

EMPOWERING FAMILIES WITH INFORMATION AND INSPIRATION

quest

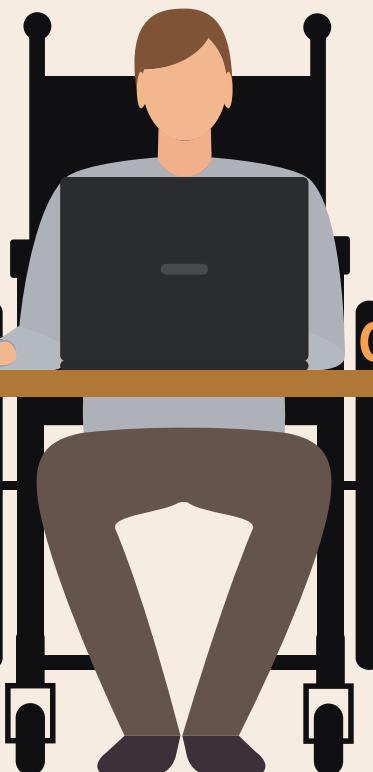
MDA.ORG/QUEST ISSUE 3 • 2021

Help Wanted

Work-from-home
makes employment
more accessible



**DISABILITY
AWARENESS**
How advocates
educate others



**ULTRA-RARE DISEASE
RESEARCH**
Overcoming challenges
to find therapies





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

<p style="text-align: center;">Patient Information EVRYSDI™ [ev-RIZ-dee] (risdiplam) for oral solution</p>	
<p>What is EVRYSDI?</p> <ul style="list-style-type: none"> 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. 	<p>Reusable Oral Syringes</p> <ul style="list-style-type: none"> 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.
<p>Before taking EVRYSDI, tell your healthcare provider about all of your medical conditions, including if you:</p> <ul style="list-style-type: none"> 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: <ul style="list-style-type: none"> 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. 	<p>What are the possible side effects of EVRYSDI?</p> <p>The most common side effects of EVRYSDI include:</p> <ul style="list-style-type: none"> For later-onset SMA: <ul style="list-style-type: none"> fever diarrhea rash For infantile-onset SMA: <ul style="list-style-type: none"> fever runny nose, sneezing, sore throat, and cough (upper respiratory infection) diarrhea lung infection vomiting rash <p>These are not all of the possible side effects of EVRYSDI. For more information, ask your healthcare provider or pharmacist.</p> <p>Call your doctor for medical advice about side effects. You may report side effects to FDA at 1-800-FDA-1088.</p>
<p>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.</p> <p>How should I take EVRYSDI?</p> <p>See the detailed Instructions for Use that comes with EVRYSDI for information on how to take or give EVRYSDI oral solution.</p> <ul style="list-style-type: none"> 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. <p>Taking EVRYSDI</p> <ul style="list-style-type: none"> 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. <ul style="list-style-type: none"> 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: <ul style="list-style-type: none"> 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. 	<p>How should I store EVRYSDI?</p> <ul style="list-style-type: none"> 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). <p>Keep EVRYSDI and all medicines out of the reach of children.</p> <p>General information about the safe and effective use of EVRYSDI.</p> <p>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.</p> <p>What are the ingredients in EVRYSDI?</p> <p>Active ingredient: risdiplam</p> <p>Inactive ingredients: ascorbic acid, disodium edetate dihydrate, isomalt, mannitol, polyethylene glycol 6000, sodium benzoate, strawberry flavor, sucralose, and tartaric acid.</p> <p>Genentech <i>A Member of the Roche Group</i></p> <p>EVRYSDI™ (risdiplam)</p> <p>Distributed by: Genentech, Inc. A Member of the Roche Group EVRYSDI is a trademark of 1 DNA Way Genentech, Inc. South San Francisco, CA M-US-00007143(v1.0) 94080-4990 ©2020 Genentech, Inc.</p> <p>For more information, go to www.EVRYSDI.com or call 1-833-387-9734.</p>

This Patient Information has been approved by the U.S. Food and Drug Administration.

Approved: 8/2020

A Summer of Promise

I hope you, your family, and your friends are enjoying the summer as we emerge from the isolation of COVID-19 and can safely enjoy each other's company again. It has been a long and challenging 18+ months for all of us, and I know we are all eager to get back to our lives.

But there are some changes that I hope we will hold on to. October is National Disability Employment Awareness Month, and one of our feature articles in this issue, "Help Wanted" (page 14), addresses how the COVID-19 pandemic has led to more remote work opportunities. I have been delighted by how this change has been embraced by so many employers that would have otherwise told employees that a work-from-home option was not possible. Many are planning to continue this practice in some form,

even when it is safe to return to the office. Not only is this a boon to individuals with disabilities, but it is also a benefit to employers. Research shows companies that embrace best practices for employing and supporting more persons with disabilities in their workforce outperform their peers. Thanks to remote work, the post-pandemic future could be a more inclusive one for all Americans.

I'm pleased to report that MDA Virtual Summer Camp was a big hit again this year. Even though we could not get together in person, our campers made new friends, enjoyed new experiences, participated in great activities, and had an all-around blast. There were also plenty of new opportunities that could only happen in a virtual environment, such as meeting fellow campers from across the country. Many thanks to our dedicated volunteers and generous sponsors for making this experience possible.

Sincerely,

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



Donald S. Wood, PhD

+WHAT DO YOU THINK ABOUT QUEST?

Please take a quick Quest reader survey at [surveymonkey.com/r/quest2021](https://www.surveymonkey.com/r/quest2021), or scan this QR code.



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.

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PUBLICATION MANAGEMENT



Kyle Bryant (left) and Sean Baumstark (right) host a podcast about living with disability.

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One in a Million

There's reason for hope in researching therapies for ultra-rare diseases.

Cover image: iStock.com/architetta; lolon

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LASTING IMPRESSION

One racing wheelchair links four MDA families.

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ONLINE



PHOTO CONTEST

Share a photo of a meaningful moment for you or a loved one with a neuromuscular disease, and it could be selected to appear in a future issue of *Quest*. Submit your photo at surveymonkey.com/r/questphoto or scan this QR code.



NEW MARKETPLACE

Check out the new *Quest* Marketplace section to find advertisements for products and services for the neuromuscular disease community in one place. Turn to **page 33**.

MDA-074

Helping Families Overcome Barriers

At MDA, we aim to bring knowledge and resources to you in accessible and understandable ways. Our community education programs give you access to a variety of important, up-to-date educational content. Our newest offering, MDA Access Workshops, are designed to address common access barriers that families in the neuromuscular disease community face.

Access Workshops

MDA's Access Workshops are on-demand, online community education programs created to provide information and resources on overcoming barriers to a variety of access topics that are relevant to the neuromuscular disease community.

Each workshop is composed of a series of modules with important information, educational videos, and interactive elements, like quizzes, that build participants' knowledge and understanding of the topic. The workshops are designed to increase health literacy, empowerment, and self-advocacy within the community.



MDA Engage Educational Events

Event	Date
Engage Webinar: Your Family. Your Future.	Sept. 14
Engage MG Symposium	Sept. 18
Engage Caregivers Seminar	Sept. 25
Engage Webinar: Newborn Screening	Oct. 6
Engage Webinar: FSHD	Oct. 21
Engage Seminar — Pediatric Care	Oct. 23
Engage SMA Symposium	Nov. 13
Engage Seminar — Adult Care	Dec. 4

Dates are subject to change.

EVENT INFO

Find more information about upcoming MDA Engage educational programming and watch past events at mda.org/engage.

"The Access Workshops are meant to be self-paced programs," says Elise Qvale, MDA's director of professional and community education. "If someone's interested in an entire topic, they may want to go through the complete workshop from start to finish to learn a lot about that topic and take away all of that material. Other people may want to go to the specific module that speaks to where they are in the process. For example, if someone just got denied a piece of equipment, they've already passed a few of the stages in our modules, so they might just go to the module on denials and pull the information they need from there."

Workshop topics

These Access Workshops are currently available:

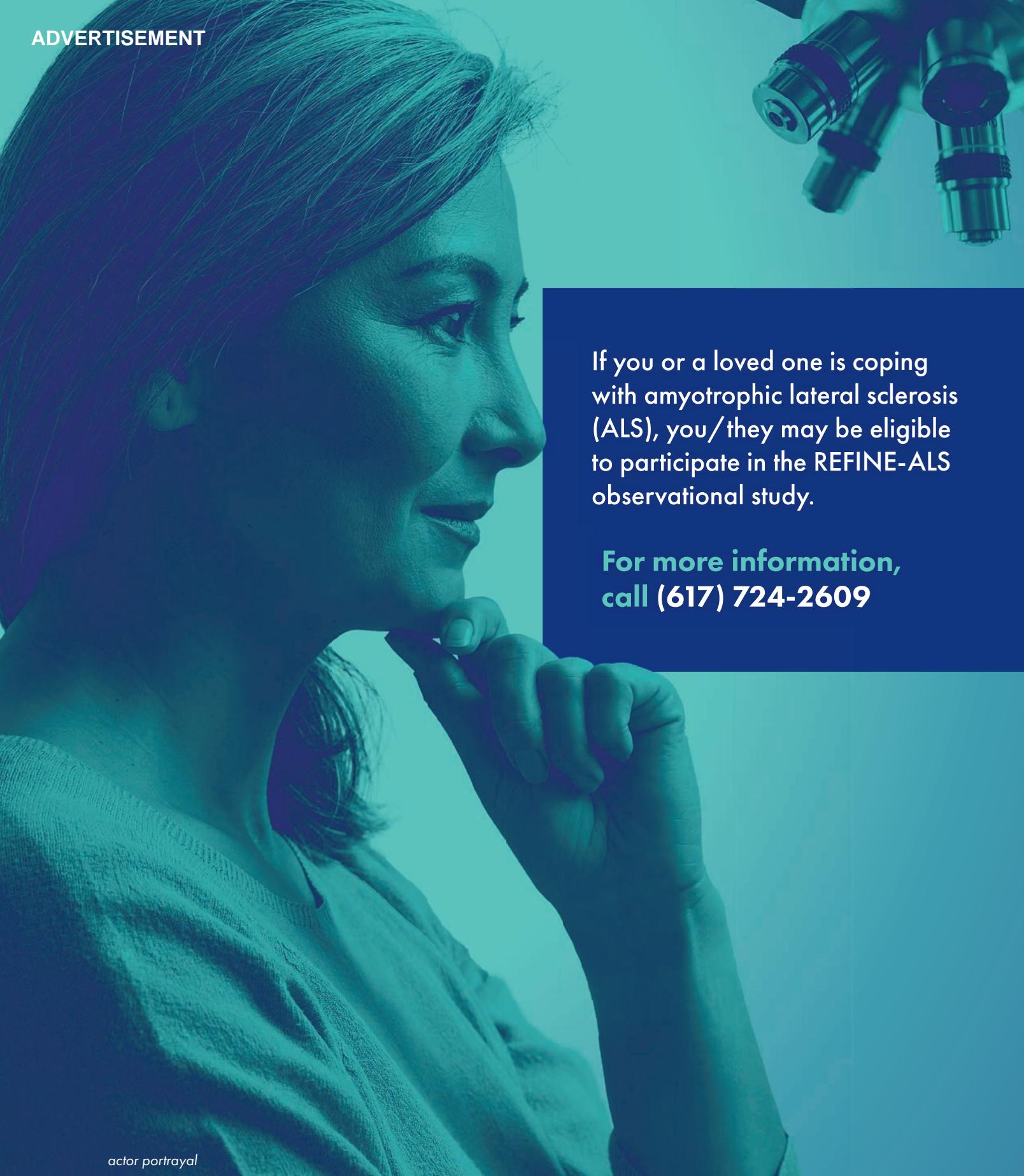
- Access to Coverage: Equipment & Assistive Devices
- Access to Education: K-12
- Access to Education:
Higher Education

Additional workshops on access to insurance and therapies will be available later in the year. [Q](#)

LEARN MORE

To find MDA's current Access Workshops and learn about upcoming ones, visit mda.org/accessworkshops. For questions about Access Workshops, email access@mdausa.org.

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If you or a loved one is coping
with amyotrophic lateral sclerosis
(ALS), you/they may be eligible
to participate in the REFINER-ALS
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actor portrayal



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REFINE  **ALS**
OBSERVATIONAL STUDY

PROGRESS NOW

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

Amyotrophic lateral sclerosis (ALS)

Participants Sought for Phase 3 Trial

Researchers at IONIS Pharmaceuticals are seeking individuals living with FUS-ALS (ALS caused by a mutation in the fused in sarcoma, or *FUS*, gene) to participate in a phase 3 clinical trial to determine the efficacy of the investigational drug ION363 (Jacifusen).

FUS gene mutations lead to the production of abnormal *FUS* proteins that form toxic clumps inside nerve cells and cause nerve damage. ION363 is an antisense therapy that interferes with the cell's protein-making machinery, leading to lower *FUS* protein levels.

The two-part study will last about two years. In part one, participants will be randomly assigned to groups designated to receive a multidose regimen of ION363 or a placebo, administered by injection into the spine, over 29 weeks. Part two is an open-label extension period during which all participants will receive ION363 for 77 weeks.

Eligibility criteria include:

- Showing signs or symptoms consistent with ALS
- Having a confirmed *FUS* gene mutation
- Having received stable concomitant medications (drugs



other than ION363) and nutritional support for at least one month prior to the start of the study

Travel support may be available.

To learn more, visit clinicaltrials.gov and enter NCT04768972 in the "Other terms" search box, or email patients@ionisph.com.



MERIDIAN Trial Enrollment Open

Researchers at Apellis Pharmaceuticals are seeking individuals living with sporadic ALS to participate in the two-year, phase 2 MERIDIAN trial to assess efficacy of the investigational drug pegcetacoplan.

Pegcetacoplan is designed to reduce the activity of the complement system (a component of the immune system) and potentially slow the progression of ALS.

Trial participants will be randomly assigned to receive either pegcetacoplan or a placebo for the first year of the study. They will self-administer their treatment at home two times a week as a subcutaneous (under-the-skin) injection. In the second year of the study, all participants will receive pegcetacoplan. Upon completion of the trial, participants will have the option to enroll in a long-term extension study.

Eligibility criteria include:

- Being at least 18 years old
 - Having a definite, probable, or laboratory-supported probable diagnosis of sporadic ALS, as defined by the revised El Escorial criteria
 - Having experienced onset of ALS symptoms within 72 weeks (18 months) prior to screening
 - Having a total Revised ALS Functional Rating Scale (ALSFRS-R) score of at least 30 at the time of screening
- Travel support may be available.

To learn more about the MERIDIAN trial, visit [clinicaltrials.gov](#) and enter NCT04579666 in the “Other terms” search box, or visit [meridiantrial.com](#).

Smartphone App Study Volunteers Needed

Researchers at Johns Hopkins University School of Medicine are seeking individuals living with ALS, primary lateral sclerosis (PLS), or a related motor neuron disease to participate in an observational study that uses a survey to assess whether disease progression can be tracked with a smartphone application.

The study uses REDCap, a secure website for medical research, to collect information and enable participants to consent and enroll.

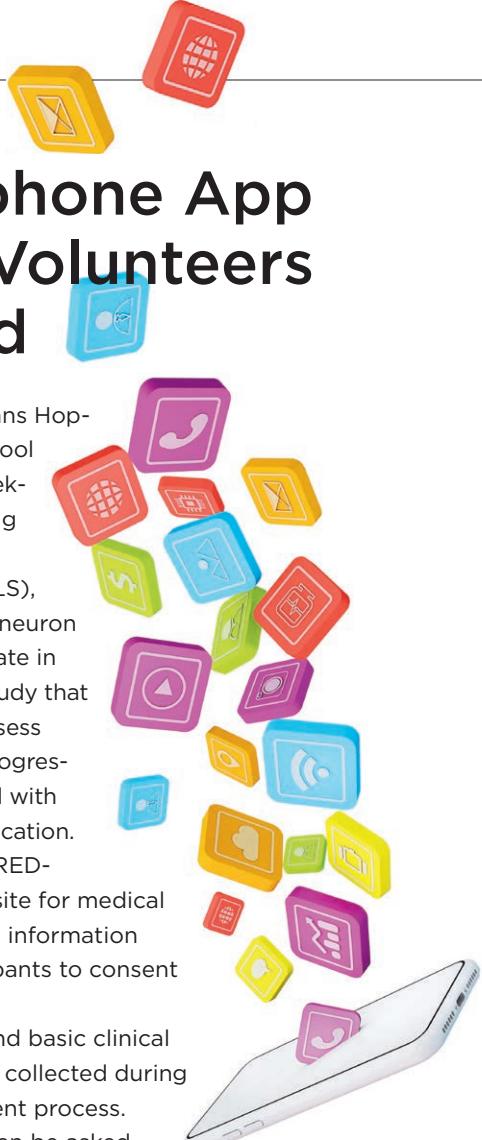
Demographic and basic clinical information will be collected during the online enrollment process. Participants will then be asked to use the Companion App for as long as they like or are able to do so. Study participants will use the app to complete four voice-recording tasks each week. In addition, participants with ALS will be asked to complete a Revised Functional Rating Scale (ALSFRS-R) questionnaire at predefined time points.

Eligibility criteria include:

- Being at least 18 years old
- Having access to a smartphone or device on which to use the application
- Having a confirmed diagnosis of ALS, PLS, or other related motor neuron disease, or being a pre-symptomatic ALS gene carrier

Healthy volunteers may also be eligible. Individuals may not be eligible to participate if they are unable to speak without an assistive device.

To learn more or enroll in the study, visit [mrprcbcw.hosts.jhmi.edu/redcap/surveys/?s=9TPWLWNCXK](#).





Patient-reported outcome measures allow the efficacy of a clinical intervention to be measured from a patient's perspective.

Duchenne muscular dystrophy (DMD)

Participants Needed for Health Survey Development

Researchers at the University of Rochester are seeking individuals to assist with the final phase of development of a disease-specific, patient-reported outcome measure for clinical trials involving people ages 11 and older with DMD. Patient-reported outcome measures are standardized, validated questionnaires that are completed by patients and designed to measure the patient's views of their health status, perceived level of impairment, disability, and health-related quality of life. For this phase of research, participants will be asked to complete an online survey twice separated by two weeks. Each survey will take approximately 20 minutes to complete and will ask some personal questions about the individual, their household, and how the disease affects their quality of life physically, mentally, and emotionally.

Participants are emailed a link to the survey to complete online. Approximately two weeks later, the participant will receive another link via email asking you to complete the survey a second time. For the purposes of this phase of the research, it is important that the second survey is taken within two days of receiving the second link.

Individuals ages 11 or older who have a diagnosis of DMD and are interested in participating, contact study coordinator Jennifer Weinstein at **585-419-5335** or email jennifer.weinstein@chert.rochester.edu.

Pompe disease

Gene Transfer Study for Late-Onset Pompe



This gene-replacement therapy is designed to address the recognized limitations of enzyme replacement therapy by targeting muscle tissues.

Researchers at Audentes Therapeutics are seeking individuals living with late-onset Pompe disease (LOPD) to participate in a phase 1/2 study to confirm safety and efficacy of the investigational drug AT845. This gene-replacement therapy may offer the benefit of long-term improvement of motor and respiratory function and quality of life in adults living with LOPD.

All participants will receive AT845 during this study. The gene-replacement therapy will be administered via a one-time intravenous infusion (slow injection into a vein) in a hospital setting.

The duration of the study is approximately five to six-and-a-half years. Participants may experience an 18-month evaluation period before receiving treatment with AT845. After treatment, there will

In total, the study will require approximately 23 to 39 scheduled visits, including an inpatient assessment of one to two days.

be a core observation period of 48 weeks with scheduled visits and assessments, followed by visits every six months to assess safety for up to five years post-treatment. In total, the study will require approximately 23 to 39 scheduled visits, including an inpatient assessment of one to two days. Travel support may be available.

To be eligible to participate, individuals must be at least 18 years old, have a documented clinical diagnosis of Pompe disease confirmed by genetic testing, and meet additional criteria.

For more information, visit clinicaltrials.gov and enter NCT04174105 in the “Other terms” search box, or visit audentestx.com/pompe-disease. To inquire about participation, contact patientadvocacy@audentestx.com.

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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.

**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

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 or 1-844-4SPINRAZA (1-844-477-4672).

MANUFACTURED FOR

Biogen, Cambridge, MA 02142



Researchers Want to Know More About Oculopharyngeal Muscular Dystrophy

A Q&A
with Anita
Corbett, PhD



Oculopharyngeal muscular dystrophy (OPMD) is rare, even among muscular dystrophies, with an incidence of 1 in 100,000 people. It is a late-onset disease, with symptoms typically first appearing after age 50.

OPMD gets its cumbersome name from “oculopharyngeal,” meaning that the disease affects the muscles of the upper eyelids and the throat. People with OPMD generally develop

droopy eyelids, called ptosis, as well as difficulty swallowing, or dysphagia. They also can experience progressive weakness in the legs.

To learn more about OPMD, we spoke with Anita H. Corbett, PhD, a professor of biology at Atlanta’s Emory University and a recognized expert on the condition.

How do you recognize OPMD?

It doesn’t hit until later in life. The first thing that sends most people to the doctor is that their eyelids are drooping. They often go to the eye doctor because they are having to tilt their heads back to see.

OPMD may be underdiagnosed because one of its chief symptoms is difficulty swallowing. There are many causes of dysphagia, and this would not be the most common diagnosis. Most clinicians probably have never heard of this disease.

How is OPMD inherited?

It's an autosomal dominant disorder, so if you have a parent with OPMD, you have a 50% chance of inheriting it.

I've met people who know the disease runs in their family and don't even go to get diagnosed when they notice symptoms, because they know what it is. Others learn about it for the first time when they are diagnosed in their 60s or 70s. They often have children — and their children have children — who could be affected. As people learn that this runs in their families, they'll need genetic testing and counseling, especially for couples deciding to have children.

How prevalent is OPMD?

In North America, OPMD seems to be more prevalent in New Mexico and parts of upstate New York, as well as in French-speaking Quebec, where the frequency is 1 in 1,000. The world's highest frequency of OPMD has been reported among Israel's Bukharan Jews (originally from Uzbekistan), with an incidence of 1 in 600.

How is OPMD currently treated?

Doctors can do surgery on the eyelids to help people see better. There's also a surgery where they take some tissue out of the throat to increase the amount of space and make swallowing easier. These procedures may need to be repeated for patients who live longer.

What causes OPMD?

The disease is caused by a genetic change marked by the abnormal expansion of the alanine-encoding (GCN)n trinucleotide repeat in exon 1 of the polyadenosine (poly[A]) binding protein nuclear 1 (*PABPN1*) gene. [Alanine is an amino acid that is used to make proteins.] Instead of the normal 10 alanines in the *PABPN1* gene, people with OPMD have 11 to 18 alanines.

It's a really modest change, and no one understands why that change causes the effects of this disease. We do know it makes PABPN1 protein stick together. This protein is critical for basic gene expression. But PABPN1 protein is in all cells of our body, so why does OPMD affect muscles? We don't know.

How did you become interested in this disease?

I got into it about 15 years ago because I study the class of proteins like PABPN1. I noticed that although people were trying to study OPMD, nobody was studying it in muscle. My colleague at the time, Grace Pavlath [MDA's former chief research officer], was a professor at Emory who worked on skeletal muscle. She and I were training to run a half-marathon together, and it came up during one of our runs. We put our expertise together to focus on this understudied condition.



Anita Corbett, PhD

What research is going on for OPMD?

Most of the current research focuses on understanding the disease mechanisms and progression in OPMD. Researchers are focused on potential treatments that can either decrease the amount of defective PABPN1 or overcome the fact that defective PABPN1 sticks together. Only one drug intervention has been explored: A 2017 trial by Bioblast Pharma that looked at the tolerability and efficacy of trehalose (Cabaletta), which would combat the self-association of PABPN1, to treat OPMD.

A total of 12 clinical trials related to OPMD are listed in the US National Library of Medicine's database, clinicaltrials.gov, indicating that there are limited studies to combat this condition. Clearly, additional efforts are required to address this disease. Currently, no global registry of OPMD patients exists, which would help researchers interested in studying the disease. [Q](#)

“

I noticed that although people were trying to study this disease, nobody was studying it in muscle.

RARE DISEASE SPOTLIGHT

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

HELP At Home

Finding accessible employment in the work-from-home era

BY KAREN DOSS BOWMAN

Sarah Schwegel, an organizing and advocacy specialist with Paraquad in St. Louis, began working from home last March as the COVID-19 pandemic emerged in the United States. Having spinal muscular atrophy (SMA), Sarah realized that she was at higher risk for complications if she contracted the illness. Her employer supported her request to work remotely.

"My supervisor said, 'Do what you need to do to stay safe and healthy,'" says Sarah, who is passionate about helping people with disabilities become as independent as possible through self-advocacy and policy change. "In the past year, I've really enjoyed being able to work from home because it adds so much flexibility, especially when scheduling attendants. It also was just a lot easier to get work done at home, where everything is set up for me."

Work-from-home gains ground

The COVID-19 pandemic disrupted life across the globe, but one silver lining emerged from this crisis: The majority of employers embraced remote work, also called telecommuting or telework, as offices around the country were shuttered to prevent the spread of infection.

Data from the Society for Human Resource Management (SHRM) shows that more than 75% of employers required or allowed employees to work from home during the pandemic in 2020. Further studies show that, for the most part, remote work was popular with workers and businesses. A survey conducted by PwC in late 2020 found a positive change

in employers' attitudes toward working from home, with 83% saying the shift to remote work has been successful for their company.

For many people with neuromuscular disease, these are hopeful signs for the possibility of expanded work-from-home opportunities in the future.

"Being able to work from home has been a long, hard-fought-for reasonable accommodation that many people with disabilities need in order to work successfully," Sarah says. "Before the pandemic, a lot of employers were reluctant to allow remote work. Fortunately, the pandemic did a great job of forcing many of us to work in fully accessible environments, like our homes. And I'm really hopeful that as we transition back to the office, working remotely stays an option; it makes the workplace so much more accessible."

What the law says

The Americans with Disabilities Act (ADA) requires employers to make reasonable accommodations to allow employees to perform essential or major functions of their positions. In addition, the US Equal Employment Opportunity Commission's (EEOC) Enforcement Guidance on Reasonable Accommodation and Undue Hardship Under the ADA states that for many jobs, working from home may be a reasonable accommodation for people with disabilities.

"Employers seem to have figured out that they can successfully manage their businesses and their employees from home and that if you allow people to work from home, your profits

UNITED





and productivity don't decline — they may well increase," says Allison Nichol, an adjunct professor at Georgetown Law in Washington, DC, and director of legal advocacy for the Epilepsy Foundation. "People work just as hard at home as they do in the office, so I think this has opened up lots of opportunities that perhaps didn't exist before."

Transforming the workplace

For many employees with disabilities, the pandemic provided their first opportunity to experience accommodations like

working from home or flexible hours. These nontraditional work styles offer numerous benefits for people with neuro-muscular diseases.

For example, eliminating the commute to an office allows them to conserve their physical and mental energy for the responsibilities of the job. For those who rely on caregivers for transportation, it takes away concerns about unexpected circumstances preventing them from getting to work.

Working from home also allows individuals with disabilities to perform their jobs in comfortable, accessible environments tailored to their personal needs. They don't have to worry about the logistics of eating lunch or going to the bathroom because they have their own accommodations — whether through assistive devices or personal care attendants.

"Working from home allows us to use the supports that we naturally have built into our homes to be more successful and supported adequately in our jobs," says Josie Badger, DHCE, CRC, of New Castle, Pa. As founder and president of J Badger Consulting, Josie provides youth development and disability consulting services to organizations.

With the growing emphasis on diversity and inclusion in corporate America, many employers are beginning to realize that removing accessibility barriers for employees with disabilities is not just the right thing to do, but it's also good for business. A commitment to a diverse and accessible workplace can be a selling point to draw talent and lead to better performance from some employees. (See "How to Find a Job That Works for You" on page 17 to learn more about finding accessible employment.)

HOW TO NAIL A VIRTUAL INTERVIEW

It's more likely than ever that your next job interview will be virtual.

"Virtual interviews can be difficult because you lack a level of personal connection that is really important in making that first impression," says Sarah Schwegel, an organizing and advocacy specialist with Paraquad. "On the other hand, they're way more accessible."

Follow these tips to be at your best on video:

- 1 Dress for the job you want.** During the pandemic, many people have become comfortable working in their PJs. Resist the urge to be casual and dress professionally — at least from the waist up.
- 2 Post a cheat sheet next to your webcam.** You'll want to look straight into your webcam so you appear to make eye contact with the interviewer, not be caught looking down at notes. Print out your talking points and post the sheet next to your webcam so you can see it while maintaining eye contact.

3 Set the mood. Find a spot with good lighting, preferably natural light near a window. Also, make sure your background is uncluttered and professional.

4 Eliminate distractions. In this new Zoom era, coworkers sometimes get to know each other's children and pets, who often appear on camera. For a job interview, however, close the door and keep your kids and furry friends out of the picture. Be sure to set your phone and computer notifications to "do not disturb."

5 Check your tech. Make sure your technology is in good working order, with a high-speed internet connection and functioning audio. Using a headset may offer the best sound. Test your equipment before the big day.



Sarah Schwegel says it's easier to get her work done at home.

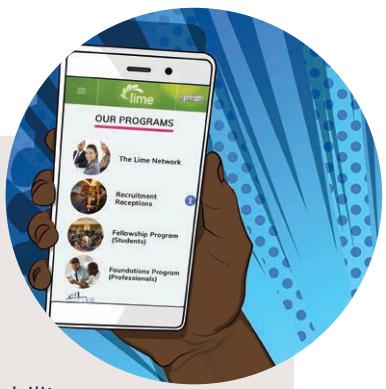
Employers are beginning to acknowledge that working from home is here to stay. Leading companies, including Google, Twitter, and Citigroup, have announced they will move to allowing permanent remote work or a hybrid schedule following the pandemic. And people with disabilities have much to gain from this transformation of the workplace.

"I think the shift to remote work during the pandemic opened employers' eyes to the fact that disabled people are incredible hires," Sarah says. "Having the opportunity to work from home during the last year has made it so much easier for people with disabilities to get jobs and for companies to see that we're great employees." **Q**

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

HOW TO FIND A JOB THAT WORKS FOR YOU

Job hunting with a disability can be intimidating. Here are tips for finding a job that plays to your strengths with an employer who recognizes your talent.



CONNECT WITH TICKET TO WORK. This free program of the Social Security Administration allows Social Security Disability Insurance (SSDI) or Supplemental Security Income (SSI) beneficiaries to explore work options without losing benefits. For some people with disabilities, income from employment could jeopardize government benefits, such as Medicaid, that help cover costs for personal care attendants, points out Josie Badger, DHCE, CRC, founder and president of J Badger Consulting. Before starting a job hunt, "you need to start those conversations, know your earning limits, and know what decisions you will have to make," she says.

Resource: Ticket to Work (choosework.ssa.gov)

SEEK COMPANIES THAT WANT A DIVERSE WORKFORCE. Browse potential employers' websites to learn about their core values on diversity and inclusion (D&I), and note how prominently disabilities and accommodations factor into their D&I policies and initiatives. You also can use online employment search tools that help candidates connect to inclusive employers. "We all need to be looking at jobs with organizations that want us," Josie says.

Resource: Disability:IN (disabilityin.org); Inclusively (inclusively.com)

BE YOURSELF. The ADA doesn't require jobseekers to disclose disabilities prior to receiving a job offer, but Sarah Schwegel, an organizing and advocacy specialist with Paraquad, recommends speaking about it early and candidly. In her experience, talking openly about her disability makes employers more comfortable and willing to have conversations about accommodations, diversity, and inclusion. "The best way to eliminate the stigma and discrimination around disability is to be real," she says. "Talk about your disability and normalize the conversation. Disability is an extremely normal part of human existence."

Resource: ADA National Network (adata.org)

PITCH DISABILITY AS A STRENGTH. Think about skills you've gained in all areas of your life, not just on the job. Sarah often mentions her disability in cover letters and interviews as a way to illustrate professional skills she has developed throughout her lifetime. "You can talk about your drive and work ethic and how you are able to problem solve in challenging situations," she says. "Every person with a disability has to do some pretty impressive problem solving, whether it's coordinating transportation, managing personal care attendants, or making sure that their world is accessible. We absolutely can use that to our advantage when we are applying for jobs."

Resource: Lime Connect (limeconnect.com)

CLEARLY STATE THE ACCOMMODATIONS YOU NEED FOR SUCCESS. There's a wide range of potential accommodations, from permission to work from home or flexible hours to voice-to-text software. Be specific in requesting the supports that will help you perform your best. "If you anticipate ever needing an accommodation, be upfront and put it in writing," Josie says. "You can talk about it first, but then follow-up with an email that outlines the conversation you had. And then save it so you have documentation."

Resource: Job Accommodation Network (askjan.org)

Creating



Navigating the impacts of a neuromuscular disease can be challenging enough, but added on top of that is the responsibility to educate others — talking to people at work, school, and in the community about disability awareness. Although having to address this issue again and again can be exhausting, the effort is crucial, believes Stephanie Bowers-Legg, who lives with myasthenia gravis (MG).

Stephanie, a servicing operations associate at the financial technology company Brex, based in Vancouver, Wash., runs an employee resource group for people with disabilities and their allies. She also set up a private Slack channel for her colleagues with disabilities and organizes in-person events where they can meet and share experiences.

Awareness

Educating others about disability can be a heavy lift, but advocates in our community say it's worth the effort

BY ELIZABETH MILLARD



Kyle Bryant (left) and Sean Baumstark (right) host a podcast about living with Friedreich's ataxia.

Stephanie discovered the need for more awareness and advocacy after she was diagnosed six years ago at age 23 and became an ambulatory wheelchair user. The confusion and occasional disbelief she encountered about why she would need a wheelchair on some days but not others made her realize that many people would benefit from disability education.

"In general, my disability tends to be an invisible one, and that makes it harder for people to understand, which is why giving them more information is crucial," she says. "I'm really open to talking about it, especially when meeting new people, because I think we need to normalize the conversation about this."

By connecting people together at her work, Stephanie helps them learn ways to talk about their own disabilities and provides much-needed support and encouragement.

"Having a shared experience can make you feel confident when you're

talking to people on your own," she says. "You know you're not alone."

Seeing the other side

When addressing disability issues, it can help to team up. That was the case for Kyle Bryant and Sean Baumstark, who both have Friedreich's ataxia (FA). The pair did a cross-country bike ride in 2010 that was filmed by a documentary film crew, and talking about what it was like to live with disability on camera inspired them to keep the conversation going.

In 2016, the friends started a podcast, Two Disabled Dudes (twodisableddudes.com), and Kyle says even after a few years, people still seem surprised that they have such a good time on the show.

"For some reason, people don't associate disability with laughing and having fun, but that's part of building awareness for us," he says. "We want to show people that maybe their assumptions about disability are more limited than they think."

Kyle, who lives in Sacramento, Calif., adds that both of them have experience being on "the other side" when it comes to disability because he wasn't diagnosed until he was 17 and Sean was diagnosed at 25. That gives them a unique perspective, Kyle believes, because in many ways, they've had to confront their own misconceptions about disability, which gives them empathy toward listeners and others. That type of self-reflection can be useful, because it can give advocacy and awareness more depth.

"It's important for us to recognize that we may have some remnants of ableist views," he says. (For more on ableism, read "Advocating Against Ableism" at mda.org/quest.) "But seeing that helps us deepen the conversation in many ways. It's sometimes a view we have to work against to make sure we're approaching things in the right way."



Stephanie Bowers-Legg spreads disability awareness at work.

FIND YOUR VOICE



If you're just getting started with talking about disability or have struggled to address it in the past, the advocates here offer some advice:

Learn about your condition. For Stephanie Bowers-Legg, researching her condition has helped her frame how to talk to others. The more you learn, the better you can articulate your thoughts, she believes. Not only can that help when talking to people in your community, but it can also be useful when building awareness among the medical professionals you see.

Keep making connections. Because Stephanie has done so much research on her condition and is a strong advocate, her doctor sometimes asks her for advice on resources to share with other patients. She sees all her conversations as an opportunity to forge more connections, and those can lead to unexpected opportunities.

Protect your energy. Although there's a great deal of work to be done around disability awareness, you have to balance out your efforts with supporting your health, suggests Bettemarie Bond. That might mean limiting how much you do, but it's an important long-term strategy.

Try to stay positive. No matter what you do and how much awareness you create, there will always be rude people. That can be exhausting when you're also navigating health issues, says Stephanie, but part of protecting your energy is trying to be positive, even in the midst of negative reactions.



Bettemarie Bond speaks publicly about disabilities.

Expanding the scope

Everyday conversations at work or in the community often are opportunities to educate others, but many who advocate for more disability awareness also set up structured talks that can reach a much wider audience.

For example, Bettemarie Bond, who lives in Levittown, Pa., with mitochondrial myopathy (MM), gives presentations to groups like first-year medical students at the University of Pennsylvania School of Medicine, attendees at meetings for major medical manufacturing companies, legislators attending Rare Disease Week on Capitol Hill, and medical professionals attending seminars for continuing education.

"I try to make the most of every opportunity," she says. "My world was turned upside down when I started experiencing symptoms and was diagnosed as a teenager. I went from being a state champion in gymnastics to needing help to push my IV pole at home. All my dreams were gone. I lost my purpose."

Although these talks and even online interactions can leave her exhausted, Bettemarie is determined to maintain a high level of advocacy, because she feels that her efforts make a difference.

Not only is there greater awareness for individuals, but there's also a potentially huge ripple effect — for example, by speaking to med students, nurses, doctors, hospital administrators, legislators, patient organizations, and equipment manufacturers, she's driving deeper understanding of neuromuscular disease in a way that will be used for patient interactions, policy change, and innovation.

"My mom taught me to advocate for myself," Bettemarie says. "It was not easy; I avoided speaking up at first because I didn't want to hurt people's feelings by challenging their

EMPOWER AND EDUCATE

MDA supports our community in learning about neuromuscular disease and spreading awareness. Here are programs and services that can help.

Access Workshops

These educational workshops provide information and resources on access to care and services and overcoming barriers. Each online workshop includes videos and activities that help you build knowledge, health literacy, 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 educational materials in English and Spanish at mda.org/education.



Engage Events

These community education programs empower individuals and families with knowledge and resources around neuromuscular disease. Find upcoming events and view recorded programs at mda.org/engage.



Facebook Live Events

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

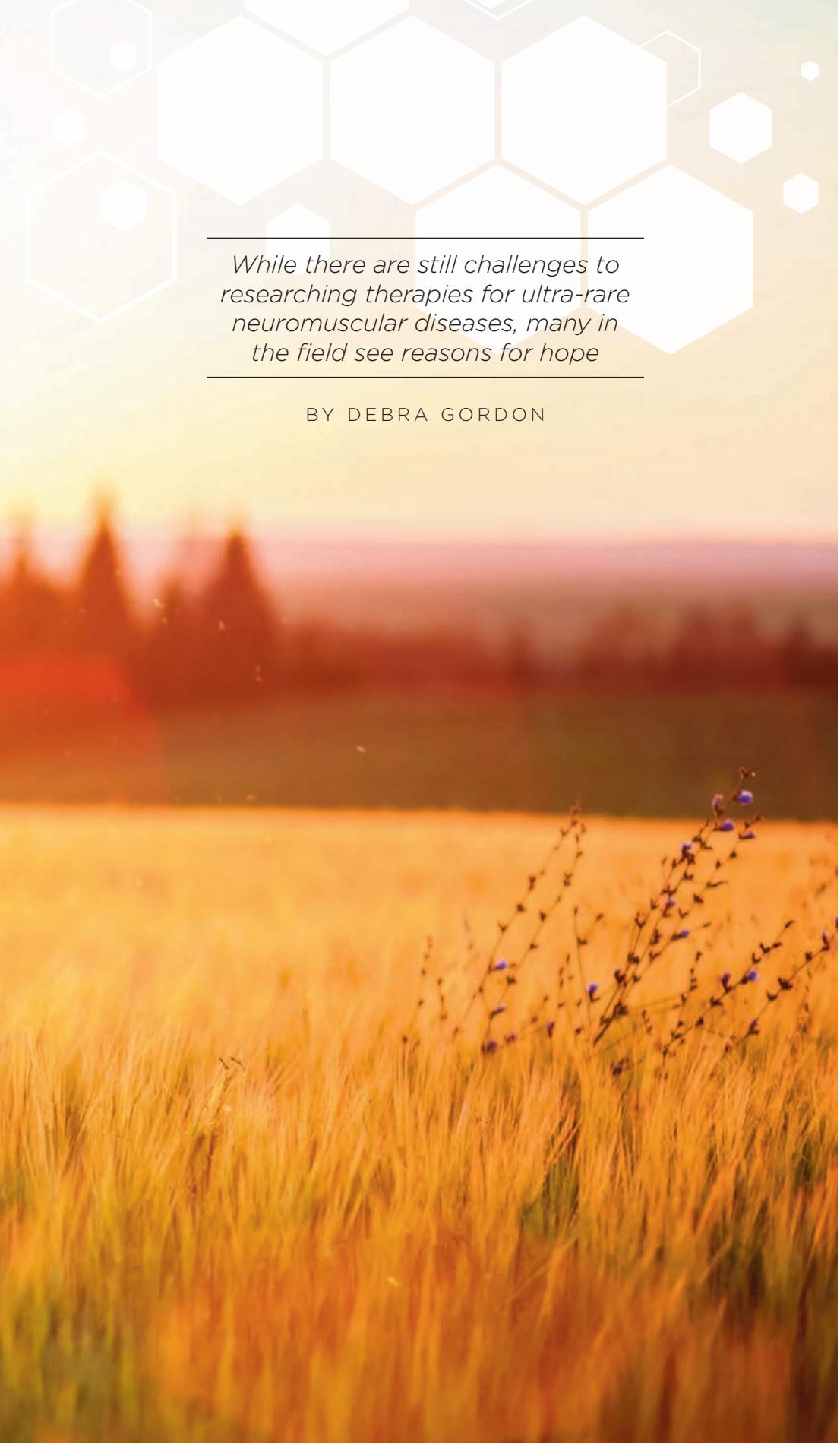


assumptions. But gradually I realized this was what they needed to hear because it helps other people with disabilities. It's important to create awareness." 

Elizabeth Millard is a freelance writer in northern Minnesota.

One in a Million





While there are still challenges to researching therapies for ultra-rare neuromuscular diseases, many in the field see reasons for hope

BY DEBRA GORDON

All the neuromuscular diseases MDA covers are considered rare, meaning they affect fewer than 1 in 20,000 individuals. But within each of those diseases, there are dozens of subtypes defined by the genetic mutations that cause them. Many, if not most, are considered ultra-rare, affecting fewer than 1 in 50,000.

For instance, Vici syndrome, which is marked by abnormal brain development as well as muscular weakness, has been diagnosed in just 100 children in the world. HACD1 congenital myopathy has been described in only 5 individuals.

“I’d say more than 80% of neuromuscular diseases fall into the ultra-rare category,” says Sharon Hesterlee, PhD, MDA’s chief research officer. “We say that MDA covers more than 43 neuromuscular diseases, but it’s really 43 categories encompassing hundreds of diseases if you start parsing them genetically.”

That makes it challenging to develop therapies. For instance, until recently, the US Food and Drug Administration (FDA) required investigational therapies for extremely rare diseases to meet the same complex requirements of any drug. Finding enough patients for a clinical trial can be difficult, particularly if they are scattered around the world. Even understanding

Genes and Neuromuscular Disease

Most neuromuscular diseases are genetic disorders. A genetic disorder is caused by changes in one or more genes. These changes are called gene variants, or mutations. Researchers have discovered mutations in more than 500 genes that can cause neuromuscular diseases, and the number is still growing. To learn more, check out MDA's educational resources, including the fact sheet "Genetics and Neuromuscular Disease," at mda.org/education.

the disease and how it progresses over time — which is critical for determining what "success" looks like in a clinical trial — can be elusive when so few people have the disease.

Practical considerations

MDA does not base its funding on the prevalence of a disease, nor does the National Institutes of Health (NIH). But large pharmaceutical companies often do. "A small number of patients doesn't mean it costs that much less to develop the drug," Dr. Hesterlee says.

On average, it costs more than \$100 million to bring an "orphan" drug to market. Orphan drugs are defined as those designed to treat diseases that affect fewer than 200,000 patients. Pharmaceutical companies are often reluctant to pursue such therapies because it is difficult to recoup the cost

of development. Thus, in 1983 Congress passed the Orphan Drug Act, which provides additional incentives to develop drugs for these diseases, including extended market exclusivity, tax credits, and federal research grants.

However, those incentives aren't always enough when it comes to ultra-rare diseases. "As things stand, it's challenging for companies to make the numbers work for diseases that affect very small numbers of people," Dr. Hesterlee says.

That's why MDA is committing to developing a pathway to get biotech companies interested in bringing novel therapies for these extremely rare diseases to market. "That means aligning the right incentives and having clear requirements in place," Dr. Hesterlee says. Potential options include tax credits, less onerous clinical trial requirements, and more public/private partnerships.

Promising progress

Families coping with ultra-rare diseases can feel frustrated as they see therapies developed for more prevalent neuromuscular diseases, like Duchenne muscular dystrophy (DMD) and spinal muscular atrophy (SMA). But there is a lot of positive news on the ultra-rare front.

Many smaller biotech companies are pursuing treatments for ultra-rare neuromuscular diseases. For instance, Leadiant Biosciences, together with the NIH, has a clinical trial underway to test a drug for the treatment of GNE myopathy, also known as Nonaka distal myopathy, an extremely rare progressive muscle disease caused by mutations in the *GNE* gene. Astellas Pharma Inc. and Dynacure are working on treatments for X-linked myotubular myopathy, and the first drug for primary hyperkalemic and hypokalemic periodic paralysis, developed by Taro Pharmaceutical Industries Ltd., was approved in 2015.

In addition, scientific advances often translate across diseases.

"The methods and platforms used to develop drugs for diseases like DMD and SMA have high promise to be applied to the super rare diseases," says Alan H. Beggs, PhD, who directs the Manton Center for Orphan Disease Research at Boston Children's Hospital (BCH).

Seng Cheng, PhD, senior vice president and chief scientific officer of the Rare Disease Research Unit at Pfizer, can point to several examples where this occurs. For instance, a gene therapy platform developed for DMD is being leveraged for other, rarer neuromuscular diseases, such as limb-girdle muscular dystrophies (LGMDs).

Another example lies in the development of animal models to test therapies for ultra-rare neuromuscular diseases. Pediatric neurologist Jim Dowling, MD, PhD, a senior scientist of genetics and genome biology at SickKids Hospital in Toronto, along with his team, pioneered the use of zebrafish





ONE FAMILY'S JOURNEY TO SUPPORTING RARE DISEASE RESEARCH

Austin Corman is like most toddlers: He loves to play and has strong opinions he's not afraid to voice. "He's joyful," says his mother, Hannah.

But Austin can't sit up, crawl, or pick up large toys, because he has LMNA-related congenital muscular dystrophy (L-CMD), an extremely rare neuromuscular disease found in less than 1 in 1 million people.

The condition affects the gastrointestinal, cardiovascular, and skeletal muscles, creating difficulties eating, breathing, and walking. Austin's doctors couldn't say how long he would live. But his parents weren't willing to leave it at that. "We wanted to see what we could accomplish if we put our minds to it," says Austin's father, Mark.

To begin, Mark read more than 1,000 journal articles about the gene responsible for Austin's condition, while Hannah started the L-CMD Research Foundation and a GoFundMe page to raise money for research. By early summer 2021, they had raised nearly \$1 million. They also connected with MDA and numerous parent-led advocacy groups for rare diseases, and they reached out directly to researchers. Now, they are not only funding research but helping researchers connect with resources and each other to further their research.

Among the researchers they support are Ignacio Pérez de Castro, PhD, at the Instituto de Salud Carlos III in Madrid, who is studying the LMNA gene; Miguel Sena-Esteves, PhD, at the University of Massachusetts Medical School, who is developing genetic therapies for rare diseases; Anne Bang, PhD, at the Sanford Burnham Prebys Medical Discovery Institute in California, who is screening FDA-approved drugs to see if any might be effective for Austin's disease; FarBiotech in Texas, which is partnering with Dr. Bang to identify additional drug candidates; the Canadian biotech company Modelis, which created animal models of the disease in worms and zebrafish; and Rarebase, a California biotech company that is looking at Austin's tissues to analyze protein expression.

"We're hopeful that we may find something that is helpful in the short-term, which could buy us some time, and in the long-term we hope that there will be gene therapy," Mark says.

"If we can extend Austin's life and give him the best opportunity to enjoy his life, that would be wonderful," Hannah says. "We know there are no guarantees, but we feel that we're here to try — if not for Austin, then for the kids who come after him."



Thanks to pioneering work by Jim Dowling, MD, PhD, and his team, **zebrafish** are used to understand disease mechanisms and identify therapies for numerous neuromuscular diseases.

as an animal model of disease to study X-linked myotubular myopathy. Today, zebrafish are used to understand disease mechanisms and identify therapies for numerous other neuromuscular diseases.

Indeed, talk to researchers of ultra-rare diseases and you hear optimism. “The barriers are really coming down,” Dr. Dowling says, thanks to support from organizations like MDA and the strong interest of families who are eager to enroll their children in clinical trials and provide strong advocacy for research. “It’s no coincidence that some of the diseases that are moving forward have very active advocacy groups,” he says. “Without that, nothing moves forward.”

Dr. Dowling also highlights the strong community of researchers dedicated

to these diseases. “There is this feeling that we’re in it together to advance the treatments and care for patients who are traditionally underserved.” In fact, he points to three ongoing clinical trials for X-linked myotubular myopathy now underway as a result of such collaboration.

Finally, Dr. Dowling is encouraged by growing support in the pharmaceutical industry, noting that two of the X-linked myotubular myopathy trials are industry-sponsored. This involvement is critical. “It’s hard to point to diseases with therapeutic advancements in which there hasn’t been industry involvement in some way,” Dr. Dowling said.

But pharmaceutical companies shouldn’t be the only funding partner. For instance, MDA, the NIH, the Myotubular Trust, the Canadian Institutes of Health Research, CuresWithinReach, and the Will-Cure and

Joshua Frase foundations have supported aspects of Dr. Dowling’s research related to the potential benefits of the breast cancer drug tamoxifen for myotubular myopathy. “It really does take a village to help get a drug into clinical trials,” he says.

Gene-targeted therapies

Another reason for optimism is that pinpointing the genetic variants responsible for diseases is far easier these days than it was even five years ago. In the “old” days, Dr. Beggs says, finding the genetic variant responsible for a disease was akin to looking for the proverbial needle in a haystack.

Enter whole genomic sequencing, which can quickly and accurately search the “haystack,” or strings of DNA sequences, to find the “needle”—the genetic variant. Without that information, gene-targeted therapies cannot be developed.

The NIH is working to promote more research into ultra-rare diseases with its Bespoke Gene Therapy Consortium, a public-private partnership devoted to fostering the development of gene therapies to treat very rare genetic diseases in populations considered too small for commercial development. Initiatives include developing a standard operational playbook for developing such therapies, with the ultimate goal of speeding these treatments to those who need them.

Today’s genetic therapies open a world of possibilities. These include antisense oligonucleotides (ASOs), which have multiple potential applications. Eteplirsen (Exondys 51), approved for DMD, and nusinersen (Spinraza), approved for SMA, are ASOs. These snippets of genetic material can direct mutated or backup genes to make the proper protein. Similar

compounds can be personalized based on the patient's individual genetic mutation.

That happened at BCH, where researchers developed an ASO to treat a 6-year-old girl with a rare form of Batten disease, in which toxic substances build up on the brain, causing a plethora of symptoms and greatly shortened life expectancy.

The girl was the only person in the world with her specific genetic mutation. Within a year, the BCH team had developed an ASO called milasen (the patient's name was Mila) and, with the help of industry advisors, commissioned it to be manufactured. Even more amazing was that the FDA, for the first time ever, allowed them to test a new drug created for a single patient. While the drug didn't reverse Mila's condition, it provided some benefits. Her swallowing and muscular function improved, and her seizures virtually stopped. She survived to age 10.

Mila's case shows the potential of using individualized therapies for these ultra-rare diseases. "It's one of the first examples of going all the way from the discovery of something new to the design and development of a new drug, assessing safety, testing it, and then actually using it," says Dr. Beggs.

Advances in ASOs spurred the creation of the nonprofit n-Lorem Foundation in 2020, dedicated to developing individualized ASOs for genetic diseases that affect fewer than 10 people in the world — even just one person. The foundation,

led by the founder of Ionis Pharmaceuticals, which co-developed Spinraza for SMA, is pursuing drugs for several neuromuscular diseases and plans to provide lifetime treatment at no cost, covering the expenses through donations and support from the pharmaceutical industry.

Such genetic treatments might only be able to treat a relatively small percentage of patients with neuromuscular disease, "but this is still a group of people who never would have had any hope of treatment previously," Dr. Beggs says. "Now, there is hope." 

READ MORE

Find more articles on rare diseases, drug development, and other research topics at mda.org/research.



Debra Gordon is a freelance medical writer based in Norfolk, Va. She has covered the intricacies of medicine and the US healthcare system for more than 30 years.

Enter **whole genomic sequencing**, which can quickly and accurately search the "haystack," or strings of DNA sequences, to find the "needle" — the genetic variant.



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In a clinical trial of 196 boys aged 5 to 15 with Duchenne muscular dystrophy, the effectiveness and safety of EMFLAZA was compared with placebo (sugar pills) and prednisone. *EMFLAZA improved muscle strength at 12 weeks compared with placebo and was numerically favored* in timed measures of patient function at 12 weeks* (time to stand from supine, time to climb 4 stairs, and time to walk or run 30 feet).

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

In a study to observe long-term effects of steroids, deflazacort was shown to delay disease progression by years



Loss of ability to stand from a lying-on-the-back position
—delayed by 2.1 years
when compared with prednisone



Loss of ambulation (ability to walk)
—delayed by 2.7 years
when compared with prednisone



Loss of hand-to-mouth function with retained hand function
—delayed by 2.7 years
when compared with prednisone

STUDY INFORMATION

This study examined the long-term effects of glucocorticoids on milestone-related disease progression across the lifespan and survival in patients with Duchenne muscular dystrophy. Comparisons between deflazacort and prednisone are not included in the approved Prescribing information for deflazacort, as prednisone is not an approved treatment for Duchenne muscular dystrophy.

Please see the full outline of study information after summary of information for Emflaza.



Emflaza®
(deflazacort)

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

Summary of Information for EMFLAZA®

What is EMFLAZA used for?

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

Who should not use EMFLAZA?

Do not use if you are allergic to deflazacort or any of the inactive ingredients in EMFLAZA.

What warnings should I know about EMFLAZA?

- **Hyperglycemia:** Corticosteroids can increase blood glucose, worsen pre-existing diabetes, predispose those on long-term treatment to diabetes mellitus, and may reduce the effect of anti-diabetic drugs. Monitor blood glucose at regular intervals. For patients with hyperglycemia, anti-diabetic treatment should be initiated or adjusted accordingly.
- **Increased Risk of Infection:** Tell your healthcare provider if you have had recent or ongoing infections or if you have recently received a vaccine or are scheduled for a vaccination. Seek medical advice at once should you develop fever or other signs of infection, as some infections can potentially be severe and fatal. Avoid exposure to chickenpox or measles, but if you are exposed, medical advice should be sought without delay.
- **Alterations in Cardiovascular/Kidney Function:** EMFLAZA can cause an increase in blood pressure, salt and water retention, or a decrease in your potassium and calcium levels. If this occurs, dietary salt restriction and potassium supplementation may be needed.
- **Behavioral and Mood Disturbances:** There is a potential for severe behavioral and mood changes with EMFLAZA and you should seek medical attention if psychiatric symptoms develop.
- **Effects on Bones:** There is a risk of osteoporosis or decrease in bone mineral density with prolonged use of EMFLAZA, which can potentially lead to vertebral and long bone fractures.
- **Effects on Growth and Development:** Long-term use of corticosteroids, including EMFLAZA may slow growth and development in children.
- **Ophthalmic Effects:** EMFLAZA may cause cataracts, ocular infections and glaucoma and you should be monitored if corticosteroid therapy is continued for more than 6 weeks.

• **Vaccination:** The administration of live or live attenuated vaccines is not recommended in patients on EMFLAZA. Live-attenuated or live vaccines can be administered at least 4 to 6 weeks prior to starting EMFLAZA.

• **Serious Skin Rashes:** Seek medical attention at the first sign of a rash.

What else should I know about EMFLAZA?

Do not stop taking EMFLAZA, or change the amount you are taking, without first checking with your healthcare provider, as there may be a need for gradual dose reduction to decrease the risk of adrenal insufficiency and steroid “withdrawal syndrome”. Acute adrenal insufficiency can occur if corticosteroids are withdrawn abruptly, and can be fatal. A steroid “withdrawal syndrome,” seemingly unrelated to adrenocortical insufficiency, may also occur following abrupt discontinuance of corticosteroids. For patients already taking corticosteroids during times of stress, the dosage may need to be increased.

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

What are the possible side effects of EMFLAZA?

The most common side effects include facial puffiness or Cushingoid appearance, weight increased, increased appetite, upper respiratory tract infection, cough, frequent daytime urination, unwanted hair growth, central obesity, and colds.

The risk information provided here is not comprehensive. To learn more, talk about EMFLAZA with your health care provider or pharmacist.

The FDA-approved product labeling can be found at EMFLAZA.com.

For medical information, product complaints, or to report an adverse event, please call **1-866-562-4620** or email at **usmedinfo@ptcbio.com**.

You may also report adverse events directly to FDA at **1-800-FDA-1088** or www.fda.gov/medwatch.

The following study support information is not included in the approved label for EMFLAZA® (deflazacort). Please talk to your healthcare professional if you have any questions.

MCDONALD 2018 STUDY SUMMARY (ABSTRACT)

Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study.

Background: Glucocorticoid treatment is recommended as a standard of care in Duchenne muscular dystrophy; however, few studies have assessed the long-term benefits of this treatment. This study examined the long-term effects of glucocorticoids on milestone-related disease progression across the lifespan and survival in patients with Duchenne muscular dystrophy.

Methods: For this prospective cohort study, male patients aged 2-28 years with Duchenne muscular dystrophy were enrolled at 20 centers in nine countries. Patients were followed up for 10 years. Comparisons were no glucocorticoid treatment or cumulative treatment duration of less than 1 month versus treatment of 1 year or longer with regard to progression of nine disease-related and clinically meaningful mobility and upper limb milestones. Kaplan-Meier analyses to compare glucocorticoid treatment groups for time to stand from supine of 5 s or longer and 10 s or longer, and loss of stand from supine, four-stair climb, ambulation, full overhead reach, hand-to-mouth function, and hand function. Risk of death was also assessed. This study is registered with [ClinicalTrials.gov](#), number NCT00468832.

Findings: 440 patients were enrolled during two recruitment periods (2006-09 and 2012-16). Time to all disease progression milestone events was significantly longer in patients treated with glucocorticoids for 1 year or longer than in patients treated for less than 1 month or never treated (log-rank p<0.0001). Glucocorticoid treatment for 1 year or longer was associated with increased median age at loss of mobility milestones by 2.1-4.4 years and upper limb milestones by 2.8-8.0 years compared with treatment for less than 1 month. Deflazacort was associated with increased median age at loss of three milestones by 2.1-2.7 years in comparison with prednisone or prednisolone (log-rank p<0.012). 45 patients died during the 10-year follow-up. 39 (87%) of these deaths were attributable to Duchenne-related causes in patients with known duration of glucocorticoids usage. 28 (9%) deaths occurred in 311 patients treated with glucocorticoids for 1 year or longer compared with 11 (19%) deaths in 58 patients with no history of glucocorticoid use (odds ratio 0.47, 95% CI 0.22-1.00; p=0.0501)

Interpretation: In patients with Duchenne muscular dystrophy, glucocorticoid treatment is associated with reduced risk of losing clinically meaningful mobility and upper limb disease progression milestones across the lifespan as well as reduced risk of death.

McDonald CM, Henricson EK, Abresch RT, et al; CINRG Investigators. Long-term effects of glucocorticoids on function, quality of life, and survival in patients with Duchenne muscular dystrophy: a prospective cohort study. *Lancet*. 2018;391(10119):451-461.



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Drink One for Dane Reaches \$10 million

In its most successful year yet, drive-through coffee chain Dutch Bros' raised \$1.9 million during its 15th annual Drink One for Dane event on May 14.

The total surpassed the goal of \$1.3 million, which was necessary to reach an all-time total of \$10 million.

"There aren't enough words to describe how grateful I am to the customers who came out to support Drink One for Dane," said Travis Boersma, who co-founded Dutch Bros with his brother Dane, who lived with amyotrophic lateral sclerosis (ALS). Dane passed away in 2009.

Dutch Bros. operates more than 450 locations in 11 states. Learn more at dutchbros.com.



Image: Dutch Bros.

Par for the Course



For years, members of the MDA community have hit the fairways to raise money at MDA golf tournaments. These events have supported families across the nation living with muscular dystrophy, amyotrophic lateral sclerosis (ALS), and related neuromuscular diseases.

"Golf is a game for everyone of all abilities," says world-ranked disabled golfer, Keegan Kilroy, who lives with limb-girdle muscular dystrophy. "The work of MDA, bringing communities together to fundraise on the golf course, is changing lives through research funding and advocacy. While we enjoy the challenges the game of golf presents, we are reminded of the critical need to increase funding for neuromuscular disease research."



Donald S. Wood, PhD, president and CEO of MDA and a golf enthusiast, added that funds raised from events like golf tournaments have led to 12 FDA-approved treatments for neuromuscular diseases in the past six years alone. "We know that more funding will lead to cures," he says.

The golfing tradition continues with two upcoming tournaments:

- **Sept. 25, 2021** — Mike Neufeldt Memorial Golf Outing at Western Lakes Golf Club, Pewaukee, Wis.
- **Sept. 29, 2021** — Boston Muscle Team Golf Outing, Pine Hills Golf Club, Plymouth, Mass.



Sign up or learn more at mda.org/get-involved/golf-events.



Tom Bell receives a check from the car club he belongs to, the Pharaohs of Music City

Flexing for a Cause



Brayden Lee

More than 2,000 car lovers and festival-goers gathered at the third annual Flex for a Cause fundraiser in Lebanon, Tenn., on June 19, raising more than \$15,000 for MDA. The festival was held in memory of Brayden Lee, who lived with Duchenne muscular dystrophy (DMD) and passed away in 2018.

"When Brayden was here, he wanted to help the other kids with DMD," says Tom Bell, the event's organizer, Brayden's dad, and a car



The car show is a highlight of the Flex for a Cause event

enthusiast. "Brayden laid the foundation by spending the last three years of his life participating in research studies, and we're building upon it."

Tom and his wife, Bekki, with the help of friends and family, organized the first Flex for a Cause in 2019. What started out as a car show that first year has grown into a full festival, with bands, a silent auction, and food vendors. Local sponsorships, car show registration fees, food vendor registration fees, and a portion of the food vendor sales benefit MDA. As a bonus this year, Tom and volunteers asked the crowd to empty their pocket change into a five-gallon bucket, raising an additional \$1,553.59 to help children go to MDA Summer Camp.

Since its first year, the event has raised more than \$30,000 for MDA. Tom, a truck driver, says he had never organized anything before the first Flex for a Cause. Now he can't wait to expand Flex with events like cornhole tournaments and a pocket change challenge with the international car club he belongs to.

"This has grown so much more beyond anything I imagined," he says. "I honestly feel that Brayden would approve of what Flex has done and where it's going. He lost his battle, but we'll continue fighting the war."

Images: Flex for a Cause Facebook page

Education Within Reach



To make planning for and going to college easier for students with neuromuscular disease, MDA launched a new online workshop

called Access to Education: Higher Education. This no-cost educational program, available on-demand, offers an overview of common considerations, tips for choosing a school, information on financial preparation, and guidance on finding

support if students encounter access barriers during their higher education experience. It also provides tools and resources to help students confidently advocate for accessibility and accommodations in housing, around campus, and in the classroom.

Jacob Smith, who lives with spinal muscular atrophy (SMA), is a landscape architecture major in his junior year at the University of Wisconsin-Madison. He says the workshop's content is tailored to students with neuromuscular disease, and he

especially likes the advice on finding a caregiver. "This is something that I had some trouble with, so informing others about how this works is very helpful," says Jacob, who credits MDA with providing the funds for research and treatments that have given him the strength and stamina to successfully attend college.

Find Access to Education: Higher Education and learn more about MDA's Access Workshops at mda.org/accessworkshops.

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MDA

Living Creatively

Releasing my first album took a lifetime of imagination and determination

BY DUSTIN CLARK



Dustin Clark

In my childhood I was a creative kid with an active imagination. I loved to play pretend with my friends. I attended art clubs and sang in the school choir. I daydreamed and made up stories in my head.

But as my imagination grew, my body deteriorated. I was diagnosed with Becker muscular dystrophy (BMD) at age 10. As I grew older, my muscles became progressively weaker. I couldn't run in gym class. I fell in the hallways at school and came home with cuts and bruises. After a particularly bad fall in which I busted my chin on the floor, my high school advised my parents to get me a wheelchair for my safety.

Creative spark

I didn't know how to cope with my disability, and my family didn't really know how to help me. As my condition worsened, I retreated inward. I felt like a burden to everyone. I stopped going out to play with friends and spent most of my time after school playing video games. I wasn't connected to my emotions, and I didn't know how to express how I felt to those around me.

But my creativity never went away. I grew up in a musical family, surrounded by voices and instruments. We all learned to play music and performed at church. Taking after my brother and my dad, I took an interest in writing songs. When I was young, my mom tried to teach me piano. I hated those lessons, but they inspired me to write my first song because I wanted to make my own music.

This creative spark came in handy for me as a disabled person. Disabled life is a highly creative endeavor, because it requires thinking outside the box to achieve your goals — from getting food out of the microwave to building a career.

Music became my outlet for this spirit of ingenuity. As a teen, I decided to pursue songwriting more seriously. My dad bought me an electric guitar and recording software, and I began to record some songs. When little else seemed to move me, music did. There was something about writing those songs that kept me pushing for new and exciting musical ideas.

That energy gave me the drive I needed to make a way for myself in the music world.

Pushing through barriers

I decided to release an album as a recording artist. I began writing songs for the album in 2011, but I didn't complete it until 2021. Needless to say, this project was quite a challenge. There were many barriers to my success along the way which required creative workarounds.

Regular guitars are too heavy for me to hold, so my dad and I, with the help of a friend who is an industrial designer, made a pulley and hoist to hang my guitars from the ceiling. I don't have the finger strength to use a keyboard, so I learned to use MIDI software to point-and-click my keyboard and drum parts one note at a time.

“

Disabled life is a highly creative endeavor, because it requires thinking outside the box to achieve your goals — from getting food out of the microwave to building a career.



Though these physical challenges required a great deal of time and patience, the mental barriers were really what held me back. When living with a disability, sometimes your body feels like your enemy — a high wall that stands between you and everything you want in life, big or small. Though efforts are being made to include disabled people in society, we always have to fight to carve out a life for ourselves in a world that really isn't built for us. It takes remarkable determination to finish what we start.

Many times, it takes a supportive friend or loved one to unlock that determination. I spent years procrastinating and agonizing over every little detail of the album, putting off the final steps needed to complete it. Then, one afternoon in 2020 my dad sat me down and challenged me to type out every step necessary to finish and release the album. He encouraged me to take action, one step at a time. That's

Dustin employs creative workarounds in his music studio, such as hanging guitars from the ceiling.

exactly what I needed to find my motivation. It didn't take much — just a little push, and the ball was rolling.

I hired an audio engineer to record bass and mix the album. Next, I planned a budget and ran a Kickstarter campaign to raise money. I even designed rewards, like CDs and t-shirts, for my Kickstarter pledgers. Finally, I organized a Zoom listening party and set up the album release on Spotify. It was one of the most productive years of my life, and all during a pandemic.

Finding success

If you are disabled, find out what you are passionate about. Pursue that passion with every ounce of drive you can muster. Use that creativity you've spent a lifetime practicing to achieve your goals. Your success may not look like someone else's, but it's going to be 100% yours.

To those who know someone with a disability, learn to honor that person's life and appreciate the grit it takes for them to exist alongside you. At the same time, don't be afraid to challenge us. It's up to us to find our own limits, but we need you to encourage us to reach new heights for ourselves. You'll be surprised what we can accomplish. **Q**

Dustin Clark, 27, is a graphic designer and musician in Columbus, Ohio, who releases music under the name Black Birch. His debut album, *Here.*, is available on Spotify and other streaming services. You can find him online at dustinclarkdesigns.squarespace.com and on Instagram at [@dustinclarkmusic](https://www.instagram.com/@dustinclarkmusic) and [@dustinclarkdesigns](https://www.instagram.com/@dustinclarkdesigns).

One Bike, Four Stories

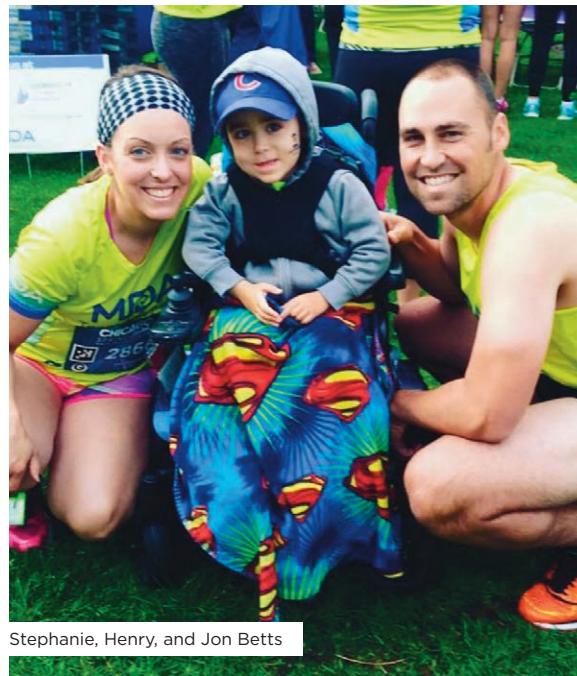
With the help of Team Momentum, a racing wheelchair connected and impacted multiple MDA families

Chris Benyo planned to push his wife, Denise, who lived with amyotrophic lateral sclerosis (ALS), over the finish line in her racing wheelchair, or duo bike, in the Bank of America Chicago Marathon in 2016. When she passed away before the race, he pushed the bike filled with mementos of her instead. He crossed the finish line holding Denise's photo. After the race, he donated the bike to MDA's Team Momentum.

Next, former MDA resource coordinator Hugo Trevino, who lives with spinal muscular atrophy (SMA), used the bike to tackle many races with his family, friends, and co-workers.

After Hugo's accomplishments, Team Momentum passed the bike to Amy Shinneman, who lives with Bethlem myopathy. She joined her husband, Jamie, for the Chicago Marathon in 2019. She then passed the bike on with the hope that it empowers others who can't participate in athletics in traditional ways. The Shinnemans now have their own duo bike and will join Team Momentum in the TCS New York City Marathon.

The bike is now with Stephanie and Jon Betts. Their son Henry, 8, lives with congenital muscular dystrophy (CMD), and they are eager to push him in the 9th Annual Henry's Hustle 5K benefiting MDA beginning Nov. 26. Taking place in person and virtually, anyone can join in the fun. To learn more, visit henryshustle.com. For more information on Team Momentum, visit mdateam.org.



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