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Evrysdi in action



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



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

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

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

What is Evrysdi?

Evrysdi is a prescription medicine used to treat spinal muscular atrophy (SMA) in adults and children 2 months of age and older.

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

Important Safety Information

- Before taking Evrysdi, tell your healthcare provider about all of your medical conditions, including if you:
 - are pregnant or plan to become pregnant. If you are pregnant, or are planning to become pregnant, ask your healthcare provider for advice before taking this medicine. Evrysdi may harm your unborn baby.
 - are a woman who can become pregnant:
 - Before you start your treatment with Evrysdi, your healthcare provider may test you for pregnancy. Because Evrysdi may harm your unborn baby, your healthcare provider will decide if taking Evrysdi is right for you during this time
 - Talk to your healthcare provider about birth control methods that may be right for you. Use birth control while on treatment and for at least 1 month after stopping Evrysdi
 - are an adult male planning to have children: Evrysdi may affect a man's ability to have children (fertility). If this is of concern to you, make sure to ask a healthcare provider for advice
 - are breastfeeding or plan to breastfeed. It is not known if Evrysdi passes into breast milk and may harm your baby. If you plan to breastfeed, discuss with your healthcare provider about the best way to feed your baby while on treatment with Evrysdi
- **Tell your healthcare provider about all the medicines you take**, including prescription and over-the-counter medicines, vitamins, and herbal supplements. Keep a list of them to show your healthcare provider and pharmacist when you get a new medicine

Genentech
A Member of the Roche Group

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

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

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

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

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

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

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

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

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

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

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

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

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

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

SMN stands for survival motor neuron.

Important Safety Information (continued)

- You should receive Evrysdi from the pharmacy as a liquid that can be given by mouth or through a feeding tube. The liquid solution is prepared by your pharmacist. If the medicine in the bottle is a powder, **do not use it**. Contact your pharmacist for a replacement
- Avoid getting Evrysdi on your skin or in your eyes. If Evrysdi gets on your skin, wash the area with soap and water. If Evrysdi gets in your eyes, rinse your eyes with water
- The most common side effects of Evrysdi include:
 - For later-onset SMA: fever, diarrhea, rash
 - For infantile-onset SMA: fever, diarrhea, rash, runny nose, sneezing, sore throat, and cough (upper respiratory infection), lung infection, constipation, vomiting

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

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

Please see accompanying brief summary for additional Important Safety Information.

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



Patient Information
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:

- are pregnant or plan to become pregnant. If you are pregnant, or are planning to become pregnant, ask your healthcare provider for advice before taking this medicine. EVRYSDI may harm your unborn baby.
- are a woman who can become pregnant:
 - Before you start your treatment with EVRYSDI, your healthcare provider may test you for pregnancy. Because EVRYSDI may harm your unborn baby, you and your healthcare provider will decide if taking EVRYSDI is right for you during this time.
 - Talk to your healthcare provider about birth control methods that may be right for you. Use birth control while on treatment and for at least 1 month after stopping EVRYSDI.
- are an adult male planning to have children: EVRYSDI may affect a man's ability to have children (fertility). If this is of concern to you, make sure to ask a healthcare provider for advice.
- are breastfeeding or plan to breastfeed. It is not known if EVRYSDI passes into breast milk and may harm your baby. If you plan to breastfeed, discuss with your healthcare provider about the best way to feed your baby while on treatment with EVRYSDI.

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

How should I take EVRYSDI?

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

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

Taking EVRYSDI

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

Reusable Oral Syringes

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

What are the possible side effects of EVRYSDI?

The most common side effects of EVRYSDI include:

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

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

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

How should I store EVRYSDI?

- Store EVRYSDI in the refrigerator between 36°F to 46°F (2°C to 8°C). Do not freeze.
- Keep EVRYSDI in an upright position in the original amber bottle to protect from light.
- Throw away (discard) any unused portion of EVRYSDI 64 days after it is mixed by the pharmacist (constitution). Please see the Discard After date written on the bottle label. (See the **Instructions for Use** that comes with EVRYSDI).

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

General information about the safe and effective use of EVRYSDI.

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

What are the ingredients in EVRYSDI?

Active ingredient: risdiplam

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

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EVRYSDI® (risdiplam)

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1 DNA Way
South San Francisco, CA
94080-4990

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



Muscular Dystrophy Association is the No. 1 voluntary health organization in the United States for people living with muscular dystrophy, ALS, and related neuromuscular diseases. For more than 70 years, MDA has led the way in accelerating research, advancing care, and advocating for the support of our families. MDA's mission is to empower the people we serve to live longer, more independent lives.

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PUBLICATION MANAGEMENT



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Strength in Community

The MDA team recently held our Annual Clinical and Scientific Conference in Nashville, Tennessee, which drew a community of more than 1,700 scientists and clinicians, in-person and virtually, from 15 countries. It is the biggest and most comprehensive conference of its kind in the world.

It's beyond exciting to hear about the rapid advances being made in genetic medicine. The fact is, at a time not too long ago, there was no hope for a cure for people living with neuromuscular disease. But MDA had the vision to push the boundaries of the medical frontier and open a new field of medicine: genetic medicine. And now, in the 21st century, MDA-supported scientists and clinicians are helping develop the first treatments for genetic diseases approved by the US Food and Drug Administration (FDA) — 15 of them in the last 12 years, and there are dozens more in the pipeline. That's why we call it the pipeline of promise.

At the conference, we had the opportunity to meet with 70 of our Care Center Directors. We're so proud that 13 new healthcare institutions have recently joined our Care Center Network, making it larger and stronger than ever. This network is what powers our MOVR (neuroMuscular Observational Research) data hub, which collects physician-input data on patients' diagnoses and progress. The quality of this data has attracted the attention of the FDA and leading pharmaceutical companies for its usefulness in getting patients into the right clinical trials. With the rapidly growing pipeline of treatments in development, the need to get patients into clinical trials has never been greater.

Speaking of clinical trials: In recognition of MDA's leading role in advancing genetic medicine, our Advocacy team has been invited to participate with the FDA in helping craft new approaches for moving clinical trials forward in genetic diseases.

While we were in Nashville, we also kicked off the Volunteer Tribute Reception, MDA's celebration of local volunteers who help support MDA all year long. (Read more about the tribute on page 38.) At MDA, families and volunteers are at the heart of our mission. We are excited to come together in communities around the country to thank you. Strength in unity. Strength in community.

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

Donald S. Wood, PhD, delivered a welcome address at the 2022 MDA Clinical and Scientific Conference.

Claiming Success

Insights on achieving your goals



Robert Pipia (right) and his wife, Maggie, married in 1997.

The Year of Independence is the perfect time to celebrate individuals in our MDA community who are shattering expectations and forging forward to achieve their goals.

The personal story of the Honorable Robert Pipia, a judge in the District Court of Nassau County, New York, who lives with an undiagnosed neuromuscular disease, proves the power of resilience and advocacy.

Whether gaining his independence as a child at MDA Summer Camp, leading his

high school wheelchair hockey team, or proving naysayers wrong early in his career, Robert has always been ready to take on his next challenge.

“If you want to accomplish something, you’ve got to advocate for yourself, and I think that’s the key: to see what level of advocacy is needed to accomplish your goal and then implement it,” he says.

Advocacy gains

As a young adult, Robert intended to become an accountant, but he quickly realized that the travel involved would make that career unrealistic for him. That fact, combined with the legal knowledge he gained advocating for himself and others in the community, led to his decision to study law.

He passed the bar in 1993. “This was only after I was forced to file a federal suit against the New York State Board of Law Examiners for Americans with Disabilities Act (ADA) violations when I was denied reasonable testing accommodations,” he says.

A more recent example of his self-advocacy was during the COVID-19 pandemic. Returning to the courtroom from remote work was not realistic for Robert, who uses a power wheelchair and has personal care attendants. “As an ADA accommodation, my court was specifically altered to allow for me to be in the courtroom virtually,” he says.

Personally thriving

Robert married his wife, Maggie, in 1997. He says his disability did not stop him from asking her to call him when they met in 1994. “She called me, and the rest was history,” he says. “People were not always supportive and asked us why we were doing this, and it was because we loved each other.”

He encourages others to take the same approach. “If you like a person and it’s appropriate, you should go for it.”

Words of wisdom

Currently a board member for Viscardi Center, Viscardi School, and MDA, Robert relishes his opportunities to make a positive impact.

“Just by being visible and telling my story, I think it helps other people with disabilities, and you help people who aren’t disabled see what’s going on,” he says.

His advice to others with disabilities who are pursuing a career? Work hard, plan everything out, and be realistic about what you’re going to face.

“When you fail, you have to get right back up and keep on going,” he says. “You can’t take the first failure as a sign that you shouldn’t be going forward. People with disabilities who are successful, their mainstay is resilience.” [Q](#)

+MORE ONLINE

Read the full Q&A with the Honorable Robert Pipia at mda.org/year-of-independence.

Fueling the Independent Spirit

I have always felt I was a living, breathing contradiction. Living with spinal muscular atrophy (SMA), I have physical limitations that cause me to be dependent on others, yet I was born with a fiercely independent spirit.



I've worked and fought for a life of independence and inclusion, despite what some along the way believed I could achieve. And I am proud to say that I have navigated roads that led to bachelor's and master's degrees, singing on television, working a 20-year career in high-tech, getting married, adopting a daughter from China (one of the least wheelchair-accessible countries on the planet, where I spent two weeks to complete the adoption), becoming a motivational speaker and a writer with my first book launching in June, and now serving all of you in my role here at MDA. Most days, my fiercely independent spirit is satisfied, but life is a never-ending journey to maintain my independence.

In developing this issue of *Quest*, I felt a responsibility to spotlight as many paths to independence as possible — adaptive vehicles, service dogs, accessible “smart” homes, MDA Summer Camp, and more — to empower this community to do more and to feed all of our independent spirits.

My dream is for the world to see possibilities instead of limitations. It begins with us. Where there is a will, there is a way. Truly.

Sincerely,
Mindy Henderson
 Director, *Quest* Editor-in-Chief
 Muscular Dystrophy Association

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Annie praises her service dog, Yardley.



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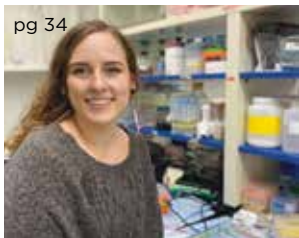
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Cover image: iStock.com/Andrey Suslov

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ONLINE

TAKE THE 2022 QUEST READER SURVEY

We'd like to know what you think of Quest magazine, the Quest blog (QuestBlog.org) and Quest podcast (mda.org/podcast). What topics would you like us to cover? Is there anything we can do better? Your feedback helps us shape the content we provide to you.

ENTER TO WIN

As a thank you for completing the survey, you can be entered in a drawing for a \$100 Visa gift card.

TAKE THE SURVEY

Go to SurveyMonkey.com/r/mdaQuest or use this QR code.



MDA-114

Biogen Supports 2022 Muscle Walks



Special thanks to Biogen for their sponsorship of 2022 Muscle Walks throughout the U.S. A highlight of this exciting partnership is onsite activation at the Quest Adaptive Lifestyle tent, which gives families and walk participants the chance to record “Man on the Street” sound bites for potential inclusion in an upcoming episode of the Quest Podcast, hosted by editor-in-chief Mindy Henderson.

GM Renews Commitment to STEM Connections



STEM Connections Program

We’re proud to announce that General Motors has renewed their support of our MDA STEM Connections Program with a \$300,000 grant. With GM’s support, MDA will provide hands-on activities at summer camp for kids to learn STEM concepts. The virtual mentoring program will also be returning in the fall to provide young adults an opportunity to explore career pathways with STEM professionals.

Shaw’s and Star Market Celebrate Shamrocks 40th



For over 28 years, dedicated associates and generous customers of Shaw’s and Star Market have been a valued partner of MDA, contributing over \$5 million to fund research, care, and advocacy. Thanks to all Shaw’s and Star Market employees, like those pictured here from the Lakewood store, for their support of the MDA mission and Shamrocks 40th birthday.

This campaign is managed by Albertsons Companies Foundation, doing business as Shaw’s and Star Market Foundation, a 501(C)(3) organization.

Sundt Construction Tees Off



For 20 years, Sundt Construction has hosted golf tournaments across the Western region to raise money for ALS research in honor of Mike Gaines, a member of the Sundt family who lost his life to ALS. To date, the golf tournaments have raised \$2 million for ALS research. MDA will be the beneficiary of this year’s golf tournaments, which are expected to raise \$100,000.



MDA Holds Tribute Celebration in the Great Volunteer State of Tennessee

In March, the MDA community came together from around the world in Nashville, TN, for the Annual Clinical & Scientific Conference. While there, we kicked off our Tribute Tour to thank partners who volunteer their time to support our families, including Dutch Bros (who just opened 7 new stores in the area), and Permobil, which is donating 20 seat elevators for power wheelchairs to MDA families whose insurance won’t cover them. The week capped off with a Tribute Reception at the Grand Ole Opry, bringing together families, volunteers, and celebrities like singer Wendy Moten of The Voice. Blake Shelton’s restaurant Ole Red gave the celebration an extra touch of local flavor!



MAKING A DIFFERENCE IN DUCHENNE MUSCULAR DYSTROPHY

**FEATURING:
JOHN BRANDSEMA, MD**

John Brandsema, MD is the neuromuscular section head in the division of neurology at Children's Hospital of Philadelphia (CHOP). He has a special interest in treating children with neuromuscular disorders and is enthusiastic about the promise that medical and scientific advances hold for people with these conditions.

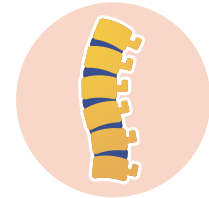
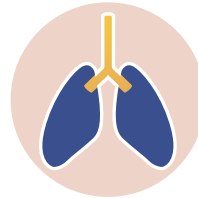
Corticosteroid treatment now and over time

Clinical studies have shown that starting boys with Duchenne muscular dystrophy (DMD) on corticosteroids soon after diagnosis can help delay, or slow, disease progression. Corticosteroids have helped boys who are still ambulatory extend ambulation and preserve muscle function.

Corticosteroid treatment can also be helpful to boys who have lost ambulation. According to care consideration guidelines, boys who are non-ambulatory should continue with corticosteroids.

Guidelines also suggest balancing the benefit of corticosteroids with proactive management of possible side effects, such as facial puffiness, high blood pressure, cataracts, abnormal behavior changes, and effects on growth and bone health.

**MULTIPLE STUDIES HAVE SHOWN
CORTICOSTEROIDS MAY HELP
PRESERVE LUNG FUNCTION AND
DELAY SCOLIOSIS**



Corticosteroid research in boys who are non-ambulatory is ongoing.

To learn more about the benefits and risks of corticosteroids, speak to your healthcare professional.

Please see the Brief Summary of Information for EMFLAZA® (deflazacort) on the following page.

“

In my decades of caring for boys and men with DMD, I've found that corticosteroid treatments, like deflazacort or prednisone, are the standard of care for the majority of patients.

”

INDICATION & IMPORTANT SAFETY INFORMATION FOR EMFLAZA® (deflazacort)

What is EMFLAZA® (deflazacort) used for?

EMFLAZA is a prescription medicine used to treat Duchenne muscular dystrophy (DMD) in patients 2 years of age and older.

When should I not take EMFLAZA?

Do not use if you have had hypersensitivity, including allergic reactions, to deflazacort or any of the inactive ingredients.

What warnings should I know about EMFLAZA?

- EMFLAZA can cause changes in endocrine function. 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.
- There is an increased risk of infection when taking EMFLAZA. Tell the healthcare provider if the patient has had recent or ongoing infections or if they have recently received a vaccine. Medical advice should be sought immediately if the patient develops fever or other signs of infection. Patients and/or caregivers should be made aware that some infections can potentially be severe and fatal. Warn patients who are on corticosteroids to avoid exposure to chickenpox or measles and to alert their healthcare provider immediately if they are exposed.
- EMFLAZA can cause an increase in blood pressure and water retention. If this occurs, dietary salt restriction and potassium supplementation may be needed.
- There is an increased risk of developing a hole in the stomach or intestines in patients with certain stomach or intestine disorders when taking corticosteroids like EMFLAZA.
- EMFLAZA can cause severe behavioral and mood changes. Seek medical attention from the health care provider if any behavioral or mood changes develop.
- There is a risk of osteoporosis with prolonged use of EMFLAZA, which can lead to vertebral and long bone fractures.

- EMFLAZA may cause cataracts or glaucoma and a health care provider should monitor for these conditions if corticosteroid therapy is continued for more than 6 weeks.
- Immunizations should be up-to-date according to immunization guidelines prior to starting therapy with EMFLAZA. Live-attenuated or live vaccines should be administered at least 4 to 6 weeks prior to starting EMFLAZA. Live-attenuated or live vaccines should not be used in patients taking EMFLAZA.
- EMFLAZA can cause serious skin rashes. Seek medical attention at the first sign of a rash.
- Rare instances of anaphylaxis have occurred in patients receiving corticosteroid therapy, including EMFLAZA.

What should I tell my health care provider?

Tell the health care provider about all medical conditions, including if the patient:

- is pregnant or planning to become pregnant. EMFLAZA® (deflazacort) can harm your unborn baby.
- is breastfeeding or planning to breastfeed. EMFLAZA may appear in breastmilk and could affect a nursing child.

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 side effects of EMFLAZA?

The most common side effects of EMFLAZA 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. These are not all of the possible side effects of EMFLAZA. Call your doctor for medical advice about side effects.

To report an adverse event, please call 1-866-562-4620 or email at usmedinfo@ptcbio.com. You may also report side effects to FDA at 1-800-FDA-1088 or at www.fda.gov/medwatch.

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PROGRESS NOW

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

Friedreich's ataxia (FA)

Gene Therapy Moves to Phase 1/2 Trial



Image: iStock.com/bernie_photo

Estimates show approximately 70% of individuals living with Friedreich's ataxia will develop FA-related cardiomyopathy.

The US Food and Drug Administration (FDA) cleared LEXEO Therapeutics' Investigational New Drug (IND) application for LX2006, an experimental gene therapy treatment for FA-related cardiomyopathy. Cardiomyopathy is a heart disease that makes it hard for the heart to deliver blood to the rest of the body. LEXEO plans to initiate an open-label phase 1/2 clinical trial of LX2006 in

individuals with FA-related cardiomyopathy in mid-2022.

Administered via intravenous (into the vein) infusion, LX2006 is designed to target the cardiac abnormalities in FA by delivering a functional frataxin gene (the gene carrying mutations in FA). This could promote the expression of the frataxin protein and restore function to mitochondria — tiny cellular “factories” that produce energy. The gene therapy is delivered into the body's cells by adeno-associated virus (AAV) vectors (delivery vehicles) that carry the frataxin gene to target cells.

In preclinical studies, treatment with LX2006 reversed cardiac abnormalities

in FA disease models and was associated with improvement in cardiac function and survival, while maintaining a favorable safety profile.

The phase 1/2 study is a 52-week, dose-ascending (amounts slightly increase with each new cohort), open-label trial of LX2006 in individuals who have FA-related cardiomyopathy. LX2006 will be administered as a one-time

intravenous infusion to trial participants in two groups, each of which contains five participants.

The FDA has granted rare pediatric disease and orphan drug designations to LX2006 for the treatment of FA.

For more information, visit lexeotx.com.



WHAT ARE AAV VECTORS?

Adeno-associated virus (AAV) vectors are a common method of delivering gene therapy. Here's how it works: Researchers turn the virus into a tiny delivery vehicle — or vector — by removing all the viral DNA that would make a person sick and engineering it to deliver a therapeutic DNA sequence to target cells. To learn more about viral vectors, go to mda.org/quest and search for the article “Express Delivery.”

Limb-girdle muscular dystrophy (LGMD)

First Human Trial of ATA-100

ATA-100, Atamyo Therapeutics' experimental gene therapy for the treatment of limb-girdle muscular dystrophy Type 2I/R9 (LGMD2I/R9) has reached multiple regulatory and financial milestones. LGMD2I/R9 is caused by mutations in the *FKRP* gene, which carries the genetic code used to produce fukutin-related protein. An authorization of a Clinical Trial Application (CTA) in Europe — the second for ATA-100 — has been granted by the

Denmark Medicines Agency. This is in addition to a CTA approval granted by the United Kingdom Medicines & Healthcare Products Regulatory Agency. Atamyo also was awarded 2 million euros in funding by the French government to support initiation of the first human clinical trial of the therapy.

ATA-100 is a single-administration gene therapy candidate designed to deliver a healthy *FKRP* gene, packaged in a modified adeno-associated

virus (AAV) vector, which delivers it into the body's cells. The cells are then able to use the genetic code carried by the *FKRP* gene to manufacture functional *FKRP* protein.

In preclinical studies involving mice with an LGMD-like disease, ATA-100 was well-tolerated and demonstrated the potential to reduce symptoms and biological markers of disease.

ATA-100 was previously granted orphan drug designation by the US Food and

Drug Administration (FDA) for the treatment of LGMD-R9, and by the European Medicines Agency (EMA) for the treatment of LGMD. Orphan drug designations by the FDA and the EMA provide for seven and 10 years of market exclusivity in the US and Europe, respectively, and confer other benefits such as tax credits, protocol assistance, and research grants.

For more information, visit atamyo.com.



Myotonic dystrophy (DM)

MARINA Trial Enrolling



Researchers are seeking adults with myotonic dystrophy type 1 (DM1) to participate in the phase 1/2 MARINA clinical trial to test AOC 1001 for the

treatment of DM. The therapy is named for the antibody oligonucleotide conjugate (AOC) used in the formula.

Developed by Avidity Biosciences, AOC 1001 is a type of ribonucleic acid (RNA) therapy designed to address the root cause of DM1 by reducing levels of a disease-related messenger RNA (mRNA) called *DMPK*. (RNA is a single-stranded

molecule that carries genetic code from DNA and is vital to the protein-manufacturing process.) In preclinical studies, AOC 1001 successfully targeted muscle cells, resulting in dose-dependent reductions of *DMPK* RNA across a broad range of muscles, including skeletal, cardiac (heart), and smooth muscles.

MARINA's primary objective is to evaluate the safety and tolerability of single and multiple ascending doses of AOC 1001 administered intravenously (into the vein). Researchers will begin to assess the activity of AOC 1001 across key DM1 biomarkers and explore clinical activity including measures of mobility and muscle strength, as well as patient-reported outcomes and quality of life measures.

To be eligible to participate, individuals must be 18 to 65 years old, have a genetic diagnosis of DM1 with a CTG repeat length greater than or equal to 100 (CTG is the DNA building blocks cytosine, thymine, and guanine), have clinician-assessed signs of DM1, be able to walk independently for at least 10 meters at screening, and meet additional criteria.

For more information on this study, visit ClinicalTrials.gov and enter NCT05027269 in the "Other terms" search box.

+ JOIN A CLINICAL TRIAL

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

Understanding Amyotrophic Lateral Sclerosis

A Q&A with Stanley Appel, MD

Amyotrophic lateral sclerosis (ALS) is a degenerative motor neuron disease typically diagnosed in adults in middle age or later. About 18,000 people in the United States are currently living with ALS, making it one of the more common neuromuscular diseases, and yet there is still a lot for scientists to learn about why it occurs and how to treat it.

ALS is about 20% more common in men. Between 80% and 90% of ALS is sporadic, meaning there is no apparent cause, while 10% to 20% of cases involve a family history of the disease. Two-thirds of familial cases can be traced to an underlying genetic cause.

We talked with Stanley Appel, MD, who co-directs the Houston Methodist Neurological Institute in Texas, about how our understanding of ALS is changing.

What do we know about the genetic underpinnings of ALS?

There may be as many as 40 genes that could be dictating the disease, but most cases of the disease are related to just a few. The major one is *C9orf72*, which accounts for about 40% of familial cases, or 2% to 3% of all ALS diagnoses. In our clinic, we also find that almost 10% of patients with no family history of the disease have mutations in this gene.



ALS is about 20% more common in men.

Image: iStock.com/GaryRadler

So, if you have mutations in this gene, you're going to get ALS?

It's not 100% certain that you will develop symptomatic ALS. While we know there are genes that predispose you to the disease, we also know there are genes that enhance resistance. So, if you're carrying resistant genes, that may prevent you from getting it, even if you're carrying the mutation.

Is there a way to shut off a disease-causing gene?

The answer is yes. It's called antisense oligonucleotides (ASOs) and one, nusinersen (Spinraza), is FDA-approved for patients with spinal muscular atrophy (SMA) after studies demonstrated significant improvements in children receiving the treatment. There are two studies ongoing using ASOs for hereditary forms of ALS. These are not easy therapeutic trials because there's a lot we keep learning as we try different ASOs. But there is a lot of promise and a lot of hope.



“

While we know there are genes that predispose you to the disease, we also know there are genes that enhance resistance.

— Stanley Appel, MD

WHERE ARE THE TREATMENTS FOR RARER DISEASES?

All neuromuscular diseases are considered rare, and it can be frustrating to hear of research and treatment advances in some diseases without progress in a disease that affects you or your family. But keep in mind that 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, director of the Manton Center for Orphan Disease Research at Boston Children’s Hospital. To learn more about the state of research into therapies for ultra-rare neuromuscular diseases, search for the article “One in a Million” at mda.org/quest.

The progress we’ve made in not just understanding the genes that cause ALS, but in translating that understanding to what the genes do and how we can potentially correct them is astounding. It’s all based on the phenomenal success we’ve had with understanding SMA and the treatments we now have for it. This approach is really changing how we manage neuromuscular diseases.

What are the most promising areas of study?

Most promising is the Healey Platform Trial, which is an exciting way to test multiple drugs against placebo rather than one at a time. Patients can switch to a different drug after six months following a washout period and being

rescreened for the master protocol. It’s an exciting way to conduct a study.

We are also looking at how to suppress inflammation, which seems to be a driving force for disease progression. MDA, as well as the ALS Association and ALS Finding a Cure, are funding some of these studies.

We encourage all our patients to participate in a study because it gives hope. Hope is the most important thing that we can provide our patients. And the hope is being realized. [@](#)

+MORE ONLINE

Find an expanded version of this article and read more Q&As about rare diseases at mda.org/quest/spotlight.

ADVERTISEMENT

Sorry we missed you at our **Biogen-sponsored Industry Forum** on SPINRAZA® (nusinersen)! However, we are excited to share a recording of the event with you. It will be available to watch at your convenience through March 2023.*

*For MDA 2022 conference registrants only.

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Connie Wolford, MSN, FNP-BC
Stanford Neuroscience
Health Center
Stanford, CA



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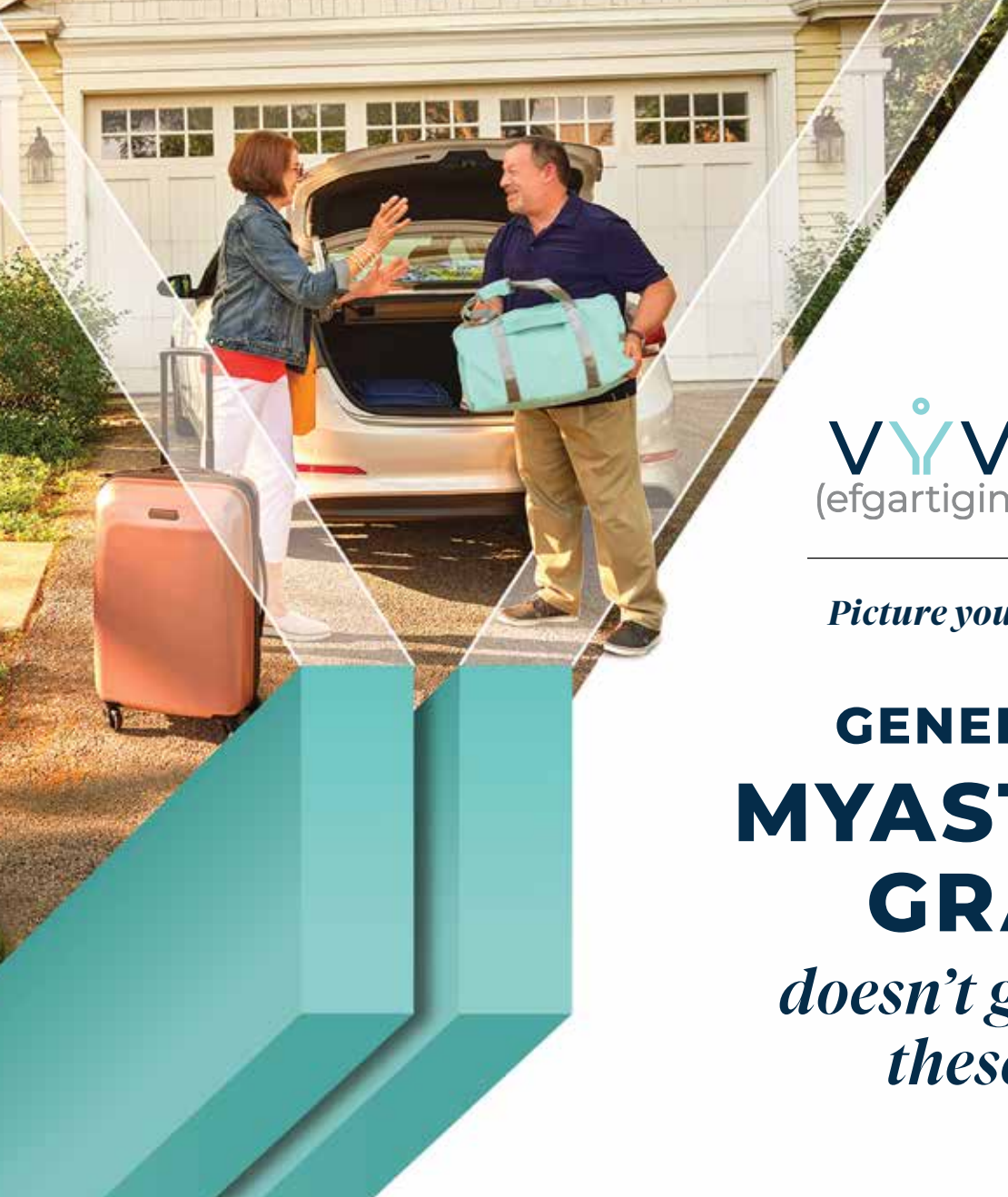
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VYVGART™
(efgartigimod alfa-fcab)

Picture your life in motion

GENERALIZED MYASTHENIA GRAVIS

*doesn't get to make
these plans*

What is VYVGART™ (efgartigimod alfa-fcab)?

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

IMPORTANT SAFETY INFORMATION

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

VYVGART may cause serious side effects, including:

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

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

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

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

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

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

Improved daily abilities

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

Reduced muscle weakness

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

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

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

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

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

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

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

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

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

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

What are the common side effects of VYVGART?

The most common side effects of VYVGART are respiratory tract

infection, headache, and urinary tract infection.

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

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

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

argenx 

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US-EFG-21-00240 V1 02/2022

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



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Funding Accessible Vehicles

Passing on lessons I learned from buying an accessible van

BY JOANNA BUONICONTI

When my mom and I had to purchase a new accessible van about two years ago, we were not in a comfortable financial situation to do so. I was going into my junior year of college, and we planned to hold off until I graduated.

Unfortunately, that plan changed when our van died for the final time a couple of days before my 20th birthday. We ended up purchasing a used Dodge Caravan that kneels to reduce the steepness of the ramp and has tie-downs to secure my power wheelchair. My mom took out a loan to help pay for it. Fortunately, the mobility dealer supplied us with a list of grants that we might be eligible for.

While the grants on the dealer's list didn't work out for us, they did point us in a direction we would not have known to look. And we ultimately learned a lot about funding accessible vehicles over the course of our journey to get back on solid financial footing with our new van.

Finding funding support

Everyone in the disability community has had to think out of the box to solve a problem. This is a helpful skill when it comes to finding vehicle funding options, which vary by state and location.

A mobility dealer often is a great place to start. It's in their interest to make an accessible vehicle affordable for you, so they generally are happy to share information about options that will cover at least some of the cost, which can include the US Department of Veterans Affairs (VA), Community Living Assistance Services and Supports (CLASS), state vocational rehabilitation services, Medicaid, and manufacturer rebates. They may also know of local organizations that provide grants.

Consider these options in your research

Health insurance: Check with your insurance provider to find out if your plan covers any adaptive equipment you need on your vehicle. Medicaid Home and Community Based Services (HCBS) waivers can help state Medicaid recipients



Author Joanna Buoniconti found funding for the accessible van she and her mom purchased.

receive funding for assistive technology, which may include vehicle modifications in some states.

State programs: Your state's vocational rehabilitation department may assist with costs if the vehicle helps you pursue educational goals or employment. Also, look for an Assistive Technology Act Program in your state at ATAPorg.org.

A mobility dealer often is a great place to start. It's in their interest to make an accessible vehicle affordable for you.



The Wilcox family found a van that accommodates their family of five and a power wheelchair.

“

I was grateful I had connections to help me find those resources, because I doubt I would have known where to look otherwise.

— Kory Wilcox

Rebates: Many vehicle manufacturers offer rebates on new vehicles when you make modifications for accessibility. Look for available rebates at [nmeda.org/consumer-resources/funding-2/rebates](https://www.nmeda.org/consumer-resources/funding-2/rebates).

Tax breaks: Some states waive sales tax on the purchase of adaptive equipment if you have a doctor's prescription. Also, the cost of your adaptive equipment may be tax deductible.

Loans: Ask your bank if they offer loans specifically for individuals with disabilities. Ask mobility dealers and conversion companies if they offer financing.

Grants: Local nonprofits that advocate for individuals with disabilities may have programs to help pay for adaptive equipment. The Mobility Resource has a list of grants and resources searchable by state at [TheMobilityResource.com/financing-handicap-accessible-vehicles/state-grants](https://www.TheMobilityResource.com/financing-handicap-accessible-vehicles/state-grants).

Community resources

The disability community itself is a valuable source for finding resources in your community.

Kory Wilcox and his wife, Elizabeth, realized they needed an accessible van when they were planning a vacation to Disney World with their family of five, including 3-year-old Asher, who has spinal muscular atrophy (SMA) and uses a power wheelchair.

They decided to buy a used RAM ProMaster and have it converted into an accessible van, because that seemed like the most affordable option. In addition to installing an in-floor ramp that extends at the push of a button, they removed the middle two seats, added tie-downs, and put down a rubberized floor to make it easier for Asher to maneuver his wheelchair in the vehicle.

“I was not prepared for when the dealership gave us the first quote for the wheelchair conversion and it was half the cost of the vehicle,” Kory says. “I never guessed it would have changed the cost profile that much.”

By doing research and talking to other parents of kids with disabilities, Kory learned about programs that would help cover the cost of the van. The Wilcox family received two grants from local charities, and Asher's Medicaid waiver program, the Missouri Children with Developmental Disabilities waiver, funded the remainder.

“I was grateful I had connections to help me find those resources, because I doubt I would have known where to look otherwise,” Kory says.

My mother and I also benefited from our community connections. A friend of mine told me about the Catastrophic Illness in Children's Relief Fund, which provides financial relief for families with large medical-related expenses in Massachusetts, where I live. A year and a half after my mom and I purchased our van, we were reimbursed for the amount left on the loan. This was such a huge weight off of both my mom's and my shoulders. Without the large loan looming above us, I could fully enjoy the vehicle and the independence it offered. [@](#)

Joanna Buoniconti is a freelance writer living with SMA in Western Massachusetts.

Do You Have Becker Muscular Dystrophy?

Are you interested in participating
in a clinical trial?

Edgewise Therapeutics is seeking adolescents and adults with **Becker Muscular Dystrophy** for a Phase 2 Trial of EDG-5506 to evaluate safety, tolerability, biomarkers of muscle damage and change in functional measures of an **investigational treatment for BMD**.

Trial anticipated to begin the first half of 2022 at multiple sites in the US, UK and the Netherlands.

To be eligible, participants must be:

- Male, aged 12 to 50 years with genetic confirmation of, and phenotype consistent with, BMD
- Able to complete functional measure testing such as the NSAA and 100-meter timed test
- Willing and able to travel to one of multiple study sites
- Willing and able to have an MRI

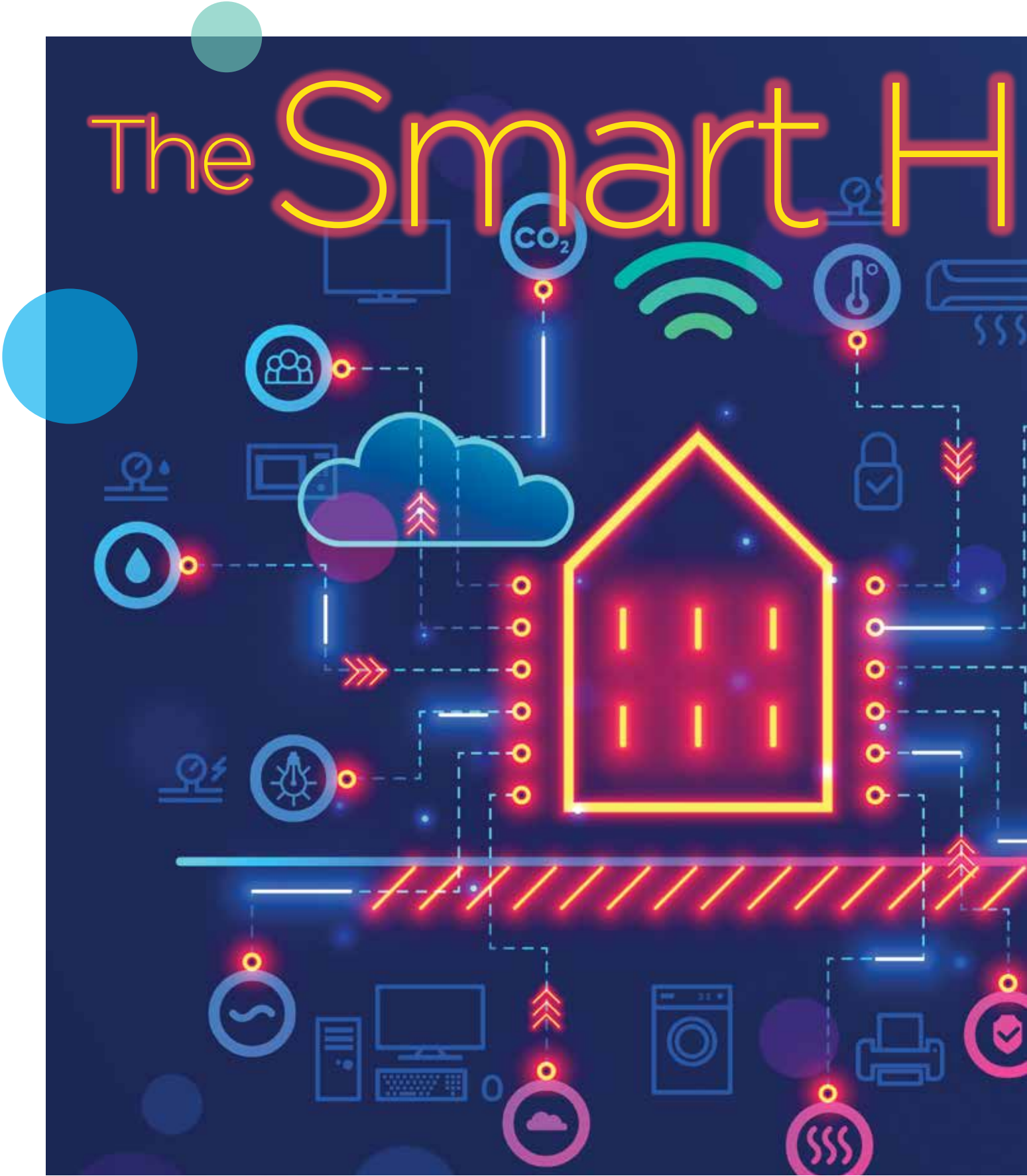
Travel expenses will be paid for by the study for eligible participants.



Taking a New Approach to Protect Muscles

To learn more, please go to www.clinicaltrials.gov, enter **NCT05291091** or contact studies@edgewisetx.com

The Smart H



Home's New Story

As home automation goes mainstream, costs come down and customization is key

BY SHAILA WUNDERLICH

You've probably heard of the "smart home." For close to two decades, it has been a regularly covered topic across all sectors of media. Each year, a story in a magazine, on prime-time news, or in an industry outlook report shows a futuristic take on a home that is automated to the point that it appears to read its occupant's mind. But what began as "the home of tomorrow" has become increasingly practical as new products and technology are rolled out on the market.

Today, the smart home is a reality. Its story, meanwhile, is less about the latest products and capabilities and more about the ways users are accessing those products and channeling those capabilities in their homes. At the forefront of this grand experiment is the population that stands to benefit from it the most: homeowners with disabilities.



Smart home hubs can corral devices and actions, such as a “Good night” button that turns off lights and locks doors.

At the fingertips

Every evening when Paul Robertson goes to bed, he touches a “Good night” button on his smartphone, triggering all the lights to turn off and the doors to lock in his Ocean City, Maryland, home. Automated lights and doors are not new to the disability community. What is relatively new is the ability to corral and control them (and oodles of other home functionalities) through a single, streamlined hub.

“My lighting is on a Lutron app. My thermostat is on a Honeywell app. My door locks are on an app called Vera,” says Paul, who lives with limb-girdle muscular dystrophy (LGMD). “But rather than having to open each app separately to control their individual functions, I have them rolled under one home-automation system that accesses and controls them all.”

Paul uses a system called ELAN, but other examples include Amazon Echo, Aeotec Smart Home Hub, and Apple HomePod. Most act as both the hub for individual apps as well as the interface through which homeowners control them. Each offers their own menu of features and app-compatibilities.

The “Good night” feature Paul relies on to close out his days is a prime example of home-automation’s other new frontier: customization. Through capabilities known as “routines,” “schedules,” “environments,” or “scenes,” homeowners can orchestrate multiple functionalities around specific daily tasks and events — whether it be the start and end of each day, arrivals and departures, caregiver visits, or mealtimes. These custom settings can be launched on command (via touchscreen, voice commands, or motion-sensor triggers) or on a schedule (via timers).

Paul marvels at the time and effort he saves from not having to walk from light to light, door to door, every time he goes to bed or leaves the house. “These kinds of things aren’t a big deal to people who aren’t mobility-challenged, but they’d take me a half-hour or more,” he says.

Favorite features

As Paul attests, if there’s one smart-home feature that appears to be gaining the most traction with the disability community,



ELAN’s home-automation system can tie together lighting, security, media, climate control devices, and more.



“Rather than having to open each app separately to control their individual functions, I have them rolled under one home-automation system that accesses and controls them all.”

—Paul Robertson



The Amazon Echo is compatible with Alexa Guard, which has added security features.



The Alexa app has touch-screen control for lighting.

it's lighting. Before Jennifer Baker, 34, had smart lighting installed in her family's Broken Arrow, Oklahoma, house, lights-out time was dictated by her mother, Patty. "What if I wasn't ready for bed at 8 p.m.?" says Jennifer, who lives with Ullrich congenital muscular dystrophy (CMD). Thanks to

Roku and Amazon Echo dashboards on her smartphone and watch, Jennifer now controls her own lights, TV — and evenings. "It makes a big difference in my sense of independence," she says.

Meanwhile, Patty favors the security provided by her daughter's tech. "She's dropped her phone several times and couldn't get to it from her wheelchair," Patty says. "Her nurse and I weren't able to reach her, and we panicked."



Smart-home capabilities make Jennifer Baker feel safer and more independent.

An Alexa voice-control plug-in on Jennifer's smart watch now permits her to call for help when she drops her phone. "It definitely makes me feel easier about leaving the house," Patty says.

As Jennifer's disease progresses, she can add features with higher levels of automation, like Alexa Guard.

Introduced in 2019, this

Echo-compatible system triggers lights, alarms, and emergency calls when it detects smoke, flooding, or the sound of doors and breaking glass.

Alexa's voice control capabilities can also be used to open and close doors in your home. In 2021, Open Sesame introduced a voice interface accessory to their line of automated door operating systems. This product allows homeowners to connect their Open Sesame door operators (both new and old models) to Wi-Fi and Alexa.

Amber Ward, OTR/L, occupational therapy coordinator at Neurosciences Institute-Neurology, Atrium Health, in Charlotte, North Carolina, notes that power wheelchairs are

Smart Spending

Depending on the specific products you choose and the method of installation (DIY or professional), costs for the average smart-home setup range from \$1,500 to \$7,000. Financial assistance options exist, but they require a fair amount of research and must be vetted before you make any purchases. Begin with the resources here, in the order listed.

- 1 Private insurance and Medicaid.** Most private insurance does not cover assistive technology, but check your specific plan to be sure. Medicaid offers some waiver or voucher options that vary by state, and both Medicaid and private insurance require prescriptions or medically authorized documentation of necessity. Look to your MDA care team for help collecting and submitting that paperwork.
- 2 Federal- and state-funded assistance.** "Every state has state or federally funded assistive technology programs that may offer anything from loaner equipment to help with installation," says Amber Ward, OTR/L, occupational therapy coordinator at Neurosciences Institute-Neurology, Atrium Health, in Charlotte, North Carolina. Visit the Association of Assistive Technology Act Programs (ATAPorg.org) to find the programs in your state.
- 3 Local vocational and service organizations.** These include grants from state-based independent living/vocational rehabilitation organizations (see ATAP site, above) and scholarships from local service clubs, such as Elks and Lyons. Check your local chapters for specific opportunities.
- 4 Private charities and causes.** "These are often local and diagnosis-specific," says Amber, who points to the Duchenne muscular dystrophy (DMD) group Team Joseph (TeamJoseph.org) as a prime example. Social media groups formed around your neuromuscular disease can be a great place to identify similar resources.



A smart light switch, such as the GE Cync, makes traditional bulbs smart.

Ring offers easy-to-set-up smart home security kits.



In 2021, Lowe’s launched “Livable Home,” an initiative to provide affordable, accessible smart-home solutions as well as expertise and installation services.

even beginning to get in on the act. Some of the latest models are equipped with infrared technology that can be used to control devices, like speakers, televisions, and tablets. “They act like universal remotes that you can operate with your hand, head, or lip, as long as you’re within line of sight from the device you’re trying to control,” she says.

Affordable access

“When I started in this industry, having voice control on a house’s lighting system could cost upwards of tens of thousands of dollars and take weeks or months to get,” says Matthew Colvin, rehabilitation technician for ImproveAbility, an Austin, Texas-based consulting company that provides assessments, recommendations, and installation services for people seeking assistive technology for their homes. “Now, it’s closer to several hundred dollars, and you could find it on the shelf at the local Home Depot.”

Indeed, home-remodeling reviewer Fixr.com estimates the sum for automating an entire home is less than Matthew’s old estimate for lighting alone — around \$5,500. The existence of Matthew’s company itself is another sign of changing times. With each passing day, it becomes easier to find professional help in choosing, installing, and even buying assistive technology for the home. In 2021, Lowe’s launched “Livable Home,” an initiative to provide affordable, accessible smart-home solutions as well as expertise and installation services.

The basic requirements for a fully automated home start with home products that can connect and interact with a user and other smart devices. These days, the list of options is long and includes light bulbs, switches, lamps, door openers, doorbells, cameras, speakers, thermostats, faucets, washers/dryers, and refrigerators. In fact, a smart plug can turn a simple device that plugs into the wall — such as a space heater, humidifier, or Christmas tree lights — into a connected device that you can control from your phone.

The next requirement is a system and interface (ELAN, Amazon Echo, Apple HomePod, etc.) that allows you to corral and control those smart products. Interfaces may be controlled by touchscreen, voice commands, or motion sensor triggers, according to a homeowner’s preference and abilities.

While outfitting an entire home at once (as Paul did) is certainly convenient, Matthew recommends homeowners get their feet wet with one or two features at first. “Do it a step at a time, get comfortable, figure out what works,” he says.

As Matthew gets to know his customers and their conditions, he can prepare their

homes for future capabilities. “We may start with a tablet you operate with your hand, but when they get to the point where they can no longer touch it, I could already have the hardware installed that allows eye control,” he says. “All they have to do is call me and I’ll launch it for them.”

And that proves the home of tomorrow truly is a reality today. [🔗](#)

Shailla Wunderlich is a St. Louis-based writer with more than 20 years’ experience in the publishing industry.



Did You Know?

Here are a few of the ways MDA makes an impact in our community.

- Funded **15** FDA-approved treatments in just seven years, with four new ones in 2021 alone.
- Awarded **\$8.5 million+** for new and continuing research projects in 2021.
- Provides grants to **150+** MDA Care Centers at top hospitals nationwide, including 48 ALS Care Centers.
- Provides care for **60,000+** individuals living with neuromuscular diseases each year at MDA Care Centers.
- Answers **10,000+** resource requests annually through the MDA Resource Center.



Apple HomePod is a smart speaker that can act as a hub for controlling your smart home.



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ALXN1210-DM-310_Quarterly Magazine Feature_V2_08Mar2022



A Clinical Research Study on Dermatomyositis (DM)

About the ALXN1210-DM-310 Study

Alexion’s mission is to deepen our understanding and transform the lives of people affected by rare diseases and who are living with devastating conditions. This knowledge allows us to innovate and evolve into new areas, where there is great unmet need and opportunity to help patients and families fully live their best lives. As part of our focus on new areas, Alexion is now researching dermatomyositis (DM) in the ALXN1210-DM-310 Study, which is enrolling approximately 180 adult participants across North America, Europe, and Asia Pacific.

The ALXN1210-DM-310 Study is a double-blind, randomized, placebo-controlled, parallel-group, multicenter study to evaluate the effectiveness and safety of ravulizumab in adult participants with DM. The purpose of this study is to evaluate the investigational medication compared to standard of care on reducing symptoms in patients with DM. This is a phase 2 study that leads to a phase 3 study.

Before enrolling in the study, interested individuals will enter the screening period to determine if they are eligible to join. Eligible participants must be at least 18 years of age with a diagnosis of DM, and they must have had an inadequate response or be intolerant to two or more DM treatments. Participants who complete the last visit in the study treatment period may have the opportunity to receive the investigational medication in the open-label extension period.

To learn more, please visit DM310.com.



Service *with a* Smile *and a* Wag

Here's what you need to know about what service dogs do and how to get one

BY BARBARA TWARDOWSKI AND JIM TWARDOWSKI, RN



Annie, a client of Canine Companions, praises her service dog, Yardley.

Image: Courtesy of Canine Companions

At Payton Rule’s high school in Missouri, the school year always ended with field day — a break from academics where kids demonstrated school spirit and played fun games on the athletic field located down a steep hill. Payton, who has Charcot-Marie-Tooth disease (CMT) and uses leg braces, missed the celebration during her first years at the school, because she could not safely maneuver the hill and the field on her own.

A few weeks before graduating from high school, Payton got her service dog, Whitt, through CHAMP Assistance Dogs in St. Louis. Holding onto Whitt’s harness gave Payton just enough counterbalance to traverse the hill and navigate the uneven terrain of the field. Finally, Payton joined her fellow students independently and enjoyed the camaraderie of field day.

Having a service dog can even the playing field for people with disabilities. These hardworking animals enhance the lives of those they serve. But having a service dog also is a responsibility. Deciding to get a service dog requires an honest evaluation of your needs and the demands of having a dog in your life.

What is a service dog?

“Assistance dog” is the general term for a guide dog, hearing dog, or service dog. A service dog works with individuals who have disabilities other than blindness (guide dog) or deafness (hearing dog).

According to Assistance Dogs International, service dogs are trained to do three or more specific tasks to mitigate the effects of an individual’s disability. They are legally allowed in public spaces with their handler.



Payton Rule is more active and independent with her service dog, Whitt.



What do service dogs do?

A service dog is not a pet. The dog has a job to do. (If you see a service dog in public, always ask permission before trying to pet it.)

These dogs are trained to follow commands to do tasks that assist people with disabilities, such as opening automatic doors, opening drawers and cabinets, picking up an object on the floor, fetching an item from another room, pushing an elevator button, turning lights on and off, helping with dressing, pulling a wheelchair a short distance, or assisting with balance when walking or standing up.

Just like people, service dogs need practice to keep their skills sharp. Payton, who is graduating from Washington University in the spring, has had Whitt at her side throughout her college career. One of Whitt’s skills is picking up an emergency phone and bringing it to her. At home, she keeps a phone with a special handle on the floor so the dog can easily reach it in a crisis. Every few months, Payton reviews this command with Whitt, so he doesn’t forget.

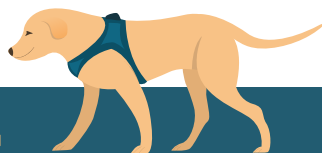
What are skilled companion dogs?

Most service dogs are paired with one person who has a disability. A skilled companion dog is an option for those under the age of 18 or an adult who may not have the physical or cognitive abilities to handle a dog. This type of service dog learns to work as part of a team.

“A skilled companion dog is primarily for someone who needs additional assistance handling the dog,” says Paige Mazzoni, chief executive officer of Canine Companions, which has six training centers across the nation. The dog is trained to work with a facilitator — typically a parent, spouse, or caregiver who lives in the same household with the recipient and cares for the dog.

The Arioto family of Gilroy, California, received a skilled companion dog from Canine Companions for their daughter, Siena, when she was 9. Siena, who has Friedreich’s ataxia (FA), was able to walk when the family got Lustra. Now, Siena is a teenager and uses a manual wheelchair. Having Lustra, who is trained to flip light switches, open and close drawers, and open automatic doors, allows Siena to be more independent.

Siena issues the commands for Lustra to perform tasks, but her parents are the dog’s facilitators. When Lustra is out in public, one of Siena’s parents has her on a leash. Lustra doesn’t attend school with Siena because a handler would need to be present with her all day.



FIND A SERVICE DOG

If you're considering applying for a service dog, start the process early. Many organizations have wait times, which can be more than a year. The Arioto family waited nine months before being matched with their skilled companion dog, and Payton Rule waited almost three years for her service dog.

There are about 100 nonprofit organizations in the United States that train and place assistance dogs. Many specialize in certain types of assistance, so compare organizations to find one that meets your needs. For example, Payton's dog is trained to assist her with her balance, but not all programs offer this type of training.

Assistance Dogs International (ADI) provides accreditation to nonprofit organizations that place assistance dogs. Search for ADI member organizations based on geographic location and additional criteria at [AssistanceDogsInternational.org](https://www.AssistanceDogsInternational.org).

Image: iStock.com/Rudzhan Nagiev

How are service dogs trained?

Service dogs typically spend two years in training before they're matched with an individual. A dog's temperament is evaluated throughout the training process. Not every dog can remain calm and responsive in a constantly changing environment or while meeting new people.

The process of obtaining a service dog can vary depending on the organization and the type of assistance needed, but it generally involves a period of on-site intensive training to bond with the dog as well as learn how to work with and care for it.

The Arioto family drove to the Canine Companions training center in Santa Rosa, California, and spent two weeks

there to be matched with Lustra and learn how to manage the dog's behavior, give her commands, and care for her.

Both the dog and the family had to show mastery of certain commands and skills before they could take Lustra home. In addition,

Service dogs are trained to do three or more specific tasks to mitigate the effects of an individual's disability. They are legally allowed in public spaces with their handler.

they took a public certification test to demonstrate they can manage the dog in public in accordance with Canine Companions and federal public access standards.

What care do service dogs need?

Caring for a service dog is a commitment. Payton has to anticipate Whitt's needs and plan accordingly. If they will be out in the evening, she brings his food and schedules his feeding. She

takes Whitt on several bathroom breaks throughout her day. And there are times when Payton opts to leave Whitt at home. For example, she did not take him to her chemistry class out of concern for his safety if chemicals were spilled on the floor.

Whitt is a furry golden retriever who requires frequent grooming. Also, just like people, Whitt needs some downtime from his job. Payton makes sure Whitt's day includes time to run and play with a ball.

What other responsibilities are involved?

Many nonprofit assistance dog organizations provide their dogs at no cost or low cost to clients. The recipient generally is responsible for travel expenses for on-site training and the costs of the dog's ongoing care. The estimated annual cost of feeding, grooming, and vet visits is \$2,000, according to Service Dogs, Inc.

Some organizations, like Canine Companions, retain ownership of the service dogs they place with families. The Arioto family meets periodically with a representative from Canine Companions, who evaluates how Lustra is doing.

Service dogs work for approximately eight years, and then they retire. When Lustra is ready to retire, the Ariotos will have the option to adopt her. If they choose not to, she'll be placed with another family to live the rest of her life as a pet.

Friend and helper

The decision to get a service dog shouldn't be taken lightly. A service dog comes with all the typical responsibilities of dog ownership, but also many rewards.

Affectionate and responsive, a service dog can boost a client's confidence and even be a bridge to social interaction.

"People mistakenly think service dogs are robots — a perfect mechanical animal that you can tell exactly what to do and they never have fun," says Paige of Canine Companions. "When they are 'released' from work, they are just like every other dog. They play. They're silly. They all have different personalities."

Payton has found having Whitt in her life is more responsibility than she'd anticipated. But their relationship is more rewarding than she ever imagined. "He's my best friend," says Payton. [@](#)

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

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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 is the only FDA-approved therapy for all patients with SMA regardless of age, including adults*

7+ years of clinical trial data across a range of SMA types
11,000+ people have been treated with SPINRAZA worldwide[†]

*Clinical studies of SPINRAZA included patients from 3 days to 16 years of age at first dose, but did not include sufficient numbers of subjects aged 65 and over to determine whether they respond differently from younger patients.

[†]Based on commercial patients, early access patients, and clinical trial participants through January 2022.

Watch SPINRAZA stories



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.

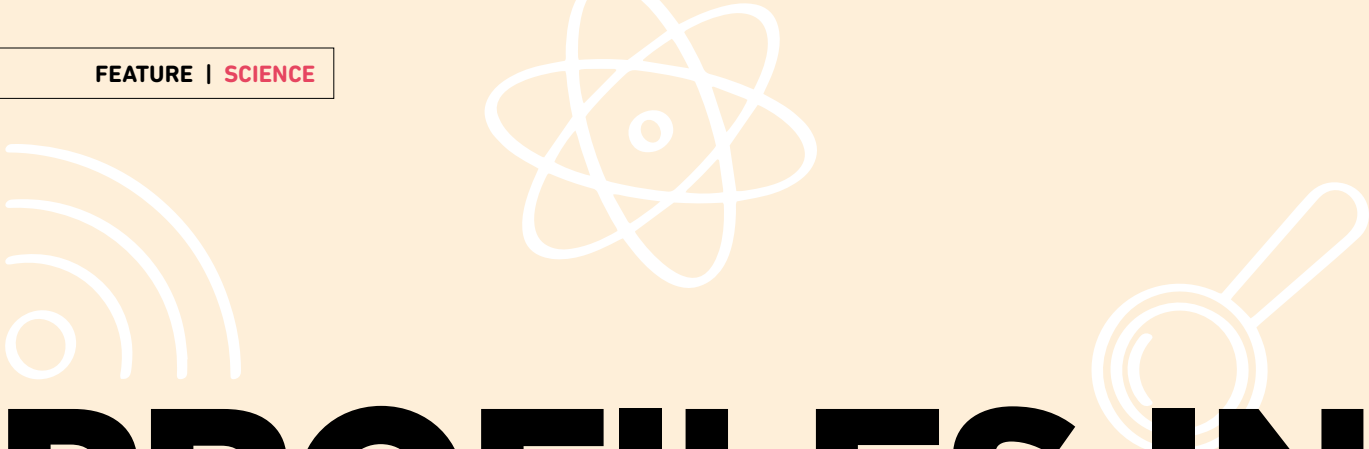
Before taking SPINRAZA, tell your healthcare provider if you are pregnant or plan to become pregnant.

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).

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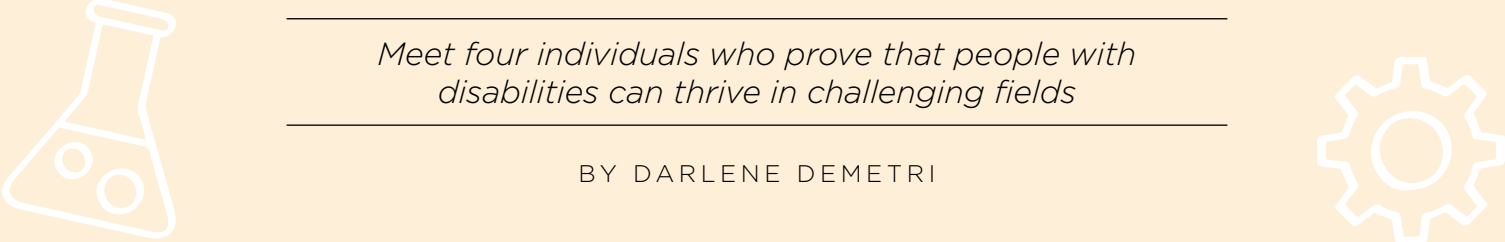


PROFILES IN

STEM

Meet four individuals who prove that people with disabilities can thrive in challenging fields

BY DARLENE DEMETRI



Although students with disabilities indicate interest and intent to pursue science, technology, engineering, and math (STEM) careers at the same rates as students without disabilities, they do not receive an undergraduate degree or pursue STEM graduate studies at the same rates. This fact was reported by the National Institutes of Health (NIH) in 2021. They also cited several barriers, for people with disabilities,

including lack of recruitment and engagement and an absence of mentors and role models.

“Research shows that people with disabilities are underrepresented in STEM and yet, at the same time there is research showing that diversity drives innovation,” says Marissa Lozano, MEd, MDA’s director of community education. “The more diversity we can bring to STEM, the more advancements we will make.”

In response, MDA launched STEM Connections in 2021, a program that exposes youth and young adults living with neuromuscular disease to STEM concepts and careers. (See “STEM for the Next Generation” on page 34.)

Here, we profile four individuals with neuromuscular disease who are breaking down barriers, specifically in STEM medical fields. We hope their stories of perseverance and words of advice

will encourage others living with neuromuscular disease to pursue their dreams, too.

Justin Moy

Justin, 21, an MDA Ambassador, lives with congenital muscular dystrophy (CMD). He is in a combined bachelor’s/master’s program at Worcester Polytechnic Institute (WPI) in Massachusetts, studying bioinformatics and computational biology, an emerging STEM field. This means he uses computer

science methods, such as software that analyzes data, to find answers to biological problems. He is applying to PhD programs and hopes to one day find a cure for CMD.

BRIGHT START: “As a little boy, I loved playing outside in the dirt, looking for worms,” Justin says. “My insatiable curiosity and the encouragement of my grandmother, who loved science, compelled me to pursue a STEM career.”

MAKING IT WORK: Justin uses a power wheelchair and finds working with computers suits his abilities. WPI’s Office of Accessibility Services has provided accommodations such as accessible on-campus housing, adjustable desks, and relocating classes to accessible classrooms.

WORDS OF ADVICE: “STEM is a great career path for people with

neuromuscular diseases, but think about and research the sort of STEM you want to go into,” Justin says. “Find a subject that interests you and look for people in your area that work on that subject, then reach out, explore, and network.”

Vovanti Jones, MD

Dr. Jones, 34, lives with limb-girdle muscular dystrophy (LGMD). She specializes in physical medicine and rehabilitation at the University of Missouri Health Care’s University Hospital in Columbia, where she directs the MDA Care Center and inpatient stroke rehabilitation unit. Among her duties, she’s currently working with patients who are in clinical trials testing new therapies for Charcot-Marie-Tooth disease (CMT) and amyotrophic lateral sclerosis (ALS). “I went into medicine to heal the sick and be at the forefront of changing health-care,” she says.



Vovanti Jones, MD, specializes in physical medicine and rehabilitation.

BRIGHT START: “Since age 4, I knew I was going into medicine,” Dr. Jones says. “I was interested in the way the body works and how we go about solving problems that happen to our bodies.” She loved attending science summer camps as a child.

MAKING IT WORK: Dr. Jones uses a scooter in the hospital because it is small and maneuverable. In other settings, she uses a power wheelchair. The university’s Office of Accessibility and ADA provides her with an adjustable desk and automatic doors in her office, a raised toilet seat in the bathroom, and a scribe for note-taking.

WORDS OF ADVICE: “To go into any advanced STEM career, such as medicine, you must advocate for yourself,” Dr. Jones says. “Don’t allow school

“ Research shows that people with disabilities are underrepresented in STEM and yet, at the same time there is research showing that diversity drives innovation.

—Marissa Lozano, MEd

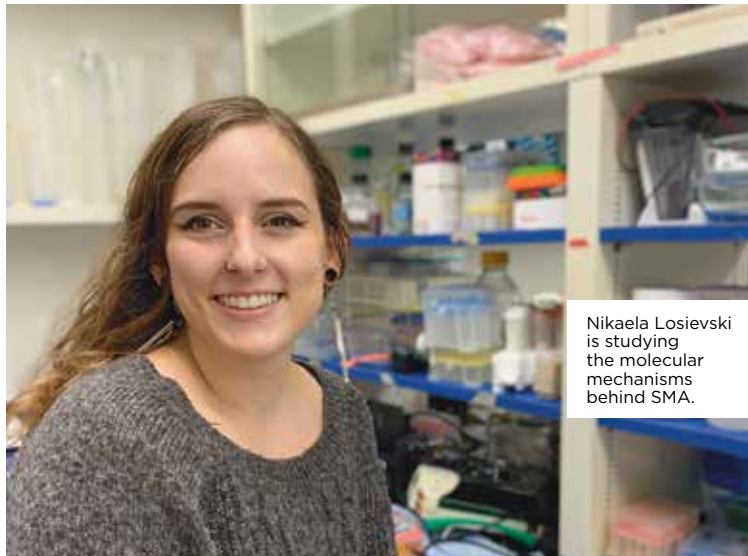


Justin Moy is studying bioinformatics and computational biology, an emerging STEM field.

administrators to box you into their ideas about what you can and cannot do. But be realistic about your limitations. There are aspects of medicine I can't do — I can't be a surgeon. But I can do rehabilitation medicine, which requires a different skill set."

Nikaela Losievski

Nikaela, 25, lives with spinal muscular atrophy (SMA). She is pursuing her doctorate in neuroscience at Ohio State University (OSU) in Columbus and is studying the molecular mechanisms behind SMA. "Scientists think the SMN protein is involved in several processes, but there's no definitive evidence for most of them," she says. She is focusing on one proposed function of SMN protein to help figure out why the disease occurs. Once she earns her PhD, Nikaela plans to continue with research, studying RNA (a molecule similar to DNA) in neurological diseases.



Nikaela Losievski is studying the molecular mechanisms behind SMA.

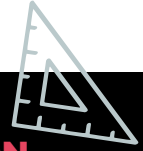
BRIGHT START: "I was always making concoctions when I was young. Whenever my mother heard the water running in the sink upstairs, she started panicking because she knew I was making a potion," Nikaela says. "I've always had an experimental mind."

Three high school science teachers further ignited her passion for pursuing a career in STEM. "Because they were women, I could see myself in their shoes," she says.

MAKING IT WORK: Nikaela uses a manual wheelchair with a smart drive. In the lab, she's found that clear, constant communication with project leaders helps her do her job. "It is vital to know what they expect of you but also that they understand your reality with a chronic illness," she says. She alerts her supervisor the moment she notices a potential barrier, so he can help advocate on her behalf.

WORDS OF ADVICE: Nikaela recommends searching organizations' websites for scientists in career paths you might be interested in, then reaching out to them for information or advice. "Once you know where to look, just ask. What's the worst that can happen?" she says.

STEM FOR THE NEXT GENERATION



MDA's STEM Connections program provides hands-on activities for kids and young adults living with neuromuscular disease to learn STEM concepts and explore STEM-based careers in a supportive environment.

This year's program involves two robust components.

MDA Summer Camp

Summer Camp is available for youth ages 8-17 in-person at more than 20 sites across the United States, as well as virtually. In either option, campers will participate in fun activities designed to expose them to different aspects of STEM.

Each camper will receive a STEM activity kit at no cost. Virtual campers will be mailed a kit containing materials and flashcards outlining each activity with links to videos, interactive content, and more.

Register for MDA Summer Camp or apply to volunteer at mda.org/summer-camp.

Fall Mentoring Program

This four-week program, offered virtually to participants ages 16-21, provides opportunities to connect with and learn from people who are thriving in STEM fields, including STEM professionals living with neuromuscular disease.

Participants attend two video sessions each week, covering topics such as job exploration, self-advocacy, and obtaining accommodations at school, college, and work. They also complete small-group projects led by mentors. Materials are provided at no cost and mailed in advance.

Volunteer to be a STEM mentor by emailing ResourceCenter@mdaUSA.org or calling (833) 275-6321. Registration for participants will open in late summer.

Marc van de Rijn, MD

Dr. van de Rijn, 34, lives with facioscapulohumeral muscular dystrophy (FSHD). He specializes in physical medicine and rehabilitation, as well as neuromuscular medicine. He currently practices physiatry at Spaulding Rehabilitation Hospital in Boston. “Physiatrists treat disease yet also focus on the whole patient, their function, quality of life, and rehabilitation,” Dr. van de Rijn says. He often works with patients with neuromuscular diseases and neurological conditions such as Parkinson’s disease.

BRIGHT START: “I enjoyed biology and chemistry in grade school, but then I got into computers in high

school,” Dr. van de Rijn says. After completing a computer programming internship, he realized he wanted to practice medicine. “My FSHD diagnosis at age 12 certainly influenced my choice of specialty,” he says.

MAKING IT WORK:

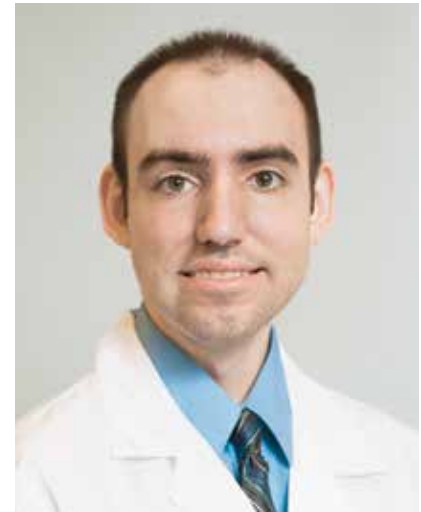
Dr. van de Rijn walks slowly because of weakness in his legs, so the hospital gave him access to the closest parking lot. He also has difficulty writing and typing, so the hospital provided speech recognition software to help him do chart documentation.

Although he’s customized the clinic to his needs, he also sees patients on a medical floor, where he has less control over the environment.

“There are in-the-moment concerns. But something you learn when you have a physical disability is that you can’t be afraid to ask for help,” Dr. van de Rijn says.

WORDS OF ADVICE:

“Being successful is not about whether you can or cannot do something, it’s about not being afraid to ask for adaptations, equipment, and accommodations to allow you to do the job at the same level as someone without a disability,” Dr. van de Rijn says. [Q](#)



Marc van de Rijn, MD, often works with patients with neuromuscular diseases.

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

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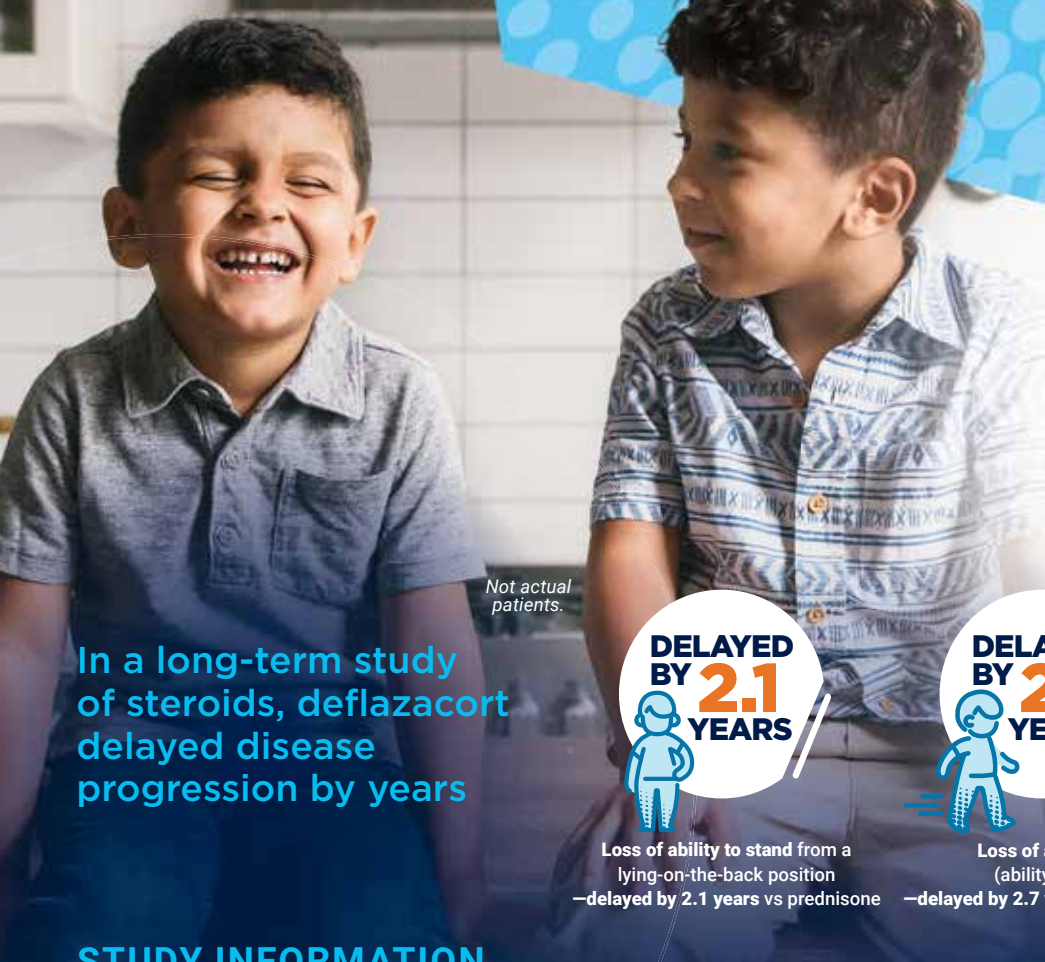
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In a long-term study of steroids, deflazacort delayed disease progression by years

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—delayed by 2.1 years vs prednisone



Loss of ambulation (ability to walk)
—delayed by 2.7 years vs prednisone



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

STUDY INFORMATION

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

Objective: This study examined the long-term effects of glucocorticoids on milestone-related disease progression across the lifespan and survival in patients with DMD

Methods: For this prospective cohort study, male patients aged 2 - 28 years with DMD were enrolled at 20 centers in nine countries. Patients were followed up for 10 years. The study measured the progression of nine mobility and upper limb milestones to compare no glucocorticoid treatment or cumulative treatment duration of less than 1 month versus treatment of 1 year or longer.

Results: 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). 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).

PLEASE NOTE This study is not in the approved prescribing label for EMFLAZA, but is consistent with the information that is included. Please talk to your son's healthcare provider if you have any questions.



Emflaza®
(deflazacort)

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



INDICATION & IMPORTANT SAFETY INFORMATION FOR EMFLAZA® (deflazacort)

What is EMFLAZA® (deflazacort) used for?

EMFLAZA is a prescription medicine used to treat Duchenne muscular dystrophy (DMD) in patients 2 years of age and older.

When should I not take EMFLAZA?

Do not use if you have had hypersensitivity, including allergic reactions, to deflazacort or any of the inactive ingredients.

What warnings should I know about EMFLAZA?

- EMFLAZA can cause changes in endocrine function. 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.
- There is an increased risk of infection when taking EMFLAZA. Tell the healthcare provider if the patient has had recent or ongoing infections or if they have recently received a vaccine. Medical advice should be sought immediately if the patient develops fever or other signs of infection. Patients and/or caregivers should be made aware that some infections can potentially be severe and fatal. Warn patients who are on corticosteroids to avoid exposure to chickenpox or measles and to alert their healthcare provider immediately if they are exposed.
- EMFLAZA can cause an increase in blood pressure and water retention. If this occurs, dietary salt restriction and potassium supplementation may be needed.
- There is an increased risk of developing a hole in the stomach or intestines in patients with certain stomach or intestine disorders when taking corticosteroids like EMFLAZA.
- EMFLAZA can cause severe behavioral and mood changes. Seek medical attention from the health care provider if any behavioral or mood changes develop.
- There is a risk of osteoporosis with prolonged use of EMFLAZA, which can lead to vertebral and long bone fractures.

- EMFLAZA may cause cataracts or glaucoma and a health care provider should monitor for these conditions if corticosteroid therapy is continued for more than 6 weeks.
- Immunizations should be up-to-date according to immunization guidelines prior to starting therapy with EMFLAZA. Live-attenuated or live vaccines should be administered at least 4 to 6 weeks prior to starting EMFLAZA. Live-attenuated or live vaccines should not be used in patients taking EMFLAZA.
- EMFLAZA can cause serious skin rashes. Seek medical attention at the first sign of a rash.
- Rare instances of anaphylaxis have occurred in patients receiving corticosteroid therapy, including EMFLAZA.

What should I tell my health care provider?

Tell the health care provider about all medical conditions, including if the patient:

- is pregnant or planning to become pregnant. EMFLAZA® (deflazacort) can harm your unborn baby.
- is breastfeeding or planning to breastfeed. EMFLAZA may appear in breastmilk and could affect a nursing child.

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 side effects of EMFLAZA?

The most common side effects of EMFLAZA 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. These are not all of the possible side effects of EMFLAZA. Call your doctor for medical advice about side effects.

To report an adverse event, please call 1-866-562-4620 or email at usmedinfo@ptcbio.com. You may also report side effects to FDA at 1-800-FDA-1088 or at www.fda.gov/medwatch.

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A Successful In-Person MDA Conference

In mid-March, more than 1,000 people gathered in Nashville, Tennessee, for MDA's 2022 Clinical & Scientific Conference.

"It's a delight to meet again in person," said MDA President and CEO Don Wood, PhD, in a welcome address. "For many of you, it's the first time on a plane in two years."

In fact, the four-day event held at the Gaylord Opryland Resort & Convention Center marked the world's largest meeting of neuromuscular disease healthcare providers, researchers, investors, patients, and caregivers, with 1,029 attendees from 15 countries.

In addition, the conference attracted 691 virtual participants via livestream and featured more than 120 speakers in 32 sessions with one full day of clinical trial updates.

The educational sessions covered timely topics, such as new gene discoveries, brain interface technology, and ethics in neuromuscular disease care, as well as the latest research on therapies for many different neuromuscular diseases.

"This is an exciting time in neuromuscular disease research ... a time when we're developing a whole new approach: genetic medicine," Dr. Wood said. "Most of the



Amy Shinneman and Carsten Bönemann, MD

diseases we cover are genetic in origin. And where there were no treatments 15 years ago, we now have 15 or 20 FDA-approved treatments, many of which originated with research funded by MDA."

Dr. Wood introduced Amy Shinneman, 47, our new MDA National Ambassador, who, along with current

TRIBUTE RECEPTION FELT LIKE A FAMILY REUNION

Building on our legacy of innovation in research, care, and fundraising, MDA brought the community together in "The Volunteer State" on March 17 for a Volunteer Tribute Reception at the Grand Ole Opry.

Tennessee Governor Bill Lee declared the date an official Day of Recognition, and MDA's celebration honored three local champions: Broadcast TV pioneer

Harold Crump; former Director of the MDA Care Center at Vanderbilt University Medical Center Peter Donofrio, MD; and former Tennessee Titan Tim Shaw, who lives with ALS. It was truly a reunion, bringing together Nashville-area families, supporters, partners, and celebrities. Dean Hayes and Kyle Gunden, buddies from MDA Summer Camp, got to see each other in

person for the first time in three years. For more photos from the celebration, visit mda.org/NashvilleTribute.



Left: Dean Hayes, left, and Kyle Gunden with Ashley Davis of Permobil

Right: Honorees Harold Crump, left, and Tim Shaw, center, with MDA's Don Wood and Kristine Welker

National Ambassador Ethan LyBrand, 12, will represent families living with neuromuscular diseases.

Amy, who lives with Bethlem myopathy, presented MDA's first Legacy Award for Achievement in Clinical Research — a new annual recognition for outstanding accomplishments in neuromuscular disease research — to Carsten Bönnemann, MD, of the National Institute of Neurological Disorders and Stroke, a division of the National Institutes of Health (NIH).

Dr. Bönnemann's accomplishments include identifying beta and delta sarcoglycan genes as causes of limb girdle muscular dystrophies (LGMD); developing preclinical animal models for congenital muscular dystrophies (CMD), which are now used to test exon-skipping therapies; establishing natural history and outcome measures for congenital myopathies toward clinical trials; and conducting the first AAV gene therapy trial for giant axonal neuropathy in humans.

In addition, Amy shared her story of living with Bethlem myopathy, a rare disease seen in fewer than



1 in 100,000 people. It affects the skeletal muscles and connective tissue, and is characterized by slowly worsening muscle weakness and joint stiffness. She lives in the Indianapolis area with her husband and two teenage sons and has been

active with MDA for much of her life.

"MDA Summer Camp turned out to be one of the best weeks of my life. I felt like I had found my people, but I really hadn't because I was still not diagnosed," she recalled. "I searched for a diagnosis for 27 years, during which time I went to college, got married, and had two beautiful boys. My husband and my boys very much led the journey searching for a diagnosis."

In 2018, Amy finally got her diagnosis. "My neurologist suggested genetic testing for myself and my parents," she said. "When we found out the answer, my family celebrated. For me, it's indescribable what it felt like. I was finally, formally introduced to myself — and I immediately started to heal from all the years of being unidentified."

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MDA Muscle Walk season kicked off on March 27 in Tempe, Arizona. Team Mogensen, pictured above, raised more than \$2,000 for MDA at the event. Gather your team and sign up for a walk at mda.org/muscle-walk.

PJ Lives On

For people on long-term corticosteroid treatment, like those with Duchenne muscular dystrophy (DMD), rapid reductions in those medications can be dangerous. But not all healthcare providers know that. The PJ Nicholoff Steroid Protocol is a clear set of guidelines they can follow in emergency situations, saving lives.



Read about the man behind these guidelines at mda.org/quest/pjs-protocol.

Image: iStock.com/bsd555

MDA's Care Center Network Expands

We're welcoming 13 new hospitals and healthcare institutions to the MDA Care Center Network. These Care Centers provide access to specialized care, clinical trials, research, treatments, and education for individuals living with muscular dystrophy, ALS, and other neuromuscular diseases. The network includes more than 150 Care Centers.

"Since MDA was founded in 1950, life expectancy and quality of life have vastly improved for individuals with neuromuscular diseases," says Donald S. Wood, PhD, MDA's president and CEO. "This is due in large part to the best-in-class, comprehensive multidisciplinary care provided to families from a wide variety of healthcare specialists at MDA Care Centers."

Find a Care Center at mda.org/care/mda-care-centers.

The new members of the MDA Care Center Network are:

- AdventHealth Orlando, FL
- Arkansas Children's Hospital, Little Rock, AR
- Baylor College of Medicine, Houston, TX
- Children's Hospital of Michigan, Detroit, MI
- Dell Children's Medical Center of Central Texas, Austin, TX
- Helen DeVos Children's Hospital, Grand Rapids, MI
- Idaho Physical Medicine & Rehabilitation, Boise, ID
- Loma Linda University, Pediatric, Loma Linda, CA
- Massachusetts General Hospital, Boston, MA
- NYU Langone Health, Pediatric, New York, NY
- Rapides Regional Medical Center, Alexandria, LA
- University of Massachusetts Memorial Medical Center, Duchenne Program, Worcester, MA
- University of Utah, Pediatric Neurology, Salt Lake City, UT

Free Educational Workshops



MDA continues to add to our library of Access Workshops, which are created to educate the neuromuscular disease community on overcoming barriers and claiming independence. The self-paced online workshops are filled with presentations and engaging activities.

Access Workshops at mda.org/AccessWorkshops:

- Access to Coverage:** Insurance — **NEW**
- Access to Coverage:** Equipment & Assistive Devices
- Access to Education:** K-12
- Access to Education:** Higher Education
- Access to Coverage:** Therapies — **COMING SOON**

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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

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

One Day at a Time

Being diagnosed with myotonic dystrophy changed my life, and I had to change with it

BY THADDEUS DOMBROWSKI

On my 28th birthday, I recognized that something was wrong. I used to be able to jump and touch the rim of a basketball hoop. But on this day, I was missing by a foot.

I remembered learning in a high school science class that a sudden loss of athleticism was a sign of a neuromuscular problem, but I didn't want a medical diagnosis altering my plans. So, I decided to shut it out of my mind until it became a real concern.

Altering plans

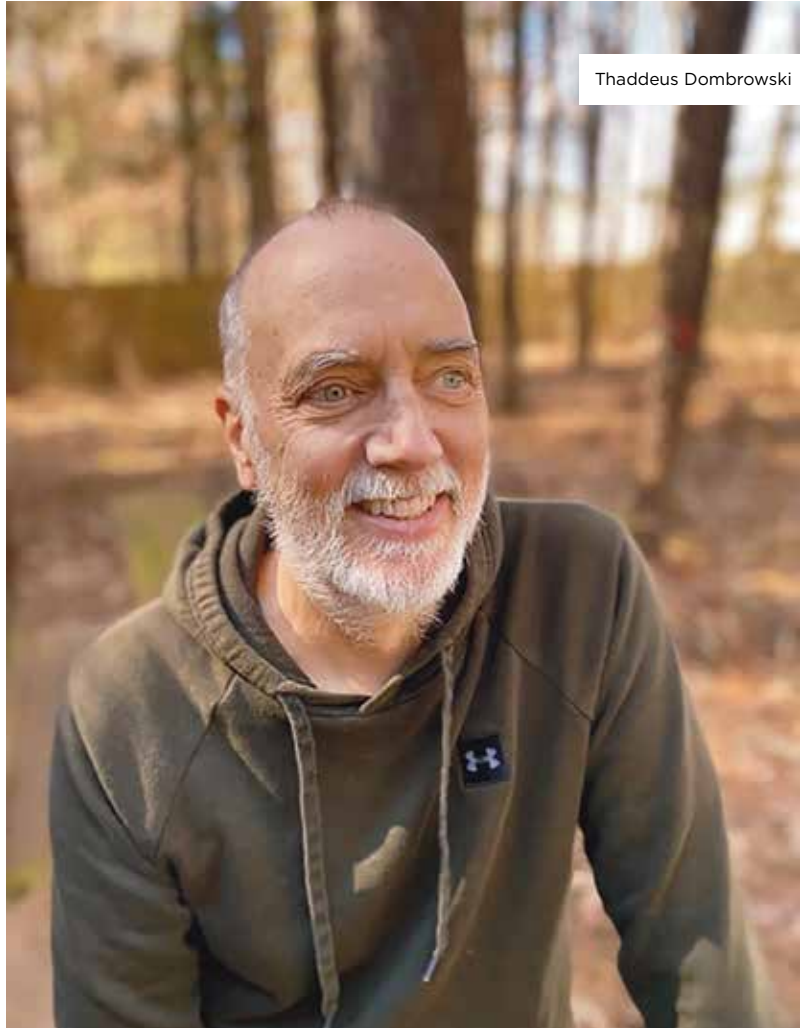
Fast forward 21 years. I was a software engineer in Arizona, and I was riding my bicycle to work, as I had been doing a few times a week for years. I loved biking, which I had taken up to strengthen my legs. But, on this day, as I locked my bike to the rack at work, my body spoke to me. In my mind I heard it say, "You are not biking anymore. I can't do this." And that was that. I still wanted to ride my bike, but my body refused.

For several years I had been keeping a journal. Lately, I found myself struggling to find words to describe what I was experiencing. Something was not right, but, until the day of my last bike ride, I didn't even know what to tell my doctor. Finally, I made an appointment. I told her about that ride and the pain I was experiencing.

“

I remember the effort it took to change my life, but also the attitudes and beliefs that needed to shift. For example, my first response to my diagnosis was simply to push harder. I didn't know what else to do. But, when I finally accepted that I have a disability, I stopped being so hard on myself.

—Thaddeus Dombrowski



At first, I was given a clinical diagnosis of motor neuron disease. In 2012, I was surprised when a genetic test revealed that I actually have myotonic dystrophy type 2 (DM2).

Advancing symptoms

Three months later, I needed a cane to walk. In medical literature, DM2 is described as mild, so I figured the cane would be the end of the story.

At work, I had accumulated a lot of sick pay. I started taking an occasional day for rest. It wasn't long before I needed a rest day each week. Then, it was twice a week.



“

After having a ramp and front deck built, I finally moved into my new home. —Thaddeus Dombrowski

Outside of work, life was becoming impossible. Dirty dishes were piled in the sink. My house was filthy. I was exhausted. I finally recognized that this was untenable. A year after my diagnosis, I declared for disability.

Within another year, I needed a wheelchair. But things began to turn around after I got into an MDA Care Center. A sleep study led to a diagnosis of central sleep apnea, which is common with DM2. I started using a bilevel positive airway pressure (BiPAP) machine, and for the first time in years, I began to sleep through the night.

Adapting to change

The year before this ordeal began, my wife and I separated. I wanted to continue to live independently, but I needed to be closer to my family. My older sister and my mom live in North Carolina. I told them that I wanted to move near them, but I couldn't give an ETA. I was just learning to adapt to my new reality, and there was so much to do.

The prospect of moving was daunting. However, by planning everything out and breaking things into achievable tasks, I made progress.

One of the first problems was finding a wheelchair accessible van. I turned to the internet to begin my search, but I knew nothing and really needed someone to talk to. I visited a mobility dealership and found that they were exactly the people to help me. They invited me to participate in a disability support group at the dealership, after hours. They also matched me with a used van within my budget.

Making a move

It took two more years until I was ready to move. To pull this off, I rented an apartment near my home and brought everything I wanted to keep to the apartment. What was left in the house was stuff to sell or give away. Then I put the house on the market. At this point, I could finally leave for North Carolina.

I rented while I was house hunting. I had no expectation of finding a home equipped for me, but I studied each option and considered the accessibility hurdles. In 2017, almost a year after moving here, I bought a home. After having a ramp and front deck built, I finally moved into my new home.

Now, five years later, I remember the effort it took to change my life, but also the attitudes and beliefs that needed

to shift. For example, my first response to my diagnosis was simply to push harder. I didn't know what else to do. But, when I finally accepted that I have a disability, I stopped being so hard on myself. I began to listen to my body and to rest more between activities. Now, I think things through more before doing them. The goal is to work smarter, not harder. I also had to become more patient and forgiving of myself and others.

Since I made these changes, I am happier and more optimistic than I have been in years. Acceptance, patience, and self-forgiveness have led me to a new confidence about my situation. [@](#)

Thaddeus Dombrowski was diagnosed with DM2 in 2012. He lives with his two cats in North Carolina, where he raises worms to support his gardening habit.

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Dynamic Duo

Teen's small business saves man's best friend

Some of the best ideas — and friendships — are born from moments of necessity. When 13-year-old Jackson Saville, who lives with spinal muscular atrophy (SMA), heard his mother's plan to keep him busy with educational activities last summer, he sprang into action.

"I had a better idea than the fun worksheets, which aren't very fun at all," says Jackson, who lives in Virginia Beach, Va. "I wanted to start my very own business." In June 2021, he created RockStar Life Designs (rockstarlifedesigns.com) and began selling personalized t-shirts, cups, and blankets, using a cutting machine, heat press, and his mother's sewing skills.

Around the same time, Jackson learned of a four-year-old golden retriever who had been hit by a car in Istanbul, Turkey, and rescued by the Southeastern Virginia Golden Retriever Rescue, Education, and Training Group. They flew the injured dog to the United States for spinal surgery. Jackson, who has gone through numerous treatments for SMA and 18 surgeries for scoliosis, immediately felt a bond with the dog, named Archie.

"I said, 'I can't just sit there and pray for Archie's surgery to work,'" Jackson says. "I must take action immediately to raise money for his surgery.'" Since then, he has sold more than 300 pet bandanas and raised more than \$1,800 to fund the dog's medical expenses.

+SERVICE DOG FAQ

Have you wondered how a service dog is different from a pet, and how to get one? Read "Service With a Smile and a Wag" on page 26.

However, Jackson and Archie's story does not end there. On Oct. 26, 2021, the Saville family adopted Archie, and now he and Jackson are inseparable. Whether Jackson is doing homework, making a custom order,

or exploring the neighborhood, he always has his trusty four-legged sidekick.

"My favorite thing about Archie is he is always there for me when I need him," Jackson says.

Jackson has big plans for the future. He wants to turn his business into a multi-billion-dollar enterprise, start an international dog rescue, and drive a shiny, red Lamborghini with a special seat in the back for Archie. [Q](#)



Jackson Saville helped rescue Archie, a golden retriever that was injured in Turkey.



Jackson is also an entrepreneur who started his own design business.



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Research: Read up-to-date management options and information about the latest science in Duchenne.



Community Voices: Watch videos showing real-life experiences and advice from patients and caregivers.



Knowledge: Access resources to help you better understand complicated subjects, including the importance of dystrophin and genetic testing, and also find questions to ask your doctor about Duchenne.



Stay in the Know about Duchenne. Sign up today.

Over 76% of people living with type 1 myotonic dystrophy indicate they are impacted by impaired sleep or daytime sleepiness.

“Long before you ever get to the muscle loss, you have the inability to stay focused and alert all day. The days are spent on my couch because I’m too sleepy to get out and do things.”

– **Susannah, living with myotonic dystrophy**

Reference: Treating sleep disorders in DM1. Myotonic Dystrophy Foundation. Accessed February 2, 2022. <https://www.myotonic.org/treating-sleep-disorders-dm1>

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Do you or someone you love have type 1 myotonic dystrophy?

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