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Welcome to
the Year of
Independence!



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Good Together

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INHERITANCE
How genetic
disease is
passed down



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Evrysdi in action

Proven to make a difference
in infants, children, and
adults 2 months and older
with spinal muscular
atrophy (SMA)



1,400+
people in the US with SMA
are taking Evrysdi, including
people up to 83 years old**



* Number of people taking Evrysdi since August 2020 (approval) through June 2021.

† 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 adults and children 2 months of age and older.

It is not known if Evrysdi is safe and effective in children under 2 months of age.

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. 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, 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
 - 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 and pharmacist when you get a new medicine

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✓ **Evrysdi helped infants with Type 1 SMA achieve a key motor milestone and delayed disease progression[‡]**

41% of infants (7/17) sat without support for at least 5 seconds after 12 months, as measured by the BSID-III gross motor scale

90% of infants (19/21) at 12 months and 81% of infants (17/21) at 23 months were alive and able to breathe without permanent support[§]

✓ **Evrysdi significantly improved or maintained motor skills in adults and children with Type 2 and 3 SMA***

Motor function improved after 12 months (average 1.36-point increase on the MFM-32 scale with Evrysdi vs average 0.19-point decrease without Evrysdi)

- 1.55-point estimated improvement versus placebo on the MFM-32 scale at 12 months (95% CI: 0.30, 2.81; $P=0.0156$)^{||}

✓ **Evrysdi is designed to help make and maintain more SMN protein**

✓ **The safety of Evrysdi is being studied in more than 450 people, from 2 months to 60 years old, with Type 1, 2, or 3 SMA[‡]**

✓ **The first and only medication to treat SMA with at-home dosing**

[‡] The efficacy and safety of Evrysdi was established in 2 main studies. FIREFISH is a 2-part, open-label study of Evrysdi in 62 infants aged 2-7 months with Type 1 SMA. SUNFISH is a 2-part study of Evrysdi in 231 children and adults aged 2-25 years with Type 2 and 3 SMA. A third study, JEWELFISH, is an ongoing safety study of Evrysdi in 174 infants, children, and adults aged 1-60 years with Type 1, 2, and 3 SMA previously treated with approved and investigational SMA medications.

[§] Permanent support was defined as having a tracheostomy (a surgery where a tube is inserted in the front of the throat into the windpipe) or more than 21 days of either noninvasive ventilation support (16 or more hours a day) or being intubated (a procedure where a breathing tube is inserted down the throat and into the windpipe) to help with breathing, in the absence of an acute reversible event.

^{||} This 95% CI (confidence interval) means that we are 95% confident that the actual average change in MFM-32 with Evrysdi will be between 0.30 and 2.81 points higher than with placebo.

BSID-III stands for the Bayley Scales of Infant and Toddler Development-Third Edition.

MFM-32 stands for the Motor Function Measure-32 Items.

SMN stands for survival motor neuron.

Important Safety Information (continued)

- 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. 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, sore throat, and cough (upper respiratory infection), lung infection, constipation, vomiting

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

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

Please see accompanying brief summary for additional Important Safety Information.

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



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

What is EVRYSDI?

- EVRYSDI is a prescription medicine used to treat spinal muscular atrophy (SMA) in adults and children 2 months of age and older.
- It is not known if EVRYSDI is safe and effective in children under 2 months of age.

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.
- 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 and 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. 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 syringes 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 syringes provided by your pharmacist (you should receive 2 identical oral syringes) 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 syringes are lost or damaged.
- Once 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, sore throat, and cough (upper respiratory infection)
 - diarrhea
 - lung infection
 - vomiting
 - rash

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.
- 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). Please see the Discard After date written on the bottle label. (See the **Instructions for Use** that comes with EVRYSDI).

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

MDA is dedicated to empowering people living with muscular dystrophy, ALS, and related neuromuscular diseases to achieve their potential.

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Building Back Stronger

It goes without saying that the world around us has changed in ways we never could have imagined over the past two years.

Fortunately, the search for neuromuscular disease treatments was able to move forward throughout the pandemic, as research laboratories and clinical trials quickly adapted to our new realities.

And while a year ago, the COVID-19 vaccine was only just beginning to be administered, this year, 100% of MDA staff are vaccinated, allowing us to continue to serve those with neuromuscular disease. That includes being able to offer MDA Summer Camp not only virtually but in person once again this summer. And that's not the only exciting news I have to share.

Just before the new year, President Biden signed the ACT for ALS into law, which invests in neurodegenerative disease research through a new FDA Rare Neurodegenerative Disease Grant Program. This law will prove to be transformative for families who are facing ALS. We'll keep you posted as developments unfold through questblog.org.

Another first: In January, I was proud to be the first MDA CEO invited to address the attendees of the International Association of Fire Fighters (IAFF) Affiliate Leadership Training Summit (ALTS). I joined IAFF's general president, Ed Kelly, on stage to announce that Fill the Boot is back in 2022, as it has been for more than 68 years. It was wonderful to meet so many fire fighters in person and to thank them for their unflagging support.

Have I used the words "in person" enough? In March, MDA's Annual Scientific and Clinical Conference for neuromuscular disease will take place in Nashville, Tennessee, as well as virtually. At this conference, the largest of its kind, clinicians and researchers will share their progress, best practices for conducting research, new diagnostic approaches, and information on treatments.

Finally, in March, we'll hit an important milestone: MDA Shamrocks turns 40. (Read more on page 34.) For four decades, individuals across America have supported our work by purchasing Shamrock pinups at their local retailers. I can't thank the employees at these establishments enough for continuing to sell Shamrocks to support MDA through the pandemic. Their hard work made 2021 a successful year, and 2022 promises to be even bigger.

A silver lining from this pandemic? The confidence born from the knowledge that no matter the challenge, the team at MDA can rise above it.

Sincerely,



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



Donald S. Wood, PhD

+YEAR OF INDEPENDENCE

We invite you to join MDA in declaring the Year of Independence. Turn the page to read about this exciting initiative and how you can get involved.

Leading the Quest for Change

For years, MDA has worked to empower people living with muscular dystrophy, ALS, and related neuromuscular diseases to reach their fullest potential. And while strides have been made through innovations in care, research, and advocacy, there is more work to do.

So this year, we're going bigger. We're declaring 2022 the Year of Independence.

This declaration is inspired by the experiences of MDA's chief-of-staff, Kristine Welker, whose son lives with muscular dystrophy. After his diagnosis, she immediately noticed the lack of people with physical disability in the workplace, in magazines, and in advertising and marketing campaigns.

This prompted her to leave her career in corporate America and join MDA. Inspired by stories of how people with disability, including her own son, are finding their personal independence, she wants 2022 to be the year disability inclusion is truly accelerated.

"I learned that there is strength in unity — that if we unite in our efforts, we can create a more inclusive world and give voice to the millions of individuals who want to work, live, and thrive without distinction," she says. "Throughout the past year, I have heard so many stories of how people with disabilities are finding their personal independence and freedom, like: Paul, living with limb girdle muscular dystrophy (LGMD), who says he feels most independent on his



Did You Know?

20,000 MDA advocates fight every day for those living with neuromuscular disease, making our voices heard by lawmakers, industry leaders, and employers. Our grassroots advocates are demanding accessible air travel for all, increased funding for medical research, better access to health care, and more. Learn how you can become an MDA advocate at mda.org/advocacy.

boat; a mother who wanted her own son to have the freedom to wear whatever he wanted and make fashion accessible to all; and Ethan, who gained his independence by attending MDA Summer Camp."

Kristine credits MDA with championing disability as diversity. "We will build on our legacy roots in advocacy and innovation to accelerate disability inclusion and leverage our own voice and platform," she says.

How will MDA lead the way?

1. We've expanded Quest into an adaptive lifestyle content platform. Extending beyond the pages of our flagship magazine, Quest now includes the Quest blog, Quest newsletter, and recently launched Quest podcast. All tell stories from our community, showcase what is possible, provide resources and information to empower the community, and reinforce the importance of equal access to independence.
2. We've joined forces with the New York City Mayor's Office for People with Disabilities and Inclusively, the online platform for job seekers, to create a DEI Coalition that will work with other Empowered Cities (empoweredcities.com) to help people with disabilities get into the workforce, establish best practices, encourage thoughtful and inclusive product development, and more.
3. MDA is committed to working with the Valuable 500 (thevaluable500.com) to help fulfill their commitment to "use the power of business to drive lasting change for the 1.3 billion people around the world living with a disability."
4. We will continue to break down barriers through public policy, advocacy, and education with our new series of on-demand programming, MDA Access Workshops, providing information and resources. (Turn to page 34 for more workshop information.) [Q](#)

+ASK US

To learn more about the Year of Independence, email us at deicoalition@mdausa.org with "Year of Independence—Tell Me More" in the subject line.

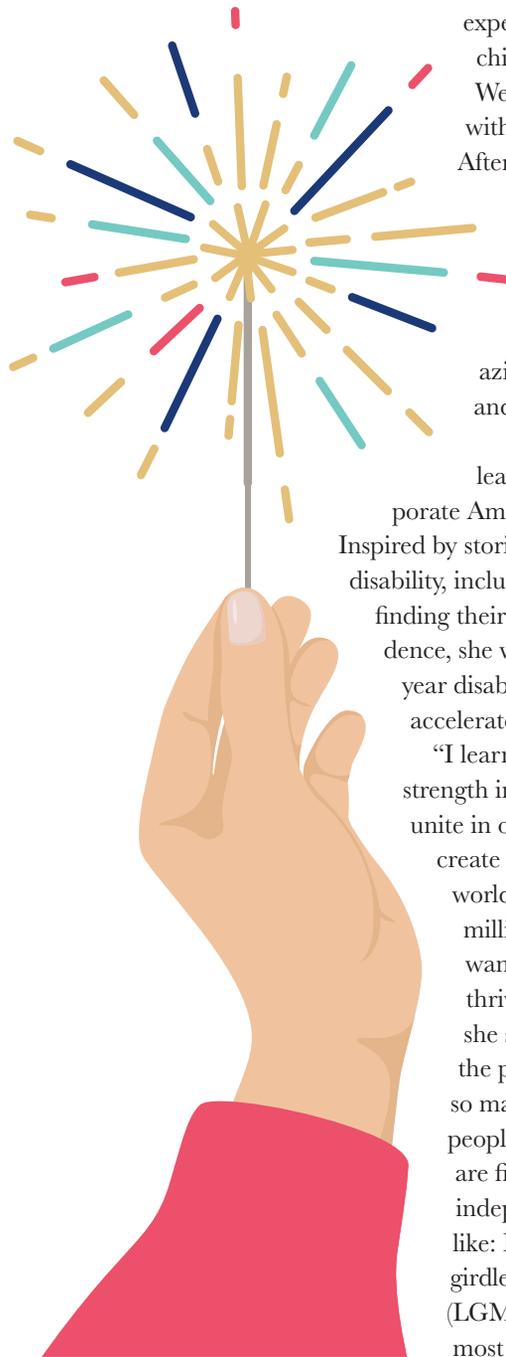


Image: iStock.com/Viktorija Lange

The Importance of Relationships



If the pandemic has taught us anything, it is the importance of relationships. Connection and belonging are basic human needs, and the quarantining and social distancing of the past two years have served to make us all more aware of how important connection is. Relationships are key to our survival, happiness, and overall mental health.

As we continue to move through the “new normal” of life with COVID-19 still among us, it is more important than ever to be intentional about nurturing our relationships. In this issue of *Quest*, we explore relationships — professional, romantic, and friendly — because the health of our relationships also is important for our survival and happiness.

My advice? Show up well in your relationships. Be your true self, and be kind. So many today are struggling with challenges we may not know about. Give others the benefit of the doubt, but demand respect and kindness for yourself in return. Seek to understand. Talk it out. Listen, but make sure your voice is also heard. Know your value, and spend time with those who also see it.

Mindy Henderson

*Director, Quest Editor-in-Chief
Muscular Dystrophy Association*



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Skye Lemmen takes in a view on her Pacific Crest Trail hike.

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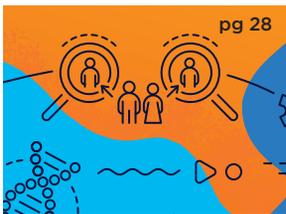
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Adults with neuromuscular disease can build healthy romantic relationships.

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Learn how genetic diseases pass from one generation to another.

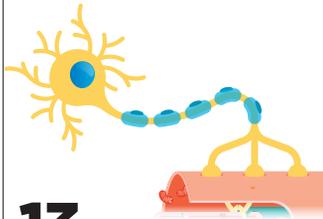
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Learn how volunteering helps build valuable skills.

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ACCESS MDA

Announcing the 2022 National Ambassadors, in-person Summer Camp, and more.

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LASTING IMPRESSION

A hiker raised funds for MDA while trekking the Pacific Crest Trail.

MORE ONLINE

Friendship Benefits

For people living with neuromuscular disease and their family members, making friends with others who have similar experiences has huge benefits. "It's just so nice not having to explain your situation over and over," says Sally Borden, who regularly chats with a friend living with the same late-onset distal myopathy she does. They met through MDA's National Connections Program.



Valerie Geerer

Valerie Geerer

Read:

"It Helps to Have a Friend Who Understands Your Neuromuscular Disease" at questblog.org.

Contact:

MDA's Resource Center to learn more about the National Connections program. [833-ASK-MDA1](tel:833-ASK-MDA1) or resourcecenter@mdausa.org.

MDA-113

Let's Celebrate Shamrocks 40th Birthday!



This March marks the 40th year of the now-iconic Shamrocks pinup program, which started in a local Irish pub in Michigan. Thousands of retail locations nationwide now participate year after year, raising more than \$330 million to date. We are grateful for CITGO Petroleum Company and all retail partners who turn the country green every March!

Learn more at mda.org/shamrocks.

MDA Kiddos Attend Fiesta Bowl!



More than 75 MDA kids got to attend the 2021 Fiesta Bowl Charities Player Outreach event on Sunday, December 26th. Paired with players from the University of Minnesota Golden Gophers, they got to play games and laser tag and then watched the Gophers win the Guaranteed Rate Bowl. Each kiddo took home an autographed card from their buddy to remember the day.

Burn Boot Camp Builds Their Muscle for MDA



Burn Boot Camp, a leading fitness franchise, raised \$510,545 last October through their annual "Be Their Muscle" campaign. Throughout the month, nearly 300 Burn Boot Camp locations across 39 states fundraised for the cause. The campaign culminated in a workout event on October 30th, where 100% of the proceeds were donated to MDA.

Dutch Bros. Launches Initial Public Offering



Join us in congratulating our longtime partners Dutch Bros., the Oregon-based coffee chain, which launched their initial public offering on September 15th. Dutch Bros. has supported MDA for 15 years through their annual Drink One for Dane Campaign, raising over \$10 million. D14D Day started in 2007 in honor of co-founder Dane Boersma, following his diagnosis with ALS.



Dr. Don Wood & IAFF Announce Fill the Boot 2022

In January, more than 1100 fire fighters from across the country gathered at the annual Affiliate Leadership Training Summit (ALTS) in Lake Buena Vista, Florida. During the General Session, IAFF General President Ed Kelly noted that MDA is the IAFF's longest and arguably proudest charitable endeavor. MDA President and CEO Dr. Don Wood thanked the audience for their steadfast efforts: "What you have done over the last 68 years is unprecedented." Fire fighters will continue to ask communities to Fill the Boot in 2022. Dedicated fire fighters will hit the streets and storefronts with boots in hand, asking pedestrians, motorists, customers and other passersby to donate to MDA, one dollar at a time.

MTPA Funds Fire Fighter Education on ALS

MDA will be launching an ALS Education initiative for Fire Fighters, a population with a two-fold risk of developing ALS compared to the general population. Through firefighters.mda.org, MDA will provide information about recognizing symptoms, finding support, and accessing the highest quality care at MDA ALS Care Centers. We are grateful to our partners at Mitsubishi Tanabe Pharma America for underwriting part of this important initiative.

PROGRESS NOW

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

MDA research in action

MDA Awards 18 Research Grants

In 2021, MDA awarded 18 new neuromuscular disease research grants totaling more than \$1.6 million. Seven development grants are providing critical support to researchers at the beginning of their careers, and 11 Idea Awards are funding bold, innovative research concepts that are expected to greatly impact our community.

“These grants do two very important things — help grow the next generation of neuromuscular disease investigators and provide seed funding for those creative, hard-to-fund projects that just need a little boost at this early stage,” says MDA Chief Research Officer Sharon Hesterlee, PhD. Here are some award highlights:

- **Alba Timón-Gómez, PhD**, of the Miller School of Medicine, University of Miami, received a development grant to study the processes underlying mitochondrial encephalomyopathies.
- **Jae-Sung You, PhD**, of the University of Illinois at Urbana-Champaign, received a development grant to study the role mammalian target of rapamycin (mTOR1) protein plays in Duchenne muscular dystrophy (DMD) pathology and discover new therapeutic targets.
- **Valérie Allamand, PhD**, of the Sorbonne Université-Inserm in France, received an Idea Award to work on a technique to correct the production of COL6 protein, which, when mutated, is associated with severe signs and symptoms in congenital muscular dystrophies (CMD).
- **Nick Menhart, PhD**, of the Illinois Institute of Technology, received an Idea Award to use a computational framework to search for new ways to edit the dystrophin message to produce better modified dystrophin proteins than are available with exon skipping.

For a complete list of 2021 grants, visit MDA’s Grants at a Glance page at mda.org/gaag.

New approval

Vyvgart Approved to treat gMG

The US Food and Drug Administration (FDA) has approved efgartigimod (Vyvgart) for the treatment of generalized myasthenia gravis (gMG) in adults who test positive for the anti-acetylcholine receptor (AChR) antibody. This subtype of myasthenia gravis may be referred to as AChR ab+ gMG. The treatment will be made available in the United States and marketed by Argenx.

An antibody fragment that targets the neonatal Fc receptor (FcRn), Vyvgart is designed to reduce the number of antibodies in the body including the autoreactive antibodies that cause gMG. Although treatment with Vyvgart will not cure gMG, it could lead to functional improvements.

The FDA based the Vyvgart approval on the positive results of the phase 3 ADAPT trial, which evaluated the safety and efficacy of Vyvgart in 167 adults with gMG.

Vyvgart was generally well-tolerated, with a safety profile comparable to that of the inactive placebo control. The trial met its primary endpoint, demonstrating that Vyvgart treatment of people with AChR ab+ gMG resulted in clinically meaningful improvements in symptom severity, as measured by the Myasthenia Gravis Activities of Daily Living (MG-ADL) score.

Find more information, visit vyvgart.com.



Amyotrophic lateral sclerosis (ALS)

Trial for Oral Drug

Researchers at Mitsubishi Tanabe Pharma Development America, Inc. (MTPA) are seeking adults living with ALS to participate in a phase 3b clinical trial to evaluate the safety and efficacy of oral edaravone (Radicava) to treat ALS. Radicava is designed to help protect cells from damage caused by free radicals and slow the progression of ALS.

Participants will be randomly assigned to receive oral Radicava or a placebo control. The total trial length will be approximately 58 weeks. Participants will be required to complete eight on-site visits and eight telephone visits.

Individuals 18 to 75 years old, with a diagnosis of definite or probable ALS according to the El Escorial revised

criteria, and who meet additional criteria, may be eligible to participate. Travel support is available for study participants and families.

To learn more about the study or inquire about participation, contact MTPA's Clinical Trials Information Desk at MT1186@iconplc.com or **800-313-9381**.

Becker muscular dystrophy (BMD)

Men Needed for Natural History Study



This study involves MRI and other tests. Exclusion criteria include use of an internal pacemaker or internal cardio-defibrillator and dependency on daytime mechanical ventilation.

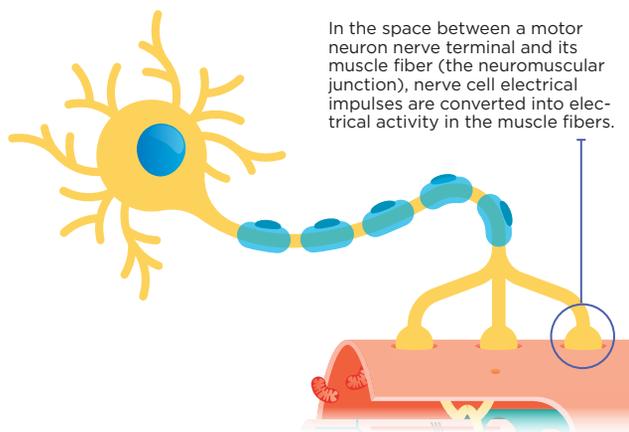
Researchers at the University of Florida are seeking men living with BMD to participate in a three-year natural history study. The objective is to gain a better understanding of the disease course and to identify biomarkers for

BMD. This is an observational study and does not involve an intervention.

Participants will be required to complete one doctor visit each year over the course of three years. During these visits, participants will be evaluated using tests such as magnetic resonance imaging (MRI), magnetic resonance spectroscopy (MRS), and a number of functional tests. Participants who can walk may perform the 6-minute walk test, 10-meter walk/run, North Star Ambulatory Assessment, and timed functional measures. All participants may be assessed using the Performance of Upper Limb test; pulmonary function tests; and analysis of blood sample, urine sample, and a one-time needle biopsy of two leg muscles.

To be eligible to participate, individuals must be male, between ages 18 and 62, have a diagnosis of BMD, and meet additional criteria.

To inquire about participation, contact study coordinator Claudia Senesac at csenesac@php.ufl.edu or **352-273-6453**.



Charcot-Marie-Tooth disease (CMT)

Observational Study Seeks Volunteers

Researchers at Ohio State University are seeking adults living with CMT type 1 (CMT-1) or type 2 (CMT-2) to participate in a one-month observational study. The CMT Establish study is designed to assess whether individuals with CMT-1 and CMT-2 exhibit altered communication between nerves and muscles, known as neuromuscular junction (NMJ) transmission, compared to healthy volunteers. No experimental treatment is evaluated in this study.

Participants will be required to attend clinic visits every one to two weeks over the course of the one-month study and will undergo muscle and nerve testing.

To be eligible, individuals must be at least 18 years old, have a confirmed diagnosis of CMT, have been on a stable regimen of concomitant medications for two months prior to enrollment, and meet additional criteria.

To learn more or to inquire about participation, contact study coordinator Amy Bartlett at **614-366-9050**.

Duchenne muscular dystrophy (DMD)

Promising Drug in Phase 2 Trial



Based on the positive results of the HOPE-2 trial, Capricor is advancing CAP-1002 to phase 3 studies and preparing for commercial manufacturing of the drug.

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Capricor Therapeutics announced encouraging results from its phase 2 HOPE-2 trial of the investigational therapy CAP-1002 for treatment of individuals in advanced stages of DMD. The primary and secondary endpoints of the study — improvement of upper limb and cardiac function — were met, with slower disease progression seen in participants receiving the drug. Because of the positive study results, Capricor is moving forward with its phase 3 trial of CAP-1002.



The phase 2 trial followed 20 primarily nonambulant (unable to walk) young men in the later stages of DMD over a 12-month time period. Participants were randomly selected to receive intravenous (IV) administration of either CAP-1002 or a placebo control every three months. Assessments included shoulder, elbow, wrist, and hand weakness and function using the validated Performance of the Upper Limb test.

CAP-1002 was found to be safe and well tolerated, with data analysis showing improved skeletal and cardiac function after receiving four doses of CAP-1002 over the course of one year.

For more information about the HOPE-2 trial, visit clinicaltrials.gov and enter NCT03406780 in the “Other terms” search box.

MIS51ON Trial Enrolling Boys

Researchers at Rare Disease Research, LLC, are seeking 7- to 13-year-old boys living with DMD to participate in the phase 3 MIS51ON trial to evaluate the safety and efficacy of high-dose eteplirsen (Exondys 51) to treat DMD. Exondys 51 is designed to promote production of partially functional dystrophin protein and may improve motor function in boys with DMD.

MIS51ON is a two-part dose-finding and comparison study. In part 1 of the study, participants will know whether they are receiving high- or low-dose Exondys 51. In part 2 of the study, participants will be randomly assigned high- or low-dose Exondys 51.

The drug will be administered as an intravenous (IV) infusion. Participants will be assessed using tests such as the North Star Ambulatory Assessment, 6-minute walk test, timed 4-step stair ascend test, pulmonary function test, and muscle biopsy.



Study participants must attend weekly appointments to receive drug treatment.

To be eligible, individuals must have a mutation of the DMD gene amenable to exon 51 skipping, have been on a stable dose of oral corticosteroids for at least 12 weeks prior to randomization, and meet additional requirements.

Travel support is available for study participants and families.

To learn more about the study or inquire about participation, contact study coordinator Deanna Baker at deanna.baker@rarediseaseresearch.com or 678-883-6897.

Translarna Trial Enrolling



Researchers at Rare Disease Research, LLC, are seeking boys between 6 months and 2 years old who are living with DMD caused by a nonsense mutation (nmDMD) to participate in a phase 2 clinical trial. It will evaluate the safety and length of effectiveness of ataluren (Translarna) to treat nmDMD. Translarna is designed to correct nonsense mutations and may slow the decline in physical functioning that occurs in boys with nmDMD.

All participants will receive Translarna for the duration of the study. The trial will last approximately eight months for each participant, and participants will be required to complete six doctor visits, including a screening visit and

post-treatment follow-up. Three of the visits will require an overnight stay.

The drug will be administered as a liquid oral suspension. Effects of Translarna will be investigated by taking and processing blood samples for gene sequencing, pharmacokinetic studies, and other assessments.

Travel support is available for study participants and families.

To learn more about the study or inquire about participation, contact study coordinator Marcial Almaraz at marcial.almaraz@rarediseaseresearch.com or 678-883-6897.

3-year-olds Needed for SRP-9001 Trial

Researchers at Stanford Neuroscience Health Center are seeking 3-year-old boys living with DMD, particularly in California, to participate in an early-phase clinical trial (a new cohort of the ENDEAVOR trial) to evaluate efficacy of Sarepta Therapeutics' investigational gene replacement therapy SRP-9001 to treat DMD.

SRP-9001 uses an adeno-associated virus (AAV) to introduce a shortened version of the dystrophin gene into muscle tissue of boys with DMD for targeted production of shortened, but partially functional, dystrophin protein. Treatment with SRP-9001 has the potential to lead to functional improvements in boys living with DMD.

SRP-9001 is administered as an intravenous (IV) infusion. The safety and efficacy of SRP-9001 has previously been evaluated in multiple studies in boys ages 4 to 7 years. This study will investigate the outcome of SRP-9001 open-label treatment in a small cohort of 3-year-old boys. Eligibility criteria include:

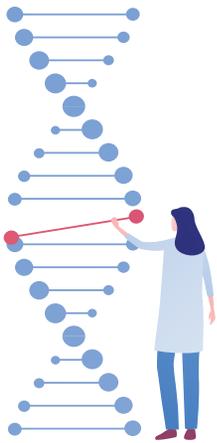
- Being male at birth
- Being 3 to 4 years old and ambulatory (able to walk) at the time of screening

- Having a definitive diagnosis of DMD prior to screening

To learn more about the study or inquire about participation, contact the Stanford Neuroscience Health Center's general portal at neuromuscularresearch@stanford.edu or **650-725-4341** and mention the ENDEAVOR trial, or contact study coordinator Lesly Welsh at lwelsh@stanford.edu or **650-497-3079**.

Myotonic dystrophy (DM)

REACH-CDM Trial Seeks Participants



Researchers at AMO Pharma Ltd are seeking children and adolescents living with congenital myotonic dystrophy (DM1), also known as Steinert's disease, to participate in the phase 2/3 REACH-CDM clinical trial to evaluate efficacy of the investigational drug tideglusib (AMO-02) to treat congenital DM1.

AMO-02 is designed to disrupt the section of repeated RNA (a chemical cousin to DNA) that causes congenital DM1, thereby rescuing a key protein,

GSK3B, that is critical for normal tissue development and function. AMO-02 has the potential to address the widespread symptoms caused by congenital DM1.

Participants will be randomly assigned to receive either AMO-02 or an inactive placebo control, administered as a strawberry-flavored liquid, over the five-month course of the study. Participants will be evaluated for various outcome measures including the primary outcome (Congenital DM1 Rating Scale, or CDM1-RS), caregiver and clinician ratings, functional abilities, and daily activities and tasks.

To be eligible, individuals must be 6 to 16 years old, have a diagnosis of congenital DM1, and meet additional requirements.

Travel support may be available for study participants and families.

To learn more about the study or inquire about participation, contact the study coordinator at info@reachcdm.com, or visit reachcdm.com.



Spinal muscular atrophy (SMA)

Communication Survey

Researchers at Russell Sage College in New York are seeking parents or legal guardians of individuals with SMA to participate in a survey about the communication abilities of affected individuals. Analysis of the current communicative behaviors used by individuals with SMA will help researchers develop communication systems that better address the needs of this population.

Approximately 50 parents or legal guardians of individuals diagnosed with SMA will be surveyed. Participants must provide consent to participate and then complete a one-hour phone or Zoom session with the investigator. During the session, the investigator will ask questions from two documents: case history and Inventory of Potential Communicative Acts (IPCA). All sessions will be recorded.

To be eligible, individuals must be a parent or legal guardian (18 years and older) of one or more individuals with a diagnosis of SMA, and the child with the SMA diagnosis must be at least 18 months old.

For more information, contact study coordinator Cheryl Ostry at ostryc@sage.edu.

A New Direction for Charcot-Marie-Tooth Disease

A Q&A with Stephan Zuchner, MD, PhD

Charcot-Marie-Tooth disease (CMT) is not one disease but a spectrum of nerve disorders. It is named after the three physicians who first described it in 1886: Jean-Martin Charcot and Pierre Marie of France and Howard Henry Tooth of the United Kingdom.

CMT affects the peripheral nerves that run through the arms and legs, connecting the brain and spinal cord to the muscles and sensory organs. It occurs in approximately 1 in 2,500 people.



Stephan Zuchner, MD, PhD

To learn more about CMT, we spoke with Stephan Zuchner, MD, PhD, a professor and chair of the department of Human Genetics at the University of Miami. Dr. Zuchner's scientific interests lie in mapping disease genes and genomic variation related to disease.

What causes CMT?

We also refer to CMT as “inherited neuropathies,” which indicates that these diseases have a genetic basis and, therefore, often run in families.

There are at least 100 different types of CMT, and every year, more disease-causing genes are being discovered. Each type of CMT is defined by the specific gene that is not working correctly. All CMT variances show similar clinical symptoms, but the severity of conditions vary with different genes.

What are the symptoms of CMT?

The most important symptoms of CMT are weakness and sensory deficits. When the nerves connected to the muscles do not work properly, individuals start having weakness in their feet and hands, which affects their ability to walk and their fine motor skills. Some end up using wheelchairs, and many struggle with basic tasks such as buttoning up a shirt or using utensils. People with CMT also struggle with day-to-day sensory functions, such as feel, touch, and temperature.

Have there been any recent advances in treatment?

Although no treatments currently can stop or reverse CMT, we have seen over the last decade increased interest within



the pharmaceutical industry and exciting new ideas for how to develop and test new therapies. There is an optimism in the field that things like genetic therapies could really begin to make a difference for CMT patients.

What treatments are being researched?

A small number of clinical trials are underway or planned for specific types of CMT, such as CMT1A and SORD neuropathy. The latter originated from our own gene discovery work and will likely go from publication to trial treatment in 24 months. There are also several projects in the research stage using genetic therapies to correct the genetic cause of CMT.

In general, genetic therapies are a fairly new type of medicine, so there are still many questions surrounding these treatments — but clearly, the industry is moving in this direction. In fact, an approved genetic therapy for spinal muscular atrophy (SMA), is a very good example of this approach, because it targets the same nerve cells that are not working correctly in CMT. [Q](#)

HOW IS CMT INHERITED?

CMT can run in a family, even when there is no obvious family history. The condition can be inherited in three different ways — autosomal dominant, autosomal recessive, and X-linked — and some of these inheritance patterns can be difficult to trace through a family tree. To learn more about inheritance of neuromuscular diseases, read “Family Inheritance” on page 28.

+RARE DISEASE SPOTLIGHT

Read more Q&As about rare diseases at mda.org/quest/spotlight.

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*Based on commercial patients, early access patients, and clinical trial participants through December 2020.

†Includes clinical trial patients.

‡Clinical studies of SPINRAZA did not include sufficient numbers of subjects aged 65 and over to determine whether they respond differently from younger patients.

§Clinical studies of SPINRAZA included patients from 3 days to 16 years of age at first dose.

§Based on commercial patients in the US (including Puerto Rico) through December 2020.

INDICATION

SPINRAZA® (nusinersen) is a prescription medicine used to treat spinal muscular atrophy (SMA) in pediatric and adult patients.

IMPORTANT SAFETY INFORMATION

Increased risk of bleeding complications has been observed after administration of similar medicines. Your healthcare provider should perform blood tests before you start treatment with SPINRAZA and before each dose to monitor for signs of these risks. Seek medical attention if unexpected bleeding occurs.

Increased risk of kidney damage, including potentially fatal acute inflammation of the kidney, has been observed after administration of similar medicines. Your healthcare provider should perform urine testing before you start treatment with SPINRAZA and before each dose to monitor for signs of this risk.

The most common side effects of SPINRAZA include lower respiratory infection, fever, constipation, headache, vomiting, back pain, and post-lumbar puncture syndrome.

These are not all of the possible side effects of SPINRAZA. Call your healthcare provider for medical advice about side effects. You may report side effects to FDA at 1-800-FDA-1088.

Before taking SPINRAZA, tell your healthcare provider if you are pregnant or plan to become pregnant.

Please see full Prescribing Information on SPINRAZA.com.

This information is not intended to replace discussions with your healthcare provider.



SPINRAZA[®]
(nusinersen) injection
12 mg/5 mL

BYRON // AGE 15

LATER-ONSET SMA, TREATED WITH SPINRAZA

Individual results may vary based on several factors, including severity of disease, initiation of treatment, and duration of therapy.

[Learn more at SPINRAZA.com](https://www.spinraza.com)

IMPORTANT FACTS ABOUT SPINRAZA[®] (nusinersen)**USES**

SPINRAZA is a prescription medicine used to treat spinal muscular atrophy (SMA) in pediatric and adult patients.

WARNINGS

Increased risk of bleeding complications has been observed after administration of similar medicines. Your healthcare provider should perform blood tests before you start treatment with SPINRAZA and before each dose to monitor for signs of these risks. Seek medical attention if unexpected bleeding occurs.

Increased risk of kidney damage, including potentially fatal acute inflammation of the kidney, has been observed after administration of similar medicines. Your healthcare provider should perform urine testing before you start treatment with SPINRAZA and before each dose to monitor for signs of this risk.

COMMON SIDE EFFECTS

- **The most common side effects of SPINRAZA include** lower respiratory infection, fever, constipation, headache, vomiting, back pain, and post-lumbar puncture syndrome (headache related to the intrathecal procedure).
- Serious side effects of complete or partial collapse of a lung or lobe of a lung have been reported.

Talk to your healthcare provider about any side effect that bothers you or that does not go away.

OTHER INFORMATION

SPINRAZA is a medication that should be administered as an injection into the lower back (a procedure called intrathecal injection) by, or under the direction of, an experienced healthcare professional.

Before taking SPINRAZA, tell your healthcare provider if you are pregnant or plan to become pregnant.

QUESTIONS?

The risk information provided here is not comprehensive. To learn more, talk about SPINRAZA with your healthcare provider or pharmacist. The FDA-approved product labeling can be found at [www.SPINRAZA.com](https://www.spinraza.com) or 1-844-4SPINRAZA (1-844-477-4672).

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The Hidden Benefits of Volunteering

Helping others can help you build valuable skills

BY DARLENE DEMETRI

Blaine, a 6-year-old black lab, is assistance dog extraordinaire to Bella Andrade, 19, who lives with spinal muscular atrophy (SMA) and uses a power wheelchair. Receiving Blaine from the Minnesota nonprofit Can Do Canines in October 2017 was a life-changing gift for Bella. With a verbal command, Blaine will open a drawer, push a handicap door access button, or retrieve a dropped pen or smart phone.

This was a big change for Bella, who was thrilled with her newfound independence.

“Can Do Canines is a unique organization because all their service animals are placed with their owners free of charge,” Bella says. “I wanted to give back to them because they changed my life for the better, and I wanted to help others with disabilities.” She volunteered for the organization’s ambassador program, which provided her with speaker training.

With Blaine at her side, Bella gave talks about Can Do Canines at the local Lions Club, children’s hospital, fire department, and elementary school. In the process, she met and built relationships with people from different walks of life.

This experience ultimately gave Bella the confidence and courage to leave her home in Minnesota to attend Arizona



State University (ASU), where she is a freshman studying biological sciences and genetics.

“Blaine really helped relieve my anxiety about going places on my own,” says Bella. “And through the speaking engagements, I realized that I’m great at public speaking and can build

HOW TO FIND THE RIGHT VOLUNTEER OPPORTUNITY

- 1. Follow your interests.** Align with a cause that’s important to you or that you want to learn more about. Dedicated volunteers make a lasting impression, and, in some cases, this could lead to a job or references.
- 2. Be direct.** Even if there is a volunteer sign-up portal, it’s a good idea to contact the volunteer coordinator directly. Their objective is to match the organization’s needs with what excites you, so be clear about your skills and interests.



- 3. Seek an inclusive environment.** Look at an organization’s website for information about diversity or disability. Consider asking the volunteer coordinator how they would make reasonable accommodations for your needs and for others with disabilities.
- 4. Find strength in numbers.** Do you have family, friends, or coworkers also looking to volunteer? Volunteering as a team is rewarding and wonderful for community building.

RESOURCES FOR VOLUNTEERING

The best volunteer opportunity is one that aligns with your interests. These organizations are a good place to start looking:

AmeriCorps
americorps.gov

JustServe
justserve.org

MDA
mda.org/volunteer

Points of Light
pointsoflight.org/for-volunteers

United Way
unitedway.org/get-involved/volunteer

VolunteerMatch
volunteermatch.org



connections with others, too. I came to ASU, where I didn't know anyone, and I've built a whole community of friends."

Résumé builder

Bella isn't the only one who's discovered that the benefits of volunteering go beyond the intrinsic value of giving back.



Carl Pettitt gained valuable work experience as a volunteer.

Carl Pettitt, 21, contacted MDA in January 2021 looking to satisfy a college volunteering requirement. A technology wiz who lives with Duchenne muscular dystrophy (DMD), Carl assisted with software applications, e-newsletters, volunteer training, and fundraising initiatives. After graduating from Blue Ridge Community and Technical College in West Virginia in the spring, Carl found that the time he spent volunteering had provided new skills and solid work experience for his

résumé. "Volunteering with MDA has greatly assisted my job search," says Carl, who has some promising leads.

According to Alison Tibbits, senior national director of volunteerism and organizational partners at MDA, one of the many benefits of volunteering is that it can help you identify new career paths by uncovering skills and passions you didn't know you had. These skills and passions may even present you in a positive light to potential employers.

Expanding opportunities

Currently, there are many worthy organizations to volunteer for, and the need for dedicated volunteers is high. As a result,

many organizations are striving to curate opportunities with inclusivity and diversity in mind. "It's no longer a one-size-fits-all approach," Alison says. "Tailoring to a volunteer's interests and skills is paramount."

In addition, the virtual landscape offers greater flexibility for volunteers and more options for organizations to reach more people.

"The COVID-19 pandemic has caused organizations to think very differently about what they can empower volunteers to do," Alison says. As with other aspects of life, tasks that involve communicating, planning, organizing, or even meeting with individuals face-to-face can be done virtually now.

"We have seen so much success when our dedicated volunteers have supported our key programs virtually," Alison says. "It really goes back to creating volunteer opportunities that are inclusive and diverse." 

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



Did You Know?

The employment rate for people with disabilities was on the rise from 2019 to 2020. Use these resources to learn more about working with a disability:

JAN (Job Accommodation Network)
askjan.org

MDA Advocacy Institute: Employment Rights for People Living with a Disability
strongly.mda.org/do-you-know-your-employment-rights

Social Security Administration: Continued Medicaid Eligibility
ssa.gov/disabilityresearch/wi/1619b.htm

Social Security Administration: Ticket to Work
choosework.ssa.gov

US Equal Employment Opportunity Commission: Disability Discrimination
eeoc.gov/disability-discrimination

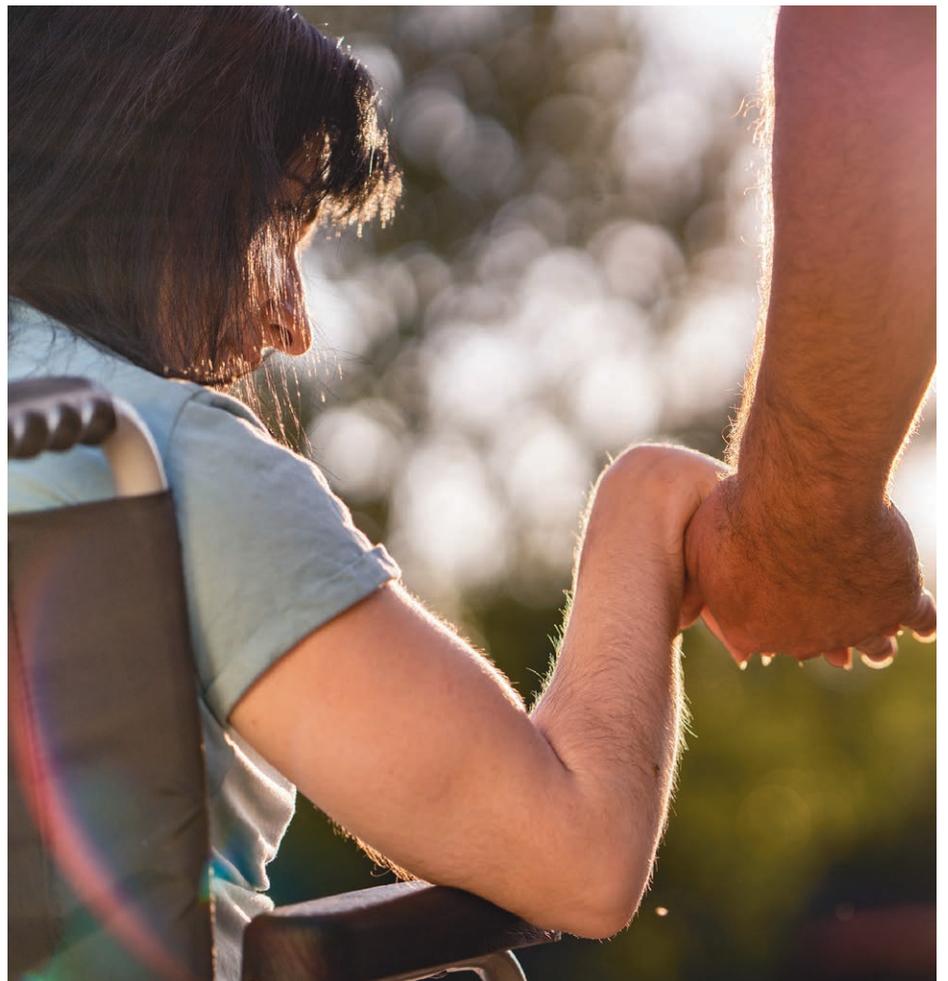


Trust. Honesty. Mutual respect. Affection. The key elements of a healthy romantic relationship are the same for all couples. When one or both partners has a disability, it can affect how they each approach daily tasks, but it doesn't change the need for constant give-and-take.

For adults living with neuromuscular disease, building and nurturing positive intimate relationships may even be especially important, because there are unique dynamics due to physical limitations or the progressive nature of the disease. Fortunately, couples can rely on tried-and-true strategies for forming healthy relationships.

Foremost among those strategies is open and honest communication.

“Share what you're thinking or feeling with your partner; holding things in just creates resentment,” says Briana Mills, an associate marriage and family therapist



Images: iStock.com/kail9; StockPlanets; Teraphim; BartCo



ve lationships

How adults with neuromuscular disease can form and nurture healthy romantic relationships

BY KAREN DOSS BOWMAN



living with SEPN1 muscular dystrophy in the Los Angeles area. “It’s important to talk about what you need and want. Also, have empathy for your partner and try to understand where they’re coming from.”

Setting and respecting boundaries helps establish the trust needed for open communication. That means valuing a partner’s identity and preferences while also supporting their needs. Having boundaries in place also can help establish roles and expectations.

“Understanding what defines a healthy relationship for you and your partner will help you work toward that — it may look different for any couple, including those living with neuromuscular disease,” says Sarah Stoney, MSW, LSW, a social worker at Children’s Hospital of Philadelphia.

A good start

Finding love is never easy. But navigating the world of dating and romance can be especially challenging for teenagers and adults living with disabilities — many of whom don’t start dating until their 20s.

“There is a prevailing stereotype that people who have a physical disability are not desirable. And whether directly or indirectly, most of us who are disabled get that message growing up,” says Briana.

The truth is, every person is capable of and deserving of love. Entering a relationship with that belief in mind helps establish it on a basis of mutual respect.

Danielle Sheypuk, PhD, a clinical psychologist living with spinal muscular atrophy (SMA) in New York City, says



“Cultivate your self-image as a person who is dateable, desirable, and has a lot to offer.”

— Danielle Sheypuk, PhD

a good starting point is to cultivate one’s “dateable” self-esteem.

“I can say that the key to successfully establishing a healthy relationship is to change your perception of yourself,” she says. “Cultivate your self-image as a person who is dateable, desirable, has a lot to offer a partner, and is capable of finding romantic love.”

This self-confidence helps a person not only stop defining themselves according to their relationship status, but once they do find someone, it helps them articulate what they want and need from their partner.

“It is important to remember that despite physical limitations, all individuals are entitled to a healthy sexuality and intimacy if that is what they desire,” Sarah says. “That’s why it is so important to talk about sexual health openly in order to destigmatize talking about sex and disabilities.”



Dual roles: partner and caregiver

For interabled couples — those in which one partner is disabled and the other is not — it is common for a romantic partner to also take on the role of caregiver.

Couples should find ways to balance the two roles. Talk about the frustrations and challenges of caregiving, but also make time for fun and intimacy — not just sex, but holding hands, hugging, and looking into each other’s eyes.

“Treat your partner lovingly, and keep the spark alive,” says Briana, whose boyfriend of six years is her caregiver.

Every couple has arguments, but they can lead to added tension when a significant other also is a caregiver. For example, what happens if a couple has a fight, and then the caregiver must help their partner to bed? Dr. Sheypuk recommends both people separating into different rooms and taking some time to calm down.

“When emotions are running hot, it’s difficult to think clearly and effectively,” Dr. Sheypuk says. “So separate and cool down — perhaps by meditating — and then reconvene to resolve the issue.”

Actively listening to each other and being willing to compromise can help partners resolve these situations and move forward.

“In a healthy relationship, the caregiver would know and understand that their role as caregiver is obviously still needed despite an argument,” Sarah says. “Failure to do so could be seen as manipulative or abusive. If you are in an unhealthy relationship and your partner is your caregiver, then certainly enlisting the assistance of either professionals or close friends and family that you trust to develop a plan to ensure your safety is of utmost importance.”



Understanding what defines a healthy relationship for you and your partner will help you work toward [setting boundaries].

— Sarah Stoney, MSW, LSW



Guarding against abuse

The Centers for Disease Control and Prevention (CDC) National Intimate Partner and Sexual Violence Survey found that about 1 in 4 women and nearly 1 in 10 men have experienced some type of abuse from an intimate partner. And, the CDC says, people with disabilities are at increased risk of experiencing these types of violence.

As a teen and young adult, Briana struggled with self-esteem, and in her early 20s she found herself in an abusive relationship with her first serious boyfriend. He was emotionally manipulative, going back and forth between professing his love and telling her he didn't love her, and he demanded physical intimacy without considering what she wanted.

"I was very naive and didn't know much about relationships," Briana says. "I had no idea at first that what he was doing was abusive because he said he loved me. I thought that this must be what love is."

Briana eventually got out of the relationship and today, as a therapist, she strives to help her clients with disabilities realize the value they bring to their relationships and empower them to recognize and escape abusive situations.

The most important way to safeguard against abuse is to know the warning signs. These can encompass a wide range of behaviors, including:

- Controlling behavior
- Name-calling or belittling
- Financial control
- Withholding medications or treatments
- Destroying medical equipment
- Inflicting physical harm
- Isolating the victim from their friends and family

These steps can help safeguard against an abusive relationship:

- **Maintain a strong network of family and friends.** Stay in touch with people who will look out for your best interests and would be willing to step in if you feel uncomfortable or afraid.
- **Use technology to stay connected.** Keep your primary communication device close by, such as your smartphone

“

Share what you're thinking or feeling with your partner; holding things in just creates resentment.

— Briana Mills



RELATIONSHIP RESOURCES

Use these resources to learn about recognizing unhealthy relationships and find support.

Centers for Disease Control and Prevention (CDC): Intimate Partner Violence
cdc.gov/violenceprevention/intimatepartnerviolence

love is respect
loveisrespect.org
866-331-9474

National Domestic Violence Hotline
thehotline.org
800-799-SAFE (7233)

National Sexual Violence Resource Center
nsvrc.org

RAINN (Rape, Abuse & Incest National Network)
rainn.org
800-656-HOPE (4673)

or iPad. If an abuser takes those away, think about alternative ways to communicate, such as using a voice-activated device, like Alexa, to call for help. A wearable device, like an Apple watch, also is a good idea.

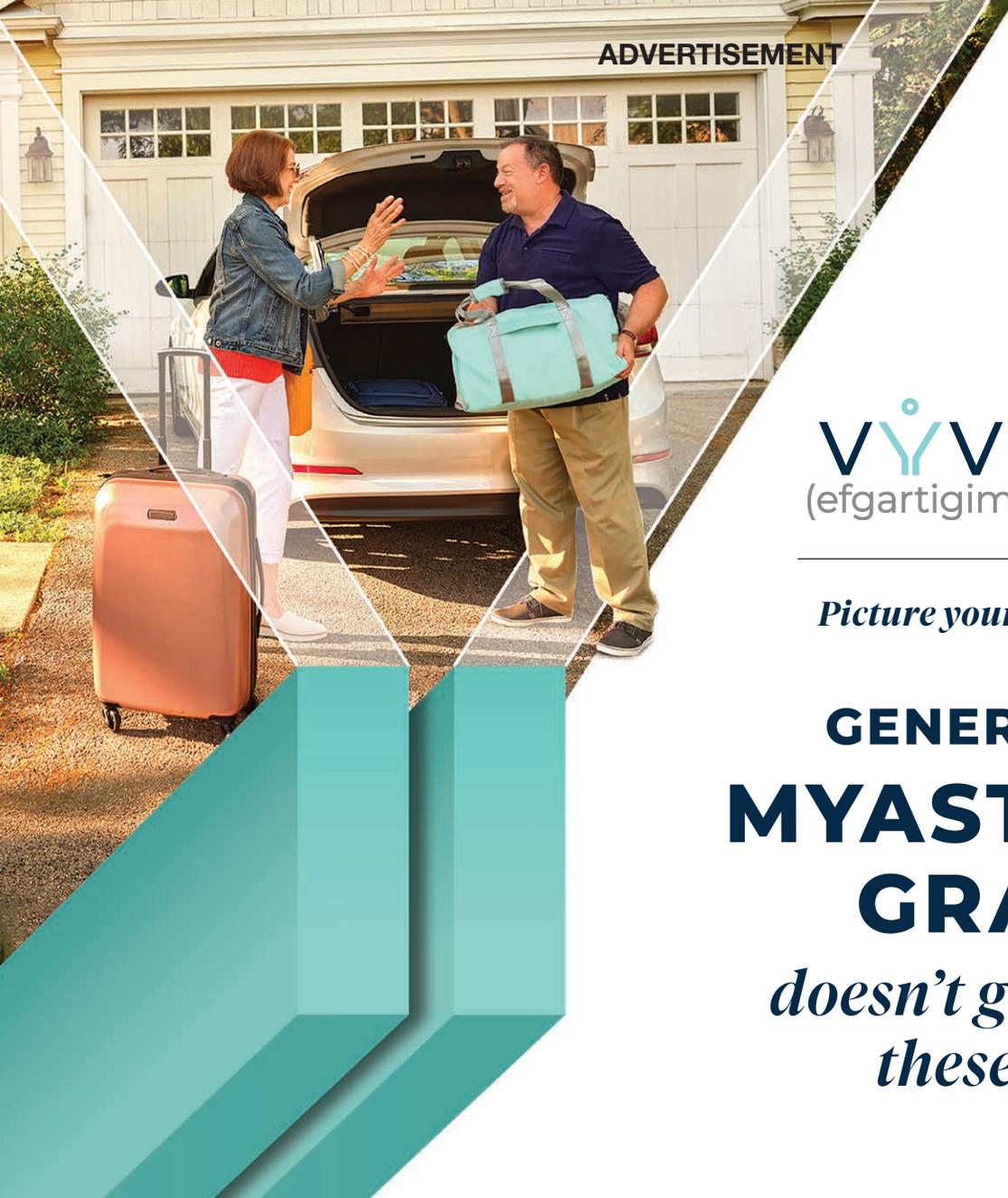
- **Seek help.** Reach out to your healthcare provider, a mental health professional, or a support organization (see "Relationship Resources" above). Or talk to a friend or family member you trust. Don't hesitate to call 911 if you feel that you're in immediate danger.

Most important, Dr. Sheypuk adds, don't settle for a relationship that feels uncomfortable or oppressive in some way. "It's important not to feel trapped in a relationship," she says. Trust your gut — if something doesn't feel right, it probably isn't.

"Sometimes people with disabilities feel like, 'I need to stay in this relationship because I'll never find someone else,' or, 'This is as good as it gets,'" Dr. Sheypuk says. "Nothing about these statements is true."

A healthy relationship built on trust, respect, and affection should make you feel good about yourself and the person you're with. "Your significant other should elevate you as a person and bring out the best of you," Dr. Sheypuk says. 

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



VYVGART™
(efgartigimod alfa-fcab)

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*doesn't get to make
these plans*

What is VYVGART™ (efgartigimod alfa-fcab)?

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

IMPORTANT SAFETY INFORMATION

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

VYVGART may cause serious side effects, including:

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

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

- **Undesirable immune reactions (hypersensitivity reactions).** VYVGART can cause the immune system to have undesirable reactions such as rashes, swelling under the skin, and shortness of breath. In clinical studies, the reactions were mild or moderate and occurred within 1 hour to 3 weeks of administration, and the reactions did not lead to VYVGART discontinuation. Your health care provider should monitor you

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

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

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

Improved daily abilities

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

Reduced muscle weakness

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

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

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

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

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

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

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

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

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

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

What are the common side effects of VYVGART?

The most common side effects of VYVGART are respiratory tract

infection, headache, and urinary tract infection.

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

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

Please see brief summary of full Prescribing Information on next page.

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



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Working Together



Anyone who lives with a neuromuscular disease that affects mobility knows that it often takes a village of caregivers to get through a week, let alone a day. For many people, personal care attendants (PCAs) provide a large part of that caregiving.

PCAs are professionals who provide home care to people with disabilities who need assistance with daily living activities. This can range from helping them get ready for the day to helping them take medication to performing chores and errands.

For Keisha Greaves, who was diagnosed with limb-girdle muscular dystrophy (LGMD) at 24, having full-time caregiving is a new experience. Her mobility declined slowly over the decade after her diagnosis, but in 2021, Keisha had a bad fall. She has not been able to walk and has relied on PCAs for daily living tasks ever since. Over the past year, she's learned that hiring PCAs, managing schedules, and navigating her own needs along with the varying personalities of caregivers is like taking on a new job. While having PCAs allows her to live independently and pursue her work as CEO of the adaptive clothing brand she founded, Girls Chronically Rock, it also is an added source of stress for her to manage.

How to establish and maintain good relationships with personal care attendants

BY JOANNA
BUONICONTI



Keisha Greaves has fun with her PCAs Aischa Badio (left) and Max Toussiant (right).

For example, on a recent morning, one of her PCAs called a few minutes after her shift was supposed to begin to say she had injured her hand and needed to visit the doctor. Keisha managed by herself until her next PCA arrived in the afternoon.

Keisha was sympathetic to the fact that her caregiver was hurt, but also annoyed that her PCA waited hours to tell her that she would not arrive at all that day. And she wasn't sure if it was appropriate — or helpful — to tell the PCA about her feelings.

"I do get upset with my caregivers sometimes," Keisha says. "But I say to myself, 'My life is in their hands.'" She holds side by side the knowledge that she is the employer in this situation, and that she is vulnerable if her PCAs don't do their job well or decide to quit.

Navigating relationships with PCAs is a tricky balance between a professional relationship and an intimate connection. Here, people with experience working with and as PCAs weigh in on how to build good relationships with your PCAs.

What is a good working relationship?

Like every other relationship under the sun, those between PCAs and clients can be complicated. To foster any healthy relationship, both parties must advocate for their respective needs. Communication is key to maintaining a good caregiver-client relationship.

It helps to establish those lines of communication from the start. "Both the PCA and client have their roles and expectations of each other," says Ann Hogan, a former service coordinator for the Pennsylvania Medicaid Waiver Program. "It's

important to break down what the roles and expectations look like for each of them and have a one-on-one conversation first to understand how to move forward."

For example, Ashleigh Peska, an MDA resource specialist who lives with LGMD and uses a power wheelchair, has been living independently with PCAs helping her full-time since she went to college in 2007. When she was a young adult, some PCAs took a "mothering" approach. She learned that one thing that's important to her is that a PCA listens to her instructions and recognizes that Ashleigh knows her care best.

From the PCA's perspective, it's helpful to know which daily tasks or routines must be followed exactly and where there's flexibility. "There's always a schedule of things, from hygiene care to medications, that I'm required to do in a shift," says Taylor Masse, who has been a PCA for more than four years while attending nursing school. "As long as everything gets done correctly, I try to make sure that my clients are in control of how their day plays out."

Having well-defined roles and routines can help both client and caregiver relax, and maybe even have fun together.

"Just having that healthy balance between me and the caregivers is important," Keisha says. "I'm still young and I like to have fun and joke around, and I enjoy doing that with my PCAs and vice versa. I am also dealing with a lot living with LGMD, so having that laughter definitely helps."

Still, boundaries must be clear. "When there's downtime, we can joke around and have fun, but when we're doing caregiving, it is important to focus on that," Ashleigh says.

Managing relationships with PCAs

Keeping a team of caregivers running smoothly can feel like a big responsibility.

"Sometimes it feels like PCAs are my number one cause of stress," Keisha says. "It's a balancing act, and that's something that I talk to my therapist about. I'm trying to work on patience and taking deep breaths when I get overwhelmed, because sometimes that helps."

Establishing effective communication, clear expectations, and predictable routines can keep managing PCAs from feeling overwhelming.

“

It's a balancing act, and that's something that I talk to my therapist about. I'm trying to work on patience and taking deep breaths when I get overwhelmed, because sometimes that helps.

— Keisha Greaves



HOW TO FIND A PCA

There are two ways to find personal care attendants (PCAs): use a home care agency or hire privately. Each has advantages and disadvantages.

Hiring privately provides more control over choosing your PCAs, negotiating pay, and arranging schedules. Many people find PCAs by asking family and friends for recommendations. Others post ads on Facebook forums or college campuses.

On the other hand, agencies can take some of the work off your hands by matching you with prescreened caregivers based on your needs and coordinating care schedules.

However you decide to find PCAs, here are resources to help:

- › MDA's Independent Living and PCA Support Resources page includes tips and resources for finding and managing PCAs. mda.org/young-adults/independent-living
- › Check out the Family Caregiver Alliance's information and resources on hiring in-home caregivers. caregiver.org/resource/hiring-home-help
- › Medicare offers a tool to find and compare home health agencies. medicare.gov/care-compare
- › Use AARP's Caregiving Checklist to gather the information you need to choose a home health agency. assets.aarp.org/external_sites/caregiving/checklists/checklist_inHomeCare.html

Image: iStock.com/Tera Vector

During the COVID-19 pandemic, Ashleigh's fiancé has been her primary caregiver, but when she had a team of caregivers in the past, she would arrange monthly meetings to review schedules and routines with all her PCAs at once. The added benefit was that her PCAs knew each other and could contact one another if they needed someone to fill their shift.

Using a home care agency can take some of that coordination off your plate. The agency will assess your needs and develop a care plan that includes caregivers qualified to provide those services. When you work with an agency, you can turn to the service coordinator or a supervisor if you're having trouble with a PCA. Some agencies will visit with the client in their home while the PCA is there to monitor interactions and mediate disputes.

However, whether you use an agency or hire PCAs privately, it helps to start by addressing any issues directly

with your PCA. Ashleigh found it helpful to approach these conversations kindly, explaining the harm that a particular issue can have on her health and offering an easy solution. For example, she once had a caregiver who bit her fingernails, which made her nails jagged. Ashleigh was on a blood thinner at the time and worried that if her PCA accidentally scratched her, the medicine would prevent normal clotting. Ashleigh explained the problem to her PCA and asked her to run a file over her nails before her shifts. The caregiver was able to incorporate this step into her routine.

When addressing tensions or conflicts, the PCA and client should work together toward a resolution. According to nursing student Taylor, most PCAs are open to constructive criticism, especially as they're becoming familiar with a client's needs. "I like to be told straight out what the issue is, and then focus on how we can fix it," Taylor says. "I don't like to tiptoe around things, since this is someone's life that I am hoping to enhance by assisting with their daily activities."

Having and maintaining positive relationships with caregivers can make a big difference in quality of life and maintaining independence. Good working relationships can reduce PCA turnover rates and stress levels on both parties. And, in some cases, it will lead to long-term, rewarding relationships between an individual who wants to live their life to the fullest and a caregiver who wants to empower them to do so.

"It's about the care that you and your client have for each other," says Asia Tucker, who has been a PCA for 11 years. "I want my clients to live their life having fun, laughing, celebrating — anything they love to do, I am willing to help them do it." 

Joanna Buoniconti is a freelance writer living with spinal muscular atrophy (SMA) in Western Massachusetts.



Ashleigh Peska enjoys a day with James Ocasio, her fiancé and caregiver.

In genetics, inheritance doesn't refer to property or financial assets. It is the process by which genetic information is passed from parents to children.

Most neuromuscular diseases are genetic disorders, meaning they are caused by changes in our genes. While these changes, called gene mutations or variants, often are inherited from parents, they don't always cause disease. For many neuromuscular diseases — and the number grows every year with continuing research — scientists have identified disease-causing gene variants. They also have been studying the inheritance patterns, meaning how a gene variant may be passed from one generation to another and how it might affect members of a family.

Knowing a disease's inheritance pattern helps us understand who else in a family is at risk for the same condition. For example, 5q spinal muscular atrophy (SMA) is an autosomal recessive condition (see "Inheritance patterns" on page 30), meaning a person must inherit a disease-causing gene variant from each parent to have the disease. A person who inherits the gene variant from only one parent is a carrier.

"With 5q SMA, we know about one in 40 to one in 50 people in the United States are carriers, and many don't know it," says Elicia Estrella, MS, LCGC, a genetic counselor at Boston Children's Hospital. "If you have a child with SMA and find out you and your partner are carriers, then you know other family members could be carriers. You can let your brothers and sisters who are of childbearing age know, and they can get genetic testing." A carrier of a 5q SMA-causing gene variant has a 50% chance of passing that gene variant to their child.



How genetic neuromuscular disease is passed from one generation to another

BY AMY BERNSTEIN



Image: iStock.com/DrAfter123

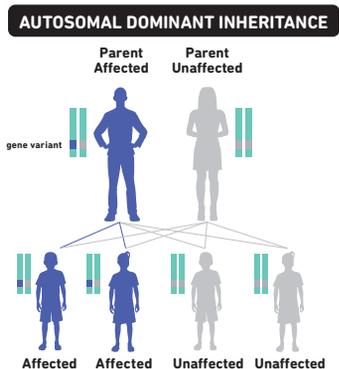
Inheritance patterns

Every person has two copies of each gene — one from their mother and one from their father. An inherited genetic condition can be passed along in the following ways.

AUTOSOMAL DOMINANT

When someone has one copy of the disease-causing gene and displays symptoms of the condition, this is called

autosomal dominant. He or she has a 50% chance of passing on the disease-causing gene variant and a 50% chance of passing on a gene without the variant to each child they have.

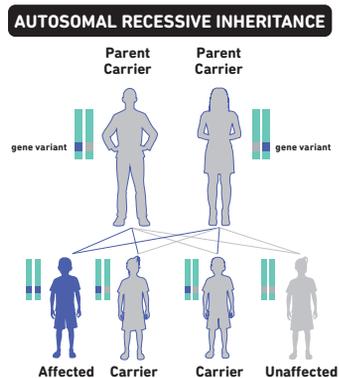


Examples: Myotonic dystrophy (DM), facioscapulohumeral muscular dystrophy (FSHD), limb-girdle muscular dystrophy type 1 (LGMD1), oculopharyngeal muscular dystrophy (OPMD), Thomsen type myotonia congenita, paramyotonia congenita, some types of Charcot-Marie-Tooth disease (CMT)

AUTOSOMAL RECESSIVE

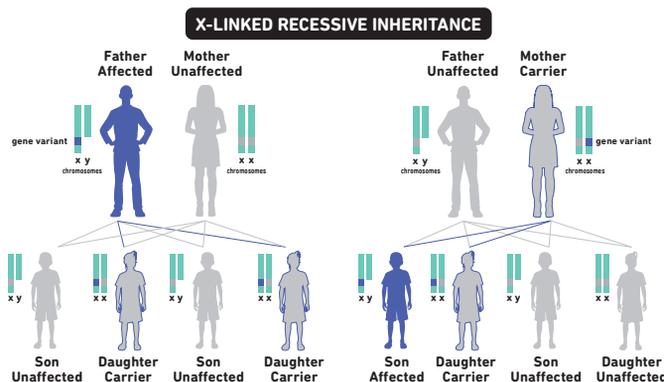
In an autosomal recessive condition, a person must have two copies of the disease-causing gene variant to have

symptoms. A carrier is a person who has one copy of the gene variant and typically does not show signs of the condition. When a mother and father are both carriers of the gene variant, each child has a 25% chance of having the genetic condition and a 50% chance of being a carrier.



Examples: most types of spinal muscular atrophy (SMA), most types of congenital muscular dystrophy (CMD), limb-girdle muscular dystrophy type 2 (LGMD2), Becker type myotonia congenita, Friedreich's ataxia (FA), most metabolic myopathies, most types of congenital myasthenic syndrome (CMS), some types of CMT

X-LINKED RECESSIVE

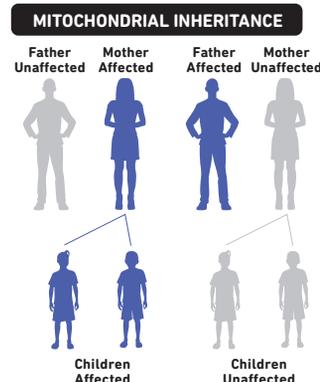


Females have two X chromosomes, and males have one X and one Y chromosome. In an X-linked recessive genetic condition, the gene with the disease-causing variant is on an X chromosome. Men pass an X chromosome to their daughters and a Y chromosome to their sons. Therefore, a father cannot pass an X-linked condition to his sons, but he will pass it to all his daughters. Women who carry an X-linked gene variant have a 50% chance of passing the variant to their sons and daughters. X-linked recessive genetic conditions generally are more severe in boys because they do not have a backup X-linked gene. Girls generally have milder symptoms or are asymptomatic carriers.

Examples: Duchenne muscular dystrophy (DMD), Becker muscular dystrophy (BMD), Emery-Dreifuss muscular dystrophy (EDMD), spinal-bulbar muscular atrophy (SBMA), some types of CMT

MITOCHONDRIAL

Mitochondria are structures found within our cells that produce energy. Each mitochondrion contains its own small piece of DNA. All our mitochondria are inherited from our mother's egg cell, so fathers do not pass on mitochondrial genes. Therefore, a woman who carries a disease-causing mitochondrial gene variant will pass it to all her children. However, many factors influence how a mitochondrial gene variant can affect a person, and the timing and severity of symptoms may differ from those of the mother.



Examples: Kearns-Sayre syndrome (KSS), myoclonus epilepsy with ragged red fibers (MERRF)

THE UNPREDICTABLE PATTERN OF MYOTONIC DYSTROPHY

Myotonic dystrophy (DM), an autosomal dominant inherited disease, is the most common form of muscular dystrophy in adults. It usually begins in the 20s or 30s, although the age of onset, severity, and prognosis can vary greatly from person to person.

This is partly because there are two types: Type 1 (DM1) is caused by a mutation, or variant, in the *DMPK* gene. Type 2 (DM2), which tends to be milder, is caused by a variant in the *ZNF9* gene. More significantly, DM1 exhibits an unusual genetic phenomenon called anticipation.

Anticipation means the symptoms of a genetic disease begin earlier in life and become more severe as the disease is passed on through generations. This genetic monkey wrench can catch some families by surprise.

“We knew nothing about myotonic dystrophy,” says Lisa Harvey-Duren, whose daughter, Kayla, was born in 2005 with congenital myotonic dystrophy, the most severe form of DM1. Kayla did not move or cry after birth and required a ventilator with a tracheostomy and feeding tube before she could go home. “We were all dumbfounded. We learned that the congenital form usually passes down from the mom, and I had been a two-sport Division I athlete in college and didn’t have any signs or symptoms of it.”

However, after Kayla’s diagnosis, it didn’t take long for Lisa to realize that her mother’s series of falls could be signs of adult-onset DM1. Lisa and her mom both had genetic tests that

looked for expanded cytosine-thymine-guanine (CTG) repeats in the *DMPK* gene, which is the gold standard for diagnosing DM1 and helps measure its severity. Her mother had 72 CTG repeats, which is considered higher than normal but not severe, and Lisa had slightly higher repeats at 75. “Kayla was diagnosed with 2,538 CTG repeats, which is just off the charts,” Lisa says. “It explained her severity. So the jump in the anticipation from me to Kayla was huge.”

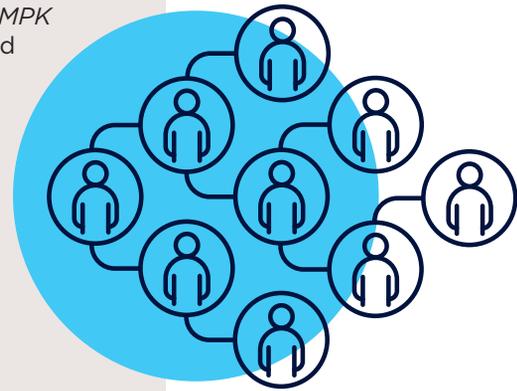
This is part of the confounding inheritance pattern of DM1. While it is known to become more severe when passed down from mother to child, it is an unpredictable advance. It can even affect siblings who inherit the disease-causing gene variant differently depending on how large the gene repeat becomes and how it alters the function of the cells.

“It’s unknown why we see anticipation expansion grow to a larger or smaller extent,” says Elicia Estrella, MS, LCGC, a genetic counselor at Boston Children’s Hospital. For many people with adult-onset DM, the signs and symptoms progress slowly and might not be recognized for years. That’s why it’s important to look at whole families when one family member is diagnosed with DM. “When we see it, we can talk to families about the risk to other family members,” Elicia says.

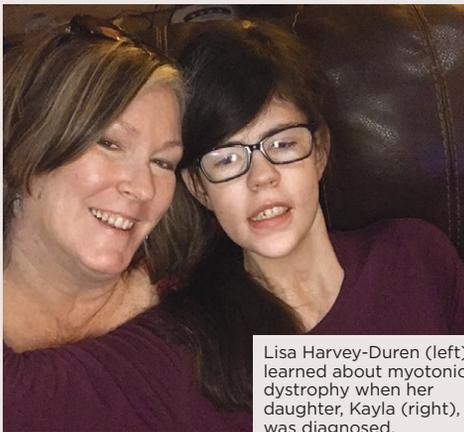
After Kayla’s diagnosis, Lisa reached out to all her first cousins on her mother’s side to let them know about the risk of passing this disease in the family. She now knows of other family members diagnosed with DM.

In addition to revealing DM in the family, which allowed them to learn about and deal with the disease, Lisa says that Kayla’s life was a gift.

Kayla was not expected to live past her first birthday, but she defied the odds, living to 13 years old and proving she was much more than her disease. “She was able to learn, she was able to thrive, she was super happy, and she made a lot of friends,” says Lisa.



Type 1 (DM1) is caused by a mutation, or variant, in the *DMPK* gene. Type 2 (DM2), which tends to be milder, is caused by a variant in the *ZNF9* gene.



Lisa Harvey-Duren (left) learned about myotonic dystrophy when her daughter, Kayla (right), was diagnosed.

+TESTING RESOURCES

For more information about finding genetic testing, including available no-cost programs, contact MDA's Resource Center at [833-ASK-MDA1](tel:833-ASK-MDA1) or resourcecenter@mdausa.org.

A parent's perspective

While knowledge is power, in the case of genetic inherited disease, it can lead to strong emotions. A parent's instinct is to protect their child. Knowing they passed down a gene that caused a disease can lead to feeling responsible for causing the disease.

"This is not something that you have control over," Elicia counsels her clients. "You have no reason to feel guilt or shame or anything for passing on your genes; it's a random event." But she acknowledges that families may need some time to come to terms with the knowledge.

"It's easy to let yourself go down a path that makes you feel like you're guilty for passing this along," says Lisa Harvey-Duren, whose daughter, Kayla, was born with myotonic dystrophy (see "The Unpredictable Pattern of Myotonic Dystrophy" on

GOT QUESTIONS ABOUT GENES?

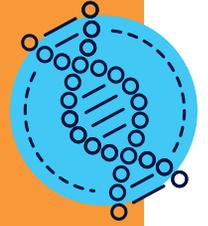
MDA has more resources to help you learn about genetics and genetic testing.

Visit mda.org/education and search for:

- Genetics and Neuromuscular Disease
- Basics of Genetic Testing
- How Genetic Neuromuscular Disease Happens

Visit mda.org/quest/research and search for:

- The Importance of Genetic Testing
- Expanding Access to Genetic Testing
- Targeting Genes



page 31). "The diagnosis was a real shock, and then I was processing that, oh my gosh, my genes have given this to my child."

Lisa turned to her family and the friends she met in the neuromuscular disease community to help her keep perspective. "That support is so critical," she says. "Talking with other families always seems to change my perspective. I was able to say, 'Instead of focusing on that aspect of guilt, I'm going to focus on what I can do to help my family and help make this disease eradicated someday.'"

Elicia affirms that knowing a disease's genetic cause and inheritance pattern ultimately is empowering. "I tell families, 'We're lucky because now we know what it is and can figure out how to treat you or your family best,'" she says. "Many families would then choose to do prenatal testing or other types of testing in the future. When we know it's a dominantly inherited versus a recessively inherited condition, for example, you may have different options for family planning."

How genetic testing helps

Many neuromuscular diseases are categorized into types and subtypes based on the gene variant that causes the disease and how that gene affects the body's cells.

Some neuromuscular diseases do not have an obvious inheritance pattern, and some have multiple possible patterns, so working with a genetic counselor to interpret genetic test results and take a family history is important.

Genetic testing not only is used to confirm a diagnosis, but it can lead to better treatment. By revealing the exact genetic cause of a disease, it can help predict how a disease will progress and give physicians vital information for targeting therapies to an individual's disease.

In addition, having a genetically confirmed diagnosis can open doors to participating in clinical trials and qualifying for new treatments.



You should consider genetic testing if you:

- Are suspected of having a neuromuscular disease but do not have a diagnosis
- Have a clinical diagnosis but not a genetically confirmed diagnosis
- Had a previous genetic test that was negative or inconclusive

Begin by talking with your neurologist and a genetic counselor. They will consider any symptoms you have and your family history to determine the appropriate tests to order.

At MDA Care Centers, genetic counselors often are part of multidisciplinary care teams. Their role is to guide and support patients and families who are seeking information about genetic conditions.

“When I give genetic test results back to families, I try to show them the variant and let them ask as many questions as they have,” Elicia says. Along with the rest of the Care Center teams, she is not just there to provide information, but also is part of the vital support system families in the neuromuscular disease community can turn to as they progress on their journeys. [Q](#)

Amy Bernstein is an editor for *Quest*.



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Many families would then choose to do prenatal testing or other types of testing in the future. When we know it's a dominantly inherited versus a recessively inherited condition, for example, you have different options for family planning.

— Elicia Estrella, MS, LCGC

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For more than 25 years, *Quest Magazine* has been telling the stories of MDA's community. With your donation to MDA, we can continue to educate and engage families and the physicians and researchers who help them with the latest news on neuromuscular disease research, health and wellness, mobility, travel, advocacy, and everyday thriving through *Quest* and our complementary educational channels.

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THANK YOU FOR HELPING TO KEEP OUR COMMUNITY INFORMED AT THIS MOST CRUCIAL TIME.

Meet our 2022 Ambassadors



Amy Shinneman (left) joins Ethan LyBrand (right) as an MDA National Ambassador for 2022. Indianapolis-based Amy, who lives with Bethlehem myopathy, is excited to start her term after serving as a local MDA Ambassador since her diagnosis in 2018. This will be the third term for Ethan, 12, who lives with Duchenne muscular dystrophy (DMD). Read more about the 2022 Ambassadors at mda.org/questblog-ambassadors.

Advocacy Rewarded

MDA's Advocacy Team was hard at work in 2021, effecting change for the neuromuscular disease community. Here are some of the highlights of last year's advocacy efforts:

- > The ACT for ALS, which would speed up therapies for those living with ALS and other neuromuscular diseases, passed Congress.
- > The Newborn Screening Saves Lives Reauthorization Act, a bill that would reapprove and strengthen the newborn screening program, passed the US House.
- > An increasing number of states are screening newborns for spinal muscular atrophy (SMA) and Pompe disease.
- > The US Supreme Court rejected a challenge to the Affordable Care Act.
- > The five-month waiting period for ALS patients to receive Social Security Disability Insurance (SSDI) benefits was eliminated.
- > Two bills were introduced into Congress to end workplace discrimination for those with a disability.



- > Work was continued to make air travel more accessible for those living with a disability with the re-introduction of the Air Carrier Access Amendments Act.

Learn how you can become an MDA advocate at mda.org/advocacy.



After transitioning to a virtual format the last two years as a response to the pandemic, MDA Summer Camp will offer the best of both worlds in 2022, with in-person and virtual camp options this summer. Registration for the upcoming season is now open to kids and teens ages 8 to 17 who live with

neuromuscular disease. To learn more or register, visit mda.org/summer-camp. **Interested in volunteering?** MDA Summer Camp would not be possible without dedicated volunteers. Volunteers must be age 18 and over. Learn more and apply today at mda.org/summer-camp/volunteer.

A Must-Have Resource

What: MDA's Access Workshops educate the neuromuscular disease community on overcoming barriers through digital presentations that include activities, videos, quizzes, and more.

Where: mda.org/accessworkshops

When: On-demand, allowing you to navigate at your own pace

Available Access Workshops:

- > Access to Coverage: Equipment & Assistive Devices
- > Access to Education: K-12
- > Access to Education: Higher Education
- > Access to Coverage: Insurance

MDA's Lucky Season



Created as a small campaign in a Michigan pub, MDA's Shamrocks pinup program has grown into one of the biggest St. Patrick's Day fundraisers in the country — raising more than \$330 million. Join in the celebration this year by purchasing a shamrock at a participating retailer or donating online. Learn more at mda.org/shamrocks.

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MDA

View From the Top

Hiker Skye Lemmen raised funds for MDA while trekking the Pacific Crest Trail



Skye enjoys the view from the top of Old Snow Mountain in Washington.

When Skye Lemmen, of Grand Rapids, Michigan, decided to hike the Pacific Crest Trail, she knew she wanted her endeavor to support MDA and the neuromuscular disease community.

So, while hiking the approximately 2,650 miles of trail, which extends from the Mexican border to the Canadian border in the Western United States, Skye raised more than \$5,000 for MDA through Donor Drive. Inspired by her dad, who lives with facioscapulohumeral muscular dystrophy (FSHD), Skye wanted to support MDA for the assistance her family has received over the years.



Skye takes a break with her dad, Walt Thebo.

“MDA helped us through the tough time when he was first diagnosed,” she says. “My dad has always been a big proponent of MDA’s

goals, and we support everything MDA does, from Summer Camp to research.”

Her dad 100% supported her ambition. “When Skye decided to do the Pacific Crest Trail and collect donations for MDA, it just made me feel really good,” her dad, Walt Thebo, says in a video on Skye’s blog, twomoredays.blog. He says that he hopes clinical trials and research will lead to advances in treatments for FSHD.

As Skye made friends and a “trail family” during the journey, she also was able to spread awareness about neuromuscular disease. “When people found out I was raising money for MDA, they asked more about it,” she says. “It helped form a bridge between people with similar struggles, and I found out about people who I didn’t even know were dealing with other diseases.”

Skye blogged throughout her journey, striving to educate readers on neuromuscular disease, and she shared breathtaking photos on Instagram ([@skye.lemmen](https://www.instagram.com/skye.lemmen)).

From watching the sun rise from the top of Mount Whitney, the highest peak in the contiguous United States, to feeling the impact of forest fires, Skye says the experiences of the trail will be with her for the rest of her life. [Q](#)

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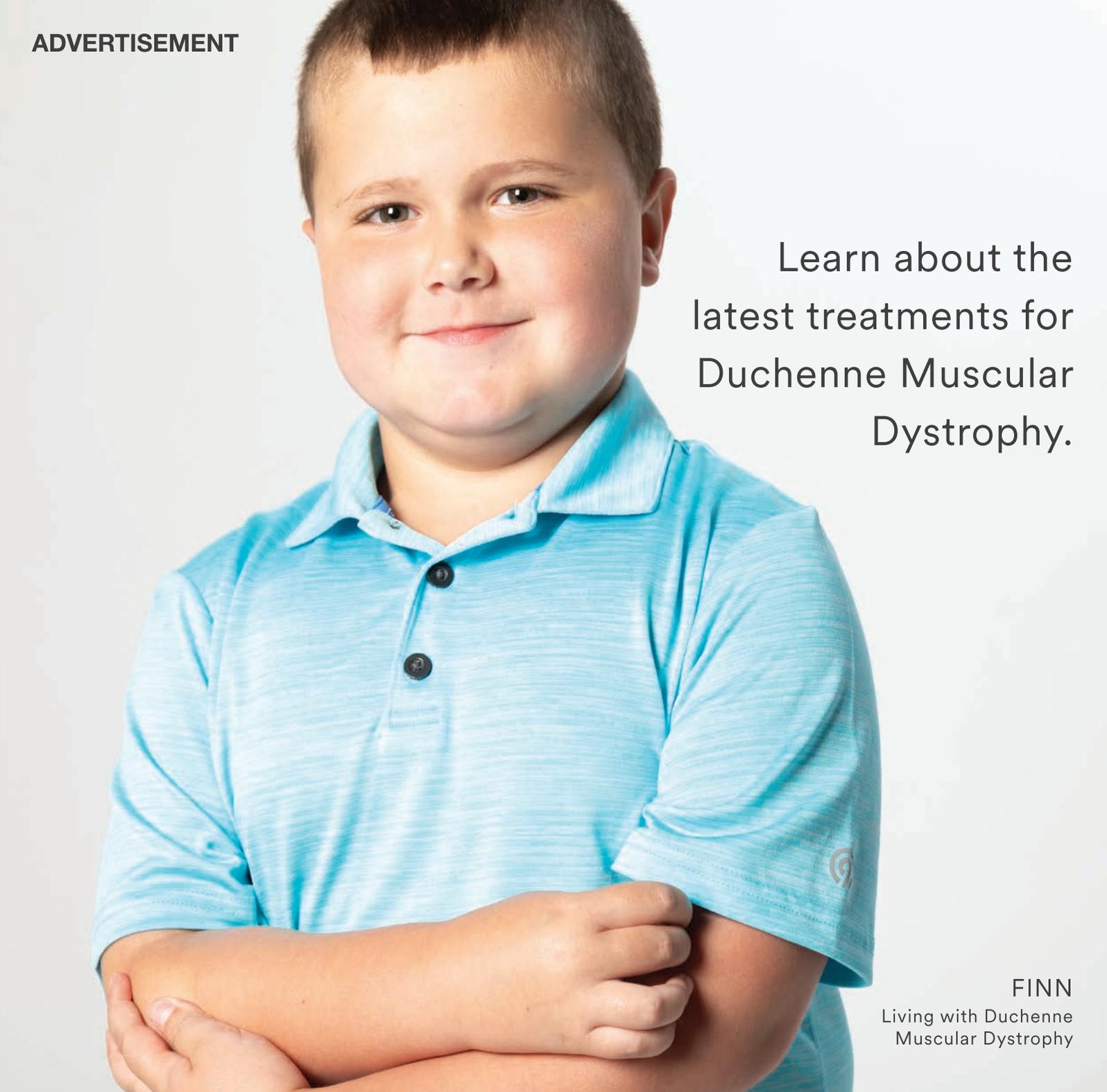
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