

EMPOWERING FAMILIES WITH INFORMATION AND INSPIRATION

Quest

MDA.ORG/QUEST ISSUE 2 • 2021

Breaking Healthcare Barriers

MDA Care Centers strive to expand access to quality care

THE RIGHT RESOURCES

How to find programs and support

EXPRESS DELIVERY

Important research on viral vectors

ADVERTISEMENT



GET TO KNOW EVRYSDI

An SMA medication for infants, children, and adults

In people 2 months and older

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

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:
 - have liver problems
 - 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
- 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.

Genentech
A Member of the Roche Group

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

What is EVRYSDI?

- EVRYSDI is a prescription medicine used to treat spinal muscular atrophy (SMA) in 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:

- have liver problems.
- 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)
 - constipation
 - 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.

Genentech
A Member of the Roche Group

EVRYSDI™ (risdiplam)

Distributed by:

Genentech, Inc.

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

EVRYSDI is a trademark of
 Genentech, Inc.
 M-US-00007143(v1.0)
 ©2020 Genentech, Inc.

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

Light at the End of the Tunnel

I hope this message finds you, your family, and your friends safe and able to enjoy each other's company. But I know that the once-in-100-year pandemic that raced around the globe this past year has touched us all. My heart goes out to those of you who have lost family, friends, or special loved ones to COVID-19. Too many of us share that pain and heartbreak.

But, there is a light coming into view at the end of the long tunnel we have traveled through together this past year. Effective vaccines have been delivered to every community in the country with estimations that there will be enough for every person by the start of summer. That is exciting news, and all of us at MDA wish to express our gratitude to those whose leadership and vision have given us hope that we — not a virus — will soon be back in control of our lives.

“

MDA's 2021 Clinical & Scientific Conference brought together more than 1,200 of the world's leading clinicians and scientists.

That sense of excitement also is true for those of us at MDA. I am writing this message on the last day of MDA's 2021 Clinical & Scientific Conference that brought together more than 1,200 of the world's leading clinicians and scientists in a virtual environment. My goal for MDA — for our professional community to gain new knowledge that will help us achieve our mission of optimizing care and treatments for all people who experience the effects of neuromuscular disease — was on active display throughout the conference.

Thank you to all who have worked tirelessly throughout this pandemic to continue advancing the extraordinary progress in achieving our mission that was reported at this year's conference and beyond.

Sincerely,



Donald S. Wood, PhD

President and CEO

Muscular Dystrophy Association



Donald S. Wood, PhD

+HOW TO GIVE

Online:
mda.org/questsupport
By mail: Quest,
 161 N. Clark St., Suite 3550,
 Chicago, IL 60601
 On checks, please write
 "Quest-4500" on the memo line.

MDA is committed to transforming the lives of people affected by muscular dystrophy, ALS, and related neuromuscular diseases through innovations in science and innovations in care.

EDITORIAL

Nancy Intrator

Acting *Quest* Editor-in-Chief

For editorial inquiries:

quest@mdausa.org

ADVERTISING

Elisa N. Beerbohm

For print and digital advertising opportunities:

mdapartners@mdausa.org

SUBSCRIPTIONS

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

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

Quest Volume 28, Issue 2

Published by
 Muscular Dystrophy Association
 161 N. Clark St., Suite 3550
 Chicago, IL 60601
(800) 572-1717
 email: quest@mdausa.org
 Available on the internet
 at mda.org/quest
 ISSN 1087-1578
 Postage paid at Bolingbrook, IL
 Nonprofit postal permit number 1446.

Postmaster: Treat as Standard
 A Mail Only

© 2021, Muscular Dystrophy Association. All rights reserved. MDA and QUEST are registered trademarks of the Muscular Dystrophy Association.

The acceptance of advertising in this magazine does not constitute or imply endorsement by MDA of any product or service. MDA accepts no responsibility for any claims made in any advertisement. *Quest* reserves the right to refuse to accept any advertisement.

Information contained in *Quest* may not be reproduced, published, transmitted or distributed in whole or in part without prior written consent of MDA.

Always consult your professional advisers as to how medical, legal or financial information in *Quest* pertains to you. MDA assumes no liability for any information in *Quest*.

FOLLOW MDA ON



PUBLICATION MANAGEMENT

glc

GLCDELIVERS.COM

MDA Care Center providers are constantly looking for ways to improve access to care.



CONTENTS

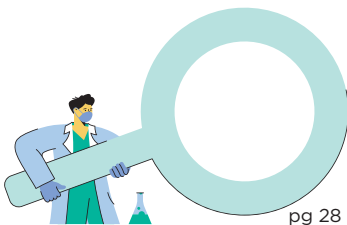
ISSUE 2 2021

FEATURES

16
Breaking Healthcare Barriers
MDA Care Centers strive to expand access to quality care.

22
Find the Right Resources
Tips to gather the tools and support you need.

26
Express Delivery
Viral vectors are an important area of research in the field of gene-targeted therapies.



pg 28

Cover image: iStock.com/Felix MAckel

DEPARTMENTS

4
MDA Q&A
The MDA Resource Center answers readers' questions.



pg 4

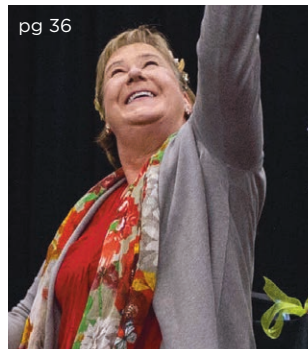
6
PROGRESS NOW
Read about recent medical research, scientific advances, and clinical trials.

12
SPOTLIGHT
Tahseen Mozaffar, MD, answers questions about GNE myopathy.

32
ACCESS MDA
MDA introduces a new spokesperson, and more.

34
FROM WHERE I SIT
Chelsea Moeller shares her experience in a clinical trial.

36
LASTING IMPRESSION
Dance has been an intimate part of Carol Cowley's healing process.



pg 36

ONLINE



GOT QUESTIONS?

WE'D LIKE TO ANSWER THEM
MDA's Resource Center wants to know what's on your mind. Their trained specialists can provide support for every part of your journey. Send your questions for the Resource Center to quest@mdausa.org and we might select your question to be answered by a Resource Center specialist in an issue of *Quest*.

How to submit questions

Email: quest@mdausa.org
Use subject line: Resource Center Q&A
Include: Your name and home state

Top of Mind

You don't have to navigate your neuromuscular disease journey alone. The MDA Resource Center is available to provide one-on-one support. Here, Resource Center specialists answer *Quest* readers' top questions.

I have oculopharyngeal muscular dystrophy (OPMD). Is COVID-19 vaccination safe for people with neuromuscular disease? Are there any side effects?

—Gretchen Sombar, Colorado

Currently, there are no specific concerns regarding the safety of COVID-19 vaccination for individuals with neuromuscular disease. In fact, many individuals with neuromuscular disease are considered at high risk for developing serious complications from COVID-19. MDA advocated for priority COVID-19 vaccine access for people living with muscular dystrophy, amyotrophic lateral sclerosis (ALS), and other neuromuscular diseases.

The known side effects of current and likely-to-be authorized COVID-19 vaccines are similar to those of the annual flu vaccine: muscle soreness at the injection site, fever, tiredness, body aches, and headache. Researchers don't expect COVID-19 vaccines to have different or more serious side effects for individuals with neuromuscular disease.

We recommend that you contact your healthcare provider for a consultation if you have any questions or concerns about getting vaccinated.

Helpful resource: Find up-to-date information about COVID-19 and neuromuscular disease at mda.org/COVID19. **Read more:** Search for the article "Are Vaccines Safe?" at mda.org/quest.

Can kids get the COVID-19 vaccine?

—Marisol Davis, Ohio

While some individuals ages 12 to 18 have participated in vaccine clinical trials, at the time of this writing, the biopharmaceutical manufacturers and US Food and Drug Administration (FDA) have only authorized the Pfizer/BioNTech vaccine for individuals 16 and older, and other currently approved vaccines for individuals 18 and older. However, clinical trials of the vaccine focused on kids are ongoing. MDA will provide updates as more is known.

Helpful resource: Find more information on COVID-19 vaccinations at mda.org/COVID19.



The clinical trials I see for Duchenne muscular dystrophy (DMD) therapies deal with exon skipping. How can I find out if any of them apply to my son, who is 31 and has DMD?

—Kathryn Gentry, Pennsylvania

Genetic testing can pinpoint the exact nature and location of a gene mutation in DMD and other neuromuscular diseases. This information will help determine whether any current therapies, such as an exon-skipping drug, would be helpful for a specific diagnosis.

Detect Muscular Dystrophy, offered in partnership with INVITAE, is a no-charge genetic testing and counseling program for individuals who meet certain criteria and are suspected of having a muscular dystrophy. A physician must order the testing kit. If you feel genetic testing would be right for your son, contact the Resource Center for information to share with his healthcare team to get started.

Helpful resource: Learn more about the no-charge genetic testing program at invitae.com/detect-muscular-dystrophy. **Read more:** Search for the article "Expanding Access to Genetic Testing" at mda.org/quest.

Has research on the therapeutic value of physical exercise for people with myotonic dystrophy (DM) shown any exercises to be effective in mitigating the effects of the disease?

—Dianne Snyder, Virginia

Exercise in neuromuscular disease is an area that is being actively studied. For DM type 1, there is evidence from some

+WE CAN HELP

MDA Resource Center specialists are available Monday through Friday, 9 a.m.-5 p.m. CT. They typically can answer questions within one to two business days. Call **833-ASK-MDA1** or email ResourceCenter@mdausa.org.

small studies that physical exercise therapy is beneficial. There also is growing evidence that an active lifestyle and low-to-moderate-intensity aerobic activity is good for neuromuscular diseases in general, but more study is needed on exercise with specific conditions.

Your MDA Care Center healthcare team can provide individualized exercise recommendations. It's also important to consult with your cardiologist before beginning an exercise program, especially if you have cardiac symptoms due to DM.

Helpful resource: Find an MDA Care Center and learn about Care Center teams at mda.org/care/mda-care-centers.

What types of resources can the MDA Resource Center connect me to?

—Stephanie Erbacher, Iowa

The staff of the MDA Resource Center will actively listen to your situation and provide information or resources in your community to assist with your needs. Some of the most requested resources that we connect individuals with are:

- General disease education
- Connecting to others living with neuromuscular disease
- Information on individualized education programs (IEPs) and education advocacy
- Caregiver resources
- Clinical trials finder
- Information on getting genetic testing
- End-of-life care resources
- Financial assistance resources
- Information on obtaining residential and vehicle modifications
- Information on where to donate gently used durable medical equipment
- Connecting with loan equipment resources in your community
- Finding an MDA Care Center
- Connecting to your local Center for Independent Living or vocational rehabilitation office



Read more: Search for the article “Support for Your Journey” at mda.org/quest.

SUPPORT MDA AND QUEST NOW

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.

To make a tax-deductible donation to MDA, visit www.mda.org/questsupport

Or send mail:
Attn: QUEST
161 N Clark St.,
Suite 3550
Chicago, IL 60601
On checks, please write
“Quest-4500” on the
memo line.

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



PROGRESS NOW

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

New approval

Amondys 45 Approved for DMD



The US Food and Drug Administration (FDA) has granted accelerated approval to casimersen (Amondys 45) for the treatment of Duchenne muscular dystrophy (DMD) in patients amenable to skipping exon 45. This is the fourth exon-skipping, disease-modifying drug approved to treat DMD in the United States.

DMD is caused by mutations in the dystrophin gene that result in little or no production of dystrophin, a protein essential to keeping muscle cells intact. Exon skipping is a treatment strategy in which sections of genetic code are skipped over during the protein-making process, allowing cells to create shortened but partially functional dystrophin protein. Administered by intravenous infusion, Amondys 45 uses Sarepta Therapeutics' exon-skipping technology to target exon 45 of the DMD gene.



Exon skipping is not a cure for DMD but potentially could slow progression of the disease, extending the length of time individuals with DMD can walk, eat independently, and breathe without assistance. It is estimated that up to 8% of patients with DMD could benefit from treatment with Amondys 45.

The FDA based its decision to grant accelerated approval to Amondys 45 on positive results from a global, randomized, double-blind, placebo-controlled phase 3 clinical trial (ESSENCE) that is evaluating the efficacy and safety of the drug in individuals with DMD. The positive results include findings that Amondys 45 has met the full statutory standards

The SareptAssist program helps individuals navigate the process of starting and continuing therapy. Sarepta's dedicated team provides information on insurance benefits, financial assistance options, treatment logistics, options for drug delivery, and ongoing education and support.

for safety and effectiveness and demonstrated an increase in dystrophin production in skeletal muscle. Continued approval may be contingent upon verification of a clinical benefit in confirmatory trials. ESSENCE is ongoing and expected to conclude in 2024.

In the last five years, three targeted exon-skipping drugs have been approved by the FDA to treat DMD: Exondys 51 and Vyondys 53 from Sarepta Therapeutics and Viltepso from NS Pharma. Approval of Amondys 45 represents another significant step forward in the development of therapies for DMD that target the root cause of the disease.

MDA-supported research — including foundational work and extensive research into the therapeutic strategy — has been central to the development of the exon-skipping approach. Laboratory development of exon-skipping therapies began in the 1990s, including the notable MDA-funded work by Steve Wilton, PhD, and colleagues, which led to what would later become Exondys 51, Vyondys 53, Viltepso, and Amondys 45.

Learn more about the approval of Amondys 45 at [sarepta.com](https://www.sarepta.com). For more information about the ESSENCE trial, visit clinicaltrials.gov and enter NCT02500381 in the “Other terms” search box.



Amyotrophic lateral sclerosis (ALS)

Observational Biomarkers Study Seeks Volunteers

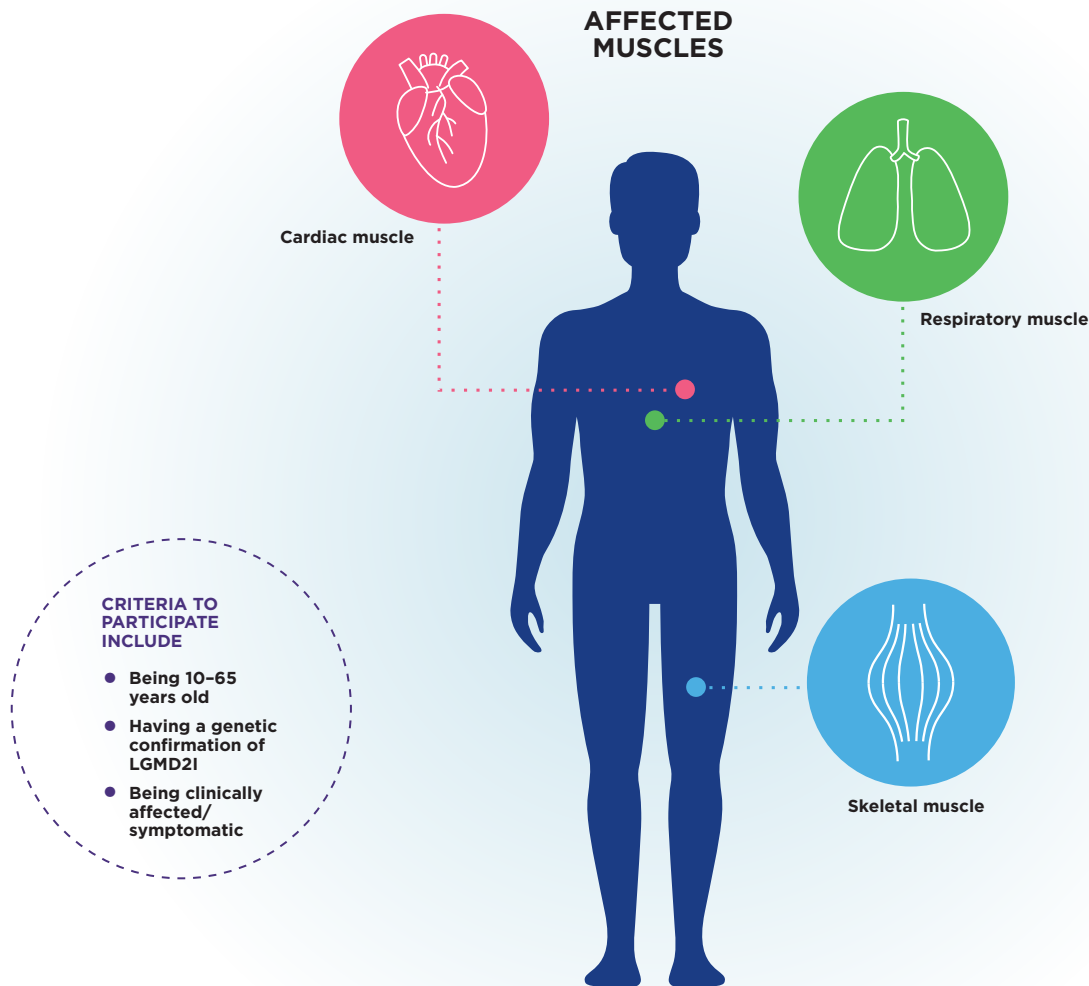
Researchers at Mitsubishi Tanabe Pharma America (MTPA) are seeking individuals living with ALS to participate in a six-month observational study designed to identify biomarkers to serve as quantifiable, biological, nonclinical measures of the effects of Radicava (edaravone) on ALS.

No experimental treatment is being evaluated in this study. Observational measures might include assessment of vitals; blood and urine collection; safety assessments; use of the revised ALS functional rating scale (ALSFRS-R); King’s clinical staging; medical history; International Classification of Functioning, Disability, and Health (ICF); and medications review.

Study participants will make a total of eight visits over six months, with in-clinic and remote options offered. Eligible participants who are prescribed Radicava will undergo biomarker testing and clinical assessments at enrollment and at one, three, and six months after enrollment.

To be eligible to participate, individuals must be at least 18 years or older at the date of enrollment, have a diagnosis of sporadic or familial ALS, and meet additional criteria. Individuals may not be eligible to participate if they have a symptom or condition that prevents them from taking Radicava, or if they are participating in an interventional clinical trial.

For a full listing of eligibility criteria, visit clinicaltrials.gov and enter NCT04259255 in the “Other terms” search box. To learn more or to inquire about participation, contact Lia Tamburello, clinical trial project manager, NCRI, at [617-724-2609](tel:617-724-2609) or ltamburello@mg.harvard.edu.



Patient engagement over the course of drug development is vital to gain an understanding of the impact of the disease on the individual, as well as on the family and community.

CRITERIA TO PARTICIPATE INCLUDE

- Being 10-65 years old
- Having a genetic confirmation of LGMD2I
- Being clinically affected/symptomatic

Limb-girdle muscular dystrophy (LGMD)

Participants Sought for Natural History Study

Researchers at ML Bio Solutions are seeking individuals with LGMD type 2I (LGMD2I, LGMD R9-FKRP-related) to participate in a 12-month natural history study for use in a phase 3 clinical trial of BBP-418 (ribitol). Natural history studies collect information about the natural progression of a disease in the absence of an intervention, and these studies can assist in drug development.

No treatment will be administered in the natural history study; however, participants will be eligible to enroll in the subsequent trial of BBP-418, a drug that may enhance diminished muscle function.

Study participants will complete baseline assessments over two consecutive days and at one-day visits at six, nine, and 12 months after enrollment. Visits may involve fine-needle muscle biopsy, muscle MRI of the

lower extremities, functional tests, lung function tests, and assessments of quality of life and activities of daily living.

Criteria to participate include:

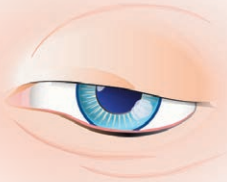
- > Being 10-65 years old
- > Having a genetic confirmation of LGMD2I
- > Being clinically affected/symptomatic

Remote enrollment may be available, and in-person visits will observe strict COVID-19 precautions.

For more information, including complete eligibility criteria, visit clinicaltrials.gov and enter NCT04202627 in the “Other terms” search box. To inquire about participation, contact Brittney Holmberg at Brittney.Holmberg@vcuhealth.org or 804-552-0014.

Myasthenia gravis (MG)

Volunteers Needed for Phase 3 Trial



Rozanolixizumab may help reduce the typical signs and symptoms of gMG, which are caused by autoantibodies that disrupt the signaling between nerves and muscles.

Researchers at UCB BioSciences are seeking individuals living with generalized myasthenia gravis (gMG) to participate in a phase 3 study designed to confirm clinical efficacy and assess safety and tolerability of rozanolixizumab.

Because it is a double-blind study, participants will be assigned randomly to a group designated to receive rozanolixizumab or an inactive placebo control, and neither the participants nor the study team will know who is receiving the drug or placebo. Upon completion, participants will be given the option to enroll in an open-label extension trial.

The 18-week study will require 14 visits. After being screened on the first visit, participants will receive six weekly subcutaneous (under-the-skin) infusions of the study drug or placebo over a five-week period, as well as routine tests and monitoring up to week six. Observational visits will continue every other week until the study is completed. Travel support may be available.

Criteria to participate include:

- > Being at least 18 years old
- > Having a documented diagnosis of gMG
- > Having a confirmed positive record of autoantibodies against acetylcholine receptor (AChR) or muscle-specific kinase (MuSK) at screening

For the full listing of eligibility criteria, go to mycaringstudy.com or visit clinicaltrials.gov and enter NCT03971422 in the "Other terms" search box. To learn more or to inquire about participation, email UCBCares@ucb.com or call 844-599-2273.

ADVERTISEMENT

Total toileting solution that hygienically cleans you then lifts you to standing.

LiftSeat®
Powered toilet lifts™



- Adjustable Seat Tilt
- Installs Easily
- Electric Washing Bidet
- \$100 Off Accessories

(877) 665-4381

liftseat.com

ADVERTISEMENT

VOICE ACTIVATED DOOR

by
OPEN SESAME

- // Releases a locked door
- // Voice or remote control activated
- // Simple to install
- // Battery backup



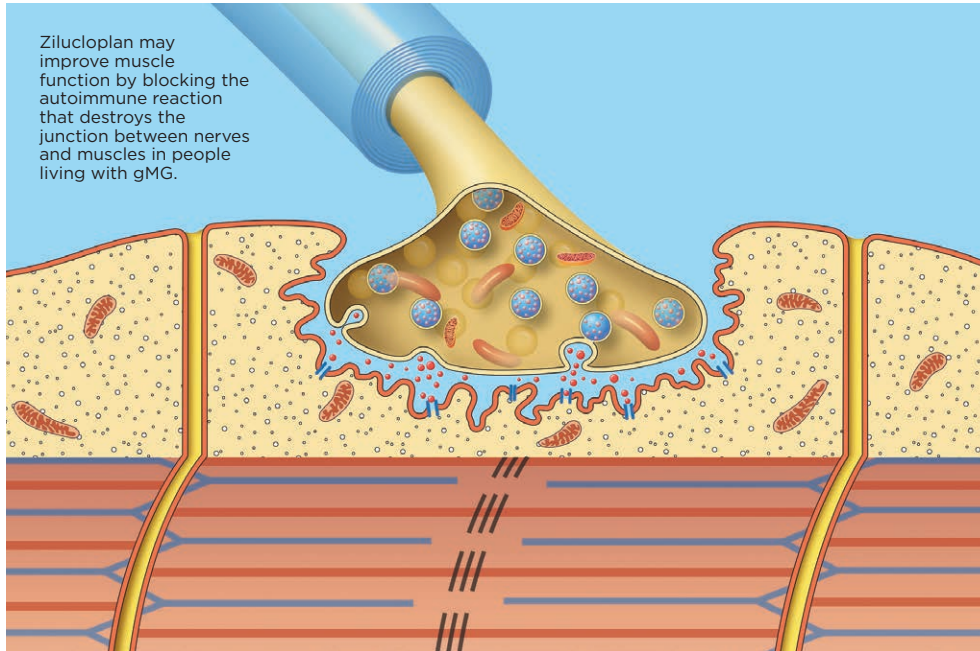
alexia
"Activate the door"

OPEN SESAME

Call for more information
888-973-7263
OpenSesameDoor.com

Myasthenia gravis (MG)

Zilucoplan Trial Participants Needed



Zilucoplan may improve muscle function by blocking the autoimmune reaction that destroys the junction between nerves and muscles in people living with gMG.

Image: iStock.com/Aldona

Researchers at Ra Pharmaceuticals (now part of UCB BioSciences) are seeking individuals living with generalized myasthenia gravis (gMG) to participate in a phase 3 study designed to confirm safety, tolerability, and effectiveness of the investigational drug Zilucoplan.

This is a blind study, meaning each participant will be assigned randomly to a group designated to receive Zilucoplan or an inactive placebo control. The drug or placebo will be self-administered at home as a daily subcutaneous (under-the-skin) injection. At

ADVERTISEMENT

PROTONE

A Professional quality fitness machine for persons with disabilities

- No hand grip strength necessary
- Adjust without assistance
- Over 16 different exercises
- Commercial quality at a home gym price
- For more information email sales@aquacreek.com

888.687.3552
www.aquacreek.com

Aqua Creek Products
Leaders in Recreation, Fitness and Ability

ADVERTISEMENT

DO YOU WANT RELIEF FROM...

- Painful Bedsores?
- Pneumonia and Other Respiratory Problems?
- Being Awakened for Turning?

AUTOMATED BODY ROTATION
 PROGRAMMABLE TIMES AND ANGLES
 SMOOTH AND SILENT OPERATION

The Freedom Bed provides many proven medical benefits PLUS the guarantee of a good nights uninterrupted sleep, every night.

The Freedom Bed is funded by private insurance plans and state programs.

Left Rotation Right Rotation

PRO BED
Medical Technologies Inc.

“ I am writing to help promote the awareness of the FREEDOM BED within the Muscular Dystrophy Community. If our son’s story can help families get the bed that will change their lives it will be worth it. The FREEDOM BED is heaven sent! It has dramatically improved our lives and allows the whole family to live normally. It is appropriately named the FREEDOM BED. ”

The Azar Family

FOR MORE INFORMATION CALL
TOLL FREE: 800.816.8243 | WWW.PRO-BED.COM

Trial participants will undergo screening and baseline assessments, and then make routine visits after enrollment.

the end of the study, participants will be given the option of enrolling in an open-label extension study.

Participants will undergo screening and baseline assessments, and then make routine visits at one, two, four, eight, and 12 weeks post-enrollment. There will also be a 40-day follow-up visit for participants

who do not enroll in the open-label extension study. Travel support may be available.

To participate, individuals must be 18-75 years of age with a diagnosis of gMG, have positive serology for acetylcholine receptor (AChR) binding autoantibodies, and meet additional criteria.

For the full listing of eligibility criteria, go to gmgstudy.com or visit clinicaltrials.gov and enter NCT04115293 in the "Other terms" search box. To learn more or to inquire about participation, email UCBCares@ucb.com or call 844-599-2273.

RECRUITING CLINICAL TRIALS

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



Image: iStock.com/Warchi

ADVERTISEMENT GUARANTEED FOOT DROP SOLUTIONS FOR ADULTS & CHILDREN

DORSI-LITE™ FOOT SPLINT

USE WITH
OR WITHOUT
SHOES



- Outstanding comfort.
- Easy On & Off
- Ultra-low profile
- Near-Normal Gait
- Will not slip off
- Also use in bath or swimming pool
- \$60/EA. + \$7 Shipping/Order
- Optional EXPRESS shipping: \$30
- Low cost replacement parts available

DORSI-STRAP™ FOR FOOT DROP

FOR USE
WITH
SHOES



Also available,
Dorsi-Strap PRO Heavy-Duty, \$45

- No Special Shoes Needed
- Nothing put into the shoe
- Outstanding comfort
- Ultra-low profile, 3OZ
- Near-Normal Gait
- Easy On & Off
- In White, Black, or Brown
- \$42/EA. + \$7 Shipping/Order
- Optional EXPRESS shipping: \$30
- Low cost replacement parts available

30 DAY MONEY-BACK WARRANTY

X-STRAP SYSTEMS
9 Stonegate Drive
Hyde Park, NY 12538

www.x-strap.com
(845) 233-4713

03/20

SPECIAL \$3 OFF/ORDER: CODE MDAX

ADVERTISEMENT LIFT YOUR LIFE TOWARD INDEPENDENCE



Ask
about our
30-Day
return
guarantee

YouTube
product
videos
on our
website

BATTERY-POWERED TOILET SEAT LIFT

- Perfect for those with muscle weakness
- Standard or mobile models available
- Lifts straight up; no forward push

BATTERY-POWERED CHAIR LIFT

- From sitting to standing in home, office or other environment
- Comfort and style for use anywhere

800-831-8580

for information and a FREE demo video

www.stand-aid.com sales@stand-aid.com

STAND AID of Iowa

Expanding Knowledge on GNE Myopathy

A Q&A with Tahseen Mozaffar, MD



Tahseen Mozaffar, MD

Tahseen Mozaffar, MD, is a neurologist and director of the ALS & Neuromuscular Center at the University of California-Irvine. He's also a leading world expert on GNE myopathy, a form of muscular dystrophy that affects an average of one person per million worldwide.

GNE myopathy is caused by mutations in the GNE gene, which is responsible for a step in the production of a sugar called sialic acid. This results in decreased attachment of sialic acid groups to skeletal muscle cells, which is thought to be why the disorder leads to muscle atrophy and weakness.

GNE myopathy is an autosomal recessive disorder, meaning it is inherited when both parents contribute a mutated gene. Genetic testing for GNE myopathy is available as part of the gene panels for neuromuscular diseases.

We spoke with Dr. Mozaffar to learn more about this disorder.

How was GNE myopathy first recognized?

It was originally described in Japan as Nonaka distal myopathy in 1992. Then, Professor Zohar Argov at Israel's Hadassah Hospital diagnosed it among Jews of Iranian, or Persian, descent. He described it as quadriceps-sparing myopathy. For the next few years, people thought these were different diseases, until they discovered the mutation and realized that it was the same disease.

The first few patients I diagnosed with this disease were in India and Pakistan. It's still referred to as a Middle Eastern disease, but it's quite common in India.

How prevalent is it in the United States?

It seems like the more you look for it, the more you find it. There are clusters in some communities in California and New York. There's an increasing realization that GNE myopathy is somewhat common and probably ranks in the top five in terms

“

There's an increasing realization that GNE myopathy is somewhat common and probably ranks in the top five in terms of muscular dystrophies. In a recent study from India, GNE mutations were the most prevalent among a cohort of 207 patients with limb-girdle muscular dystrophy (LGMD).

of muscular dystrophies. In a recent study from India, GNE mutations were the most prevalent among a cohort of 207 patients with limb-girdle muscular dystrophy (LGMD).

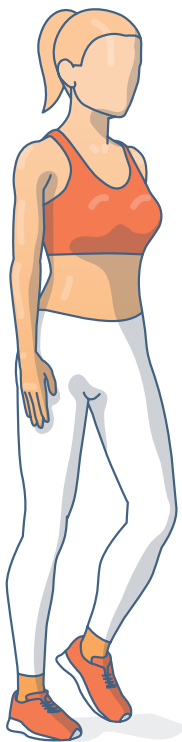


Image: iStock.com/Oleksii Bulgakov

GNE myopathy symptoms usually start with difficulty lifting the front part of the foot, called foot drop.

How does GNE myopathy progress?

It starts with foot drop, which usually presents in the late teens or early 20s. The leg muscles get progressively weaker. In some families, the disease is more aggressive, and individuals begin using a wheelchair within 15 years; other individuals can walk for a much longer period. Eventually, the disease involves the upper extremities, and some individuals have difficulty holding their necks up.

How do you treat GNE myopathy?

We just started the first-ever GNE myopathy clinic at the MDA Care Center at the University of California, Irvine, and because of the expansion of telehealth, we are able to see patients from all over the country. We are holding this clinic six times a year, and the Neuromuscular Disease Foundation is planning to spread this model to other parts of the United States, as well as Israel.

Currently, treatment consists of neurological evaluations and physical and occupational therapy. In terms of medications, we don't have anything to

offer at this point. Because people with GNE myopathy have low levels of sialic acid, a number of individuals are taking sialic acid supplements from the internet, but we don't even know how reliable those supplements are. [The US Food and Drug Administration (FDA) does not review dietary supplements for safety and effectiveness before they are marketed.]

Are any treatments being researched?

A new trial (NN109) through the NeuroNEXT consortium is about to start to evaluate the efficacy of

N-acetyl-D-mannosamine monohydrate (ManNAc), which is a precursor for making sialic acid. ManNAc is found where the GNE enzyme works, and some investigators believe that ManNAc would work better than extended-release sialic acid, which was tested previously with disappointing results. This new trial involves close to 150 patients at 13 sites across the United States. We are working with Ultragenyx to possibly bring another drug to research testing.

What about a potential gene therapy to treat GNE myopathy?

A group of researchers in Japan, Israel, Holland, and the United States are studying this, but there are a few problems. We don't have a reliable mouse model or good validated antibodies that can detect the GNE protein in cell cultures, so a lot of the focus is on developing strategies to have better mouse models. Pharmaceutical companies would only be interested in developing a therapy if there's a reliable animal model or a marker of the disease that they can measure. [Q](#)

+RARE DISEASE SPOTLIGHT

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

THIS MOMENT

COMES
FROM WITHIN

CAMDEN // AGE 4

PRESYMPTOMATIC SMA, TREATED WITH SPINRAZA



NICOLE // AGE 45

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.

Victories are personal for the **11,000+** who have been treated with SPINRAZA worldwide.*

3700+
ADULTS

Thousands of adults have been treated with SPINRAZA worldwide*

3-80
DAYS YEARS

There's someone from almost every age group who has taken SPINRAZA^{†‡§}

7+
YEARS

Safety and efficacy evaluated in the longest clinical trial in SMA to date[§]

*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).

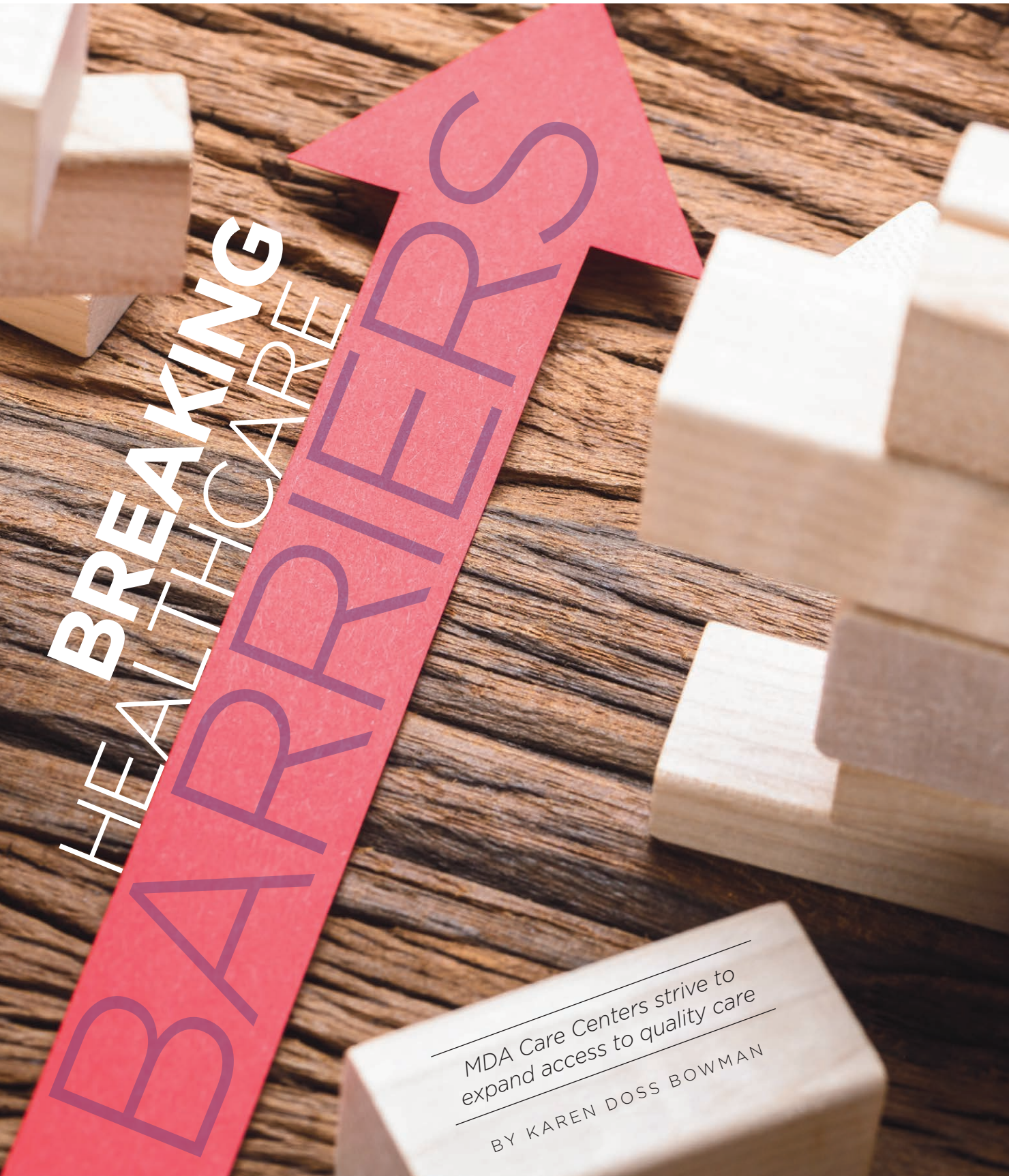
MANUFACTURED FOR

Biogen, Cambridge, MA 02142





When she lived in Dallas, Kelly Pagano regularly visited her local MDA Care Center and felt confident in the way her medical team managed her condition, limb girdle muscular dystrophy (LGMD). Along with regular check-in appointments, the clinic provided a wealth of information about her disorder and research updates, connected her with local resources, and offered participation in clinical trials. She also participated in local support groups and MDA events. >>



HEALTHCARE
BREAKING

BARRIERS

MDA Care Centers strive to
expand access to quality care

BY KAREN DOSS BOWMAN

Image: iStock.com/AndreyPopov



Kelly Pagano noticed a difference in access to care for her LGMD after moving to a rural town.

+FIND A CARE CENTER

MDA Care Centers provide expert clinical care for individuals living with muscular dystrophy, ALS, and many other neuromuscular diseases at more than 150 of the top healthcare institutions across the United States. To find an MDA Care Center, visit mda.org/care/mda-care-centers.

Since moving to rural Nolensville, Tenn., two years ago, however, Kelly has felt alone in dealing with her disorder. “Currently I am not getting care for my LGMD, which is a scary place to be,” she says.

Neuromuscular conditions encompass a wide range of diagnoses, symptoms, and management strategies. People living with these disorders benefit from regular, specialized care management. However, neuromuscular diseases often take years to diagnose, and expert care is not always accessible for a variety of reasons: distance from a clinic, inability to take time off work, or lack of childcare or transportation, to name a few.

MDA supports a nationwide network of Care Centers — the nexus of expert clinical care and medical research — and provides an array of programs and services, including the National Resource Center, educational resources, community activities, and Summer Camp for kids. We recognize that many individuals, like Kelly, struggle to connect with providers who understand their conditions and are knowledgeable about the best care practices. Healthcare providers in the MDA Care Center network are constantly, actively looking for ways to improve access to care.

Serving rural communities

For Aravindhan Veerapandiyan, MD, assistant professor of pediatric neurology and director of the comprehensive neuromuscular disease program at Arkansas Children’s Hospital, expanding access to care is a personal quest. As a child, he watched his father, also a physician, care for patients in his family’s rural Indian community, which sparked his own desire to work with underserved communities.

“This feels personal to me because I grew up in a place where I saw people traveling miles and miles to get medical care,” says Dr. Veerapandiyan, known as Dr. Panda to his patients and families. “I have witnessed how draining that is for families and the emotional burden they carry. This is critical because I strongly believe that everyone should get equal care.”

In 2019, Dr. Veerapandiyan helped expand the Arkansas Children’s neuromuscular disease clinic in Springdale, located in the northwest corner of the state. This clinic helps bridge gaps in access to care in a state where families often travel up to four hours for their loved ones to receive specialized neuromuscular disease care. Just like the main location in Little Rock, it is staffed by a multidisciplinary care team with expertise and commitment to patients and their families living with neuromuscular disease.

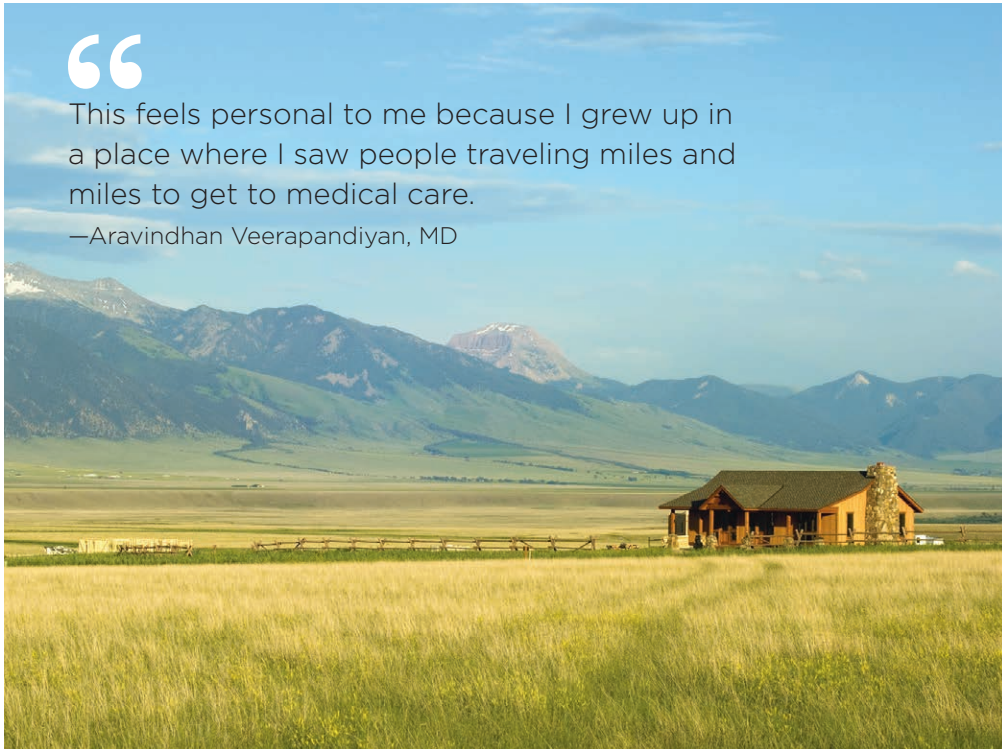
“The satellite clinic gives an opportunity for these kids to have the best quality of life that they can.” Dr. Veerapandiyan says. “Living in a rural area shouldn’t prevent them from getting the best quality care that they deserve and need.”

“

This feels personal to me because I grew up in a place where I saw people traveling miles and miles to get to medical care.

—Aravindhan Veerapandiyan, MD

image: iStock.com/Natalia Bratslavsky



During the COVID-19 pandemic, when the expanded use of telehealth visits by video or phone allowed many people to get healthcare without leaving home, Dr. Veerapandiyan found that some of his patients lacked high-speed Internet, or even reliable phone service.

“Parents often had to drive 30 minutes to an hour away from their homes to get to a public place where they could access free Wi-Fi to connect with telemedicine,” Dr. Veerapandiyan says.

Arkansas Children’s is working on expanding telehealth access by offering telemedicine workstations in primary care physicians’ offices. Families can go to these providers’ offices to have telehealth visits with specialists.

Between the satellite clinic and expanded telemedicine options Dr. Veerapandiyan has been able to reconnect with some families he hadn’t seen in years.

“It’s not that parents and caregivers don’t want to give their children the care they need,” Dr. Veerapandiyan says. “Usually there’s something preventing them from coming. With telemedicine we can’t do the hands-on exams that often are necessary, but at least it’s a way to bring them back to us and monitor their conditions.”

Educating the providers

Neuromuscular diseases encompass a wide range of symptoms, and their occurrence rates range between relatively rare and extremely rare. Most primary care physicians won’t

see more than a handful of patients with neuromuscular diseases over the course of their entire careers. When they do see them, they might be slow to recognize the symptoms or unfamiliar with the best course of care.

For this reason, Edwardo Ramos, MD, a sports medicine and spinal cord specialist at Instituto de Rehabilitación del Caribe, an MDA Care Center in San Juan, Puerto Rico, regularly reaches out to primary care providers, as well as physical therapists and occupational therapists — specialists who are likely to see individuals experiencing weakness or mobility issues — to educate them about neuromuscular disease.

“I try to give them confidence in terms of how they can identify a patient who

“

[MDA Care Centers] are updated on all the available treatment options and also any clinical trials that are available to patients.

—Edwardo Ramos, MD





Image: iStock.com/Hispanolistic

Education in Spanish

MDA is committed to helping our community access the care they need. Our disease fact sheets and other community education materials are available in English and Spanish. To view and download educational materials, visit mda.org/education. Turn to page 22 to learn about more great MDA resources.

might have a neuromuscular disease, and I tell them that our clinic can serve as a place for confirming the diagnosis,” Dr. Ramos says. At a recent annual meeting for local primary care doctors, he gave a presentation about potential signs of neuromuscular disease and genetic testing options. Many of the healthcare providers he meets in Puerto Rico know very little about neuromuscular diseases and are unaware that they could refer a patient they suspect might have a neuromuscular disease to one of the three MDA Care Centers on the island.

“It’s important for providers to understand that the MDA clinics are the primary centers for diagnosing and managing patients with different neuromuscular diseases,” Dr. Ramos says. “These clinics are updated on all the available treatment options and also any clinical trials that are available to patients.”

Building trust

Access to care isn’t always a matter of proximity to a high-quality medical center. In New York City, for example, residents are close to a large number of major healthcare institutions in and around the city. However, individuals can still experience challenges related to language barriers and economic stressors, such as housing and job concerns.

Leslie D. Delfiner, MD, pediatric neurologist and co-director of the MDA Care Center at Montefiore Medical Center in the Bronx, says her team strives to provide flexible care that recognizes the impact of those challenges. For example, if a parent cannot take the whole morning off work for their child to see multiple specialists in one visit, they might break the

appointment up over two shorter visits.

To address language barriers, Montefiore offers phone interpreters and on-site American Sign Language translators for any family who needs them. The Care Center’s social worker helps identify families’ needs and provide support and services for patients and families.

“We do not force families to fit into a single model for scheduling and for access to care,” says Dr. Delfiner. “It’s important to identify the challenges that impact a family’s ability to engage in care and address each of those concerns at every visit. Medical appointments can be stressful if a family has trouble traveling to visits or is concerned that a parent may lose their job because of time away from work. Sometimes in clinic, we spend a significant amount of time addressing these issues and creating a family-specific care plan that accommodates their needs.”

Dr. Delfiner recognizes that she and other MDA Care Center providers can play a critical role in helping to break down healthcare barriers for individuals living with neuromuscular disease. “As clinicians, we need to engage with patients and their families as partners in care,” she says. [Q](#)

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

ADVERTISEMENT



If you or a loved one is coping with amyotrophic lateral sclerosis (ALS), you/they may be eligible to participate in the REFINE-ALS observational study.

**For more information,
call (617) 724-2609**

actor portrayal



Mitsubishi Tanabe Pharma America

REFINE^{ALS}
OBSERVATIONAL STUDY

FIND THE RIGHT Resource

Follow these tips to gather the tools and support you need to live well

BY
CHARMAINE
DYMOND



Illustrations: iStock.com/fourSage

MDA COMMUNITY RESOURCES

MDA is committed to transforming the lives of people living with neuromuscular disease. Here are programs and services that can help you find the resources you need for everyday life.

Access Workshops

These educational workshops provide information and resources on access and overcoming barriers. Each workshop includes online activities, videos, quizzes, and more that help you build health literacy, empowerment, and self-advocacy skills. They're available on demand, allowing you to work through them at your own pace. Find workshops at mda.org/accessworkshops.

Advocacy Center

Find resources and webinars that will help you make your voice heard on issues that are important to the neuromuscular disease community. Visit mda.org/advocacy.

Community Education

From disease fact sheets to in-depth guides on genetics and clinical trials, MDA provides materials to help educate our community about the fundamentals of neuromuscular disease. Find materials in English and Spanish at mda.org/education.

es



Amy Curran, 38, lives in Philadelphia with GNE myopathy, as do her three older siblings. Considering the high cost of accessible vehicles, this close-knit family decided to pool their resources and buy one wheelchair-accessible van. They use an online calendar to keep track of who's using the van when. The van fits two power wheelchairs, so they often take turns going on outings in pairs.

This inventive approach means the four siblings get the use of a much-needed accessible vehicle at a fraction of the cost, which has helped Amy maintain her social life and independence. "Without the van, my partner takes me to social events in his car with a transfer chair, and I have far less independence without my power wheelchair," she says.

Living with neuromuscular disease can mean using a variety of resources, equipment, and support. But as Amy and her siblings have discovered, accessing these resources isn't always easy, and getting the help you need may require thinking outside the box.

"Resources can turn up in unexpected ways. It's important to think creatively and recognize the gems when you find them," says Deborah Wais, LCSW, a social worker at the MDA Care Center at the University of California San Diego.

Here are some ideas to help you find the right resources.

Be prepared

The first step to finding the resources you need is knowing what you need, which can be more challenging than it sounds. That's why it's important to understand how your disease might progress and be proactive in your medical care. Deborah recommends talking to your care team about your diagnosis and what your future needs might be.

Mike Myers lives in Elmer, NJ, with his 21-year-old son, Jacob, who has Duchenne muscular dystrophy (DMD). He and Jacob have learned that understanding possible future needs makes it easier to plan ahead. "It can take six months to get a new piece of equipment when it comes to dealing with insurance and approvals," Mike says. "We try to be



Amy Curran

Engage Events

MDA Engage community education programs empower individuals and families with knowledge and resources around neuromuscular disease.

Some are focused on adults or children, specific diseases, or caregivers. All are free to attend. Find upcoming events and view recorded programs at mda.org/engage.

Facebook Live Events

Through Facebook Live, MDA tackles timely and engaging topics like the COVID-19 vaccine and accessible fashion. Follow MDA at facebook.com/mdaorg. Find past events on MDA's YouTube channel at youtube.com/mda.

MDA Care Centers

We support a network of Care Centers at more than 150 of the top healthcare institutions across the United States. To learn about MDA Care Centers and find a location, visit mda.org/care/mda-care-centers.

National Resource Center

The Resource Center provides one-on-one support via phone or email for individuals and families looking for information about neuromuscular diseases, services, activities, and more. Resource Center staff are available Monday through Friday, 9 a.m. to 5 p.m. CT, and are typically able to answer questions within one to two business days. Call **833-ASK-MDA1** or email resourcecenter@mdausa.org.

ahead of the game and get the ball rolling before Jacob absolutely needs something.”

When Jacob was younger, Mike found that talking to other parents of kids with neuromuscular disease was helpful in understanding what Jacob might need in the future.

“We get a lot of information from networking with other parents and hearing what they’ve been through and what tools they’ve used,” Mike says.

MDA’s National Connections Program offers a way for people in the neuromuscular disease community to connect with other individuals living with neuromuscular disease, caregivers, parents, spouses, or siblings. To

join the program, contact MDA’s Resource Center at **833-ASK-MDA1** or **resourcecenter@mdausa.org**. You can request to be matched with others based on criteria including diagnosis, age, or interests.

Like many people in the neuromuscular disease community, Mike is happy to pass on the support he’s received to

others. “The other day, a man I met at an MDA event called me. His son is 8 and has started to go through some of the same things Jacob did at that age,” Mike says. “We’ve been doing this long enough that we can help other people find the resources they need now.”

Be proactive

Josh Cueter, 23, lives with spinal muscular atrophy (SMA) in Troy, Mich. He’s found that being proactive is the best way to get access to available resources.

“It’s a lot of phone calls, research, and trial and error,” says Josh, who does research before speaking with caseworkers and social workers who help him navigate programs such as Medicaid. “I’ve had the most success doing a fair amount of legwork on my end and then just laying it all out there for them. It helps to maximize the chance of success.”

Josh also took a proactive approach to finding personal care attendants (PCAs) while in college. After posting a job advertisement on campus, he recruited and trained a network of PCAs to meet his needs. “I had the most success at finding caregivers among my fellow students,” he says. “A lot were health science students who could use their work for me as a resume builder, but I also found good people through word of mouth and social networks.”

Talk to experts

Sometimes the challenge is in knowing what help is available. This was Josh’s experience when he started college. “There are a fair number of resources to help people with disabilities create their own independent lifestyle through work and school, but it was hard to understand what the different agencies provided and how it all fit together,” he says.



Jacob Myers

HELPFUL RESOURCES

Use the links below to find national and local services and resources



Community support and independent living

Area Agencies on Aging and Disability
eldercare.acl.gov

Local Centers for Independent Living
ilru.org/projects/cil-net/cil-center-and-association-directory

211 Network
211.org

Administration for Community Living
acl.gov

Patient Resource, by Amy Curran
prezi.com/view/Qb5LRvdjzHk0uDOaInb

Accessibility

ADA National Network
adata.org

Wheelchairs and adaptive technology:
National Seating and Mobility
nsm-seating.com

NuMotion
numotion.com

State Assistive Technology Centers
at3center.net/stateprogram

Employment services

Social Security Ticket to Work Program
choosework.ssa.gov

Lime Connect
limeconnect.com

Disability:IN
disabilityin.org

MDA's National Resource Center is staffed by specialists, many of whom have personal experience navigating school and employment with neuromuscular diseases. Resource Center specialists can provide information on many aspects of living with neuromuscular disease, from getting genetic testing to getting a service animal. They also can direct you to national, regional, and local resources. (Call the MDA Resource Center at **833-ASK-MDA1** or email resourcecenter@mdausa.org.)

Mike has found that MDA Engage educational events (mda.org/engage) also are a useful source of information. Engage programs offer the opportunity to hear from and interact with presenters, which include experts and individuals living with neuromuscular disease.

In January, Mike attended an Engage webinar on “Psychosocial Support for Living with a Chronic Illness,” which has helped him deal with the added stress of the COVID-19 pandemic. “While we miss the social aspect and spontaneity of traditional MDA events, Engage online has been an effective way to share important information,” Mike says.

Make your own luck

When the resources you need aren't available, it's time to think creatively, which is one of Amy's specialties.


For Amy, finding resources that help her continue creative pursuits, like painting, is crucial to her well-being. “Losing my ability to be creative feels like I'm losing my sense of self,” she says. She talks to people with all kinds of disabilities to get ideas, which is how she learned about a gripping tool designed for people with rheumatoid arthritis that helps her hold a paint brush.

“I make it a personal mission to find creative tools and give everybody I know access to tools that help them keep their

personal identity as well,” Amy says.

With that mission in mind, she used Prezi to create an online virtual presentation on adaptive equipment and resources (see Helpful Resources, below). “It's full of different things that can help you with everyday tasks and living creatively so that you don't have to scour the internet,” she says.

Amy also is involved in advocacy work. Her current goal is to change a rule in Pennsylvania that doesn't allow spouses to be paid caregivers. “My partner and I are not married because if we were, he could not be my paid caregiver,” Amy says. She is researching policies in other states and plans to bring that information to her state legislators.

Finding, adapting, or even creating the resources you need isn't always easy, but planning, persistence, and creative thinking can help. “It can be hard work, but if you keep searching and being proactive, something will stick, and it'll pay off in the end,” Josh says. 



Josh Cueter

Charmaine Dymond is a freelance writer in Halifax, Canada.

Job Accommodation Network
askjan.org

State Vocational Rehabilitation Agencies
askearn.org/state-vocational-rehabilitation-agencies

Healthcare services

Center for Medicare Advocacy
medicareadvocacy.org

National Organization for Rare Disorders
rarediseases.org

Patient Services Incorporated
patientservicesinc.org

Pharmaceutical Assistance Programs
medicare.gov/pharmaceutical-assistance-program

Caregiver services

Caregiver Action Network
caregiveraction.org

Caregiver Resource Network
caregiverresource.net

Family Caregiver Alliance
caregiver.org

Financial services

ABLE National Resource Center
ablenrc.org

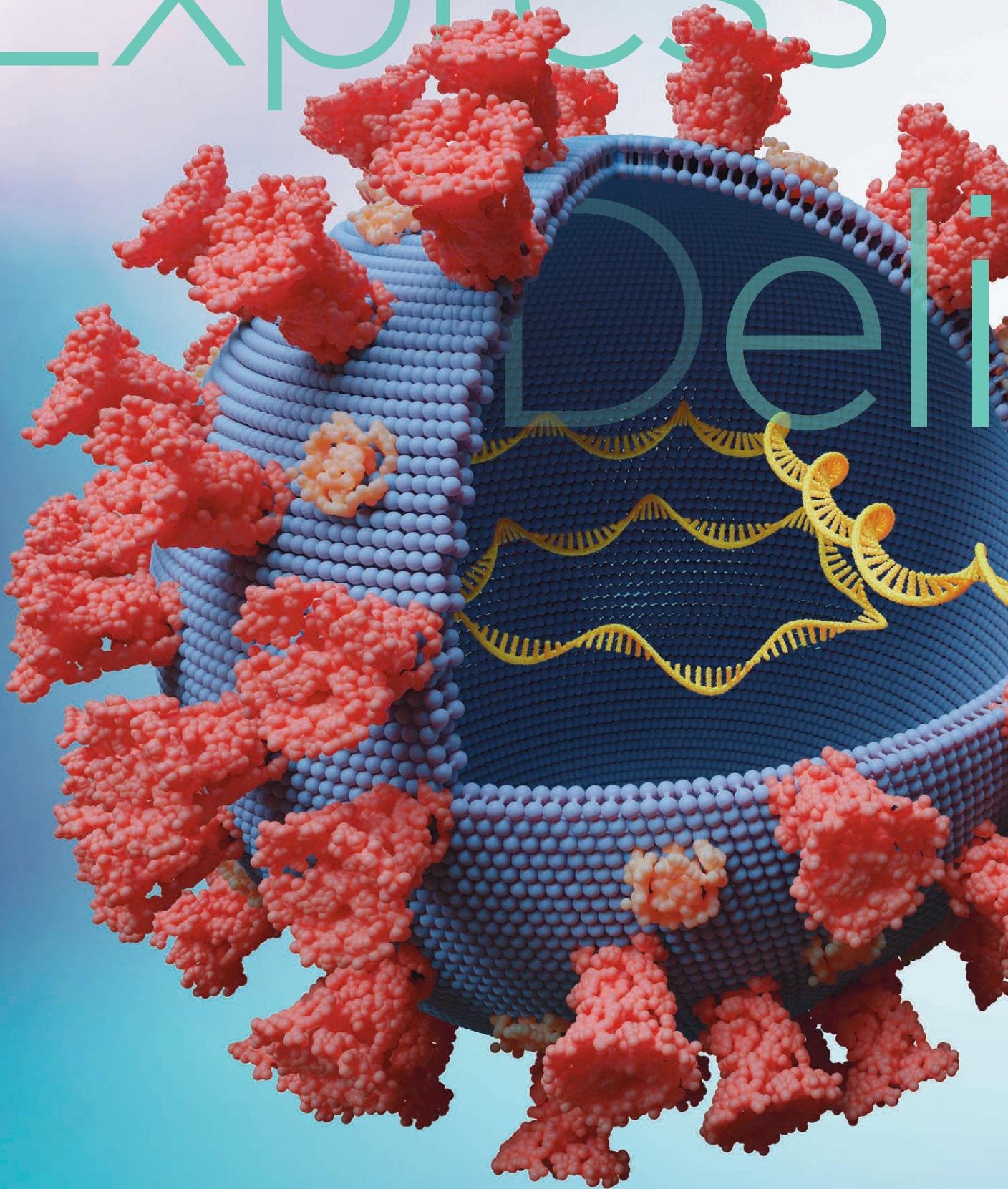
National Disability Institute
nationaldisabilityinstitute.org

NCOA Benefits Checkup
benefitscheckup.org

Prosperity Now
prosperitynow.org



Express



Dei

Viral vectors are an important area of research in the exciting field of gene-targeted therapies

BY LARRY LUXNER

very



Usually, viruses are bad news. But lately, they're getting good press, thanks to the COVID-19 vaccines created by AstraZeneca, Johnson & Johnson, and Immunity Bio, which use viral vectors to deliver instructions to our cells to fight off the novel coronavirus.

Delivering immunity isn't the only thing viral vectors can do. They also hold great promise in gene-targeted therapies for neuromuscular diseases.

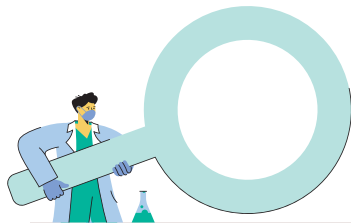
The usefulness of viral vectors in gene therapy has already been demonstrated by Zolgensma, the first drug based on an adeno-associated virus (AAV) vector to be approved by the US Food and Drug Administration (FDA). Zolgensma treats spinal muscular atrophy (SMA) by using a modified virus to deliver a functional SMN gene to target cells. SMA is caused when the SMN gene is missing or mutated. Neurologists around the world are now giving Zolgensma to babies born with SMA to stop the progression of the disease.

So, how are scientists turning viruses into lifesaving therapies? >>

Inside job

“Viruses are very good at entering your body,” says Jeff Chamberlain, PhD, a professor of neurology, medicine, and biochemistry at the University of Washington and director of the Sen. Paul D. Wellstone Muscular Dystrophy Cooperative Research Center of Seattle. “Normally, a virus tries to get into various cells and drop off its own genes in order to make copies of that virus. For gene therapy, the only part of the virus we keep is the shell, because it has all the information you need to get into the body. It can latch onto muscle cells, then break open and release its contents.”

Basically, researchers turn viruses into tiny delivery



WORDS TO KNOW

DNA A long molecule that carries a cell’s genetic information.

genes Segments of DNA that act as blueprints for making proteins for virtually every function and structure in the body.

gene-targeted therapy The introduction, removal, or change in the content of a person’s genetic code, with the goal of treating or curing a disease.

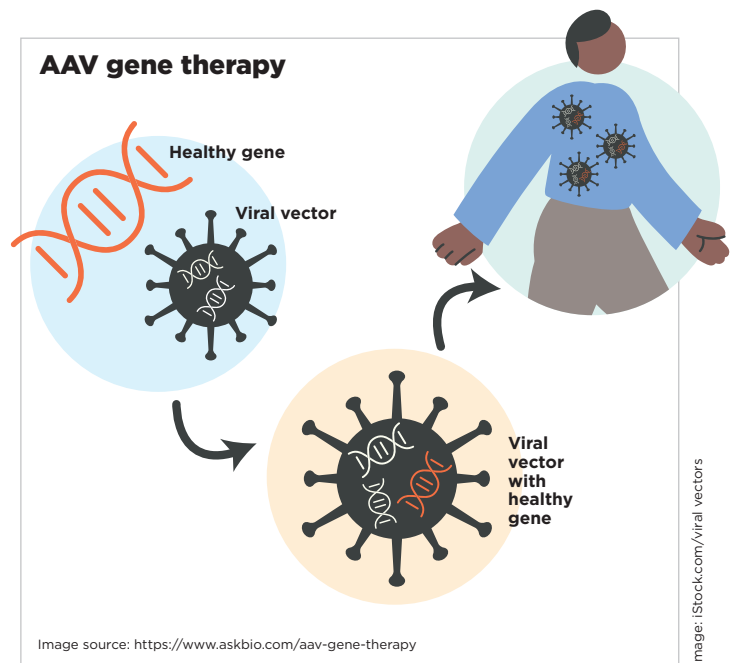
genetic mutation A flaw in a sequence of DNA that alters gene function.

nanoparticle A microscopic particle whose size is measured in nanometers.

nanotechnology Technology that deals with dimensions of less than 100 nanometers, especially the manipulation of individual atoms and molecules.

vector A delivery vehicle that carries new genes to cells.

Image: iStock.com/miakiev



vehicles — or vectors — by removing all of the viral DNA that would make people sick and engineering them to deliver therapeutic DNA sequences to target cells.

“It’s surprisingly simple to grow the virus in a lab in such a way that you get the outer infectious shell, but the only DNA packed into that shell is your therapeutic gene,” says Dr. Chamberlain, who started testing various types of AAV 20 years ago. Much of his work has been supported by MDA grants.

“We thought AAV might be a good delivery vehicle, but there was a problem: It’s a very small vector, so it cannot carry much DNA. That’s a problem with Duchenne muscular dystrophy [DMD] because dystrophin [which is lacking in boys with DMD] is the largest-known gene in nature. All of the early work with AAV involved single injections directly into one muscle, and it didn’t spread.”

In 2004, researchers made a key breakthrough: They found that certain AAV vectors delivered into the bloodstream at high doses were able to transfer new genes to muscles throughout the body. And through trial-and-error mouse studies, scientists came up with a synthetic version of the dystrophin gene that was one-third the size of normal dystrophin — meaning it was small enough to fit into an AAV vector.

“We’ve actually made dozens of these micro-dystrophins and found that some of them worked amazingly well, some not,” he said. “When you combine that with the AAV delivery vehicle, we were able to show that we could deliver a new dystrophin gene to every muscle of the body.”

Micro-gene therapy is now under extensive clinical evaluation for DMD, showing promising early results, and it also is in preclinical testing for dysferlinopathy.

Key challenges

While viral vectors are the leading platform for gene therapy delivery, there are challenges to overcome before these therapies can be distributed to large numbers of people.

One of the biggest challenges is that people may have pre-existing antibodies to the virus used as the vector. For example, AAV is found naturally in many animals, including humans. It does not cause a known illness, so a person wouldn't know if they're infected. But, if that person is given an AAV-based gene therapy, their pre-existing antibodies will neutralize the AAV vector, making the therapy ineffective.

"Between 20% and 80% of the population may not be eligible for a specific gene therapy because they have pre-existing antibodies," says Sharon Hesterlee, PhD, MDA's executive vice president and chief research officer. "There's no way to know if you've been exposed until you're tested for antibodies." Currently, individuals considering receiving gene therapy or entering a clinical trial that involves a viral vector must be tested for antibodies. Having too many of those antibodies makes them ineligible for the treatment or trial.

Pharmaceutical companies have been able to produce large quantities of the COVID-19 vaccine from a different virus, called adenovirus (AV), very quickly because only tiny amounts of the vaccine are required to stimulate the immune response that protects against COVID-19 when it is injected into a muscle. But gene therapy requires a large amount of viral vectors to carry the therapeutic gene to cells, and it must be introduced into the bloodstream. This process not only delivers the therapeutic gene, but it also causes the body to mount an immune response to the viral vector.

That immune response causes another challenge: People who have been treated with a particular gene therapy once cannot be redosed.

"When you use a virus to deliver a healthy gene, you might as well give a person a vaccine against that same virus. The body mounts an immune response to that virus and remembers it," Dr. Hesterlee says.

Finding solutions

Researchers are looking for workarounds to these challenges. One solution is to design therapies that only require one dose, like Zolgensma. "If the first dose is effective, you don't really need a second dose because it's a long-lasting treatment," Dr. Chamberlain says.

He also says researchers are getting closer to solving the problem of redosing. "There is good data in animal models showing there are ways to give a second dose down the road," he says. "Right now, it's a brand-new technology. Manufacturing facilities are not in place, and it's very expensive to make experimental medicines that have never been made before. But the more patients we treat, the less these treatments will cost."

The treatment methods developed for the most common neuromuscular diseases often benefit more rare diseases. "The first studies showing that AAV could be an effective delivery system really came out of the DMD work we were doing, and it's since been applied

+LEARN MORE ABOUT GENES

Genes tell our cells how to grow and work. If you'd like to learn more about the role genes play in neuromuscular disease, download the fact sheet "Genetics and Neuromuscular Disease" at mda.org/education. To learn more about gene-targeted therapies, search for "Targeting Genes" at mda.org/quest.



“

We've actually made dozens of these microdystrophins and found that some of them worked amazingly well, some not. When you combine that with the AAV delivery vehicle, we were able to show that we could deliver a new dystrophin gene to every muscle of the body.

—Jeff Chamberlain, PhD



“

Sometimes there's an overblown immune response, causing a severe reaction. We have to select people who are not likely to have an immune response to the vector and learn how to prevent, monitor, and treat side effects better.

—Sharon Hesterlee, PhD

Advances in nanotechnology are making it possible to develop nanoparticles that can **deliver gene therapy** without triggering an immune response.

to hundreds of diseases,” Dr. Chamberlain says.

One of those diseases is X-linked myotubular myopathy. Audentes Therapeutics has a promising phase 1/2 clinical trial for an AAV-based gene therapy to treat that rare condition. Sarepta Therapeutics is running clinical trials for three different types of limb-girdle muscular dystrophy (LGMD), and AskBio is planning to start a clinical trial for another LGMD therapy.

Closely monitoring how these therapies perform in clinical trials is important, because viral vectors have been linked to rare, but serious, side effects.

“Sometimes there's an overblown immune response,

causing a severe reaction,” Dr. Hesterlee notes. “We have to select people who are not likely to have an immune response to the vector and learn how to prevent, monitor, and treat side effects better.”

Research continues

To avoid the complications of viral vectors, researchers are working on nonviral methods to deliver genes to cells.

Advances in nanotechnology are making it possible to develop nanoparticles that can deliver gene therapy without triggering an immune response, though that technology has not yet advanced to clinical trials. Some pharmaceutical companies are developing their own nonviral gene therapy platforms, including Myosana Therapeutics and Dyne Therapeutics, which are both pursuing nonviral means to treat DMD.

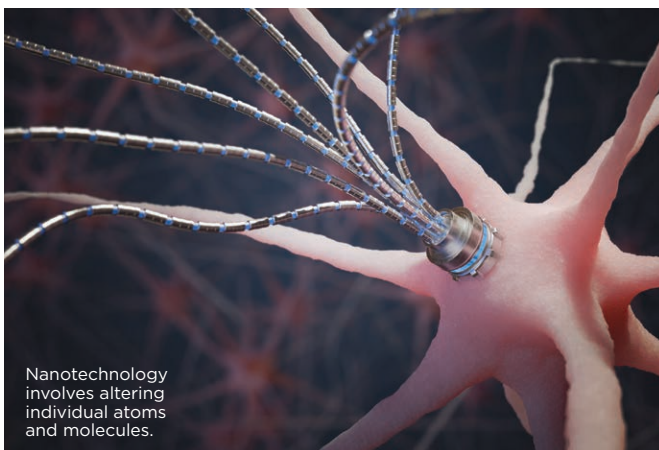
Developing new technologies and therapies is expensive and resources are limited, so research in emerging areas focuses on the most common neuromuscular diseases. But eventually, those technologies will be used in treatments for other diseases. MDA has invested more than \$125 million in the development of gene-targeted therapies that have the potential to benefit the more than 40 diseases under MDA's umbrella.

Despite the challenges, researchers are not giving up on viral vectors. “The benefits are far greater than the safety concerns, and the companies doing these trials now are learning new things every day about how to precondition the body to have less of a reaction,” Dr. Chamberlain says.

In addition, there's still a lot to discover in the AAV space. “There are thousands of kinds of AAVs, and new synthetic versions are looking much better,” Dr. Chamberlain says. “We can also now use higher doses with fewer side effects because of recent advances in how to make these AAV vectors. It's still the early days.”

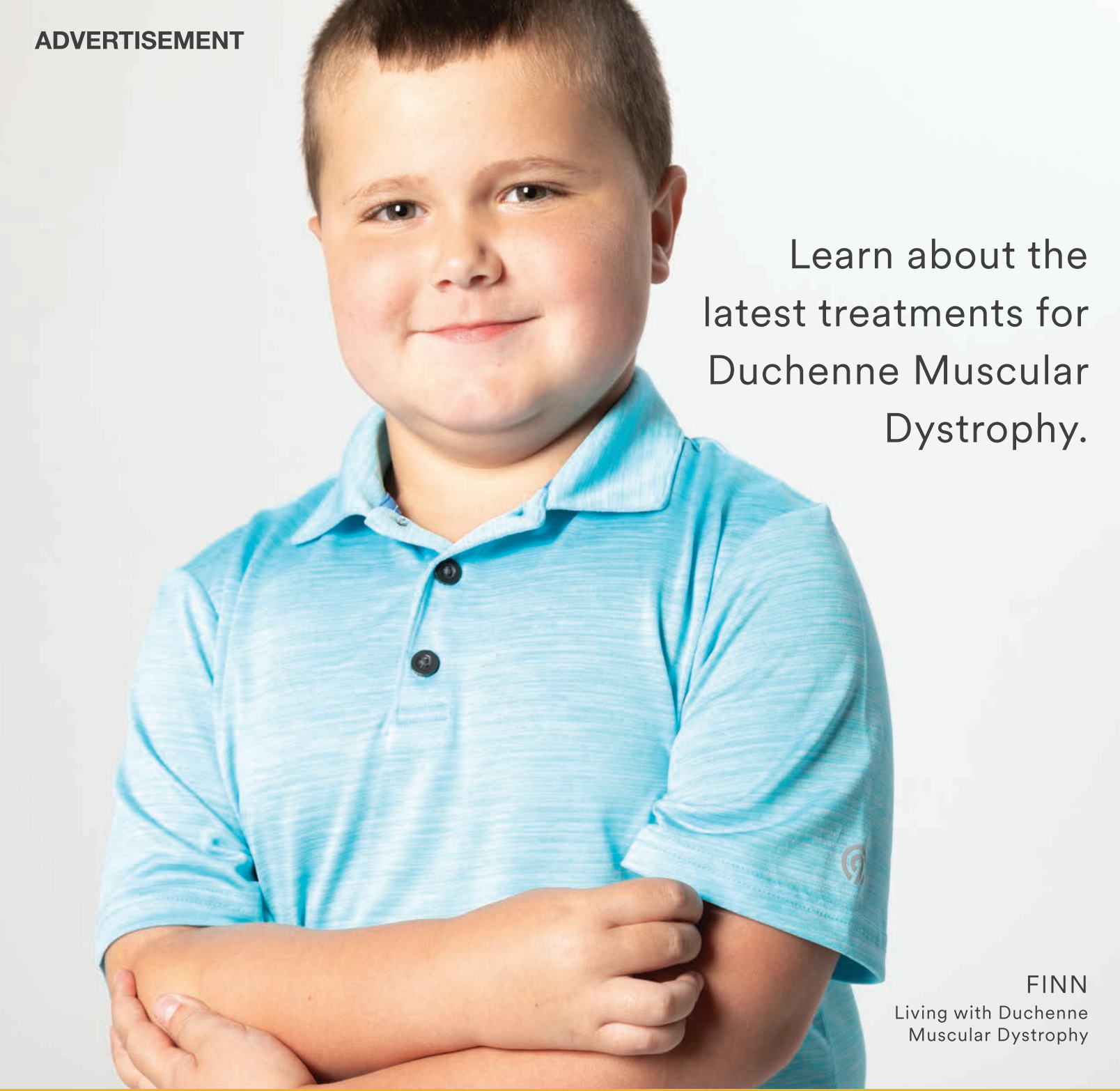
As the days go on, researchers surely will have more treatment methods — both viral and nonviral — they can apply to more diseases. [Q](#)

Larry Luxner is a journalist based in Israel.



Nanotechnology involves altering individual atoms and molecules.

Image: iStock.com/vchal



Learn about the
latest treatments for
Duchenne Muscular
Dystrophy.

FINN
Living with Duchenne
Muscular Dystrophy

Our long-term goal is to provide therapies to 100% of individuals with Duchenne muscular dystrophy by unlocking the full potential of RNA technologies, gene therapy, and gene editing.

FOR MORE INFORMATION, PLEASE VISIT WWW.SAREPTA.COM

Virtually Rewarding

Whether in-person or online, MDA Summer Camp offers invaluable experiences



Davis Cury is an MDA Summer Camp volunteer.

Davis Cury volunteered to be an MDA Summer Camp counselor for the first time in 2019 at in-person camp. When Davis, 21, signed up to help again in 2020, he knew that virtual Summer Camp would offer some interesting challenges. Would campers be able to

make connections on the computer instead of poolside? Could the rewarding moments of Summer Camp be translated online? The answers: Yes.

“It was interesting to figure all of this out, but we were able to take the Summer Camp experience and make it fun virtually,” says Davis, a biochemistry and philosophy

major at the University of Florida, Gainesville. “In fact, I think the virtual platform expanded a lot of boundaries because campers with more advanced medical needs were able to attend.”

Microsoft Teams chat rooms replaced in-person activities, but campers still enjoyed round-robin games, trivia, and — a Summer Camp favorite — arts and crafts.

Davis also was pleasantly surprised that, even while working within HIPAA rules regulating how much personal information could be shared on the digital platform, campers were able to interact and make the hallmark connections of Summer Camp.

“The bonds the kids formed empowered them and really broke down barriers,” he says. “I’m eternally grateful for being able to be a part of that.”

To learn about MDA Summer Camp 2021 or sign up, visit mda.org/virtual-camp.

Running to a Cure

For Nyheim Hines, MDA’s 2021 spokesperson, supporting the mission is personal

Indianapolis Colts running back Nyheim Hines, whose mother, grandmother, and uncle live or have lived with limb girdle muscular dystrophy (LGMD), has been a longtime supporter of MDA. Now, as MDA’s official 2021 spokesperson, he will make an even greater impact in the quest to find a cure.

“Having family members with muscular dystrophy has always motivated me to spend my free time trying to raise awareness, so we can find cures and provide the best care for families across the country living with these diseases,” Nyheim says.

As spokesperson, Nyheim will collaborate with MDA to bring attention to campaigns and fundraisers throughout the year, including MDA Shamrocks, the St. Patrick’s Day-themed pinup and online fundraising campaign.



Nyheim Hines, center, stands with, from left, his sister, Nyah; his mother, Nannette; and his father, Darrin.

Prior to his time as spokesperson, Hines supported MDA through the NFL’s “My Cause My Cleats” campaign, an opportunity for players to wear custom cleats promoting an organization of their choice. His cleats included the MDA logo and the names of his mother, grandmother, and uncle, and his participation in the program brought attention to the neuromuscular disease community.

“I want to use the platform I have to make this world a better place,” he says.

Devoted to Muscle Walk

Ever since Siena Arioto, 12, of Gilroy, Calif., was diagnosed with Friedreich's ataxia (FA) five years ago, MDA has been a huge part of her family's support system. They have enjoyed giving back to MDA by joining Muscle Walk and holding poker tournaments, garage sales, raffles, and even a luau, to raise funds. The tradition continues this year when they hold a socially distanced golf tournament and Facebook fundraiser.

"We will continue to do everything we can to support MDA," says Laura,



Siena with her service dog, Lustra.

Siena's mom. "They are not only working hard to save lives but also make life more enjoyable."

Join the Muscle Walk fun virtually this year by walking, running, doing a yoga class, or moving in any way you choose.

Learn more and sign up at mda.org/muscle-walk.

TIME TO ENGAGE

MDA's Engage community education program provides important, up-to-date, and relevant educational content to the neuromuscular disease community.

Date	Topic
May 20	Gene Therapy and Duchenne Muscular Dystrophy
May 22	Adult Neuromuscular Disease Care
June 5	Limb Girdle Muscular Dystrophy
June 10	Creating Strong Connections and Relationships
July 20	Medical Equipment and Assistive Devices for Neuromuscular Disease (Part 1)
August 10	Medical Equipment and Assistive Devices for Neuromuscular Disease (Part 2)
August 14	Duchenne Muscular Dystrophy

● WEBINAR ● SEMINAR ● SYMPOSIUM

To register for upcoming MDA Engage activities and watch past events, visit mda.org/engage.

THE BOOT IS BACK

Fire fighters are ready with boots in hand — in-person and virtually — for the annual Fill the Boot fundraiser. Over the past 67 years, the International Association of Fire Fighters has helped MDA raise over \$672 million to help find a cure for neuromuscular disease and to provide support and resources to the MDA community. The COVID-19 pandemic has made this fundraiser even more critical than ever.



Donate now by visiting mda.org/ftb.

Image: iStock.com/bortonia

HONOR A FAMILY MEMBER
by making a tribute gift to MDA in your will or trust.

To learn more, visit mdalegacy.org.
For more info, visit mdausa.giftlegacy.com

MDA

A Little Piece of the Future

My experience participating in an FSHD clinical trial

BY CHELSEA MOELLER



Chelsea Moeller and her mother, Laura, both participated in the phase 2b Fulcrum ReDUX4 clinical trial last year.

Of the 80 patients in the phase 2b international Fulcrum ReDUX4 clinical trial, my mom and I are two. To the trial, we are data — nameless numbers in different age groups from states separated by half the country that met all of the eligibility requirements. To us, we are doing something together to advance science on behalf of everyone living with facioscapulohumeral muscular dystrophy (FSHD).

The ReDUX4 trial is testing whether the drug losmapimod will suppress the expression of the troublemaking DUX4 gene. In the human embryo, DUX4 has a brief starring role in igniting the development process. Then, it's supposed

to quit and never be heard from again. In those with FSHD, it doesn't quit, and proteins run amok, attacking and killing healthy muscle tissue. It's a degenerative slope that alters lives.

Saying yes

When the trial enrollment opened, I felt urgency with an edge of avoidance. This would be my first clinical trial. I wanted it, and I didn't.

There were a lot of unknowns — whether I would qualify, whether there would be a slot left if I did, whether I could meet the demands of the trial. It also meant acknowledging my condition and its trajectory in a way I had yet to do. With so many aspects out of my control, I resolved to let eligibility be the answer. If they said yes, so would I.

To be eligible for the trial, my muscles had to be in an active state of decline with enough muscle-to-fat ratio to assess the presence of DUX4 protein in tissue samples. My screening visit was at the University of Massachusetts (UMass) Medical School in Worcester, a three-hour drive from my home in central Maine. It involved a battery of push-pull strength evaluations and an MRI.

Eleven weeks after my first conversation with the trial coordinator, I was confirmed eligible. Four weeks after that, I traveled to Maryland for my first muscle biopsy at the Kennedy Krieger Institute, where they were accustomed to performing a less invasive small-needle biopsy. The beginning of the trial was a dizzying order of operations including the coordination of a million moving parts: the trial schedule, where things were often due within a set window of time; the UMass team's schedule, as well as mine and my husband's; and the availability of diagnostic equipment.

It quickly became evident that the UMass clinical team and I were learning together. I was one of two patients enrolled in the trial at this site. The trial required that all sites follow specific instructions from Fulcrum, which they often read from a notebook while completing. It felt competent while also feeling very "trial."

ReDUX4 is a double-blind, placebo-controlled, multicenter study. Participants were randomly assigned to take pills that either contain the drug or a placebo. No one knows who's getting what until the very last participant has completed the trial and the data analysis, which is happening now, is done. The hope is that results show the drug is more effective than the placebo.

In addition to taking two pills twice a day and logging it into a phone app, the trial involved two muscle biopsies, two partial-body MRIs, check-in visits at specific intervals, blood draws, EKGs, strength assessments, surveys, and physical therapy evaluations. The visits were intended to be spread out over 48 weeks.

Giving thanks

Three months into the trial, the coronavirus pandemic threw a wrench in things, and Fulcrum pivoted: in-person visits were replaced with phone calls and backyard blood draws by mobile phlebotomists, and medication showed up by courier in cryogenic-looking packaging with timers and temperature control.

I was thankful the pandemic didn't fully disrupt trial progress. I was thankful I didn't have to choose between continuing the trial or taking COVID-19 precautions. In fact, my mom and I have had a real sense of gratitude throughout this entire process — grateful to be part of it and for the many people we'll never know who are devoting resources to finding treatments; grateful to our doctors, trial coordinators, physical therapists, and MRI technicians who were kind and calm, even with all the changes. We were treated with respect and appreciation.

In addition to gratitude, my mom and I also felt our privilege throughout. My experience showed me that clinical trials often are a thing for privileged people — people who can accommodate the appointments, travel, and lack of control over what happens and when. I could take off work every few weeks for the overnight trip to UMass. I could travel to Maryland for a biopsy. I had people to watch my daughter when I wanted my husband by my side, and I'm still independent enough to travel alone when needed.

Fulcrum worked to remove as many barriers to participation as possible, paying for travel and accommodations and

“

I was thankful the pandemic didn't fully disrupt trial progress. I was thankful I didn't have to choose between continuing the trial or taking COVID-19 precautions.



providing stipends for appointments. Even so, I can see how trial participation is daunting, because nothing else pauses while you fight for futures like your own.

Looking ahead

From where I sit, participating in this and, I hope, future trials is the least I can do in the face of a condition that will increasingly let me do so little.

I stopped speculating about whether I was in the placebo group early on. On the other side of those 48 weeks and a couple of months into the “open label” portion of this trial, where all participants can opt in to taking losmapimod, I am not the Hulk. I don't feel like I've gotten a lot worse in the last year either, which could be the drug working, or it could just be the disease doing its erratic, slow dance of deterioration.

If you have a progressive condition, though, you know that pausing the process preserves a little piece of the future. Real or imagined, losmapimod or not, I'll take it. [Q](#)

+FIND A TRIAL

Learn more about clinical trials and use MDA's Clinical Trials Finder tool at mda.org/clinical-trials.

Chelsea Moeller, 39, was diagnosed with FSHD in 2012. She lives in Maine with her husband, 4-year-old daughter, and Newfoundland dog. Her mother, Laura, lives in the Midwest, and the two are happy to have each other as they travel the long and winding road of FSHD.

Dancing Through the Journey

The art of movement has been an integral part of Carol Cowley's healing process

For Carol Cowley, 62, dance has been the common thread throughout her life. She began dancing as a teen and continued into adulthood. In her mid-40s, her life was profoundly altered by symptoms related to central core disease (CCD), a type of congenital myopathy. Finding a way to keep dancing helped her to accept and adapt to her changing body.

“Along with being a nurse practitioner for the last 32 years, dance was such a big part of my identity,” says Carol, who lives in Louisville, Colo.

During the years leading up to her CCD diagnosis, Carol struggled to find a cause for her unexplained muscle pain and weakness. Faced with misdiagnoses, unnecessary surgeries and providers who discounted her symptoms, Carol struggled with fear and hopelessness, retired on disability, and stopped dancing.

Then, a few years after her CCD diagnosis, she tried a Dance for Parkinson's class, which is open to anybody with mobility or other chronic health challenges. “It's wonderful,” Carol says. “It's all about adapting, feeling safe, and experiencing the joy of moving with others.”

During the pandemic, Carol continues to take



Carol Cowley performs a dance she choreographed.

Photo: Heather Gray

dance classes from her wheelchair via Zoom. She credits the power of dance with helping her return to work part-time.

“I will never walk barefoot on the sand or backpack again, but I'm learning to let go of what is no longer physically possible for me,” Carol says. “There's a flipside to living with a (dis)ability: it has taught me to be more compassionate with myself and to discover new ways to connect with the things that bring my life joy and purpose — while still honoring my body's limitations.” [Q](#)

+LIFE STORY

If you'd like to share your story with Quest, email us at quest@mda.org.



**TALK TO YOUR DOCTOR
IF YOU HAVE EXPERIENCED
ANY OF THE FOLLOWING:**

- Morning headaches
- Difficulty climbing stairs
- Trouble getting up from a chair
- Unexplained weight loss
- Difficulty breathing—especially while lying down
- Sleep disorders (sleep apnea)
- Excessive daytime fatigue

**IT COULD BE
POMPE
DISEASE**

The average Pompe disease patient goes undiagnosed or misdiagnosed for up to **12 years.**

**Not an actual patient*

Know the Pompe **Spectrum**
at pompe.com

Genetic Testing is Vital.

No two cases are the same.
Check with your doctor about testing options for Pompe disease.

There are many questions about

ALS

You can help find answers.

The National ALS Registry

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



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

Collecting and analyzing data

Striving to better understand the disease

Helping researchers find possible risk factors

Your participation can make a difference.



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



U.S. Department of Health and Human Services
Centers for Disease Control and Prevention
Agency for Toxic Substances and Disease Registry