson, CFP, a financial planner who lives with SMA, advises beginning holiday budget planning early in the year. "Christmas comes every year, so be proactive instead of reactive," he says. "Saving for gifts can be just like any other financial goal, such as retirement — you set a goal and follow a plan to reach that goal."

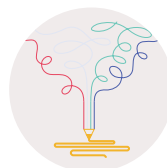
While saving for holiday expenses can take some of the stress out of the season, those in the neuromuscular disease community know it can lead to other challenges. "My greatest frustration is the savings limit faced by those of us receiving SSI or similar assistance. I know how to save and plan for my family's gifts, but I would be over my limit and face penalties," Jonathan says. "My little life hack is to pay over the balance on my credit card, then have a credit on that account for future purchases."



Jonathan Greeson, CFP

Jonathan also has a hack for gift shopping: "I really like the wish list tool on Amazon," he says. "The prices change often, so you can usually get a better deal. I scroll through my list at least once a week. Sometimes a \$25 book is marked down to \$7 one day, so patience pays off."

Finally, Jonathan cautions that, although this is the season of giving, we must accept our limits. "It's great to want to give every child in your community a gift, but it's okay if you can't," he says. "Maybe you can organize a fundraiser, so the whole community can make that dream come true. You don't have to do it all yourself." [@](#)



### SEASONAL SUPPORT

It's true: You don't have to do it all on your own. Find a dozen ways to get assistance at [MDAQuest.org/12-ways-to-ask-for-help](http://MDAQuest.org/12-ways-to-ask-for-help).

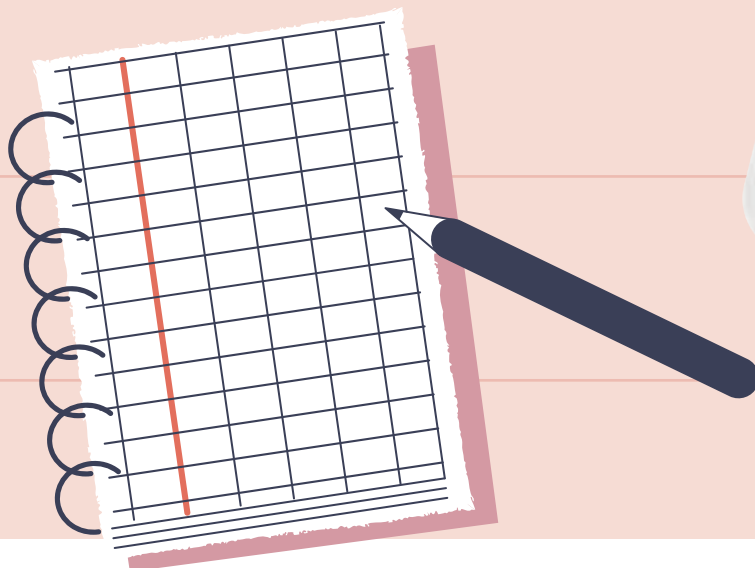


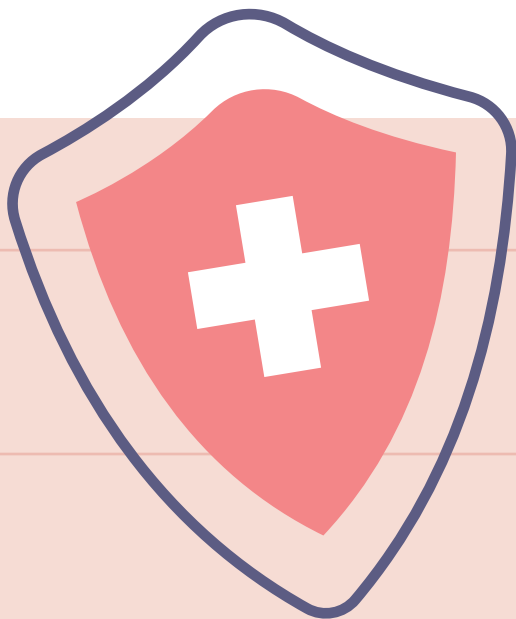
# Next Steps

*How to make a smooth transition from pediatric to adult healthcare*

BY KAREN DOSS BOWMAN

When Madison Parotta's healthcare team at Nemours Children's Hospital in Wilmington, Delaware, initiated conversations about transitioning her to the adult setting around the age of 18, she was ready. Diagnosed with spinal muscular atrophy (SMA) at around 10 months old, Madison had practiced advocating for herself from a young age — from asking questions at medical appointments to speaking up during her IEP meetings at school.





### Your Travel Checklist

When planning for a trip, you want to make sure you are prepared. This can range from making a packing list to budgeting. The same rules apply to your transition to adult care. Review the list below to help you feel ready for this new and exciting journey.

	Yes!	Halfway to yes!	No, but I want to learn!
I can give my full name, date of birth, and full address when asked.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can give the name of my disease or condition and what parts of my body are affected.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can tell you what my allergies are.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know the names of my medications.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can tell you what medications I am taking.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can tell you the doses of my medications.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I can tell you which doctor I see.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know how to refill my medications.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I understand basic insurance terms, such as deductible and co-pay.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I feel comfortable talking to my healthcare provider.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I feel comfortable talking to my pharmacist about tobacco, drug, and alcohol use.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I have discussed my travel plans with my healthcare provider.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
I know how to use a travel kit.	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

### New Roles and Responsibilities

When setting out on a new adventure, it is helpful to have a plan A as well as a plan B, just in case of an unexpected event. As the leader of your journey, it is your responsibility to delegate tasks to your support team to ensure all goes smoothly.

**Start by making a list of your support people and what each can help with.**

Your support people may offer advice, financial or transportation assistance, emotional support, and more. Be sure to discuss the support you need with the individuals on your list.

Name	Support type	Contact information

**Next, think about different areas of your life and set goals for what you want to accomplish:**

**Housing**  
 Where do you want to live? \_\_\_\_\_  
 What will you need to live independently? \_\_\_\_\_  
 Which support people can help you achieve this goal? \_\_\_\_\_  
 How are they willing and able to assist? \_\_\_\_\_

**Transportation**  
 How can you get to the grocery store, doctor's appointments, and social engagements?  
 Is there anyone you can rely on for transportation needs? What if you are in trouble and need quick assistance? \_\_\_\_\_



*“People with neuromuscular conditions have to grow up fast, but in their late teens, they still deal with constraints from parents and caregivers. So now, my adult doctor has given me full autonomy. She gives me her recommendations, but she emphasizes that I know my body best.”*  
 — **Madison Parotta**



Madison Parotta likes the autonomy of the adult care setting.

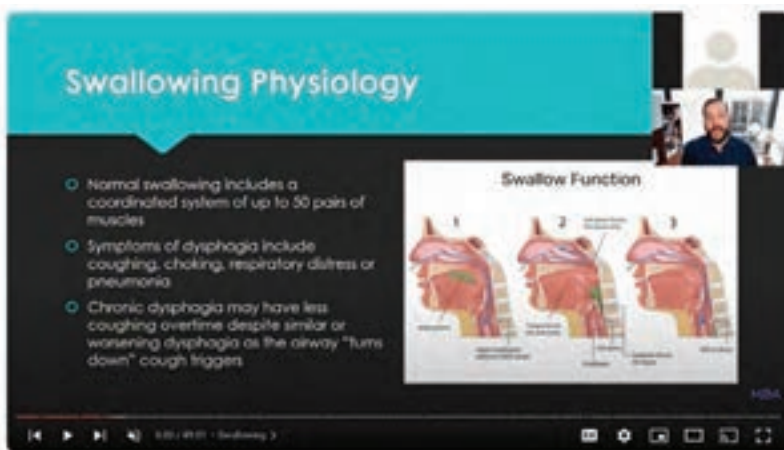
“My parents were always encouraging me to do things for myself,” says Madison, 28, who now receives care at Penn Medicine in Philadelphia. “That definitely helped prepare me to advocate for my healthcare needs.”

Madison recalls that her providers at Nemours Children’s gave her a checklist of services that she would probably need to pursue moving forward — such as personal care assistance and Medicaid. One of the biggest challenges she faced in aging out of pediatric care was the loss of funding for services such as physical therapy and assistive technology.

Even so, Madison enjoys the autonomy she feels attending an adult clinic.

“In a way, I feel more comfortable with my adult neurologist than I did with my childhood doctors, so that’s a big

plus for me,” Madison says. “People with neuromuscular conditions have to grow up fast, but in their late teens, they still deal with constraints from parents and caregivers. So now, my adult doctor has given me full autonomy. She gives me her recommendations, but she emphasizes that I know my body best. If I disagree with her, I feel free to do what is right for me.”



MDA’s Transitions in Care online seminar covers relevant health topics for teens, young adults, and their families.

**What age to transition?**

Achieving independence is an essential rite of passage for launching teens into adulthood. Like Madison, many children with neuromuscular diseases share the same longing for independence as their peers without disabilities.

Making the transition from the world of pediatric healthcare to the adult setting is a critical part of the journey. This can be a time of anxiety for young adults as they learn to navigate a new healthcare environment, manage changes to health insurance, and balance expanded responsibilities — often while adjusting to major



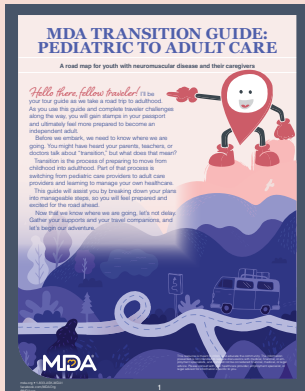
## PREPARE FOR THE TRANSITION WITH MDA



Like many milestones in life, moving from pediatric to adult care is best met with preparation. These resources can help.

### MDA Transition Guide: Pediatric to Adult Care

Geared for adolescents and young adults, this transition guide is chock full of information, exercises, and checklists that are helpful for individuals, as well as parents, caregivers, and healthcare providers. To view or download the guide, go to [mda.org/education](http://mda.org/education) and click on “K-12 Education Resources.”



### Transitions in Care seminar

MDA’s online Transitions in Care seminar is designed to give you the knowledge, skills, and resources you need to successfully manage the change. The seminar is divided into five sessions, each under one hour, available on-demand and free of charge.



Scan this QR code to watch the seminar.

### Access Workshops

MDA’s Access Workshops provide in-depth online learning on topics related to healthcare and independence, also free of charge. Find the Access Workshops series at [mda.org/AccessWorkshops](http://mda.org/AccessWorkshops).

life changes such as starting college, holding down a job, or moving out of their parents’ home.

When does a patient age out of pediatric care and make the move to adult care? There is no magic age of transformation, experts say.

“The transition away from pediatric healthcare is a process,” says adult neurologist Hani A. Kushlaf, MD, director of the MDA Care Center at the University of Cincinnati (UC). “It’s not a one-point-in-time decision.”



Hani A. Kushlaf, MD

Ideally, the transition from pediatric to adult care happens gradually, starting around a child’s 13th birthday. Over time, their pediatric care team helps them build skills by asking them questions and giving them a voice in medical decisions.

Anne Connolly, MD, Chief of the Division of Neurology at Nationwide Children’s Hospital in Columbus, Ohio, believes the groundwork for this transition can be laid at an early age. She begins asking patients health-related questions when they are around 5 years old — depending on their maturity and developmental abilities.



Anne Connolly, MD

“Children obviously are not transitioning at 5 or 6 years old, but they’re able to answer very simple questions for themselves, such as, ‘Does anything hurt?’ or ‘What kind of stretches do you do each day?’” says Dr. Connolly, who also is a professor of pediatrics at The Ohio State University College of Medicine. “It’s around age 13 to 15 that I suggest a brief period where the parents step out of the room for a few minutes so the child can feel free to talk to me about anything. That is how we begin developing a doctor-patient relationship that isn’t completely absorbed by the parents.”

### Planning for change

During childhood, parents manage all aspects of their children’s healthcare: making appointments, refilling prescriptions, managing health insurance, and more. Transitioning to an adult care model can be overwhelming as young adults get used to new clinicians, new routines, and new expectations. Additionally, adults have different social and medical needs and life goals. And because they are no longer growing, there are no new motor milestones to track.

How can families know if their child is ready for the transition to adult healthcare?

Ideally, the transition from pediatric to adult care happens gradually, starting around a child’s 13th birthday.

*“As a neurologist, I’m directing the care of children with neuromuscular diseases and helping them connect with adult specialists — including neurologists, cardiologists, and pulmonologists — to orchestrate all the pieces of this big puzzle.”*

— Andre Prochoroff, MD

Just as there is no magic age for switching from pediatric to adult care, there are a variety of care models that may be used depending on the patient’s needs and a medical institution’s practices.

For example, some individuals with Duchenne muscular dystrophy (DMD) may continue with their pediatric providers into adulthood because of specialized nutritional or other needs. Other young adults may not have the intellectual ability to manage their own healthcare. Each patient’s medical team can help assess the person’s ability and willingness to make the transition.



Andre Prochoroff, MD

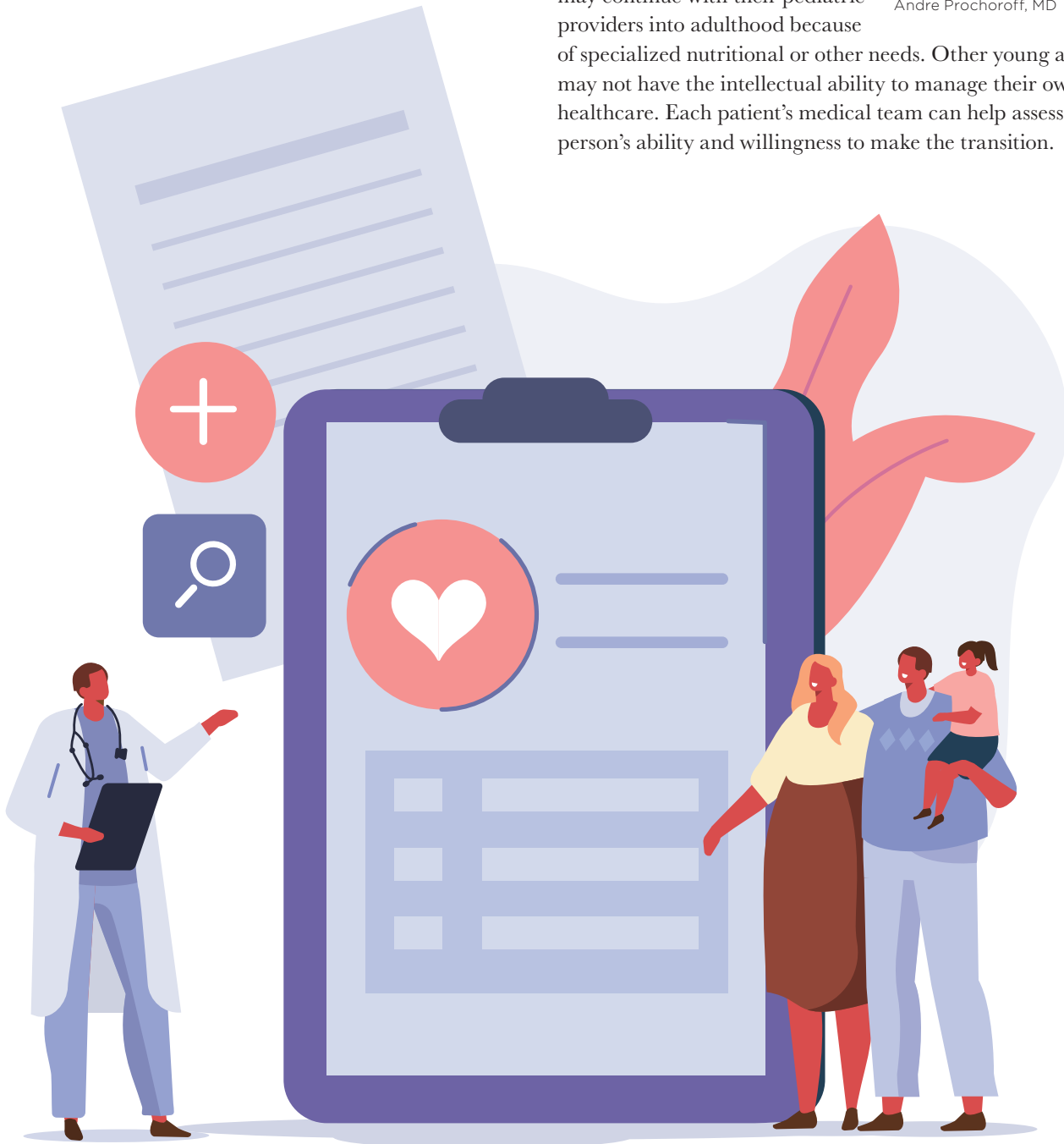


Image: iStock.com/Irina\_Strelnikova



Andre Prochoroff, MD, a pediatric neurologist at MetroHealth in Cleveland, Ohio, oversees multidisciplinary neuromuscular care throughout childhood. He compares his role in promoting the transition to adult care to being “the director in an orchestra.”

“We are fortunate at MetroHealth to be a larger system that offers all the components here to make a good orchestra,” says Dr. Prochoroff, who also is a faculty member at Case Western Reserve University School of Medicine. “As a neurologist, I’m directing the care of children with neuromuscular diseases and helping them connect with adult specialists — including neurologists, cardiologists, and pulmonologists — to orchestrate all the pieces of this big puzzle.”

### Hybrid transition model

Dr. Kushlaf, an associate professor of neurology and pathology at UC, participates in a hybrid transition model in partnership with Cincinnati Children’s Hospital.

Though he is an adult practitioner, he begins seeing patients 15 years old and up at Cincinnati Children’s. This makes


the transition process easier for teens and their families. When they become young adults and move to the adult care setting, they’ve already established a rapport.

“The advantage of patients seeing me at Cincinnati Children’s first is that they get to know me,” Dr. Kushlaf says. “So, when I tell them I want to start seeing

them across the street at the adult clinic, they know it’s only the location that has changed. That takes away a lot of their anxiety.”

### Getting started

Lynne Wood, MD, a pediatric neurologist at Billings Clinic in Montana, encourages individuals and their families to use Got Transition ([GotTransition.org](http://GotTransition.org)), an online toolkit that helps teens and their parents prepare for the transition from pediatric to adult healthcare. MDA’s downloadable Transition Guide ([mda.org/education](http://mda.org/education)) is another good resource.

“Start the conversations with your child’s doctor early,” Dr. Wood says. “Don’t be afraid to be the squeaky wheel. Don’t be afraid to advocate for your child if you feel like something is missing in their care. We do a lot of hand-holding at the pediatric stage, but adult providers generally rely on patients to bring more to the table. It’s okay to ask for what you need.” 



*“Don’t be afraid to be the squeaky wheel. Don’t be afraid to advocate for your child if you feel like something is missing in their care.”*

— Lynne Wood, MD

**Some individuals with Duchenne muscular dystrophy may continue with their pediatric providers into adulthood because of specialized nutritional or other needs.**

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



**D**aniel Barvin always wanted to be a parent. “It was just something that was in me,” says Daniel, 35, of Houston, Texas. “My wife and I met when we were quite young, and five years in I was like, ‘OK, let’s start having kids.’ We weren’t even married yet, but I just knew I wanted to be a father.”

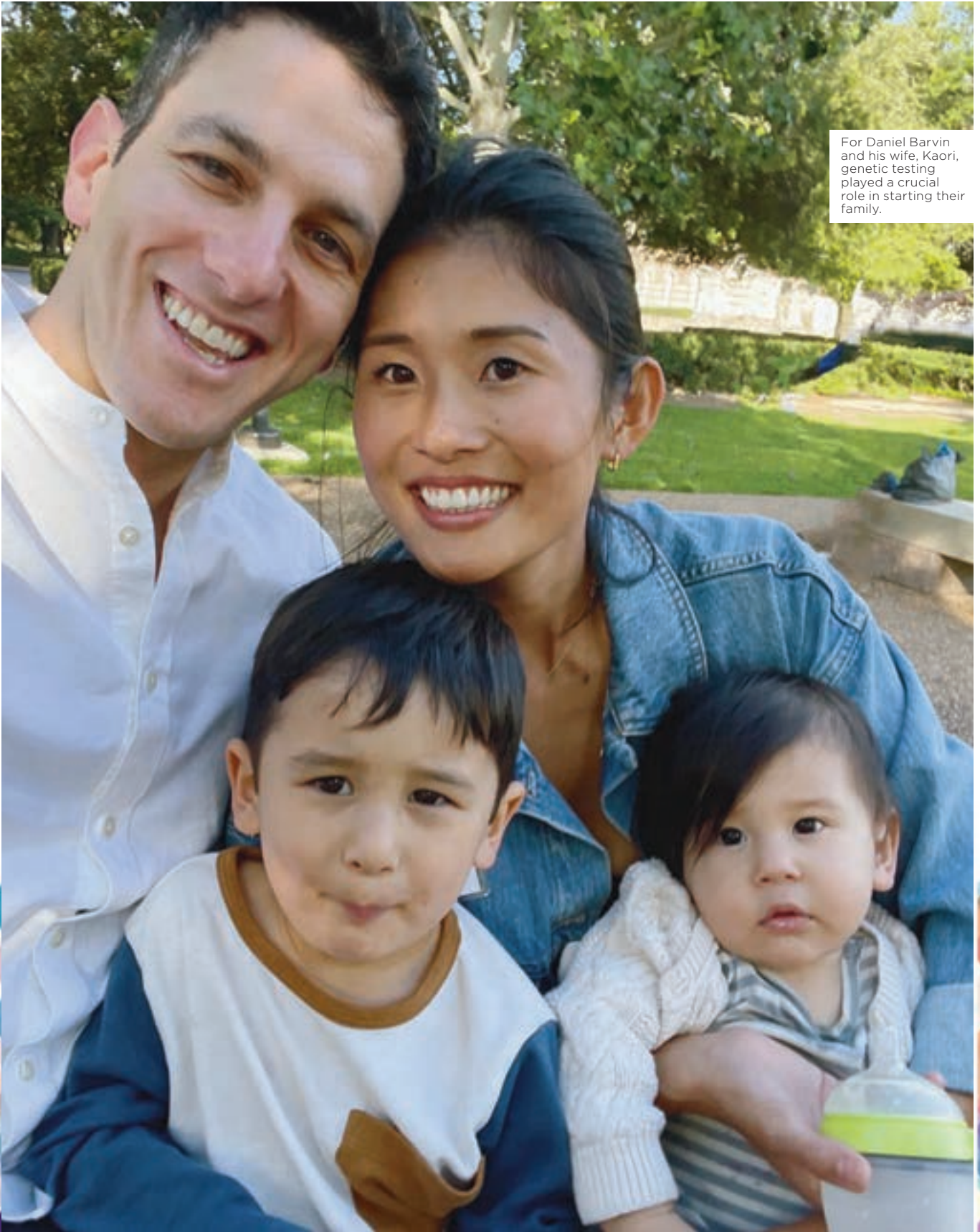
But fatherhood wasn’t the only thing that was “in” Daniel. He also carries the *C9orf72* gene mutation that can lead to familial amyotrophic lateral sclerosis (ALS), frontotemporal dementia (FTD), or both.

# FAMILY

*Genetic testing can help current  
and aspiring parents*

BY MATT ALDERTON

Image: iStock.com/natrot



For Daniel Barvin and his wife, Kaori, genetic testing played a crucial role in starting their family.

## FREQUENTLY ASKED QUESTIONS



### **My doctor gave me a diagnosis. Why should I get a genetic test?**

In many cases, genetic testing can confirm and improve the accuracy of a clinical diagnosis by pinpointing an individual's disease-causing mutation. This can lead to eligibility for certain therapies or medications and clinical trials, as well as a better understanding of how the disease will progress. This can also facilitate accurate genetic testing for affected or unaffected members.



### **Can't I order a genetic testing kit on the internet?**

There are many DNA testing kits on the market that claim to reveal your ancestry or provide health insights. But over-the-counter genetic testing kits are not medical tests, can't match the specificity of the tests ordered in a clinical setting, and don't offer the expert interpretation and counseling your care team delivers with test results.



### **I already had a genetic test. Do I need another?**

Genetic testing is advancing quickly. If your previous genetic test was negative or inconclusive, keep asking about new testing. Anyone considering a gene therapy or participating in a clinical trial also should ask if their testing needs to be repeated.



### **Is genetic testing covered by health insurance?**

Many health insurance plans will cover genetic testing and carrier screening when it is ordered by a physician. Check with your plan for coverage and reimbursement details.



### **If I have a diagnosis, should my family be tested?**

Talk with your genetic counselor about who in your family should be tested based on the type of disease and who else is showing symptoms. Relatives may be interested in genetic testing to find out if they are at risk of developing symptoms later in life or if they're carriers. Carriers have a chance of having children with the same genetic disease.

In people with FTD, damage to the brain's temporal and frontal lobes can cause dramatic changes in language, personality, and behavior. Approximately 15% of people with FTD also develop ALS.

Although doctors didn't recognize it at the time, Daniel's grandfather died of FTD when Daniel was 8 years old. Later, his father developed early-onset dementia and an uncle and an aunt died of ALS.

"It was brutal," recalls Daniel, who says doctors never raised the specter of heredity. "This was before genetic testing was prevalent, so every doctor told us these illnesses were sporadic and we had nothing to worry about."

When Daniel's father died following a dementia episode in 2016, his family ordered an autopsy and discovered he had the *C9orf72* gene mutation. Suddenly, Daniel's tragic family history made a lot more sense. From that clarity, however, came an unforeseen crisis of conscience: Knowing the gene was in his DNA, Daniel asked himself whether he could still become a father — and whether he should.

He isn't alone. Daniel is among millions of aspiring and current parents whose genes could predispose their offspring to hereditary neuromuscular conditions. However, unlike the generation before them, they don't have to wonder helplessly about the future. Thanks to genetic testing and family planning, they can understand risks, prepare for them, and take action if they choose to.

### **What is genetic testing?**

Genes tell your cells how to grow and work. When you have genetic testing, medical professionals examine your genes for changes — called mutations or variants — that may cause diseases.

"Persons with a wide variety of symptoms could have those symptoms as the result of an underlying genetic change, and identifying and documenting that change increasingly is the way that we make a diagnosis," explains Jennifer Roggenbuck, a licensed genetic counselor at

**Genes tell your cells how to grow and work. When you have genetic testing, medical professionals examine your genes for changes — called mutations or variants — that may cause diseases.**



The Ohio State University College of Medicine, where she specializes in neuromuscular diseases.

Although the underlying science is similar, the clinical genetic tests one gets in a medical setting are different from the commercial genetic tests one might order from the internet to research their ancestry or their predisposition to baldness. “Those tests use a different technology,” continues Jennifer, who says commercial tests examine fewer gene variants with less rigor. “With medical genetic testing, there’s an element of clinical expertise to ordering the genetic test and interpreting the results that’s completely different than what you might get from direct-to-consumer genetic testing.”

And yet, medical and commercial tests have one thing in common: They’re non-invasive. “Sometimes we need blood samples and sometimes we need saliva samples. Often, we’re just using a very simple cheek swab,” says Kristin Engelstad, a certified genetic counselor in neurology at Columbia University.

### Proactive parenthood

Genetic testing can be diagnostic or predictive — it can help you understand the cause of symptoms you have or your risk for developing a genetic disease when you don’t have symptoms. In both cases, the results of a genetic test may qualify you for life-enhancing and life-saving therapies, medications, and clinical trials.

That’s what initially motivated Daniel to seek genetic testing in 2018: He wanted to know if he carried the same genetic mutation for ALS and FTD that had affected his relatives, and what he could do about it if he did.

“I found out that there is a 95% chance that I will develop the disease,” Daniel says. “I had prepared myself for that news, and I approached it with a mindset of action. The generation before me had no idea this was coming. They were blindsided by the disease, but I have the ability to prepare for it.”

The ability to prepare was especially important to him as he pondered parenthood. “When my wife and I eventually met with a genetic counselor, the key takeaway was that we could do family planning, which we had never heard about,” continues Daniel, who learned that he has a 50% chance of passing down the *C9orf72* gene mutation to a child. “That became a torch that I carried forward. This had decimated my father’s generation, and who knows what it will do to mine. But I decided to do everything I could to save the next generation.”



*“The generation before me had no idea this was coming. They were blindsided by the disease, but I have the ability to prepare for it.”*

— Daniel Barvin





Any prospective parent — regardless of family history — may want to consider carrier screening, according to Ellen Moran, a certified genetic counselor at the Center for Children at NYU Langone’s Hassenfeld Children’s Hospital in New York. Comprehensive carrier screening encompasses broad genetic screening for autosomal recessive disorders like spinal muscular atrophy (SMA), X-linked disorders like Duchenne muscular dystrophy (DMD), and autosomal dominant disorders like forms of limb-girdle muscular dystrophy (LGMD). With carrier screening, future parents may identify potential risk factors.

Armed with this information, couples can decide whether to pursue family planning options, which may include pre-implantation genetic testing (PGT) with in vitro fertilization (IVF) or using donor eggs or sperm.

With PGT and IVF, eggs are retrieved from the mother and fertilized in a laboratory with sperm from the father. Fertilized eggs can be tested for dominant and recessive genetic mutations, and healthy embryos can be implanted in the mother.

When using donor eggs and sperm, Kristin emphasizes the importance of working with reproductive centers that routinely conduct carrier screening. Genetic counselors might be able to refer couples to reproductive centers that understand and appreciate genetics.

“There are a lot of options. It just depends on what people are comfortable with, what their insurance will cover, and what they can afford out of pocket,” Kristin says.

Daniel and his wife opted for PGT and IVF and now have two children, a 1-year-old daughter and a 3-year-old son — neither of whom carry the *C9orf72* gene mutation.

“I’m in a much better position than the previous generation to take steps that protect my children’s future,” says Daniel, whose personal experience inspired him to start a career at Coya Therapeutics, a biotechnology company

## AFFORDABLE GENETIC TESTING

As technology advances, genetic testing is becoming more affordable. In addition, some sponsored programs offer genetic testing or carrier screening for specific neuromuscular conditions at no cost. Generally, your doctor will need to verify your eligibility and order the test.

To learn about sponsored genetic testing programs and other resources, contact MDA’s Resource Center at **833-ASK-MDA1** or **ResourceCenter@mdaUSA.org**.

developing therapeutics for ALS, and to establish a nonprofit called End The Legacy: Genetic ALS and FTD. “My kids don’t have this gene, but I’m going to be incredibly proud to tell them the story of their ancestors and the proactive stance we’ve taken against ALS and FTD.”

### Genetic testing for children

In cases where there is a known risk of genetic disease after conception, parents have options to be prepared to welcome their baby.

“We can check the genetic status of the fetus with a variety of tests,” notes Jennifer, who says the most common tests are chorionic villus sampling, which involves genetic testing of the placenta beginning at 12 weeks of pregnancy, and amniocentesis, which involves genetic testing of the amniotic fluid around the fetus starting at 15 weeks of pregnancy.

Parents who learn of a neuromuscular disease in their family after having children should consult with a genetic counselor to find out if their children should be tested.

For children with no symptoms, genetic testing typically can wait. “We do not recommend genetic evaluation for children who are currently healthy,” says Jennifer, who points out that genetic testing in childhood can have personal and



### ONLINE EXCLUSIVE KNOW BEFORE YOU TEST

Find out what you need to know about your rights regarding your genetic status and how it could impact you in “Can Genetic Testing Make You Uninsurable?” at **MDAQuest.org/insurable**.

financial implications in adulthood when individuals apply for insurance, start relationships, etc. “We want to respect the child’s autonomy. So, unless there’s a medical reason to do genetic testing early, we usually say the child can make that decision when they’re 18 or older,” she says.

Families of children showing symptoms may want to pursue testing. “Genetic testing could help you get an accurate diagnosis,” says Ellen, who explains a genetic diagnosis is necessary to provide accurate genetic counseling to the family and determine eligibility for clinical trials and treatments, including disease-modifying therapies that tend to be more effective when they’re given early. “The sooner you start, the better the prognosis is for treatment.”

When it comes to getting a diagnosis, parents are their children’s best advocates. “If you have a child with a mysterious medical condition that your physician doesn’t understand — particularly one that’s lasted a long time and was present from an early age — it might be worth asking, ‘Could my child benefit from genetic testing?’” Jennifer says. “Don’t wait for your doctor or pediatrician to bring it up because it may not be at the forefront of his or her mind.”

### Where to start

Individuals seeking genetic testing for family planning should see a genetic counselor. Often, the first step is getting a referral from your primary care physician or neurologist. Alternatively, you may be able to make an appointment directly with a genetic counselor. (Use the National Society of Genetic Counselors “Find a Genetic Counselor” tool at [FindaGeneticCounselor.nsgc.org](https://www.nsgc.org/Find-a-Genetic-Counselor)).

“Ideally, you want to see a genetic counselor who specializes in neuromuscular disorders,” advises Jennifer, who says demand for specialists outstrips supply. “Fortunately, many



*“Persons with a wide variety of symptoms could have those symptoms as the result of an underlying genetic change, and identifying and documenting that change increasingly is the way that we make a diagnosis.”*

—Jennifer Roggenbuck

genetic counselors offer telemedicine appointments as long as you’re in the same state.”

It’s helpful to understand what a genetic counselor does, according to Jennifer, who says there’s more to genetic testing than the actual test. Genetic counselors also help patients determine what kind of genetic test to get, what the implications of testing might be, and what actions they can take in response to their test results.

“Genetic counselors can help you review the medical landscape surrounding genetic testing, as well as the psychological, emotional, and family issues that come into play with these types of decisions,” Jennifer explains. “We’re patient-focused, so we’ll never tell you what to do. Our role is to provide risk assessment, education, and support to individuals as they determine what steps are right for them at a particular time in their lives.”

Whether your motivation is your own health or that of your future children, genetic testing can be fraught but fruitful.

“It can be a scary process, but we try to impress upon families that they’re not in this alone,” Ellen concludes. “There’s a whole team of people there to work with you as you go through this.” [Q](#)

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Matt Alderton is a Chicago-based freelance writer who frequently covers health topics.

Parents who learn of a neuromuscular disease in their family after having children should consult with a genetic counselor to find out if their children should be tested.

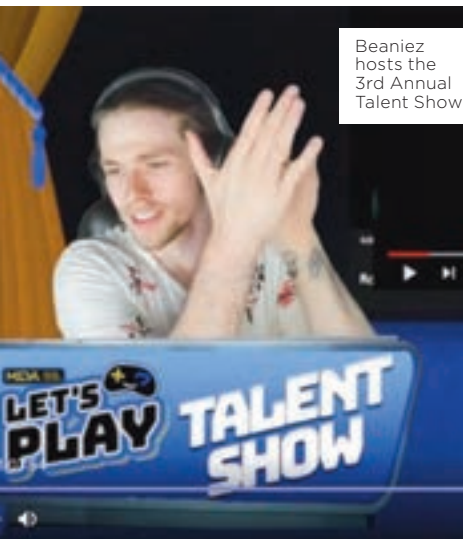




# 2023 in Review



## MDA Let's Play Summer of Fun



Beaniez hosts the 3rd Annual Talent Show.

Known for streaming and gaming six days a week, the MDA Let's Play community dials up the fun in the summer. Over the summer, Let's Play:

- Hosted its 3rd Annual Talent Show featuring more than three hours of singing, art, magic, gaming, and full-costume lightsaber duels. MDA National Ambassador Leah Zelaya kicked off the event by speaking with our community and sharing her adaptive skiing skills.
- Traveled to New Jersey, where host Beaniez met community members Zebra, Zebukiel, and Harleyq to do the Hot Ones spicy chicken wing-eating challenge. The MDA fundraising event was livestreamed on the MDA Let's Play Twitch channel.
- Joined in the fun of MDA Summer Camp, where Beaniez and other MDA Let's Play community members participated as counselors and campers.

Join the MDA Let's Play community for game nights, karaoke, barbecues, fireside chats, and all sorts of entertainment. Learn more at [mda.org/lets-play](https://mda.org/lets-play).



In August, Marissa Lozano, MEd, MDA's Director of Community Education, and Amanda Sweet, Account Director for Consumer Fundraising for MDA, toured the Comcast Accessibility Lab in Philadelphia. They learned more about the Xfinity Web Remote, a web-based remote control that allows someone with no audible voice to use voice commands. To read an interview with the Comcast Accessibility team, visit [MDAQuest.org/accessibility-comcast](https://MDAQuest.org/accessibility-comcast).

## Family Fun

This year, MDA added another exciting option to the recreation activity lineup: Family Getaways. Like MDA Summer Camp, participants of all ages can take part in things like outdoor activities, campfires, arts and crafts, and family and community bonding activities. MDA created these unique, accessible events to promote fun, independence, and adventure, and to provide a chance for families to experience the magic of camp together. Angee, mom to two boys who live with neuromuscular disease, says her kids gained new confidence during the Family Getaway they attended in Oregon. "They felt seen and valued. Their lights get dim in the world. They are so bright right now," she says.

To find upcoming Family Getaways, visit [mda.org/family-getaways](https://mda.org/family-getaways).

# Happy Campers



Thanks to many MDA Summer Camp volunteers, medical team members, and campers this year, we were excited to hold all planned in-person camp sessions. We continued our partnerships with many accessible campsites across the country that have experience serving medical populations. In 2023, 673 campers attended in person, enjoying activities like fishing, tie-dying, scratch art, friendship bracelets, swimming, zip lining, horseback riding, talent shows, kayaking, socializing around the campfire, and so much more. MDA also held two virtual camp sessions, where 142 campers enjoyed activities from the comfort of their homes.

To learn more about MDA Summer Camp, visit [mda.org/summer-camp](https://mda.org/summer-camp).



From rock climbing to horseback riding, everyone had a great time at Summer Camp.

## SUMMER CAMP IN THEIR OWN WORDS

### Campers say they had fun ...

“MDA Summer Camp has been the highlight of my summers since I was 8. It has taught me to come out of my shell and make new friends. Those friendships have now become permanent and ones that I look forward to re-establishing every summer. Camp is where I feel most comfortable and where I want to be.”

— Abby, 16, camper in Minnesota

“We cannot thank you all enough. The counselors, volunteers, and staff cannot know how much this means to these kids. Finding a place where they can participate and meet kids like them, along with giving a break to families at home, is priceless. Noah said he just wishes he could go more weeks!” — Kelsey, mom to a camper in Ohio

### ... and so do volunteers

“Camp is pure magic. If you’ve been once, it’s the easiest decision in the world to return year after year.” — Volunteer, Illinois

“It was honestly the best week of my life. The happiness brought to the kids was so contagious that I’ve never felt this happy before. I will definitely be coming back next year and would recommend everyone to be a part of this beautiful experience.”

— Volunteer, Florida





# MDA Advocacy Updates

MDA Advocacy has celebrated many victories in 2023, and the team is still hard at work.

## Accessible air travel

Recently, the Advocacy team released two powerful videos showing firsthand accounts of the inaccessibility of air travel. The brief documentaries follow MDA advocates Madison Lawson and Mindy Henderson (Quest's editor-in-chief) as they travel to Washington, DC, facing barriers and challenges along the way.

Watch their stories on MDA's YouTube channel at [youtube.com/mda](https://youtube.com/mda).

MDA's Advocacy team is working to improve air travel for people living with disabilities. Thanks to thousands of advocates making their voices heard, we are seeing results:

- The US House of Representatives passed its version of legislation to reauthorize the Federal Aviation Act (FAA). This bill mandates better training for airline staff, updates Department of Transportation (DOT) standards for responding to complaints about lost, broken, or damaged

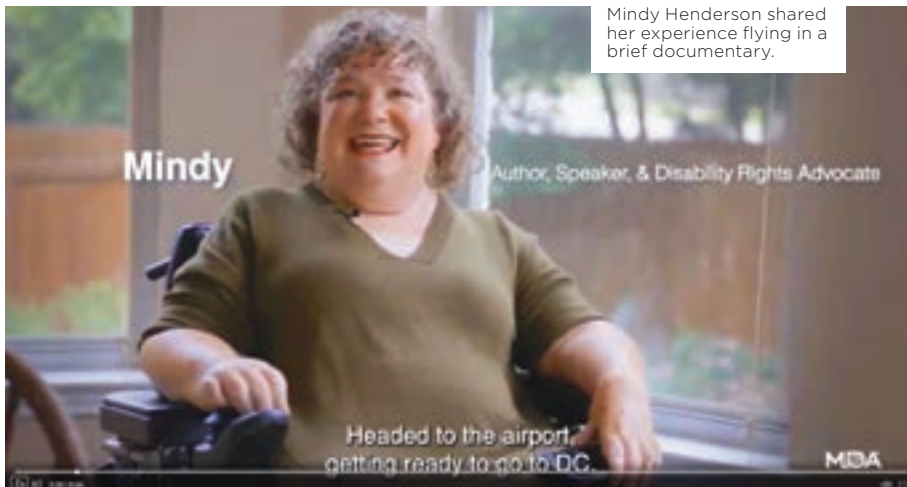
wheelchairs, and sets a concrete timeline and plan to allow people to fly in their wheelchairs. FAA reauthorization happens every five years and is our best opportunity to make meaningful change to air travel, including much-needed accessibility reforms. MDA is hopeful the Senate will pass an equally strong bill before sending it to the president.

- The DOT released a regulation to mandate accessible lavatories on single-aisle airplanes. Soon, passengers with disabilities will be better able to use restrooms on flights.
- MDA's CEO, Donald Wood, PhD, was invited to the White House to advocate for better accessibility.

## Newborn screening

MDA continues to make progress on adding Duchenne muscular dystrophy (DMD) to newborn screening programs. The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) voted to move the nomination of DMD for the Recommended Uniform Screening Panel (RUSP) to the next stage of review, bringing us one step closer to screening newborns for DMD.

Mindy Henderson shared her experience flying in a brief documentary.



MDA's Advocacy team is working to improve air travel for people living with disabilities. Thanks to thousands of advocates making their voices heard.

MDA's documentaries highlight the importance of accessible air travel.



MDA is working on getting DMD added to newborn screening panels.

Image: iStock.com/greenleaf123



# Quest Marketplace

## NOTICE TO OUR READERS:

MDA does not endorse products, services, or manufacturers. Such names appear here solely because they may provide valuable information. MDA assumes no liability for the use or contents of any product or service mentioned.

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Powered toilet lift & electric washing bidet



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## Introducing the Quest Media Adaptive Lifestyle Website

**Quest**  
Media

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



# My Myotonic Dystrophy Journey

*My parents don't have this inherited disease, but I do*

BY EMILY BLUME

Can you remember anything unusual from your childhood?" my neurologist probed. As I sat in the white-walled doctor's office with my mom, neither of us could come up with anything. Dustin Smyth, DO, a neuromuscular disease specialist, was looking for indicators that may have gone unnoticed at the time.

I didn't know it then, but his advice and care would be life-changing for me in the best possible way.

In terms of my childhood health, it was seemingly uneventful. A full, happy, charmed childhood without a blip on the radar. They say ignorance is bliss, and it was for the first three decades of my life.

The initial signs of the disease surfaced in my late 20s, when I was working as a morning-shift television reporter in Davenport, Iowa. My alarm went off at 1:30 a.m. Monday through Friday, and I made it into the newsroom 45 minutes later. One morning in the fall of 2019, while reporting live, I suddenly struggled to finish the sentence that was scrolling across the teleprompter in front of me. I couldn't pronounce my words completely. It felt like my tongue froze. It was so unprecedented that I brushed it off and felt grateful nobody seemed to notice.



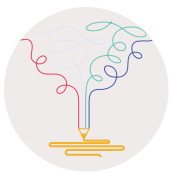
Emily Blume's journalism background helped her investigate the cause of her symptoms.

## More than stress

In the years that followed, it happened more frequently, in different environments, and usually while I was under some sort of pressure. So, I chalked it up to stress. My neurologist would later explain that while stress triggered the myotonia (muscle spasm) in my tongue, it was not the root cause. At the time, however, myotonia was not part of my lexicon, so I'd refer to this phenomenon as "the tongue thing."

Months later, "the tongue thing" materialized again, but this time it was my hands that froze. My heart dropped. "How could stress contribute to this?" I wondered.

By this point, I started to give more thought to something that previously never took up even an iota of space in my mind: My parents had used a sperm donor to have me. Could this be related?



## FAMILY INHERITANCE

Neuromuscular diseases can be passed from one generation to another in a variety of ways. For an overview of neuromuscular disease inheritance patterns, visit [MDAQuest.org/family-inheritance](https://mdaquest.org/family-inheritance).

## Following the clues

After “the tongue thing” manifested as a muscle spasm in my hands, I figured it was time to see a new doctor. The symptoms were enigmatic to all the health professionals in my close circle, and Google wasn’t providing any clear answers, either.

I walked into what would be the first of many neurology appointments with a lofty list of possible clues. I discussed everything I thought could be related — including the sperm donor, whose medical history was a mystery to me. I left that appointment with the words “myotonic dystrophy” (DM) scribbled across the top of my after-visit summary. This wasn’t a diagnosis, but it was a start.

In the weeks that I waited on the results of my genetic test, I reached out to the sperm bank my parents had used in hopes of finding answers. Myotonic dystrophy type 1 (DM1) is always inherited from the mother or father, and for each generation that inherits it, the symptoms manifest in more pronounced ways. This is called anticipation. Because my symptoms were on the mild side and emerged later in life, whoever passed it down to me was likely asymptomatic. So, at this point, inheriting it from my mom was just as strong a possibility as inheriting it from the donor.

The genetic counselor explained to me that they didn’t test donors for DM 30 years ago — and more alarming, they still don’t. Next, she started her search to get in touch with the man who donated more than 30 years ago.

The sperm bank was able to track down the donor, and he agreed to be tested for DM1. His cytosine-thymine-guanine (CTG) repeat count came back at over 50, which is just above the baseline for the asymptomatic form of this disease, explaining why he never knew he had it. My mom was also tested, and her results came back negative.

## Managing my health

When my genetic counselor presented my positive test for DM1, I was relieved. Now that I know what it is, I can learn how to manage it, I told myself.

With this newfound knowledge, I tracked down the aforementioned Dr. Smyth, a provider in Coeur d’Alene, Idaho, who has extensive experience with DM patients. He provided tactics and advice and prescribed me critical medications — mexiletine and modafinil — that control my myotonia and daytime fatigue.

“You can live into your 90s and die of something entirely unrelated,” Dr. Smyth reassured me. This has become my



mantra on the days when fears of the unknown transpire.

I recognize I’m lucky beyond measure when compared to others who face greater health struggles. In the time since my diagnosis, I’ve learned the intricacies of what contributes to my symptoms and what I can do to manage my health as best as possible.

Thankfully, I haven’t noticed any further disease progression.

Dr. Smyth’s care has been life-changing for me, and that’s a far cry from the “I’m sorry, there is nothing we can do” I got from the first neurologist I was referred to. We must be our own advocates. If your provider isn’t giving you answers or solutions to help, keep looking for someone who will.

While ignorance may have been bliss for some time in my life, knowledge is power today — and with that knowledge, I feel fortunate to have organizations like MDA that empower those of us living with neuromuscular diseases to live our best lives. [Q](#)

Emily Blume is a freelance journalist living in Spokane, Washington, with her golden retriever, Gary.

**Myotonic dystrophy type 1 is always inherited, and for each generation the symptoms manifest in more pronounced ways. This is called anticipation.**





## Blazing Trails

*Photo contest winner Leslie Krongold finds joy in nature*



Congratulations to our 2023 Lasting Impression Photo Contest winner, Leslie Krongold, 61, of Mendocino, California. In this photo, Leslie, who lives with myotonic dystrophy type 1 (DMI), enjoys the accessible trails in Hendy Woods State Park in Northern California. “This is the first park I’ve been to since I’ve been using a wheelchair that has an extensive raised boardwalk that meanders around the park,” she says. Leslie loves the park’s thoughtful design, which includes wheelchair-friendly trails through old-growth redwood groves and accessible picnic and bathroom facilities.

“My smile and enthusiasm in the photo are authentic — no posing. I’m very sensitive to heat, but the forest shade was perfect, and I felt incredible joy and freedom zooming along in this park.”

Since beginning to use a wheelchair two years ago, Leslie began working with local organizations to make trails more accessible. She urges others who use mobility devices to contact land and trail preservation organizations and share

their points of view. “Most people want to help and often don’t realize how many things are inaccessible until a relative, friend, or they themselves become mobility-challenged,” Leslie says. “I have

learned so much more about accessibility in the last two years, even though I started facilitating support groups for adults with neuromuscular diseases 25 years ago. I just didn’t ‘get it’ until I needed to.”

She now organizes monthly walks to introduce these accessible trails to others ([GlassHalfFull.online/leslies-accessible-walks](https://GlassHalfFull.online/leslies-accessible-walks).)

“I want to meet people who both appreciate the raw beauty we have surrounding us and will benefit from a more accessible ground to walk or roll on,” she says. [Q](#)

### +MORE PHOTOS

Congratulations to the runners-up in our reader photo contest: Tammy Stoneburner of Decaturville, Tennessee; Kristin Secor of Owego, New York; and Andrew Shelley of Claremont, California. See their photos at [MDAQuest.org/2023Photos](https://MDAQuest.org/2023Photos).



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

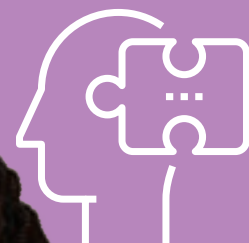
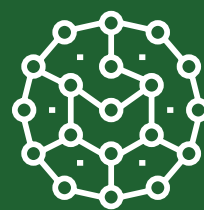
The National ALS Registry is a program that allows people with ALS to fight back and help defeat the disease.

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

Collecting and analyzing data

Striving to better understand the disease

Helping researchers find possible risk factors



**Your participation can make a difference**

Ask us about the Registry today.  
For more information, call **800-232-4636** or visit **[cdc.gov/als](https://cdc.gov/als)**.



# 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](https://clinicaltrials.gov) (NCT05291091) or contact [studies@edgewisetx.com](mailto:studies@edgewisetx.com)



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