

Quest

EMPOWERING FAMILIES WITH INFORMATION AND INSPIRATION

MDA[®] Muscular
Dystrophy
Association

MDA.ORG/QUEST
FALL 2018

from
lab
to pharmacy

**How scientists develop
safe and effective
drugs to treat
neuromuscular diseases**

TAKE TO THE SKIES
How MDA advocates
for accessible air travel

THE NEW BLACK
Adaptive fashion goes
mainstream

ADVERTISEMENT

Biogen discovers, develops, and delivers therapies for the treatment of neurodegenerative and rare diseases.



WWW.BIOGEN.COM



©2017 Biogen. All rights reserved • 12/17 SMA-US-0310 • 225 Binney Street, Cambridge, MA 02142

About the Muscular Dystrophy Association

MDA is committed to transforming the lives of people affected by muscular dystrophy, ALS and related neuromuscular diseases through innovations in science and innovations in care. Since our inception, MDA has committed more than \$1 billion to accelerate the discovery of therapies and cures. MDA supports the largest network of multidisciplinary clinics providing best-in-class care at more than 150 of the nation's top medical institutions and serves the community through MDA Summer Camp; the Resource Center; and educational conferences, events and materials for families and health care providers.

For advertising opportunities:

Patty MacEwan

National Director of Corporate Partnerships
pmacewan@mdausa.org

For editorial queries:

quest@mdausa.org

To update personal information and your Quest subscription status, contact the MDA Resource Center at **833-ASK-MDA1**.

Published quarterly by

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

Postmaster: Treat as Standard A Mail Only
© 2018, Muscular Dystrophy Association. All rights reserved. MDA and QUEST are registered trademarks of the Muscular Dystrophy Association.

The acceptance of advertising in this magazine does not constitute or imply endorsement by MDA of any product or service. MDA accepts no responsibility for any claims made in any advertisement. Quest reserves the right to refuse to accept any advertisement. For advertising information, call (703) 582-8476.

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

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



PUBLICATION MANAGEMENT



Generous Partners Help Us Reach Our Goal

MDA's mission is to transform the lives of people affected by muscular dystrophy, ALS and related neuromuscular diseases through innovations in science and innovations in care. It's an ambitious goal, and one we couldn't possibly aspire to without the commitment and generosity of our partners.

MDA has been proud to team up with leading corporations, organizations and brands who generate tens of millions of dollars each year through integrated cause marketing campaigns, community outreach initiatives, employee engagement programs, patient and provider educational programs, and year-round special events. With their support, not only is MDA accelerating the development of therapies and cures, but also providing much needed programs and services that enrich the lives of people with neuromuscular disease.



We are especially grateful to our largest partners:

- > IAFF
- > CITGO Petroleum Corp.
- > Harley-Davidson
- > Acosta
- > Albertsons Companies
- > Casey's General Store
- > Circle K
- > Dutch Bros
- > Kroger
- > National Association of Letter Carriers
- > Sailormen/Popeyes
- > Petroleum Marketing Group
- > 7-Eleven
- > Andeavor
- > Bojangles'
- > Burger King
- > Harris Teeter
- > Jiffy Lube
- > Mansfield
- > Price Chopper
- > Publix
- > Applebee's
- > C.N. Brown Company
- > DECA
- > ERA Real Estate
- > Fareway Meat & Grocery
- > Fishing for MD
- > RelaDyne
- > Shaw's/Star Market
- > United Refining Company
- > Wendy's

Thank you, too, to our pharmaceutical industry partners:

- > Biogen
- > Sanofi-Genzyme
- > AveXis
- > Mitsubishi Tanabe Pharma America
- > PTC Therapeutics
- > Santhera Pharmaceuticals
- > Sarepta Therapeutics

With gratitude from the whole MDA team,



Adam Cotumaccio
EVP, Chief Impact and Philanthropy Officer



fall 2018

CONTENTS

DEPARTMENTS

04 SHARING OUR STRENGTH

MDA knows the power of working together.

06 PROGRESS NOW

Read the latest news on medical research, scientific advances and clinical trials.

14 THRIVE 365

Scholarships help lessen the financial load of higher education.



pg 50



38 ACCESS MDA

Team Momentum memorable moments, fundraising tips and more.

50 FROM WHERE I SIT

A young woman finds her happy place in the alternative music scene.

52 LASTING IMPRESSION

A father and son celebrate a milestone with Harley-Davidson.

FEATURES

16 OPENING DOORS

Family caregivers find new life paths.

22 HOW DRUGS ARE DEVELOPED

It takes a lot of time and effort for a new drug to get to the pharmacy shelf.

28 TAKE TO THE SKIES

Access to air travel should not be denied based on physical ability.



34 ADAPTIVE IS THE NEW BLACK

People living with disabilities have a passion for fashion.



pg 14
Cody
Schoppe

NOV. 27 is #GIVINGTUESDAY™

Join in this global day of giving and help MDA make 2018 our biggest Giving Tuesday ever. You can participate by giving your time or a donation on Nov. 27, then tell your family, friends and community about it using #GivingTuesday.

To make a donation to MDA, visit mda.org/donate, or call us at 800-572-1717.



PROVIDING *FREEDOM*

through innovative mobility solutions

Wheelchair Accessible Minivans



Wheelchair Accessible SUV



With a variety of wheelchair-accessible vehicles available, VMI can accommodate almost any wheelchair user. Each one of our solutions offers flexibility, interior space and reliability.

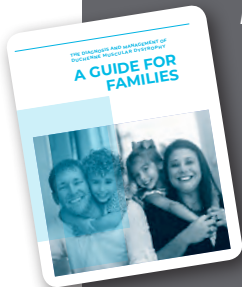
Contact VMI to learn more about the industry's most spacious wheelchair-accessible vehicles.

vmivans.com | 855-VMI-VANS



Proud Partners

At MDA, we take a big-picture perspective across the full spectrum of neuromuscular diseases to uncover breakthroughs that accelerate treatments and cures. That's a big job, and we know we can't do it alone. Often, we combine strengths with other organizations with common goals. Here, remarks on the Strongly blog (strongly.mda.org) and elsewhere illustrate the power of working together.



Earlier this year, MDA partnered with DMD patient advocacy groups, including TREAT-NMD, the World Duchenne Organization and Parent Project Muscular Dystrophy, to produce the new *Duchenne Guide for Families*. This guide serves as a roadmap to new medical care considerations for DMD families. Robin Geiger, vice president of MDA Care and Clinical Services, wrote this on the Strongly blog:

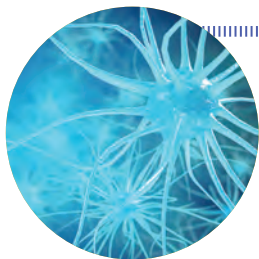
"My strongest drive within the organization is to provide the most efficient and standardized processes, creating health equity for patients. And along the way, I hope to improve quality of life for families and caregivers."

MDA in the Lab

MDA partners with researchers and organizations around the world by funding cutting-edge research aimed at finding treatments and cures for neuromuscular diseases. Turn to page 6 to learn about our latest grants, or visit our Grants at a Glance page at mda.org/gaag.

In September, MDA joined with the Charcot-Marie-Tooth Association and the Hereditary Neuropathy Foundation to host a patient-focused drug development meeting for CMT. Lucas Kempf, M.D., acting associate director of the U.S. Food and Drug Administration's Rare Disease Program, made these closing remarks:

"There are 2.6 million people worldwide [with CMT], so the impact of the 300 people here will have a huge effect across the entire world."



MDA is teaming up with Answer ALS, a nationwide consortium assembling a comprehensive clinical, genetic, molecular and biochemical assessment of amyotrophic lateral sclerosis (ALS). Through the partnership, Answer ALS will build a dataset of disease measures and develop new approaches to analyzing the massive amount of data, which will be shared openly with the global research community. Amanda Haidet-Phillips, Ph.D., MDA scientific portfolio director, said this on the Strongly blog:

"Mining this rich, complex dataset with the goal of identifying ALS disease subtypes requires sophisticated computational tools. The project investigators will develop a series of tools to specifically interrogate and interpret the combined Answer ALS datasets, leading to an improved understanding of ALS."

Know the Pompe Spectrum



Pompe disease presents as a spectrum of symptoms and variable rates of disease progression. Symptoms vary from person to person and may begin at birth all the way to adulthood. Many signs and symptoms overlap with other disorders as seen in the table below.

→ SHARED SIGNS AND SYMPTOMS WITH POMPE DISEASE

DISEASE PREVALENCE	MUSCLE WEAKNESS	ABNORMAL GAIT OR DIFFICULTY WALKING	ELEVATED CK*	RESPIRATORY INSUFFICIENCY	DIFFICULTY SWALLOWING	EXERCISE INTOLERANCE
Myopathy, General	●	●		●	●	●
Duchenne MD 1.7 – 8.2 per 100,000	●	●	●	●	●	●
Limb-Girdle MD 0.8 – 6.9 per 100,000	●	●	●			
Myasthenia Gravis 20 per 100,000	●	●		●	●	●
Inflammatory Myopathy 14 – 25 per 100,000	●		●		●	●
Polymyositis 2.7 – 70 per 100,000	●	●	●		●	●
Pompe disease 2.5 per 100,000	●	●	●	●	●	●

Testing for Pompe Disease (Acid Maltase Deficiency) is available through a number of laboratories. If you suspect Pompe Disease talk to your doctor about testing options available to you.

*CK = Creatine Kinase (a marker of potential muscle damage)



progress now

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

Each of MDA's 34 new grants will impact neuromuscular disease research in different ways – from better understanding the underlying mechanisms of a particular disease to uncovering therapeutic targets to building clinical research network infrastructures that will expedite clinical trials.

MDA Research in Action

MDA Awards 34 New Research Grants

Research projects seek to further our understanding of neuromuscular diseases and aid the development of treatments

In August, MDA announced the award of 34 new grants totaling \$9.9 million for the Summer 2018 grant cycle. These new grants represent our continued commitment to funding groundbreaking research that will one day lead to treatments and cures for the diseases in our program.

Of note in this latest round of funding are two grants awarded to set up clinical research networks for facio-scapulothoracic muscular

dystrophy (FSHD) and limb-girdle muscular dystrophy (LGMD), respectively. Additionally, a research infrastructure grant was awarded to fund the development of computational tools that can integrate and analyze complex amyotrophic lateral sclerosis (ALS) data sets.

This round of funding also includes 24 research grants awarded to established, independent investigators; five development grants awarded

to investigators at the beginning of their careers and who are on the brink of becoming independent investigators; one clinical trial travel grant to help alleviate the financial burden on individuals and families traveling to participate in clinical research; and one clinical research training scholarship to a physician in the early stage of his or her medical career who is interested in pursuing a career in clinical

neuromuscular research.

The Summer 2018 grant cycle builds upon decades of MDA-backed research that has led to several FDA-approved treatments and a clinical pipeline of therapies more robust than ever before, in addition to providing funding for novel discovery research.

 **Read more about these exciting new grants at strongly.mda.org.**

Amyotrophic lateral sclerosis (ALS)

REFALS ALS Study

Researchers will assess effects of levosimendan on respiratory function



Researchers at Orion Pharma are looking for people with ALS to participate in REFALS, a phase 3 study designed to assess the effects of oral levosimendan (ODM-109) on respiratory function in individuals with ALS.

Study participants will be randomized to receive either levosimendan or placebo, and total study duration for each patient will be approximately 48 weeks. At eight required clinic visits, participants will undergo tests to assess respiratory function. Patient function will be evaluated using the ALS Functional Rating Scale Revised (ALSFRS-R).

Participants taking riluzole and/or edaravone may qualify for enrollment, but the dose must be stable at least four weeks prior to the screening visit, and treatment may not be changed or started during the study.

To be eligible to participate, individuals must have an ALS diagnosis, and 12 to 48 months must have passed since symptom onset. In addition, participants must:

- > Be able to obtain a full electromyogram (EMG) report consistent with ALS
- > Be able to swallow treatment capsules throughout the duration of the study
- > Meet defined respiratory and additional study criteria

Throughout the study, participants will continue to see their regular doctor for routine care.



To learn more about this study, visit clinicaltrials.gov and enter NCT03505021 in the "Other Terms" search box.

Analyzing ALS Data

MDA and Answer ALS team up to enhance understanding of disease mechanisms

MDA has awarded a research infrastructure grant totaling \$550,000 to Jeffrey Rothstein, M.D., Ph.D., at Robert Packard Center for ALS Research, Johns Hopkins University School of Medicine. Answer ALS is a nationwide consortium assembling one of the most comprehensive clinical, genetic, molecular and biochemical assessments of ALS to date.

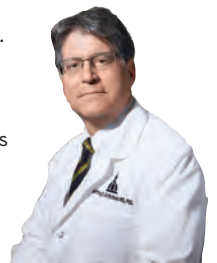
The Answer ALS and MDA partnership will develop a series of computational tools, known as probabilistic graphical models (PGMs), that can integrate and analyze complex ALS disease networks, identifying and quantifying changes in cellular pathways.

Through the Answer ALS program, 1,000 ALS patients across the United States will be monitored for one year. Disease measures such as breathing function and muscle strength will be captured at clinic visits, but participants will also use an app that will allow them to track disease progression at home.

Biospecimens such as blood and cerebrospinal fluid will also be collected, allowing for analysis of DNA, RNA, protein and cellular pathways, as well as for generation of stem cell lines from ALS patients used to model "disease in a dish."

The comprehensive Answer ALS dataset will generate more than 20 trillion datapoints, and MDA's grant will fund the development of new data analytics approaches to support the integration and analysis of this massive amount of data, which will be shared openly with the global research community.

The overarching goal of the program is to better understand and ultimately end ALS.



Jeffrey Rothstein, M.D., Ph.D.



Read more about this exciting new project at strongly.mda.org.

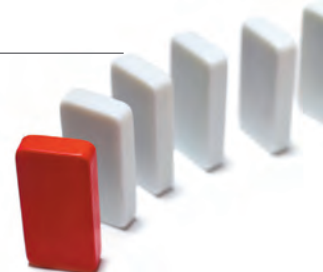
Charcot-Marie-Tooth disease (CMT)

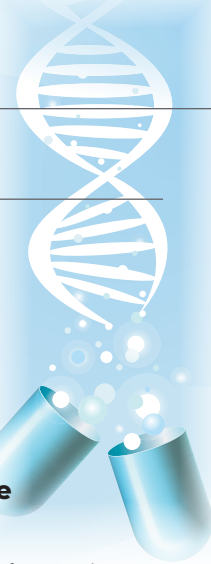
MDA Grant Boosts CMT Biomarker Development

\$1 million investment aims to speed CMT clinical trials

MDA awarded a human clinical trial grant for development of a critical biomarker for CMT to Mary M. Reilly, M.D., at UCL Institute of Neurology and National Hospital for Neurology and

Neurosurgery in London, England. The investment, totaling \$1 million, will fund Reilly's work to evaluate a new MRI protocol designed to detect disease-related changes in muscles over time >





in children with CMT1A, as well as in the other three most common types of CMT: CMT1B, CMT2A and CMTX.

Currently, one of the major barriers to developing safe and effective treatments for CMT is a lack of outcome measures, or biomarker tests that can accurately reflect the progression of disease during a short period of time. Because CMT typically progresses slowly, it can be difficult for scientists to detect drug effects in clinical trial participants within the relatively short duration of most clinical trials.

With colleagues, Reilly aims to develop a new sensitive outcome measure that will allow researchers conducting clinical trials to reliably detect any positive effects of a candidate treatment within a one- to two-year time frame.

If successful, the project could shorten the length of clinical trials, reduce the number of individuals needed to participate and hasten drug development for CMT.

Duchenne muscular dystrophy (DMD)

Encouraging Results in DMD Gene Therapy Trial

Gene therapy addresses genetic cause of disease

Sarepta Therapeutics has reported encouraging preliminary results from a phase 1/2a gene therapy trial designed to assess the investigational drug AAVrh74.MHCK7. micro-dystrophin in boys with DMD.

After 90 days in the trial, the first three participants all showed robust expression of micro-dystrophin – a shortened version of the protein that is absent in DMD-affected muscles. Participants also showed a decrease in levels of serum creatine kinase (CK), an enzyme biomarker strongly associated with muscle damage caused by DMD.

Principal investigator Jerry Mendell, M.D., at Nationwide Children's Hospital in Columbus, Ohio, in collaboration with Louise Rodino-Klapac, Ph.D., developed AAVrh74.MHCK7.micro-dystrophin specifically for DMD. The drug is designed to address the genetic cause of the disease via the delivery of highly miniaturized micro-dystrophin replacement genes that enable production of a functional protein



Learn more about this exciting new project at strongly.mda.org.

Aqua Creek Products
THE LEADERS IN RECREATION, FITNESS, AND ABILITY

THE SCOUT 2 LIFT

UL CERTIFIED

f t p in

1.888.687.3552
www.aquacreek.com

Granny Jo Products
For Dignity and Independence

wrap yourself...


...in comfort

Visit us online at
www.GrannyJoProducts.com
or call 863-698-9130

to substitute for the dystrophin missing in people with DMD.

Since results were announced, Sarepta's trial was put on clinical hold due to a manufacturing issue. No safety issues were reported for participants in the trial, and Sarepta submitted plans to address the U.S. Food and Drug Administration (FDA) concerns. In September, the FDA lifted the clinical hold, allowing the trial to resume.

Of note: Two other gene therapy trials are underway for DMD. Pfizer currently is testing PF-06939926 in a phase 1 trial, and Solid Biosciences is testing SGT-001 in a phase 1/2 trial. Interim results have not yet been made available for either study.

 **For more information, visit clinicaltrials.gov. Enter NCT03375164 in the "Other Terms" search box for Sarepta's AAVrh74.MHCK7.micro-dystrophin trial. Enter NCT03362502 to learn about Pfizer's trial and NCT03368742 to read about Solid Biosciences' trial.**

Translarna Phase 3 Study

Boys with a genetic DMD diagnosis may be able to participate

Researchers at PTC Therapeutics are looking for individuals with DMD to participate in a phase 3 study to evaluate the safety and efficacy of ataluren (brand name Translarna).

Translarna is designed to promote the formation of full-length and functional dystrophin protein in boys with a nonsense (premature stop codon) mutation. Trial participants will be randomized to receive either ataluren or placebo to determine whether treatment with ataluren is associated with stabilized or slowed disease progression.

Total study duration for each patient will be approximately 144 weeks. At 12 clinic visits (one every three months), disease progression will be assessed with a series of functional tests including a six-minute walk test, timed function tests, the North Star Ambulatory Assessment and magnetic resonance imaging (MRI), as well as blood tests and physical examination to monitor for adverse events.

In order to be eligible to participate, individuals must be male, 5 years or older, have a genetic diagnosis of DMD caused by a nonsense mutation in the DMD gene and meet additional study criteria.

Throughout the study, participants will continue to see their regular doctor for routine care.



 **To learn more or to inquire about participation, contact Mary Frances Harmon at 908-912-9256 or mharmon@ptcbio.com.**

obi

The first dining robot of its kind.



EFFECTIVE FOOD CAPTURE

Obi's patented features allow for optimal and efficient food delivery



SIMPLISTIC OPERATION

Obi can be operated by any part of the body that can activate an accessibility switch



DINING FREEDOM

Obi is battery operated and weighs only 7 lbs., so it's perfect for dining on the go!

Ask us about our
FREE trial program!



844-435-7624
www.MeetObi.com

Duchenne muscular dystrophy (DMD)

Ezutromid Development for DMD Discontinued

PhaseOut DMD trial data showed no clinical benefit

Summit Therapeutics has announced that it is discontinuing development of ezutromid for DMD, following reports that primary and secondary endpoints were missed after 48 weeks of treatment in the company's PhaseOut DMD trial.

PhaseOut DMD was a phase 2, multicenter, open-label clinical trial of ezutromid, a utrophin-modulation therapy. Thirty-eight boys enrolled in the trial completed the 48-week regimen.

Although well-tolerated, ezutromid did not provide the desired therapeutic effect in trial participants. As a result, Summit is winding down the study and focusing efforts on other therapeutic areas going forward.



If your child participated in PhaseOut DMD, you should contact their study doctors for more information.



Facioscapulohumeral muscular dystrophy (FSHD)

MDA Grant Supports FSHD Clinical Research Network

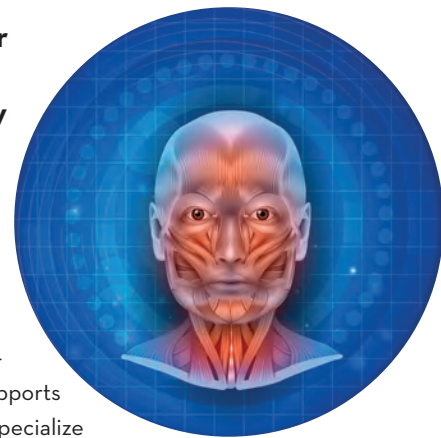
Network aims to spur advances in FSHD research and therapy development

MDA awarded a clinical research network grant to develop and maintain a core FSHD Clinical Trial Research Network (CTRN). The investment, totaling \$1.2 million, supports seven medical centers that specialize in FSHD research and clinical care and is targeted to spur advances in FSHD research and speed the development of new therapies.

Recent breakthroughs in research include the identification of the underlying molecular cause of FSHD: the abnormal activation of a gene called DUX4, which normally is silenced or inactive. Armed with this knowledge, a number of drug companies have begun working to develop and test targeted therapeutic strategies to treat FSHD. Consequently, an urgent need exists to establish the tools that will be necessary to execute high-quality clinical trials quickly and efficiently.

Successful FSHD clinical trials depend on several factors, including the ability to recruit patients, patient access, a precise understanding of the natural history of FSHD and the major contributors to its variability, and reliable outcome measures that are sensitive to change in FSHD.

MDA funding will enable the FSHD Clinical Trial Research Network to build a team of trained clinical evaluators, all working with standardized approaches, and develop regulatory strategies optimized to streamline drug trials.



Learn more about this exciting new project at strongly.mda.org.

OPEN Your Door to Independence

Now you can open your own door to independence with the new, residential door opener from Power Access. Empower all the doors in your home or apartment, from front to interior, with this easy-to-install, lower-cost unit. Use our convenient remote control, or use ECU, sip, puff & other controls to effortlessly access your space.

800-344-0088
handicap-access.net

POWER ACCESS
AMERICA'S DURABLE DOOR OPENER™





QUANTUM® 4FRONT®

The number one solution for
traction and performance.

Test drive the 4Front® today.

FEATURING
SMOOTH RIDE
SUSPENSION



- AVAILABLE WITH 10" POWER ADJUSTABLE SAFE SEAT ELEVATION AT UP TO 3.2 MPH
- CASE (CASTER ANGLE SENSOR ENCODER) TECHNOLOGY
- SUPERIOR, AUTOMOTIVE-GRADE SRS (SMOOTH RIDE SUSPENSION)
- LED FRONT AND REAR FENDER LIGHTS



QUANTUM®

(US) 866-800-2002

(Canada) 888-570-1113



Limb-girdle muscular dystrophy (LGMD)

MDA Funds Creation of LGMD Clinical Research Network

Network aims to accelerate treatments for LGMD

MDA awarded a clinical research network grant to Nicholas Johnson, M.D., at Virginia Commonwealth University in Richmond, to establish the Limb-Girdle Muscular Dystrophy Clinical Research Network.

The \$700,000 investment supports seven U.S. care centers with significant expertise in LGMD research and clinical care and is targeted to facilitate the development of tools and infrastructure needed to efficiently and effectively conduct

clinical trials and accelerate treatments for LGMD.

Therapy development for LGMD could significantly benefit from rich natural history data to describe the clinical course of the different forms of LGMD, as well as from validated clinical outcome assessments that can be used to determine whether a drug has provided treatment benefit.

An initial step in the development of the network infrastructure is to

conduct a longitudinal study of candidate clinical outcome assessments and develop disease-specific patient-reported outcomes. Investigators will initially focus on four different genetic LGMD subtypes and will collect DNA, genetic diagnosis and clinical information to help characterize the natural course of the disease.

Through the coordinated activities and enhanced communication among this



Nicholas Johnson, M.D.

new network of LGMD clinics, Johnson and colleagues aim to standardize approaches and develop clinical outcome assessments for use in future clinical trials.



Read more about this exciting new project at strongly.mda.org.

Savant Headrest

i symmetric designs
www.symmetric-designs.com | **tf 1.800.537.1724**

DO YOU WANT RELIEF FROM...

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

AUTOMATED BODY ROTATION
 PROGRAMMABLE TIMES AND ANGLES
 SMOOTH AND SILENT OPERATION

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

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

IR CONTROL OPTION AVAILABLE!

Left Rotation Right Rotation

PRO BED
Medical Technologies Inc.

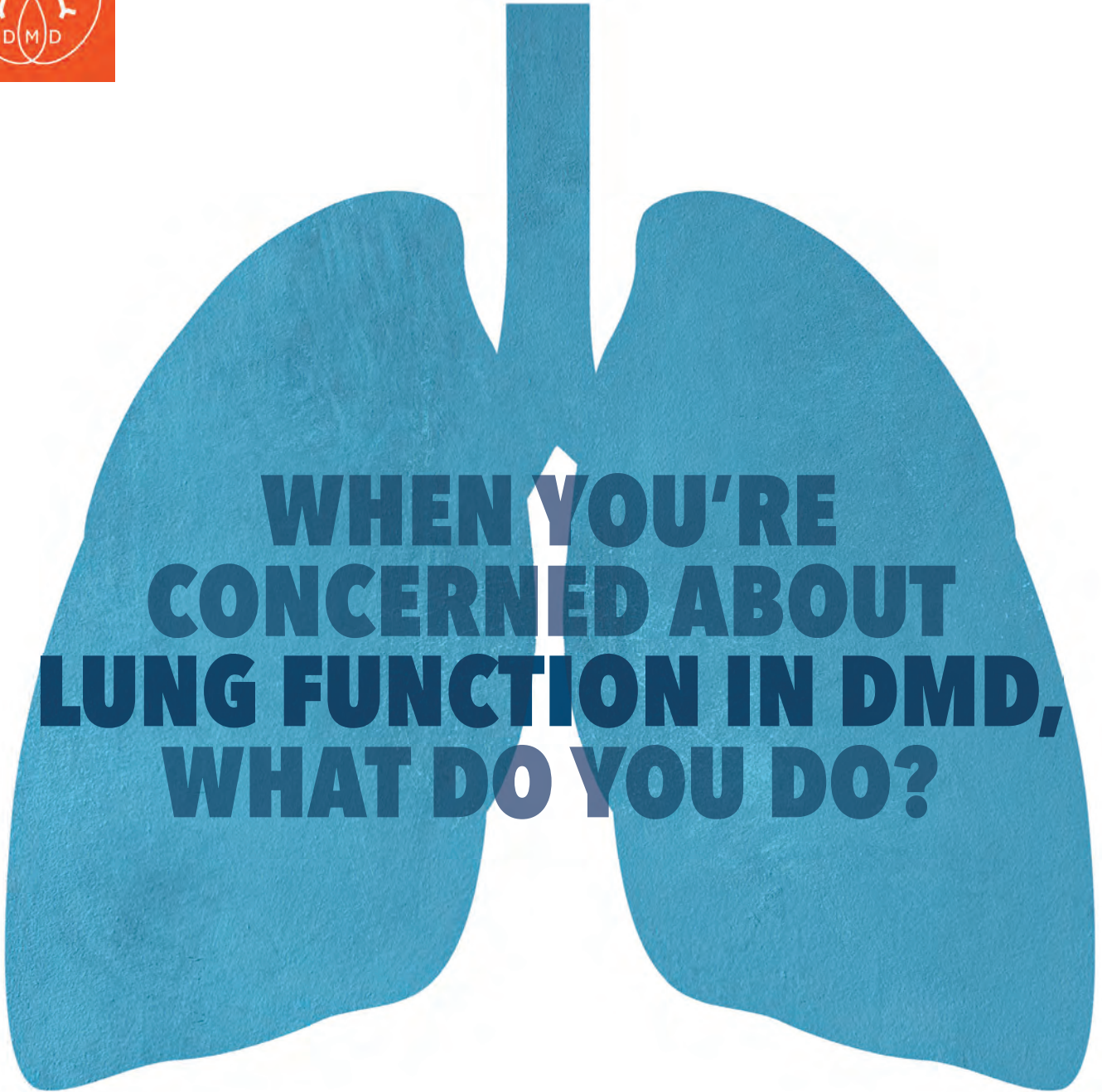
“ I am writing to help promote the awareness of the FREEDOM BED within the Muscular Dystrophy Community. If our son’s story can help families get the bed that will change their lives it will be worth it. The FREEDOM BED is heaven sent! It has dramatically improved our lives and allows the whole family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

The Azar Family

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



ADVERTISEMENT



**WHEN YOU'RE
CONCERNED ABOUT
LUNG FUNCTION IN DMD,
WHAT DO YOU DO?**

First, take a breath.

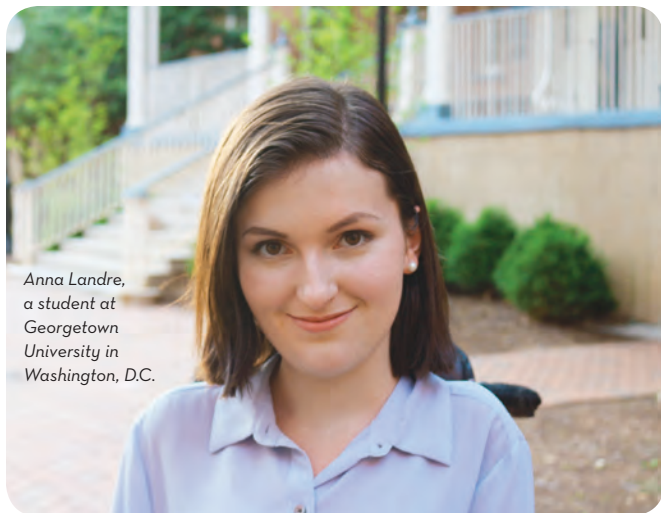
Lung function is important. So is feeling supported. Having access to respiratory information can make you feel empowered. On TakeabreathDMD.com, you can read about ways people with Duchenne muscular dystrophy (DMD) can help manage their lung function and their well-being. We'll provide helpful information and keep you informed about the news in DMD.

Visit TakeabreathDMD.com for respiratory information
and to sign up for news updates.

Dollars for Scholars

Scholarships help lessen the financial load of higher education. Here's how students with neuromuscular disease find and win them.

BY CHERYL ALKON



Anna Landre, a student at Georgetown University in Washington, D.C.

The cost of higher education has soared over the past two decades. According to U.S. News & World Report, in-state tuition and fees at public universities have increased 243 percent, while they have jumped 168 percent at private institutions.

For many prospective college students, winning scholarships is key to making education more affordable. The trick is thorough research. There are lots of scholarships out there, many of which are earmarked for individuals with particular backgrounds, students applying to particular schools, students pursuing particular fields of study and myriad other considerations. The best advice for students seeking financial assistance? Think broadly about how your situation applies to scholarships' requirements and, above all, don't be afraid to apply.

BE CURIOUS

Cody Schoppe of Cameron, Texas, found a perfect fit in an unexpected place. Sarepta Therapeutics, a medical research and drug development company that focuses on Duchenne muscular dystrophy (DMD), established the Route 79 Duchenne Scholarships program earlier this year, and Schoppe is one of the first recipients.

Francesca Nolan, director of investor relations and corporate communications for Sarepta, says the scholarship applicants who stood out were authentically themselves and vastly curious, and they sought to both inspire others and be inspired by the world around them.

In his application, Schoppe, a 21-year-old who lives with DMD, wrote about Project Maynardstang, an effort he started in 2011 to balance his love of the Ford Mustang and his dedication to raising awareness of neuromuscular disease. ("Maynard" is Schoppe's nickname.)

His family's 1993 Mustang, Ugly Betty, "is loud, and it's mainly to cruise around and show off in," Schoppe says. The car is painted black on top and red below, bisected by a bright blue line to represent muscular dystrophy awareness. Schoppe and his father take Ugly Betty to cruise-ins and car shows, where he sells Project Maynardstang merchandise and takes donations, giving all proceeds to MDA.

The Route 79 Scholarship is helping to support Schoppe as he studies website design at Temple College in Texas. He hopes to make Project Maynardstang a full-time job in the future.

BE REAL

Nikaela Losievski, 22, of Granger, Ind., will graduate from Michigan State University in December with a bachelor's degree in science with a concentration on neuroscience, and she is applying for doctorate programs in cell biology and neuroscience. She plans to teach and do research on spinal muscular atrophy (SMA), which she was diagnosed with at 16.

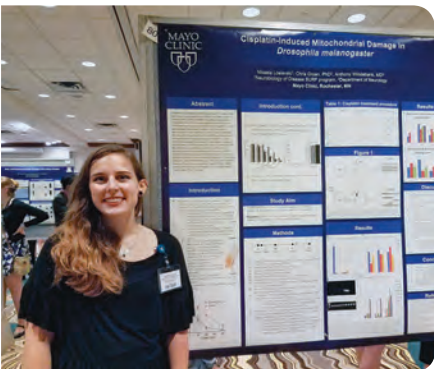
Losievski won several academic and other scholarships, including one from her elementary school and one for students from St. Joseph County, Ind., and she feels that writing candid essays was key.

"I encourage people to take every opportunity to get involved in everything," she says, noting that participating in extra-curriculars in high school gave her experiences to write about. She also advises not shying away from the realities of life with a neuromuscular disease, even if it means writing about missing tests because of doctor



Cody Schoppe, a student at Temple College in Texas

Nikaela Losievski, a student at Michigan State University



appointments or how pain makes it hard to study.

“Show how having a disability is a disadvantage,” she says. “With muscular dystrophy, a lot of it is about energy — we don’t have the capacity, so it’s easy to tire out.”

While your essay should reflect you, looking to others for help can give you an edge. “Don’t just do an essay and submit it; I had multiple people edit my stuff,” she says. That extra feedback helped her strengthen her scholarship applications.

THINK OUTSIDE THE BOX

Look for scholarships that focus on diversity, says Anna Landre, 19, a sophomore at Georgetown University’s School of Foreign Service majoring in regional and comparative studies.

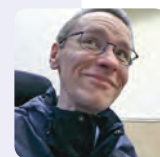
Landre, who lives with SMA, was valedictorian of her high school. But because Georgetown doesn’t award academic scholarships, Landre searched online for scholarships and won several, including one from her high school, one from the Lep Foundation and one from her local Division of Vocational Rehabilitation in Ocean County, N.J.

TOP TIPS FOR APPLICANTS

Scott Hatley, 39, is executive director of Incight (incight.org), a Portland-based non-profit that helps people overcome barriers to education, employment and independence. Living with Duchenne muscular dystrophy (DMD), Hatley won scholarships for college, and today he helps decide which candidates receive Incight scholarships, which range from \$500 to \$2,500.

Hatley has some advice for scholarship applicants:

- > **Start early.** “Some applications take a lot of effort.”
- > **Broaden your search.** “People don’t realize how many scholarships are out there, such as from the local Kiwanis, Rotary Club, your own high school, community foundations and so on.”
- > **Be focused.** “It helps to have a core focus or goal for what you want to pursue, so you can find the right scholarships for the path you are taking.”



Landre is thinking about working with the United States Foreign Service after graduation. She recommends taking a broad approach when considering scholarship applications.

“If a scholarship is looking for diversity, disability is diversity — it’s not just race or religion,” she says. “People shouldn’t be afraid to use their experience as a person with a disability to empower them to get their education. Talk about the ways that your life has been challenging, but always end in the ways that you have grown and been able to accomplish things.” [Q](#)

Cheryl Alkon is a freelance writer based in Massachusetts.

Education Resources

Find resources to help you choose, prepare for and pay for higher education at mda.org/young-adults/education.



INNOVATIVE PRODUCTS FOR SENIORS AND PEOPLE WITH DISABILITIES

Take it with you. Not ON you.

Carry your phones, keys, documents and many personal items in NuprodX easy to-access cases. No more bags in your lap or searching for lost items!

The HOLSTER



The CADDY



Now your phone is always at hand: Swivel mount keeps it out of the way, and swings out for easy access when you need it. Don’t miss another call!

Keep your keys, wallet, important papers—even your iPad® with you. Easy slide-out mount keeps it out of the way except when you need it.

Visit our web site for complete information about our products.
nuprodX.com
 855-221-5171

Available at 

*Current and former
caregivers find new
career paths that
tap into their life
experiences*

BY DONNA SHRYER

OPENING



Three mornings a week, Karen Toenniss, 50, grabs the car keys and dashes off to her part-time position as coordinator for the MDA ALS Care Center at Houston Methodist Hospital. Nestled in her heart, she carries her husband's memory. "Every time I save someone even a little bit of frustration as they deal with ALS, it feels like Mike is with me," she says. >>





Karen Toeniss, right, gained a passion for ALS while caring for her late husband, Mike, center.

Taking the reigns as the clinic's coordinator wasn't a planned career move, although the opportunity did acknowledge Toeniss' passion, which grew from her life experience. "I left my job as a neonatal and pediatric intensive care unit nurse in 1995, two years after my husband Mike was diagnosed with ALS. I was his caregiver until he passed in 2006," Toeniss recalls. "In 2007, I thought, 'What am I supposed to do now?' I had a 12-year gap in my resume."

That's when a door swung open. Stanley Appel, M.D.,

co-director of the Houston Methodist Neurological Institute and Mike's neurologist, phoned Toeniss. "Dr. Appel asked me to run his ALS clinic," Toeniss says. "I had no experience managing a clinic, and I wasn't planning a new career based on my years as Mike's caregiver. But Dr. Appel said, 'I can teach you how to run a clinic; I cannot give you the passion you have for ALS.' That was 11 years ago."

FITTING THE PIECES TOGETHER

Not everyone's professional journey is like Toeniss', with a door fortuitously opening at just the right moment. More often, if your caregiving experience beckons you to follow a new career, you'll need to open your own doors. A good place to begin is with three key questions, suggests Kerry Hannon, an expert on career transitions and author of "Great Jobs for Everyone 50+." Your answers will signal if the time's right to make a change.

1. Are you physically fit?

"When we devote a large part of our life to caring for another, we sometimes forget to take care of ourselves. Potential employers want to see that you have the energy and endurance to get the job done," Hannon stresses. Regardless of how strong your desire is to pursue a new calling, you may need to step back and buck up your stamina and health. Grant yourself this time.

2. Are you financially fit?

Do research on typical salaries in your chosen field or organization. Hannon suggests

FILL THE GAP

When you've been a caregiver for a loved one, you probably took time away from active employment. But don't think of it as a gap in your résumé, urges Kerry Hannon, a career transitions expert. Instead, think of it as time when you developed new skills that will help you in the professional world.

Here are two strategies for building a solid résumé:

1 EXPRESS EXPERIENCES AS PROFESSIONAL SKILLS.

"Your mission as a job seeker is to drill down into the business side of caregiving," Hannon says. That means translating your duties into business terms whenever possible. For example, you didn't help a child get a wheelchair; you served as a patient advocate, spearheading a complex negotiation process to secure needed equipment.

At different points during your caregiving experience, you were likely also a project manager, financial manager and personnel director as you hired and oversaw caregivers and medical experts.

2 EMPHASIZE ACTIVE LEARNING.

"Employers want someone who's not stuck in their ways and is an active learner," Hannon says. That makes education and certifications strong résumé features – but look deeper, Hannon urges. People with experience in caring for someone with a neuromuscular disease likely have many experiences that count as active learning. For example, although John Mower left his job as a high school counselor in 2012, the years when he was full-time caregiver to his wife, who had ALS, were packed with impressive active learning.

"While I cared for Robin, she and I were heavily involved with MDA and the ALS Association. We raised funds for research, Robin spoke at MDA gatherings, we attended ALS walks and we spoke about ALS to first-year medical students at the University of Toledo Medical Center," John says. "For five years, we traveled across the United States in a motor home, raising awareness, attending ALS-related events and helping others diagnosed with ALS."

Mower's experience demonstrates an adaptability and willingness to learn that go well beyond classroom walls. After all, showing can be much more effective than telling.

More often, if your caregiving experience beckons you to follow a new career, you'll need to open your own doors.

writing down a budget. "You want to be financially nimble, so you can do the job you want to do," she says.

3. Are you spiritually fit?

"This takes serious soul searching — especially when you're coming off of years dedicated to caring for a loved one," Hannon says.

And don't be surprised if your spiritual fitness takes

sudden twists and turns, as John Mower, 56, explains. Mower's story began in 2009, when his wife, Robin, was diagnosed with ALS. At first, Mower worked as a high school counselor by day and cared for his wife by night. In 2012, he retired to care for Robin full-time. After his wife passed in 2017, John felt a need to reach out to other families struggling with ALS — but then suddenly it became too painful.

"I know I can help people with neuromuscular disease, but after everything I went through, I suddenly wasn't sure if I could be there emotionally," John says.

If John chooses to return to the ALS community, either professionally or as a volunteer, Hannon strongly recommends that he — or

CAREGIVING AND EMPLOYMENT IN THE UNITED STATES




Approximately **43.5 million** caregivers provide unpaid care to an adult or child.

More than **75 percent** of all caregivers are female, and they may spend as much as 50 percent more time providing care than men.

 About **17 percent** of full-time workers act as caregivers.

39 percent of caregivers leave their job to have more time to care for a loved one. 

 **34 percent** of caregivers leave a job because their work does not provide flexible hours.

Sources: National Alliance for Caregiving, AARP, Institute on Aging, Gallup-Healthways

ADVERTISEMENT

HOPE-2

DUCHENNE CLINICAL TRIAL

Currently Enrolling



 Capricor

Visit ClinicalTrials.gov (NCT 03406780)
and hope2trial.com for more details

 @Capricor   @CapricorTherapeutics

www.capricor.com



Jennifer Shumsky (right), pictured with her family, was happy to find a new career helping other Duchenne families.

anyone in a similar situation — makes time in his life to take care of his spiritual fitness. “It can be religion, meditation, yoga, long walks with your dog, or anything that keeps you centered and replenishes your soul.”

RETHINK YOUR RÉSUMÉ

Jennifer Shumsky is a primary caregiver for her 17-year-old son, Xavier, who was diagnosed in 2006 with Duchenne muscular dystrophy (DMD). She also is a Personalized Access Coordination Team manager at the Little Hercules Foundation Duchenne Family Assistance Program, a nonprofit dedicated to improving the lives of those diagnosed with DMD.

Shumsky’s path to this position meant a career

change, and to promote herself she needed to take a fresh look at her professional skills.

Shumsky began as a registered nurse (RN) and eventually joined a medical management company in Minnesota, where she served as an RN reviewing claims and prior authorization requests. In 2016, the family moved to Ann Arbor, Mich., and Shumsky set out to find a job.

Writing a résumé posed a dilemma. She did not want to diminish her career as a nurse, but she did not want to remain in nursing.

“During my years with the medical management company, I learned so much that helped me as a Duchenne parent,” Shumsky recalls. “I wanted to help families that didn’t have my knowledge base.”

Shumsky revised her résumé to put less emphasis on nursing and more emphasis on the skills she learned, including a keen understanding of the equipment, medications, insurance and appeals specific to Duchenne families.

The tactic worked, and as Shumsky says, “I’m finally able to put all my skills together and use them to help families living with Duchenne.”

FOLLOW YOUR PASSION

Sometimes it feels as if your path is chosen for you. If caregiving helped you find a new focus for your life, that deserves top billing in your résumé and during interviews.

Take 42-year-old Erin Hill, who is area director of MDA South Dakota. Her career choice evolved after her father was diagnosed with ALS in 2011.

“I shared caregiving with my family, and after Dad passed away in 2012, I felt a pull to help the neuromuscular disease community. I wasn’t ready to stop fighting back against ALS,” Hill says.

As Hill settled into her position, she met several co-workers who had also lost someone to neuromuscular disease. “Like me, they were pulled to MDA, so they could continue the fight,” she says.

Nearly four years into her job, Hill says her drive has only grown stronger. “My goal is to help raise the research funds so we can find a cure as soon as possible. Then I’ll be out of a job!” she says with a chuckle — followed quickly with a heartfelt sigh. “I can’t wait for that day.” **Q**

Donna Shryer is a freelance writer in Chicago.



Erin Hill enjoys spending time at MDA Summer Camp as part of her role with MDA South Dakota.

ADVERTISEMENT

A close-up photograph of two hands, one from a child and one from an adult, gently holding a small green seedling with two leaves as it is planted into dark brown soil. The background is softly blurred, showing more soil and another seedling in the distance.

IMAGENE IF

GENE REPLACEMENT THERAPY COULD HELP CHANGE LIVES.



GENE REPLACEMENT THERAPY is changing the way we see genetic diseases. By targeting faulty or missing genes, this innovation is creating a new world of opportunities and potentially helping people living with genetic diseases.

Discover more about this scientific advancement at [ExploreGeneTherapy.com](https://www.ExploreGeneTherapy.com)

HOW DRUGS ARE DEVELOPED

It takes a lot of time and effort for a new drug to go from the scientist's lab to the pharmacy shelf. Here's the process in a nutshell.

BY AMY MADSEN



Have you ever wondered what has to happen for the scientific community and pharmaceutical industry to develop a new drug and get it on the market, where physicians can prescribe it to treat a neuromuscular disease?

What does it cost? How long does it take?

Estimates vary, but it's safe to say that, on average, out of 10,000 experimental compounds, one drug may make it to pharmacy shelves, and it will take at least 10 years and more than \$1 billion to get it there.

Time and costs are directly associated with the number of steps involved in the drug development process — all of which help ensure that treatments that reach the market are safe and effective.

STEP 1: DISCOVERY

Researchers first need to identify the underlying gene alterations or other causes of a disease to know where to start in developing a therapy. This can be a long and complex process involving, for example, studying tissues from patients living with the disease, creating lab mice that model the disease, and identifying and validating targets to see how modulating those targets alters the disease course.

While thousands — or tens of thousands — of compounds may be possible candidates for developing into a drug, early testing helps researchers determine which are most promising and worthy of further study.

In a process called optimization, each compound, or series of compounds, is tested until the developers decide that they have the optimal compound selected — the

one with the best chance of working and the lowest chance of causing side effects. This compound is called the development candidate.

STEP 2: PRECLINICAL DEVELOPMENT

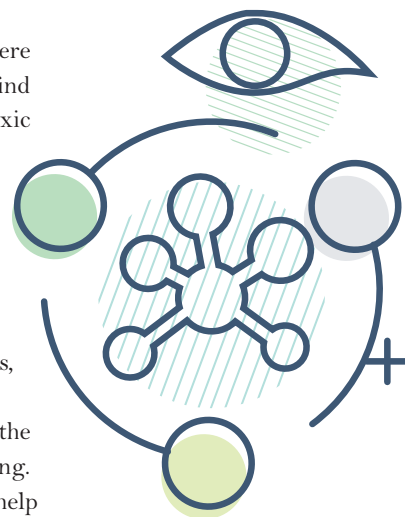
During this time, researchers conduct a series of tests on the development candidate to assess its effectiveness and toxicity (the potential to cause serious harm). These tests include:

- Animal models of disease to assess whether the drug is able to alter the disease course. These studies provide early insights on whether the drug might work in people.
- Absorption, distribution, metabolism and excretion (ADME) studies to assess a compound's stability, what happens if it degrades, where it goes in the body and what happens to it once it's there. If a compound fails ADME studies, researchers may return to optimization in order to try a different development candidate.
- Pharmacokinetics and pharmacodynamics studies to investigate what happens to the drug after delivery and

help determine how much drug should be given (dosage) and how often. These studies typically are done in animal models, and there are calculations that predict how to convert the amount of drug given to an animal to the amount that should be given to humans.

- Toxicology studies, where developers attempt to find an upper dose that is toxic to determine how high a dosage they can use in people. Sometimes, signs of a side effect may not be recognized until a drug is administered to patients, but toxicology studies are designed to reduce the chances of this happening.

Results of these tests help developers determine how a drug will be administered so that it gets to the specific area of the body where it is needed and at the right concentration. They also help developers determine the formulation, or how the drug is made. In addition to the active ingredients, the formulation could include packing components to turn it into a pill, liquids to turn it into a fluid, or other components that help the drug





For his work to develop Spinraza, Adrian Krainer was recently awarded the 2019 Breakthrough Prize in Life Sciences.

get to where it needs to go or disperse through the tissues.

At the preclinical stage, developers have to be able to show that they can make exactly the same drug substance every time, even when they make much larger quantities. Pharmaceutical companies must demonstrate

that they have a robust protocol to make their drug and the ability to make enough uniform drug for clinical trials.

STEP 3: CLINICAL TRIALS

Clinical trials are experiments done in humans to see if a potential drug is safe

and effective. They are very different from routine medical care (including prescribed use of drugs approved by the U.S. Food and Drug Administration, or FDA), because it's unknown exactly how the treatment will affect people.

There are three stages of clinical trials, from early, small-scale, phase 1 studies to late-stage, large-scale, phase 3 studies.

As developers design a clinical study, they will consider what they want to accomplish for each of the different clinical research phases, and they begin the Investigational New Drug (IND) process.

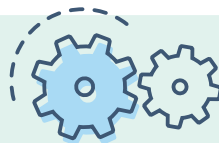
The FDA's primary concern is to protect trial participants. The agency will require that the developers begin testing the drug at very low doses to look for any signs of toxicity, often in healthy volunteers instead of people with the disease, before moving up to doses that might be effective.

The FDA also makes sure that the trial is well-designed, so as to have the best chance of yielding useful information about the drug with minimal risk to the trial participants.

Drug developers must submit an IND application to the FDA before beginning clinical research. Included in the application are animal study and toxicity data, manufacturing information, clinical protocols, data from any prior human research and information about the investigator.

The FDA IND review team has 30 days to review the IND submission. They may accept the IND and grant approval to begin clinical trials, or they may issue a clinical hold to >

SPINRAZA: THE STORY BEHIND A LIFE-CHANGING DRUG



Adrian Krainer, Ph.D.

Adrian Krainer, Ph.D., St. Giles Foundation professor and program chair of Cancer & Molecular Biology at Cold Spring Harbor Laboratory in New York, led research to develop Spinraza for spinal muscular atrophy (SMA). MDA funded foundational work in SMA and invested nearly \$750,000 in awards to Krainer for early-stage development of the breakthrough drug. Here, Krainer shares the story of Spinraza's development.

Spinraza's entire discovery, development and approval process took about 12 years, not counting previous foundational work on RNA splicing, antisense technology, SMA natural history and mechanisms, mouse models, clinical measures, etc.

In 2004 we began to collaborate with Ionis Pharmaceuticals to conduct a screen for antisense

oligonucleotides that increase correct splicing of SMN2. This was followed by a great deal of lead optimization, characterization of the mechanism of action and extensive testing in SMA mouse models.

We published the development candidate, which is the same as Spinraza, in 2008. Ionis then carried out rigorous ADME and toxicology studies, including in non-human primates, and selected intrathecal bolus administration [injection into cerebrospinal fluid] as the optimal route.

After the IND application was approved, the first-in-human dose was given in November 2011.

Ionis partnered with Biogen for the later clinical trials. These lasted about five years, until FDA approval came on Dec. 23, 2016. They involved hundreds of SMA patients and many clinical centers in multiple countries.

This may seem like a long time, but each trial needs to be rigorously designed, regulatory and institutional approvals have to be obtained, and

eligible patients have to be recruited, who don't all enroll at the same time. The later-stage trials have to wait for completion and evaluation of the earlier trials.

Overall, this was a highly efficient process, considering how many unknowns there were at the start of the trials: Could SMA be effectively treated after disease onset? Which SMA type(s) would be most likely to respond to treatment and show clear effects of the drug? How much drug should be given and how frequently for maximum efficacy within a safe range? What should be the primary outcome measures, and for how long should how many patients be treated before a statistically significant change in these measures might be observed?

Happily, the clinical studies were extremely well designed, and the two pivotal phase 3 trials achieved the efficacy endpoints a year ahead of schedule. The results were compelling, and as a result, FDA approval for the first SMA drug was granted in record time.

The Battery Matters



Trusted by leading power wheelchair manufacturers for 35 years.
When consistent quality, long-term reliability and value matter –

ACCEPT NO IMITATIONS.



800-372-9253 • MKBATTERY.COM

© MK Battery 2018

DRUG DEVELOPMENT IN A NUTSHELL



delay or stop the investigation if they think participants will be exposed to unreasonable or significant risk.

The developers are responsible for informing the review team about new protocols, as well as serious side effects seen during the trial. This information ensures that the team can monitor the trials carefully for signs of any problems. After the trial ends, researchers must submit study reports.

This process continues until the developers decide to end clinical trials or file a marketing application.

STEP 4: FDA REVIEW

If clinical trial data indicates a drug is safe and effective for its intended use, the pharmaceutical company files an application to market the drug. The New Drug Application (NDA) includes all known data about the drug and is intended to demonstrate that the drug is safe and effective for its intended use in the population studied. In addition to clinical results, the NDA contains proposed labeling, safety updates, drug abuse information, patent information, data from studies outside the United States, institutional review board compliance information and directions for use.

Once the FDA receives an NDA, the review team decides if it is complete. If it is not complete, the review team can refuse to file the NDA. If the NDA is accepted, the review team has six to 10 months to decide whether to approve the drug.

Often, the NDA contains sufficient data for the FDA to determine the safety and effectiveness of a drug. Sometimes, though, questions arise that require additional consideration. In these cases, the FDA may consult with one of its advisory committees to get independent, expert advice and to permit the public to make comments.

During the review process, the review team conducts a full analysis of the application; FDA inspectors travel to clinical study sites to conduct routine inspections for evidence of fabrication, manipulation or withholding of data; and the project manager assembles all reviews and other documents into an “action package,” the official record of the FDA review. The review team issues a recommendation, and a senior FDA official makes a decision.

Often, some issues need to be resolved before a drug can be approved. Sometimes, the FDA requires the developers

to address questions based on existing data. In other cases, the FDA requires additional studies. At this point, developers can decide whether or not to continue further development. If a developer disagrees with an FDA decision, there are mechanisms to make a formal appeal.

In cases where the FDA determines that a drug has been shown to be safe and effective for its intended use, the applicant must develop and refine prescribing information for it to be approved. This is referred to as “labeling.” Labeling accurately and objectively describes the basis for approval and how the drug should be used.

STEP 5: FDA APPROVAL

Although the drug may be made available on the market now, the true results of its safety and efficacy will be determined over its lifespan in the marketplace.

It may be frustrating to see potentially lifesaving drugs take so long to go from start to finish, but drug development is an intricate process conducted with safety in mind and everyone’s best interests at heart. [Q](#)

Amy Madsen is a science writer and frequent contributor to Quest.

Rooting for Research

MDA is a strong advocate for policies that advance biomedical research and ensure that safe and effective treatments are developed and delivered as quickly as possible. Learn about the policies and issues that impact our community and how you can get involved at mda.org/advocacy.

Q700M

SEDEO® ERGO™



BE SEEN.

FOR YOUR STYLE, YOUR CHARISMA,
YOUR CONFIDENCE.

Shawn H., Motivational Speaker

 QUICKIE

ADVANCED ENGINEERING.
CUSTOMIZED FOR YOU.
SUNRISEMEDICAL.COM

TAKE US TO THE



BY BRITTANY JOHNSON HERNANDEZ

Access to air travel should not be denied based on physical ability. Here's what MDA is doing about it.





CHECK IN



Every day, millions of people board flights bound for destinations across the United States and around the globe. Unfortunately, navigating sprawling airports and negotiating cramped planes makes traveling by air an uncomfortable experience for most passengers, and it can prove to be downright daunting for those with mobility challenges and other disabilities. >>





KNOW YOUR RIGHTS

Because of the Air Carrier Access Act (ACAA):

- ▶ You can't be refused a plane ticket because of a disability.
- ▶ You can't be required to sit in a certain seat or area of the plane because you have a disability.
- ▶ You are entitled to assistance boarding and making connections.

For more information, visit [transportation.gov/airconsumer/passengers-disabilities](https://www.transportation.gov/airconsumer/passengers-disabilities).

Many people living with diseases and conditions that limit mobility, like muscular dystrophy and ALS, avoid air travel altogether because of the constraints and shortcomings of the system, but every person should have the opportunity to travel by air, especially those who need to travel long distances for specialized medical care. MDA is working to ensure that air travel is a reality for those living with limited mobility, from business travelers to sightseers and everyone in between.

LEGISLATIVE SOLUTIONS

Many people do not realize that the protections of the Americans with Disabilities Act (ADA) do not extend to air travel and that this method of transportation is governed by another law entirely. Thirty-two years ago, the Air Carrier Access Act (ACAA) was signed into law by President Ronald Reagan. The ACAA prohibits discrimination against individuals with disabilities who travel on any airline operating in the United States. The

act's passage laid the groundwork for the ADA four years later. While the ACAA was a good start toward the goal of equalizing access to air travel, the current system still leaves much to be desired for travelers with disabilities.

MDA often partners with other organizations to boost our advocacy efforts, and the issue of accessible air travel is no exception. By joining forces with other groups representing disability communities, we have been able to amplify the voices of travelers with disabilities before Congress, the Federal Aviation Administration (FAA) and the airline industry. As Congress considers legislation regarding the FAA, MDA has taken a seat at the table to advocate for provisions that would make air travel easier for people living with neuromuscular diseases. One of these measures would create a diverse advisory committee that would include individuals from the disability community to investigate and report on the needs of all passengers with disabilities. If created, this committee would provide the FAA with valuable insight on the real-world travel requirements and constraints of people with disabilities. This type of direct input from the disability community would help shape policy going forward, and MDA will continue to urge Congress to pass this legislation.

MDA is also working on a more specific initiative to lay the groundwork for longer-term changes to the aviation industry that would make it easier to travel with assistive devices. Traveling with manual or power

wheelchairs can present many challenges to passengers, especially when the wheelchair must be checked in the airplane baggage compartment. Recent accounts of expensive, specialized mobility devices being nearly destroyed in the baggage handling process have no doubt discouraged many people from boarding an airplane. No one wants to find that their checked item has been damaged in flight, but when that item is an absolute necessity, like a wheelchair, the consequences can be life altering until the problem is resolved.

MDA is working with members of Congress on an initiative that would require the FAA to undertake a study on the use of in-cabin wheelchair restraints. If people could travel in their wheelchairs on the plane, instead of being required to check them, damages to equipment would likely be minimized and air travel would become a more pleasant experience. While the process of conducting a study and evaluating results may take some time, MDA continues to work toward this long-term goal. ▶

Traveling with manual or power wheelchairs can present many challenges to passengers, especially when the wheelchair must be checked in the airplane baggage compartment.

Be Informed

One way to improve your air travel experience is to know what resources and supports are in place to assist passengers with disabilities. MDA's Accessible Air Travel Resource Center helps you find important information about airports, airlines and your rights as a traveler. Visit [cqrcengage.com/mda/accessibleairtravel](https://www.cqrcengage.com/mda/accessibleairtravel).



The National ALS Registry: Get The Facts

The National Amyotrophic Lateral Sclerosis (ALS) Registry enables persons with ALS to fight back and help defeat ALS (Lou Gehrig's Disease). By signing up, being counted, and answering brief questions about your disease, you can help researchers find answers to critical questions.

Learn more at www.cdc.gov/als or (800) 232-4636



Who can sign-up?

Anyone with ALS

No computer? Don't worry! A family member, caregiver or friend with a computer can help you. You can also contact your local ALSA chapter or use the computer at your public library



What do I need?

- A computer with an internet connection
- An email address



What if I need help?

Caregivers and others can help you in person or even over the phone



What kind of information is collected?

- Basic demographics (e.g., age, sex, height, weight)
- Military history
- Physical activity
- Family history



Do I need to update my information?

YES! Every six months – you'll get an email reminder



Will my information be private?

- YES! Only approved registry scientists can see it, NOT employers or insurers
- You CANNOT be looked up in the registry by name



YOU JOINING

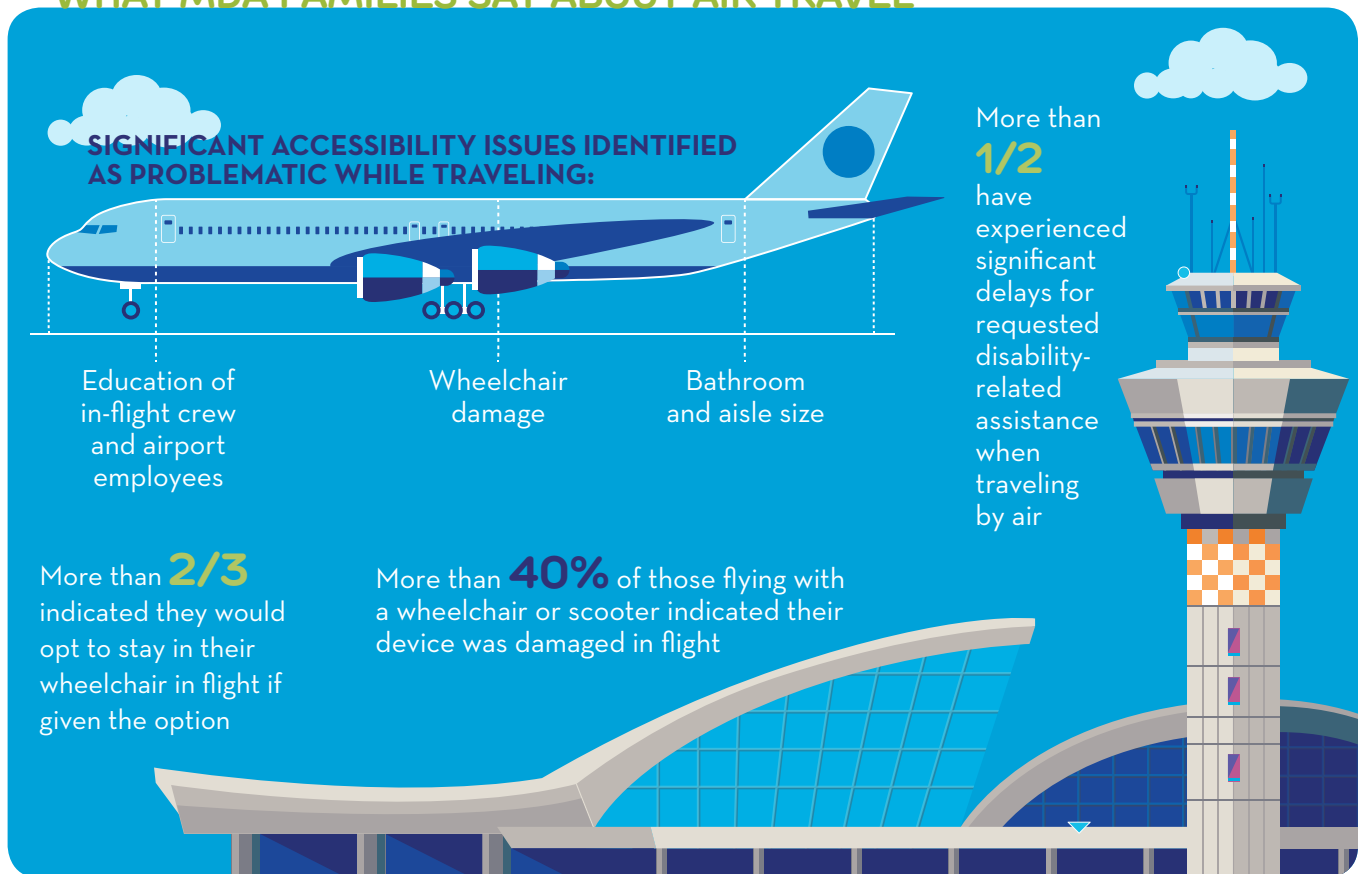


More information for research

A better understanding of ALS

The chance to help create a better future for persons with ALS

WHAT MDA FAMILIES SAY ABOUT AIR TRAVEL



FINDING OUR VOICE

There is recourse for passengers when travel plans go awry, including damage to wheelchairs and other assistive equipment, but many people either don't know what to do or don't feel that the process will meaningfully fix the problem. MDA created the Accessible Air Travel Resource Center to help people with neuromuscular disease navigate the current travel landscape, but there is significant room for improvement on the part of the government and airlines.

In a 2016 survey of more than 2,000 people living with neuromuscular disease, more than 70 percent indicated that they have experienced problems with accessibility while traveling by air, and 40 percent said their mobility device

was damaged during travel.

These numbers are unsettling alone, but our concerns are further compounded by the fact that fewer than 4 percent had ever filed a complaint with the Department of Transportation (transportation.gov/airconsumer) and more than half of respondents did not even know they could file a complaint about the problems they experienced.

We were disappointed to see these results, but we were not surprised by them. MDA has and will continue to use the findings of this survey as a powerful tool to advocate for positive changes to the aviation industry in our work with the government and airlines.

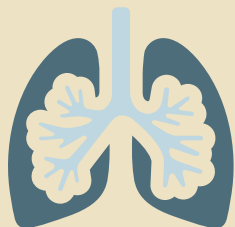
However, our advocacy efforts are only as powerful as the voice of our community.

As MDA works toward our goal of making the skies friendly for all travelers, we depend on the individuals and families we represent to help us make our case to members of Congress, the Department of Transportation and air carriers. Start by visiting MDA's advocacy page (mda.org/advocacy) to get up to speed on the latest developments and find out how to advocate for accessible air travel and a number of other issues. When we work together, the sky's the limit. **Q**

Brittany Johnson Hernandez became MDA's director of advocacy in July 2018. She is a professional public health advocate who previously served as the principal transportation staffer for a member of the U.S. House Committee on Transportation.

Be an Advocate

Together we can ensure that the voice of our community is heard. That is why we ask you to join MDA's network of advocates, families, volunteers and partners. Visit mda.org/advocacy and click on "Become an Advocate."



A chance to **advance DMD therapy**

Join a study of respiratory function in Duchenne muscular dystrophy (DMD).

The *SIDEROS* study

is a clinical trial that will study whether a therapy called idebenone is safe and effective at delaying the loss of breathing function in boys and men with DMD.

The study will compare the efficacy of idebenone to placebo in those currently on steroids (either prednisone or deflazacort).



The therapy

Idebenone is an oral tablet developed by Santhera Pharmaceuticals, a specialty company focused on developing novel treatments for DMD.

Who can participate?

- Males with DMD, any mutation
- Age 10 or older
- Ambulatory or non-ambulatory
- On corticosteroids for at least 12 months
- Forced Vital Capacity between 35% and 80%

*Find out more about the study or who can participate at **SiderosDMD.com** or by emailing us at **sideros@santhera.com**.*



The mainstream success of adaptive clothing shows that people living with disabilities have a passion for fashion

BY ANDREW CONNER



ADAPTIVE

IS THE

new

BLACK

This fall, New York Fashion Week opened with a bang: A runway show called Fashion Revolution presented numerous types of adaptive clothing worn by 30 models with disabilities. Organized by the Runway of Dreams Foundation — a nonprofit created by Mindy Scheier, whose son, Oliver, has a rare type of muscular dystrophy — the show highlighted the foundation's goals of dispelling common fashion industry misconceptions about people with disabilities.

IMAGES COURTESY OF TARGET



Features like hidden snap closures and wrap-around zippers make adaptive clothing easy to wear.

IMAGE COURTESY OF BILLY FOOTWEAR

fashion TIP #1

Plan ahead. Jenny Imhoff, whose daughter, Reagan, has SMA, recommends researching before heading to a store, especially when traveling to larger cities. “When we travel to New York or Chicago, obviously those are bigger markets, so we can find more adaptive clothing in stores,” she says. “We’ll sometimes send an email or go to the company’s website to find out which locations have adaptive clothing.”

“The first misconception is that people with disabilities are a niche demographic. In fact, people with disabilities are the largest minority on the planet,” Scheier says. “The second is that these people don’t care how they look and don’t spend money on clothing.”

YOU ARE WHAT YOU WEAR

Scheier’s experience with her son and her background in the fashion industry cemented her outlook on adaptive clothing early on.

“What you wear is core to who you are as a person, and it has a direct correlation with your confidence,” she says. “In my own world, it has given Oliver such a sense of being included. Even being able to wear a simple pair of jeans — what that did for him is unimaginable.”

Kim Anderson, MS, OTR/L, an occupational therapist at the MDA Care Center at Children’s Hospital of Philadelphia, has seen

how easy-to-wear clothing that looks and feels good can affect a person’s life.

“It gives them a sense of independence,” Anderson says. “Having a patient be able to do things themselves is a major thing we work toward. It gives them confidence, and they can

choose to fit in or stand out in a way that they want to by getting clothing they like, instead of sweat pants or oversized T-shirts.”

For Ben, a 13-year-old with spinal muscular atrophy (SMA), it was all about sporting a pair of high-top sneakers.

“We had a hard time getting Ben’s braces in high-tops,” says Page Laska, Ben’s mom. “So I began searching for high-tops, and I came across BILLY shoes. When we got them, we happened to have a family dinner. He put them on and showed everybody his new shoes with a beaming smile.”

NOT JUST NICHE

As companies realize the demand for adaptive clothing, some are making it part of their business plans. The

recent Fashion Revolution show featured clothing designed by Nike, Target and Tommy Hilfiger, the last of which collaborated with Scheier to create its adaptive clothing line. Adaptive clothing is also available at Kohl’s, Sears, J.C. Penney and online retailer Zappos.

Consumers hope more brands will take note of the growing market for adaptive clothing.

“I’ve been with the neuromuscular disease clinic for four years, and fashionable adaptive clothing didn’t even exist when I started,” Anderson says.



Reagan, 13, shows off a jacket from Target’s adaptive clothing line.

FINDING ADAPTIVE CLOTHING

Start your fashion journey with these brands and retailers that offer adaptive clothing online:



BILLY

Footwear:

This company's flip-top zipper sneakers are highly adjustable and easy to take on and off. billyfootwear.com



Target:

Check out Cat & Jack adaptive clothes for boys and girls as well as their women's and men's adaptive lines for adults. target.com

Tommy Hilfiger:

The Tommy Hilfiger Adaptive line brings the designer's signature preppy style to jeans, sweaters, polos and more. usa.tommy.com/tommy-adaptive



Zappos.com: This online retailer organizes their shoes and clothing into helpful categories, such as orthotic-friendly shoes and shirts with magnetic closures. zappos.com/adaptive



Runway of Dreams founder Mindy Scheier (standing, right) onstage with models during Fashion Revolution

PHOTOS BY NOAM GALA/GETTY IMAGES FOR RUNWAY OF DREAMS FOUNDATION



Target's Cat and Jack display during the Fashion Revolution event

Reagan, a 13-year-old former MDA Ambassador who lives with SMA, says access to adaptive clothing is improving, but she still doesn't have as many options as her peers without disabilities.

"It's getting better, but it's still something you have to search for because there are only a few stores that offer it — but it's great that they do," she says.

Reagan's mom, Jenny Imhoff, explains that when it comes to finding clothing in stores, Target has had the most to offer.

"Last year, we got Reagan two different coats at Target,"

she says. "In the past, we would have to make her something, and as she got older it wasn't cool and it looked so different. With Target's line, she loved them — they looked just like the other kids', and they're made specifically for wheelchair users."

LOOKING FORWARD

Even with retailers like Target bringing adaptive clothing to brick-and-mortar stores, for now the internet offers the most options. However, as the adaptive clothing market grows, Scheier believes adaptive fashion will become easier to find everywhere.

fashion TIP #2

Search for style. Google and other search engines can be great tools for finding clothing that fits a specific need, whether it's easy on/off shoes or adjustable-waist pants. Page Laska, mother of a 13-year-old with SMA, recommends adding words like "trendy" or "stylish" to your search to weed out more old-fashioned options. You can also try limiting your search to recent results so you're likely to find newer products.

"I hope the future of adaptive is in-store," she says. "It was important to start online to demonstrate there is a market out there and that disability knows no age or socioeconomic background, but if we really want to be mainstream, it will be in stores. I do think that will happen in our lifetime." **Q**

Andrew Conner is a writer and editor for Quest.

In 2018, Muscle Walks across the country united friends, neighbors and communities in celebration of the strength of MDA families.



MDA[®] Muscular Dystrophy Association
Muscle Walk

mdamusclewalk.org

Thank you to our National Sponsors:





access MDA

Your guide to the MDA community, from news briefs to inspiring profiles

Building Momentum

A lifelong MDA supporter runs in a memorable Boston Marathon with MDA Team Momentum



Joe deMello running the 2018 Boston Marathon

MDA has always been a part of Joe deMello's life. Neuromuscular disease runs in his family, with Charcot-Marie-Tooth disease (CMT) affecting his father and aunt and Duchenne muscular dystrophy (DMD) affecting one of his cousins.

"When we were kids my dad would drive around and we would donate to the Fill the Boot drives," he says. "Every time I go by a fire station I think about MDA." DeMello also remembers participating in Hop-a-Thon fundraisers for local MDA telethons in Plymouth and Needham, Mass.

So when deMello watched the Boston Marathon in April 2017, while he and his sister both attended Northeastern University, he got inspired and wanted to try it – even though he didn't consider himself a runner.

"I said to my sister, 'It would be cool to run it,' and she said, 'You should see if MDA does anything,'" deMello says. That conversation gave him the spark he needed to reach out to MDA Team Momentum and apply to run the 2018 Boston Marathon.

After being accepted in October 2017, deMello was

swept up in a whirlwind of fundraising and endurance training until the big race in April 2018.

“One word I would use to describe the experience is ‘humbling,’” he says. “From training to working with MDA and all of my donors – I had a certain amount of self-confidence, but I wasn’t expecting certain people to donate, like my friends at school. As a college student, there’s only so much you can give, and the fact that I could raise about \$15,500 – it was eye-opening.”

However, even after putting his all into raising money and training, deMello could have never prepared for the conditions on race day.

“I’ve been told that I had a very unique experience because of the weather,”

deMello says. “They called it Monsoon Monday.”

Runners were hit with heavy rain, wind speeds that reached more than 25 miles per hour and temperatures that were among the coldest recorded at a Boston Marathon in the past 30 years. Through all of it, deMello remembered what he was running for and maintained a sunny disposition.

“It was one of the best days of my life,” he says. “You’re on a cloud when you’re running, and I can vividly remember most of the race. In a lot of the pictures you’ll see me smiling. I was so happy seeing my family hold up signs for me. Then, crossing the finish line and seeing the MDA team – that was just awesome, and it was one of the most memorable moments of my life.”




Joe deMello's family braves the weather to show their support.

While deMello registered for the marathon thinking it would be a once-in-a-lifetime event, he does plan on running again – and raising money for MDA – in the future.

“A lot of people in my training group said, ‘You’ll want to come back,’ and I told them running long-term wasn’t for me. But sure enough, the second I got on the course they were right,” he says. “The

feeling of crossing that finish line was absolutely electric, and whether it’s this spring or in five or 10 years, I’d love to do it again.”

 **Cross the most meaningful finish line of your life with MDA Team Momentum. Registrations are now live for 2019 events. Find upcoming races and join the team at mdateam.org.**

Introducing

DuchenneAndYou.com

a comprehensive Website for information about Duchenne muscular dystrophy and support for you and your family



ADVERTISEMENT

Get the latest on:

Understanding Duchenne

Understanding genetics

Living with Duchenne

Support resources

Visit DuchenneAndYou.com Today!

Register to receive information, news, and updates

PTC1602US016UB

PTC
THERAPEUTICS™
Translating Science • Transforming Lives

WHAT COULD SPINRAZA UNLOCK?

The **first** treatment for children and adults with spinal muscular atrophy (SMA)

SPINRAZA was evaluated in a well-controlled study of 126 individuals with later-onset (Types 2 and 3) SMA. The results are supported by an open-label study of 28 individuals with later-onset (Types 2 and 3) SMA, aged 2 to 16 years at first dose. Limitations of the open-label study included differences in dosing compared with the approved regimen and the lack of an untreated group.



Maintaining motor function over time

Individuals taking SPINRAZA continued to improve or maintain motor function over nearly 3 years, regardless of their level of function before starting treatment.



Attend a SPINRAZA event

Visit [SPINRAZAevent.com](https://www.spinrazaevent.com) or call **1-888-615-4343** to connect with your local SMA community.

INDICATION

SPINRAZA 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.

For additional Important Safety Information, please see brief summary of full Prescribing Information on the next page.

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



Lauren
age 20
 Later-onset (Type 3) SMA
 treated with SPINRAZA

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

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.

QUESTIONS?

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

MANUFACTURED FOR

Biogen
 Cambridge, MA 02142

revised 05/18





Colin Batty running for Team Momentum.

Keep It Simple

An MDA Team Momentum veteran shares his secrets for fundraising success

Boston resident Colin Batty joined MDA Team Momentum as a way to honor his relatives living with Charcot-Marie-Tooth disease (CMT). His mother's side of the family – including his mom, aunts and uncles – all have a form of the condition.

A group fitness instructor, Batty has run the Boston Marathon twice with Team Momentum, raising nearly \$23,000 to date, and in 2018 he received the CITGO Fueling Good Award. He found the greatest fundraising success by keeping the process simple.

"No event is too small," Batty says. "You don't have to host a big, grandiose event. My most successful events were renting out a room at a local tavern, inviting friends out and raffling off prizes, and also splitting profits at my family's local IAFF Legion Hall's bingo night."

Batty credits the supportive MDA Team Momentum staff with helping make the process manageable. "They really prepare you for success, from fundraising to training," Batty says. "You don't have to go it alone."



Learn how MDA Team Momentum gets you beyond the finish line at mdateam.org. MDA also provides tools to support fundraisers of all kinds, from bowling parties to bake sales, at mda.org/yourway.

Raz-CAT

REHAB SHOWER COMMUNE CHAIR

Tilts

- 40°

Grows

- Adjustable from 11" – 16" deep
- 14", 16" and 18" back widths
- 15" and 18" back heights

Positions

- Accepts all Raz positioning options
- Modular recline available



Nothing else comes close

info@razdesigninc.com (877)720-5678

SEE ALL OF OUR MODELS AT
razdesigninc.com



Abilities
Serving the Community
Since 1979
EXPO

Products ✓

Answers ✓

Fun ✓

Abilities Expo Checks All the Boxes:

- ✓ Latest products and services
- ✓ Access to community experts
- ✓ Informative workshops
- ✓ Tools for seniors
- ✓ Adaptive sports
- ✓ Inclusive dance
- ✓ Assistance animals
- ✓ Fun activities for kids of all ages

FREE
ADMISSION



#AbilitiesExpo



www.AbilitiesExpo.com
Register online today. It's free!

San Mateo
October 26-28, 2018

Los Angeles
February 22-24, 2019

Toronto
April 5-7, 2019

New York Metro
May 3-5, 2019

Chicago
June 21-23, 2019

Houston
August 2-4, 2019

Boston
September 13-15, 2019



YOUR TREATMENT NEEDS ARE OUR PRIORITY

PTC Cares™ is a patient support program that provides personalized case management support before your first prescription and throughout treatment.



PTC Cares is
here to help with:

- Benefits investigation support
- Information about EMFLAZA® (deflazacort) and PTC Cares services
- Securing access to EMFLAZA

Call 1-844-4PTCCARES
(1-844-478-2227)

8 AM - 8 PM EST Monday - Friday
to speak with a
PTC Cares case manager.

Visit www.PTCCares.com
for more information.

EMFLAZA is indicated for the treatment of Duchenne muscular dystrophy in patients 5 years of age and older.

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

Please see Indication and Important Safety Information on the next page and accompanying brief summary.


Emflaza®
(deflazacort)

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

INDICATION & IMPORTANT SAFETY INFORMATION FOR EMFLAZA® (deflazacort)

INDICATION

EMFLAZA® is indicated for the treatment of Duchenne muscular dystrophy in patients 5 years of age and older.

IMPORTANT SAFETY INFORMATION

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

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

- **Hyperglycemia:** Corticosteroids can increase blood glucose, worsen pre-existing diabetes, predispose those on long-term treatment to diabetes mellitus, and may reduce the effect of anti-diabetic drugs. Monitor blood glucose at regular intervals. For patients with hyperglycemia, anti-diabetic treatment should be initiated or adjusted accordingly.
- **Increased Risk of Infection:** Tell your healthcare provider if you have had recent or ongoing infections or if you have recently received a vaccine or are scheduled for a vaccination. Seek medical advice at once should you develop fever or other signs of infection, as some infections can potentially be severe and fatal. Avoid exposure to chickenpox or measles, but if you are exposed, medical advice should be sought without delay.
- **Alterations in Cardiovascular/Kidney Function:** EMFLAZA can cause an increase in blood pressure, salt and water retention, or a decrease in your potassium and calcium levels. If this occurs, dietary salt restriction and potassium supplementation may be needed.
- **Behavioral and Mood Disturbances:** There is a potential for severe behavioral and mood changes with EMFLAZA and you should seek medical attention if psychiatric symptoms develop.
- **Effects on Bones:** There is a risk of osteoporosis or decrease in bone mineral density with prolonged use of EMFLAZA, which can potentially lead to vertebral and long bone fractures.
- **Effects on Growth and Development:** Long-term use of corticosteroids, including EMFLAZA may slow growth and development in children.
- **Ophthalmic Effects:** EMFLAZA may cause cataracts or glaucoma and you should be monitored if corticosteroid therapy is continued for more than 6 weeks.
- **Vaccination:** The administration of live or live attenuated vaccines is not recommended. Killed or inactivated vaccines may be administered, but the responses cannot be predicted.
- **Serious Skin Rashes:** Seek medical attention at the first sign of a rash.
- **Drug Interactions:** Certain medications can cause an interaction with EMFLAZA. Tell your healthcare provider of all the medicines you are taking, including over-the-counter medicines (such as insulin, aspirin or other NSAIDs), dietary supplements, and herbal products. Alternate treatment, dosage adjustment, and/or special test(s) may be needed during the treatment.

Common side effects that could occur with 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.

Please see the accompanying full Prescribing Information

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

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

EMFLAZA® (deflazacort) Consumer Brief Summary of the FDA-Approved Product Information Initial US Approval: 2017

What is EMFLAZA?

EMFLAZA® is a corticosteroid indicated for the treatment of Duchenne muscular dystrophy (DMD) in patients 5 years of age and older.

When should I not use EMFLAZA?

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

What should I tell my healthcare provider before taking EMFLAZA?

It is important to tell your healthcare provider if you have had recent or ongoing infections, develop a fever, have recently received a vaccine or are scheduled for a vaccination, or experience any other side effects.

What warnings should I know about EMFLAZA?

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

For patients already taking corticosteroids during times of medical stress, the dosage may need to be increased.

Cushing’s Syndrome: Cushing’s syndrome occurs with prolonged exposure to exogenous corticosteroids, including EMFLAZA. Symptoms include high blood pressure, truncal obesity and thinning of the limbs, purple striae, facial rounding, facial plethora, muscle weakness, easy and frequent bruising with thin fragile skin, posterior neck fat deposition, osteopenia, acne, amenorrhea, hirsutism, and psychiatric abnormalities.

Hyperglycemia: Corticosteroids can increase blood glucose, worsen pre-existing diabetes, predispose those on long-term treatment to diabetes mellitus, and may reduce the effect of anti-diabetic drugs. Monitor blood glucose at regular intervals. For patients with hyperglycemia, anti-diabetic treatment should be initiated or adjusted accordingly.

Increased Risk of Infection: Medical advice should be sought immediately if you develop a fever or other signs of infection as some infections can potentially be severe and fatal. Avoid exposure to chickenpox or measles, but if you are exposed, medical advice should be sought without delay.

Alteration in Cardiovascular/Kidney Function:

EMFLAZA can cause an increase in blood pressure and water retention or a decrease in your potassium or calcium levels. If this occurs, dietary salt restriction and potassium supplementation may be needed.

Behavioral and Mood Disturbances: There is a potential for severe behavioral and mood changes with EMFLAZA and you should seek medical attention if psychiatric symptoms develop.

Effects on Bones: There is a risk of osteoporosis or decrease in bone mineral density with prolonged use of EMFLAZA, which can potentially lead to vertebral and long bone fractures.

Effects on Growth and Development: Long-term use of corticosteroids, including EMFLAZA may slow growth and development in children.

Ophthalmic Effects: EMFLAZA may cause cataracts or glaucoma and you should be monitored if corticosteroid therapy is continued for more than 6 weeks.

Vaccination: The administration of live or live attenuated vaccines is not recommended. Killed or inactivated vaccines may be administered, but the responses cannot be predicted.

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

What are the side effects that could occur with EMFLAZA?

- facial puffiness or Cushingoid appearance
- weight increased
- increased appetite
- upper respiratory tract infection
- cough
- frequent daytime urination
- unwanted hair growth
- central obesity
- colds

What other medications might interact with EMFLAZA?

Certain medications can cause an interaction with EMFLAZA. Tell your healthcare provider of all the medication 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 treatment. Do not take EMFLAZA suspension with grapefruit juice.

The information presented is not comprehensive. Talk to your healthcare provider for more information or see www.EMFLAZA.com for the full FDA-approved product information.

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

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



Patrick Cusick with his family

A New Direction

MDA Executive Director Patrick Cusick found a new career after his diagnosis

Patrick Cusick, executive director of the Columbus, Ohio, MDA office, was diagnosed with Becker muscular dystrophy (BMD) when he was 29. At the time, he was working in sales, and he and his wife were expecting their first child.

The diagnosis was difficult for Pat to accept, but it eventually led him to his current career path. Now, Pat is a friend to families facing new diagnoses like his own.

"The best thing is meeting parents of boys with Becker," he says. "I try not to give advice — just share my own experience. Being a grown man who has the same diagnosis and has got a job and is married and has kids ... I hope parents think, 'Maybe it's not as bad as we thought. Maybe there is another path forward.'"

Most of all, he encourages families to support loved ones living with BMD in pursuing their passions.

"I coach tackle football and walk with a cane. I can't physically show [the plays], but I'm good at explaining things that are interesting to me," he says. "Don't sell yourself or your child short about what you're able to accomplish."

 Read stories from around the MDA community at strongly.mda.org.

LIFT YOUR LIFE TOWARD INDEPENDENCE



POWER TOILET AID™

Battery-powered toilet seat lift

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



NEW FREEDOM Chair™

Manual or battery-powered lift

- Rehab facility model with power height adjustment available
- No transfer required! Stand while directly lifted from wheelchair

Standing program provides many physical and psychological benefits

Battery-powered chair lift

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

Ask about our 30-Day return guarantee

800-831-8580 for information and a FREE demo video
www.stand-aid.com sales@stand-aid.com

YouTube product videos on our website

Why I Walk

“I walk for my son James, who received a congenital muscular dystrophy diagnosis at 5 months old. When my husband and I learned about this disease, we were absolutely devastated and scared for what this meant for our future. We came to realize that the most important thing we can do for ourselves and for James is to harness strength and positivity, and do everything that we can do to advocate for him, for this community and to give James the absolute best life.” –Molly Parisi



The James Gang, one of the highest fundraising MDA Muscle Walk teams in the country this year, brought in \$30,114 for MDA. The team was formed to support James (sitting front and center with his parents, Molly and Anthony Parisi), who was diagnosed with congenital muscular dystrophy (CMD) at 5 months old.



Find MDA Muscle Walk events and teams at mdamusclewalk.org.

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

LiftSeat®
Powered toilet lifts™



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

(877) 665-4381

liftseat.com

GUARANTEED FOOT DROP SOLUTIONS

DORSI-LITE™
FOOT SPLINT



USE WITH OR WITHOUT SHOES

DORSI-STRAP™
FOR FOOT DROP



FOR USE WITH SHOES

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

- Outstanding comfort.
- Easy On & Off
- Ultra-low profile, 30Z
- Near-Normal Gait
- Will not slip off
- Also use in bath or swimming pool
- \$60/EA. + \$7 Shipping/Order
- Optional EXPRESS shipping: \$25
- Low cost replacement parts available

- No Special Shoes Needed
- Nothing put into the shoe
- Outstanding comfort
- Ultra-low profile, 30Z
- Near-Normal Gait
- Easy On & Off
- In White, Black, or Brown
- \$42/EA. + \$7 Shipping/Order
- Optional EXPRESS shipping: \$25
- Low cost replacement parts available

30 DAY MONEY-BACK WARRANTY

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

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

09/17

SPECIAL \$3 OFF/ORDER: CODE MDAX

Leave Your Legacy.

By making a gift to MDA in your will or trust, you can help free kids and adults from the harmful effects of muscular dystrophy, ALS and related life-threatening diseases.

Learn more about how a gift in your Will or Trust can improve the lives of families for current and future generations at mdalegacy.org or call 312-260-5976.



LIFT YOUR LIFE TOWARD INDEPENDENCE

Battery-Powered Chair Lift



From sitting to standing in home, office or other environment

Comfort and style for use anywhere

Ask about our 30-Day return guarantee

800-831-8580

for information and a FREE demo video

www.stand-aid.com

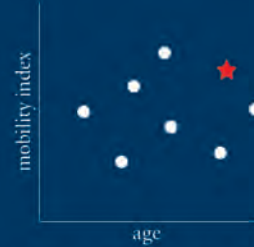
sales@stand-aid.com

YouTube product videos on our website

IBMR

INCLUSION BODY MYOSITIS REGISTRY at Yale

The Personalized IBM Index Calculator



An online tool designed to help you monitor your experience with inclusion body myositis

To learn more about this ongoing study, please visit us at

www.ibm.yale.edu



REDUCED SURGERIES & FALLS



Specializing in CMT Bracing

Improve Balance, Comfort Walking & Running!

CHECK OUT OUR YOUTUBE CHANNEL TO LEARN HOW WE CAN HELP YOU!



Email: CMTBracing@aol.com
www.DynamicBracingSolutions.net

Check for a Licensed DBS Clinician Closest To You!

(866) 999-WALK (9255)

*Also Provide Solutions For: Foot Drop, Foot & Ankle Pain, Multiple Sclerosis, and Various Neuromuscular Disorders

Music Makes It Better

By going out of her comfort zone, a young woman finds her happy place in the alternative music scene

BY KELLY BERGER



PHOTO BY NICKY PARRY

Kelly Berger loves the inclusive atmosphere and behind-the-scenes access of music festivals.

Whether it's through headphones, blasting out of the van speakers or playing live right in front of me, music has been my constant source of happiness.

After years of misdiagnoses, I was recently diagnosed with a form of congenital muscular dystrophy called collagen VI (with a bunch of letters and numbers after it that make me sound like a robot). Through the ups and downs of medical woes and lifestyle changes — from walking to using a wheelchair — the one thing that has consistently put a smile on my face is music in any way, shape or form.

No matter what I faced, I could press play on my iPod

and instantly escape. It's brought such a healing release for me to just get lost in a song, album or entire discography and forget all the daily struggles or what may come down the road. I can also turn to my ever-growing collection of ticket stubs, guitar picks, set lists and confetti from the concerts I've attended and get lost in the memories, reliving each one over again in my mind with the cheesiest grin plastered on my face.

LIVING LOUD

There's always been something about live music that truly captivated me. Seeing a band in action onstage, feeling the bass pump through you and kick-start your heart, singing along with complete strangers in

the moment — there's simply nothing like that connection to music. That post-concert adrenaline high is hard to even put into words. Such a freeing feeling — like I can do and conquer anything — that I wish lasted forever. It makes me forget about the wheelchair and any other annoying daily medical matters and be at one with the music, if only for a couple hours.

I remember back in my early teenage years persistently bugging my mom to let me go to an alternative rock show, a genre outside of my normal comfort zone. After finally wearing her down, I went to my first rock show in 2004. It was an arena show with designated ADA accommodations, so I knew my wheelchair and I would be safe. I was amazed to see the sense of community within the stadium, the craziness

“There were endless high fives, fist bumps and hugs from embrative fans. I was hooked, and I still have the same feeling of excitement every time I go to a show.”

Share Your Story

If you have a “From Where I Sit” story idea, email us at quest@mdausa.org.

You can read more stories from around the MDA community at strongly.mda.org.



of crowd-surfers and mosh pits breaking out, and yet everyone was taking care of each other, still having the best time. There were endless high fives, fist bumps and hugs from embrace fans. I was hooked, and I still have the same feeling of excitement every time I go to a show.

I decided to push the boundaries of what I felt comfortable with at a live music show. That summer I attended my first ever VANS Warped Tour. Boy, was I in heaven. I got to get up close and personal with tons of bands all at once in a sweaty, action-packed day. This touring punk rock music festival mainly takes place around arenas or in parking lots and campgrounds, making the terrain a challenge for a wheelchair. My biggest fear was navigating the crowd without getting trampled or entering a spontaneous mosh pit. Again, I was pleasantly surprised by how positively receptive the alternative scene was toward me. Strangers were happy to lend a hand when my chair needed an extra push or clear a path when things got insanely cramped. The VANS Warped Tour was what I looked forward to most every summer. I've seen some of my

favorite bands play, interacted with them and met the most amazing lifelong friends over the years. I've gone now for more than a decade and a half.

I'm also extremely lucky that my wonderful city of Cincinnati has Bogart's, an iconic, welcoming music venue that feels just like home immediately as you enter. The staff has always gone above and beyond my needs, clearing crowds, securing patrons in the ADA section and assisting with drink orders. It's a really tight-knit group that I can graciously say is like my second "concert" family.

PURSuing HAPPINESS

I know it can be scary or overwhelming to get out and find a public venue that you feel confident in or talk to people about your specific needs. With every instance, what I was so afraid of turned out to be easy to overcome, even if it required a little help from strangers. And to think of all the concerts and festivals I would have missed had I not ventured out ...

Sometimes the biggest thing holding us back is our own fear. I encourage you to break down those walls that may be keeping you from trying something new, wheelchair

or not. Go outside of your normal comfort zone — I promise you, it's not as terrifying as you'd think. Find a release that helps put you at ease and allows you to forget all the hardships. Who knows — you might just discover your new favorite activity or meet new friends with similar interests.

Whatever the case, pushing ourselves even when we can't actually physically push ourselves leads us to the happiest place of all. I've found my happy place in music. [Q](#)

Kelly Berger, 29, lives in Cincinnati and freelances in social media and public relations. She's an avid music festival and concertgoer. Catch her at a show or follow her journey at thekellyberger.com.

“Strangers were happy to lend a hand when my chair needed an extra push or clear a path when things got insanely cramped.”

OPEN SESAME

REMOTE CONTROL DOOR SYSTEM

Automatically opens and closes a door from any mobility device

- Release a locked door
- Simple to Install
- Adaptable/Portable for most doors

OPEN SESAME

CALL FOR OUR BROCHURE
1-888-973-7263
www.opensamedoor.com

Hog Heaven

MDA family celebrates a milestone with Harley-Davidson



On Labor Day weekend, Harley-Davidson and MDA hit the streets of Milwaukee to celebrate Harley's 115th anniversary — and the 38-year partnership between the iconic American motorcycle maker and MDA.

More than 6,500 riders and bikes convened for Harley's 115th Anniversary Parade on Sept. 2. Within the parade was MDA's Parade of Heroes, a group of MDA families and

supporters who joined the event in the spirit of both Harley's and MDA's ideals of freedom and independence.

One of those heroes was Jace, 5, riding in the side car of his dad, Ben Dorer's, Harley-Davidson Street Glide motorcycle. Jace, who lives with spinal muscular atrophy (SMA), traveled from Michigan to Milwaukee with his family, including his grandfather, who also rode in the parade.


"The most memorable thing was riding with my dad and my son and seeing how much fun they both had, waving and giving high fives to kids during the parade," Dorer says. **Q**

Top and bottom left: Three generations ride together. Above: Jace and his dad, Ben Dorer.

Revved Up

Since 1980, Harley-Davidson riders have logged thousands of miles to raise millions of dollars for MDA to fuel our mission to transform the lives of people affected by neuromuscular diseases through innovations in science and care. Find the latest stories about our partners' amazing work at strongly.mda.org/fundraising-events/partners.





Difficulty swallowing
Falling
Difficulty climbing stairs
Weakness
Breathing problems
Trouble walking
Easily fatigued
Droopy eyelids
Slurred speech

Looking for people with LEMS, CMS, or MuSK-MG

You Can Help Make a Difference

Catalyst Pharmaceuticals is conducting several important clinical trials in Lambert-Eaton myasthenic syndrome (LEMS), congenital myasthenic syndromes (CMS), and myasthenia gravis with antibodies to muscle-specific kinase (MuSK-MG) to study the safety and efficacy of the oral investigational medicine amifampridine phosphate (sold in Europe as FIRDAPSE®). Your participation can help bring an FDA-approved treatment to other people struggling with these rare diseases.

If you or someone you know has one of these conditions, your help is needed.

CMS Patients

This study is continuing to enroll patients ages 2 years and older who have a body weight greater than or equal to 10 kg (22 lb) and who have been diagnosed with CMS, including those with certain genetically confirmed defects. The study is being conducted in Atlanta, Baltimore, Boston, Columbus, and Los Angeles. All travel-related costs will be covered for the patient and a companion.

MuSK-MG Patients

This clinical study, to be conducted in the United States, is now enrolling patients ages 18 years or older and diagnosed with MuSK-MG. All travel-related costs will be covered for the patient and a companion. For study sites, please go to <https://clinicaltrials.gov/ct2/show/NCT03304054>, which will list study sites as they are approved, or contact us as noted in Learn More.

LEMS Patients

Patients ages 18 years or older with a confirmed diagnosis of LEMS may be eligible to enroll in the Expanded Access Program (EAP).

Learn More

For more information about our clinical studies in **CMS** or **LEMS**, contact Catalyst at EAP@catalystpharma.com or call **1-844-347-3277**. For more information about **MuSK-MG** contact Catalyst at MuSKMG@catalystpharma.com or call **1-844-347-3277**. More information about our trials is also available at www.catalystpharma.com.

Catalyst Cares

A pharmaceutical company focused on developing treatments for rare neuromuscular and neurological diseases, Catalyst is dedicated to providing education to patients and physicians. For more information about our work, please visit www.catalystpharma.com.



JUST GOT COOLER



NOW YOU CAN ENJOY ALL OF THE BENEFITS of the patented, award-winning CoolCore® fabric with your ergonomically-designed carbon fiber or aluminum ADI positioning back. As with the seat cushions, the CoolCore fabric of the ADI back cushion wicks moisture away from your body, helping keep you dry and comfortable. The chemical-free fabric makes both the back and seat cushions cool better and last longer—a perfect combination for your busy life. Stay chill with CoolCore and ADI.



Learn more at StealthProducts.com