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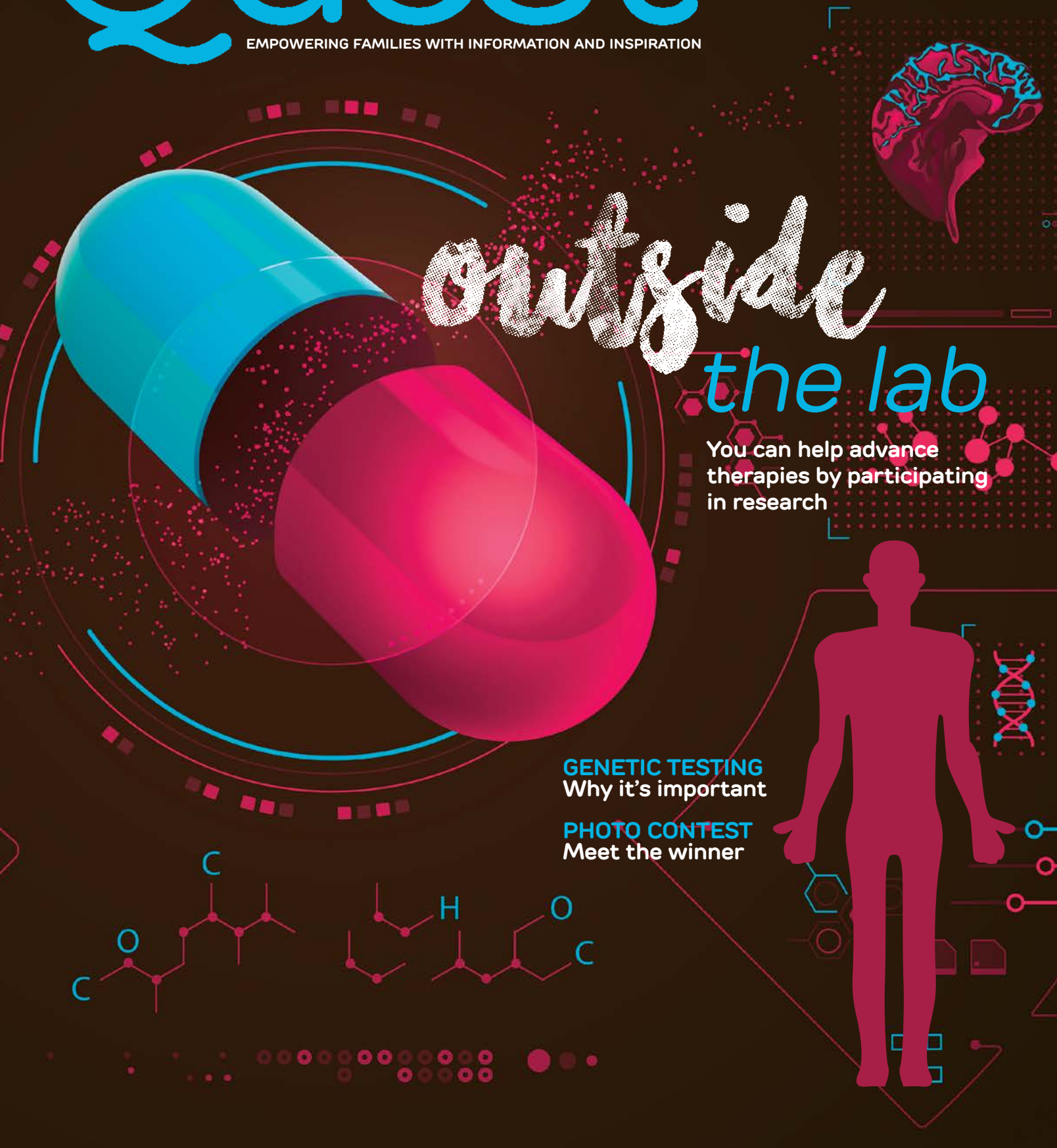
EMPOWERING FAMILIES WITH INFORMATION AND INSPIRATION

MDA Muscular Dystrophy Association



outside the lab

You can help advance therapies by participating in research

GENETIC TESTING
Why it's important

PHOTO CONTEST
Meet the winner

NEW

zolgensma®
(onasemnogene abeparvovec-xioi)
suspension for intravenous infusion

Photograph is not of an actual SMA patient and caregiver.

I'll always remember the day

we received the one-time-only dose for SMA

ZOLGENSMA® (onasemnogene abeparvovec-xioi) is a prescription gene therapy used to treat children less than 2 years old with spinal muscular atrophy (SMA). ZOLGENSMA is given as a one-time infusion into the vein. ZOLGENSMA was not evaluated in patients with advanced SMA.

To learn more, talk to your child's doctor about **the one-time-only dose** and visit ZOLGENSMA.com.

Indication and Important Safety Information

What is ZOLGENSMA?
ZOLGENSMA is a prescription gene therapy used to treat children less than 2 years old with spinal muscular atrophy (SMA). ZOLGENSMA is given as a one-time infusion into the vein. ZOLGENSMA was not evaluated in patients with advanced SMA.

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

- ZOLGENSMA can cause acute serious liver injury. Liver enzymes could become elevated and may reflect acute serious liver injury in children who receive ZOLGENSMA.
- Patients will receive an oral corticosteroid before and after infusion with ZOLGENSMA and will undergo regular blood tests to monitor liver function.
- Contact the patient's doctor immediately if the patient's skin and/or whites of the eyes appear yellowish, or if the patient misses a dose of the corticosteroid or vomits it up.

What should I watch for before and after infusion with ZOLGENSMA?

- Viral respiratory infections before or after ZOLGENSMA infusion can lead to more serious complications. Contact the patient's doctor immediately if you see signs of a possible viral respiratory infection such as coughing, wheezing, sneezing, runny nose, sore throat, or fever.
- Decreased platelet counts could occur following infusion with ZOLGENSMA. Seek immediate medical attention if a patient experiences unexpected bleeding or bruising.

What do I need to know about vaccinations and ZOLGENSMA?

- Talk with the patient's doctor to decide if adjustments to the vaccination schedule are needed to accommodate treatment with a corticosteroid.
- Protection against respiratory syncytial virus (RSV) is recommended.

Do I need to take precautions with the patient's bodily waste?
Temporarily, small amounts of ZOLGENSMA may be found in the patient's stool. Use good hand hygiene when coming into direct contact with bodily waste for 1 month after infusion with ZOLGENSMA. Disposable diapers should be sealed in disposable trash bags and thrown out with regular trash.

What are the possible or likely side effects of ZOLGENSMA?
The most common side effects that occurred in patients treated with ZOLGENSMA were elevated liver enzymes and vomiting.

The safety information provided here is not comprehensive. Talk to the patient's doctor about any side effects that bother the patient or that don't go away.

You are encouraged to report suspected side effects by contacting the FDA at 1-800-FDA-1088 or www.fda.gov/medwatch, or AveXis at 833-828-3947.

Please see the Brief Summary of the Full Prescribing Information on the next page.

US-ZOL-19-0230 08/2019

avexis

IMPORTANT FACTS ABOUT ZOLGENSMA® (onasemnogene abeparvovec-xioi)

USE

ZOLGENSMA is a prescription gene therapy used to treat children less than 2 years old with spinal muscular atrophy (SMA).

- ZOLGENSMA is given as a one-time infusion into the vein.
- ZOLGENSMA was not evaluated in patients with advanced SMA.

WARNINGS

Acute Serious Liver Injury and Elevated Liver Enzymes

- ZOLGENSMA can cause acute serious liver injury. Liver enzymes could become elevated and may reflect acute serious liver injury in children who receive ZOLGENSMA.
- Patients will receive an oral corticosteroid before and after infusion with ZOLGENSMA and will undergo regular blood tests to monitor liver function.
- Contact the patient's doctor immediately if the patient's skin and/or whites of the eyes appear yellowish, or if the patient misses a dose of the corticosteroid or vomits it up.

Decreased platelet counts could occur following infusion with ZOLGENSMA. Caregivers should seek immediate medical attention if a patient experiences unexpected bleeding or bruising.

OTHER IMPORTANT INFORMATION

Patients should be tested for the presence of anti-AAV9 antibodies prior to infusion with ZOLGENSMA.

Vaccination schedule should be adjusted where possible to accommodate treatment with an oral corticosteroid. Caregivers should talk with the patient's doctor to decide if adjustments to the vaccination schedule are needed during corticosteroid use. Protection against respiratory syncytial virus (RSV) is recommended.

Viral respiratory infections before or after ZOLGENSMA infusion can lead to more serious complications. Contact the patient's doctor immediately if you see signs of a possible viral respiratory infection such as coughing, wheezing, sneezing, runny nose, sore throat, or fever.

Temporarily, small amounts of ZOLGENSMA may be found in the patient's stool. Use good hand hygiene when coming into direct contact with bodily waste for 1 month after infusion with ZOLGENSMA. Disposable diapers should be sealed in disposable trash bags and thrown out with regular trash.

COMMON SIDE EFFECTS

The most common side effects that occurred in patients treated with ZOLGENSMA were elevated liver enzymes and vomiting.

These are not all the possible side effects. Talk to the patient's doctor about any side effects that bother the patient or that don't go away.

QUESTIONS?

To learn more, talk to your doctor and you can visit www.ZOLGENSMA.com for Full Prescribing Information.

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AveXis, Inc.

Bannockburn, IL 60015 US-ZOL-19-0181 06/2019

The Year of the Advocate

Brittany Johnson Hernandez

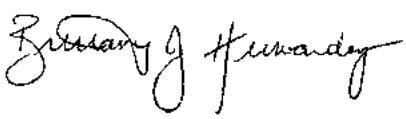
It was a banner year for MDA's advocacy operation in 2019. Our small but mighty advocacy team, based in Washington, DC, advocated for initiatives important to the neuromuscular disease community before policymakers in the US Congress and many federal agencies, including the Food and Drug Administration, Department of Transportation, National Institutes of Health, and Centers for Medicare and Medicaid Services. We promoted initiatives across the policy spectrum, from ensuring robust funding for medical research to guaranteeing affordable health coverage and making air travel more accessible, just to name a few.

We couldn't do any of this successfully without the partnership of our advocates across the country. The importance of sharing the personal impact that policies have on the lives of everyday Americans cannot be understated in MDA's efforts to promote the needs of our community.

One of the best ways to do this is through in-person meetings with policymakers, and we were able to do just that at our Public Policy and Advocacy Conference in October. At this three-day meeting, we hosted individuals and families who traveled from across the country to Washington, DC, for intensive education sessions on issues related to newborn screening, accessible air travel, therapeutic development, and employment issues for people living with disabilities.

After these policy sessions, our advocates took to Capitol Hill, where they held more than 100 meetings with Congress members and their staff to lobby on behalf of themselves and the broader MDA community. Advocates who weren't able to join us in person took action from home via MDA's virtual advocacy engagement website, echoing the loud calls to action from the individuals and families at the Capitol.

While the conference lasted three exciting days, MDA's advocacy operation is active all year long, and you can get involved today. Becoming an advocate is easy, and taking action helps the whole MDA community. Join today so we can make a difference together for everyone living with a neuromuscular disease.



Brittany Johnson Hernandez
Senior Director of Policy and Advocacy
Muscular Dystrophy Association

Become an Advocate

Join the MDA advocacy network at **mda.org/advocacy**.

MDA 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 healthcare providers.

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

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A CLINICAL STUDY IN MYASTHENIA GRAVIS

ALEXION

The CHAMPION MG STUDY

Alexion is currently recruiting patients with anti-acetylcholine antibody receptor positive generalized myasthenia gravis (MG) 18 years of age or older for a Phase 3 study of ravulizumab-cwvz, called the CHAMPION MG Study. The study will assess ravulizumab, compared to placebo, on the improvement of MG symptoms (MG activities of daily living). Participants may continue on their current medicines*, as long as they are stable, and after a 26-week study treatment period all participants can receive ravulizumab-cwvz for an additional follow up period of up to 2 years. For more information and to learn if you are eligible for the CHAMPION MG Study, please contact **ClinicalTrials@alexion.com** or go to **MGCHAMPION.com.**

*Except for other complement inhibitors, rituximab, chronic Plasma Exchange or Intravenous Immunoglobulin

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DEPARTMENTS

FEATURES

IMAGE: ISTOCK.COM/PROCIO

MORE ONLINE

NEWBORN SCREENING: WHAT YOU NEED TO KNOW

"Newborn screening is one of the most important and impactful public health programs in the United States," says Dr. Rodney Howell, pediatrician and former chairman of MDA's Board of Directors. Learn about the latest changes to the newborn screening program and how it benefits children in a Q&A with Dr. Howell. **Visit mda.org/quest for this and other stories.**

COVER IMAGE: ISTOCK.COM/JACKIE NIAM

MDA-930

Looking Forward to Learning

At MDA, we believe in the power of community and the importance of building relationships with others going through similar experiences.

If you are living with a neuromuscular disease, we invite you and your loved ones to attend our MDA Engage education events taking place across the country.

EVENT SPOTLIGHT

In March 2020, the Pompe disease community will come together in Hyattsville, Md., for MDA's Engage Pompe Disease Symposium.

MDA Engage disease-specific symposia are one-day events empowering individuals and families with knowledge and resources. This symposium will feature experts in the Pompe disease field providing updates on research and clinical trials, best practices in care, a drug development roundtable, and more.

MDA Engage Pompe Disease Symposium
When: March 8, 2020
Where: Hyattsville, MD
Register: mda.org/pompe-symposium20

Erik K. Henricson, PhD, MPH (right), moderates a panel discussion at an MDA Engage Duchenne Muscular Dystrophy Symposium.

Find an Event

For a complete list of MDA Engage events and to register, visit **mda.org/care/mda-engage**.

Thank you to these sponsors for supporting MDA Engage educational events.

ALEXION
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MAKING THE MOST OF EVERY MOMENT

The role of a caregiver

By Grace Grutter

The moment you become a parent of a child with spinal muscular atrophy (SMA), everything changes.

My daughter, Nella, was diagnosed with SMA when she was 11 weeks old, and the doctors warned me and my husband that she would only get weaker over time since her condition was terminal.

We cherished every moment that we had with Nella, and it was important to us to spend time seeking out possible treatment and care, even though we were told treatment for SMA was not available at the time of her diagnosis. For months, Nella struggled to swallow or move unaided. She had to sleep with a ventilator to help her breathe throughout the night. For such a young life, it seemed unfair that she had so many challenges to face.

In 2016, we first heard about a clinical trial for SPINRAZA® (nusinersen), a potential treatment for SMA. The trial was taking place in Orlando, Florida, and would have required our family to move across the country, so we did not have Nella participate. Instead, I decided to immerse myself in the SMA community and follow the journeys of those accessing treatment. It was through this community that I learned the trial would be expanding its access to a hospital near our home in St. Louis, Missouri. We consulted with our doctor to discuss the risks and benefits, and in December 2016, Nella received her first loading dose of SPINRAZA as part of the trial. Weeks later, SPINRAZA was approved by the FDA for treatment in adults and children. I get chills thinking about the enormous feelings of hope and gratitude I felt as Nella received her first injection of SPINRAZA. At her diagnosis, doctors had told us that there was no treatment and to prepare for our daughter to pass. And yet, here she is – alive, receiving treatment, and reaching milestones we never thought possible. In June 2018, Nella completed her eighth SPINRAZA injection and shortly afterwards, she celebrated her fifth birthday. She continues to surprise us – recently during physical therapy she turned her head almost 180 degrees all on her own. Some may consider this to be a small movement, but it isn't small for us – it's everything!

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

Every caregiver has their own unique experience, but despite these different experiences I know there is one thing we all have in common – it's that we make the most of every moment with our loved ones.

INDICATION

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

IMPORTANT SAFETY INFORMATION

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

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

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

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

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

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.

SPINRAZA® (nusinersen) injection 12 mg/5 mL

Biogen is compensating Grace for sharing her and Nella's story. This content has been reviewed for compliance with FDA guidelines. Please keep in mind these are Nella's experiences with SMA Type 1 and SPINRAZA, and others may have different experiences. For more information about SPINRAZA, visit SPINRAZA.com.

IMPORTANT FACTS ABOUT SPINRAZA® (nusinersen)

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

WARNINGS
Increased risk of bleeding complications has been observed after administration of similar medicines. Your healthcare provider should perform blood tests before you start treatment with SPINRAZA and before each dose to monitor for signs of these risks. Seek medical attention if unexpected bleeding occurs.
Increased risk of kidney damage, including potentially fatal acute inflammation of the kidney, has been observed after administration of similar medicines. Your healthcare provider should perform urine testing before you start treatment with SPINRAZA and before each dose to monitor for signs of this risk.

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

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

OTHER INFORMATION
SPINRAZA is a medication that should be administered as an injection into the lower back (a procedure called intrathecal injection) by, or under the direction of, an experienced healthcare professional.
Before taking SPINRAZA, tell your healthcare provider if you are pregnant or plan to become pregnant.

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

MANUFACTURED FOR
Biogen
Cambridge, MA 02142

revised 11/18

SPINRAZA® (nusinersen) injection 12 mg/5 mL

Biogen

IMAGE: ISTOCK.COM/KOTO_FEJA

progress now

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

Amyotrophic lateral sclerosis (ALS)

$2.5M to Fund ALS Research

Study focuses on leveraging patients' own immune cells to fight ALS

MDA, along with The ALS Association and ALS Finding a Cure, announced that they jointly awarded a clinical trial grant of more than $2.5 million to leading investigators at the Houston Methodist Neurological Institute and Massachusetts General Hospital. The grant will fund research to expand upon the promising results from the first in-human study to leverage patients' own immune cells to treat ALS.

Regulatory T-lymphocytes (Tregs) are a type of white blood cell that reduces immune system activation. People with ALS have fewer Tregs in their blood, and those Tregs have decreased function, allowing harmful immune processes to occur.

Building on preclinical results showing it is possible to restore Treg function outside the body, the first clinical trial in humans demonstrated that Tregs could be extracted from ALS patients, expanded *in vitro*, and safely infused back into the same individuals. Researchers observed a significant improvement in Treg suppressive function and an apparent slowing of disease progression in the three people who received this treatment with "expanded Tregs."

Enrollment is open for the phase 2a clinical trial, in which researchers will study the biological activity, safety, and tolerability of expanded Tregs.

Principal Investigator Stanley Appel, MD, is one of the country's foremost experts on ALS.

For more information on this trial, including enrollment information, visit ClinicalTrials.gov and enter NCT04055623 in the "Other Terms" search box.

Charcot-Marie-Tooth disease (CMT)

Grant to Clinical Research Network

Aim is clinical trial readiness for CMT

Michael Shy, PhD, at the University of Iowa was awarded an MDA Clinical Research Network Grant totaling $423,413 over three years to further develop the Inherited Neuropathies Consortium (INC), a network of clinical investigators dedicated to creating the infrastructure necessary to evaluate therapies for people with inherited peripheral neuropathies collectively known as CMT.

The award will help expand the INC's natural history data to become "clinical-trial-ready" for common and rare forms of CMT; identify, characterize, and facilitate treatments for novel forms of CMT; train future investigators in the field; and share INC data with scientists, physicians, and patients in an easily accessible manner.

The primary aim of the INC is to establish a large longitudinal cohort of patients with CMT whose data will be used to study disease progression, look for genetic modifiers to CMT1A, and identify new genetic causes for axonal forms of CMT. In addition, consortium scientists will develop and test outcome measures to determine the natural history of patients with CMT, as well as train the next generation of CMT researchers.

IMAGE: ISTOCK.COM/IPOPBA

Clinical outcome assessments, biomarker data, and natural history studies are essential to run and interpret clinical trials in CMT.

To learn more about the Inherited Neuropathies Consortium, visit rarediseasesnetwork.org/cms/inc.

Duchenne muscular dystrophy (DMD)

Vamorolone Results

Trial shows benefits with fewer side effects

IMAGE: ISTOCK.COM/HISPANOLISTIC

ReveraGen announced positive results from its ongoing phase 2 clinical trial of vamorolone in boys with DMD. Vamorolone, a "dissociative steroid," is an anti-inflammatory compound that researchers hope will convey the same benefits of traditional glucocorticoids, such as prednisone and deflazacort, without the unwanted side effects of those drugs.

The trial showed that treatment with vamorolone significantly improved two motor function outcome measures — velocity of the 10-meter run/walk and four-stair climb — compared to matched historical controls.

Importantly, boys with DMD who were treated with vamorolone had normal growth rates, and they had less weight gain and fewer

Cushingoid features than boys with DMD who were treated with prednisone and deflazacort in a previously published study.

ReveraGen's phase 2b Vision-DMD trial is currently recruiting boys with DMD, ages 4 to 7 years, to evaluate the safety and efficacy of vamorolone as compared to the current standard of care.

Learn more about Vision-DMD at vision-dmd.info/2b-trial-information or visit ClinicalTrials.gov and enter NCT03439670 into the "Other Terms" search box.

Grant for Novel Drug

Increasing sarcospan could slow muscle damage

Rachelle H. Crosbie, PhD

MDA awarded MDA Venture Philanthropy (MVP) funding totaling $389,463 over two years to Rachelle H. Crosbie, PhD, professor and chair of Integrative Biology and Physiology at the University of California, Los Angeles. The award will support the development of a small molecule drug that increases expression of sarcospan, a protein that may help prevent the muscle damage that occurs in DMD.

Sarcospan is naturally present in muscle, but Dr. Crosbie's laboratory showed that increasing the amount of sarcospan can have a protective effect on skeletal and cardiac muscle in mice with muscular dystrophy. Her team has recently identified a set of small molecules that increase the levels of sarcospan, and with support from the MVP award, she will chemically optimize these lead compounds and test them in DMD mice and DMD human cells.

If successful, this work may lead to the development of a novel drug that could slow the loss of function in limb muscles, respiratory muscles, and the heart.

Read more at mda.org.

Friedreich's ataxia (FA)

Omaveloxolone Study

Drugmaker to seek regulatory approval

Reata Pharmaceuticals announced positive results of its pivotal phase 2 clinical trial (MOXIe) of omaveloxolone in patients with FA. Patients treated with omaveloxolone showed a statistically significant improvement in neurological function after 48 weeks of treatment compared to placebo, and the drug appeared to be well tolerated.

Based on the trial results, Reata plans to submit regulatory filings for approval in the United States and worldwide. If successful, omaveloxolone would be the first approved therapy designed to slow the progression of the disease.

The US Food and Drug Administration (FDA) and the European Medicines Agency have granted Orphan Drug designation to omaveloxolone for the treatment of FA.

To learn more about the clinical trial, visit ClinicalTrials.gov and enter NCT02255435 in the "Other Terms" search box.

Myotonic dystrophy (DM)

Research Network Grant

$1M awarded for testing new treatments

Charles Thornton, MD, professor of Neurology at the University of Rochester, was awarded an MDA Clinical Research Network Grant totaling $1,118,673 over three years to continue to lead the development of the Myotonic Dystrophy Clinical Research Network, which comprises six medical centers specializing in research and clinical care of DM types 1 and 2.

Established in 2013 and supported by funding from MDA and other patient advocacy groups, the National Institutes of Health, and pharmaceutical company Biogen, the network aims to gain a more detailed understanding of the DM disease process and collect necessary clinical trial data to determine the most appropriate outcome measures, biomarkers, and endpoints.

Since the network was formed, the six participating sites (in California, Florida, Kansas, New York, Ohio, and Washington, DC) have developed standardized equipment and procedures for obtaining and analyzing needle muscle biopsies, quantifying measures of myotonia, and taking measurements of muscle strength and motor function. Currently, the network is recruiting for a variety of studies.

Visit myotonic.org to learn more.

Spinal muscular atrophy (SMA)

Spinraza Results

Trial data supports early treatment

In clinical trials, participants who received treatment with Spinraza experienced life-changing outcomes, including the achievement of major motor milestones.

Biogen announced positive long-term results from its ongoing phase 2 NURTURE clinical trial evaluating Spinraza for treatment of SMA. New data demonstrated that, after almost four years, infants who were treated with Spinraza before developing symptoms of SMA continue to achieve and maintain motor milestones that babies with SMA types 1 and 2 normally would not be expected to achieve, and they show no signs of loss of motor function.

Spinraza is a disease-modifying antisense oligonucleotide designed to treat the underlying genetic defect in SMA. NURTURE is the longest study of infants with SMA who received treatment before symptom onset.

The overall findings of this study support the efficacy of Spinraza across a range of SMA patients and appear to support the early initiation of treatment.

For more information about the NURTURE clinical trial, visit ClinicalTrials.gov and enter NCT02386553 in the "Other Terms" search box.

IMAGE: ISTOCK.COM/NICOLAS_HETMANSTOCK, TTSZ

Spinal muscular atrophy **(SMA)**

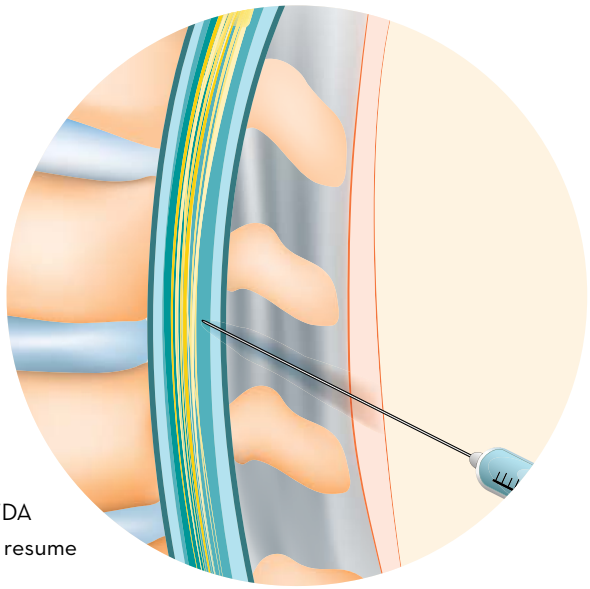
STRONG Trial

Partial hold on testing AVXS-101

In October, the US Food and Drug Administration (FDA) placed a partial hold on clinical trials for intrathecal (IT) administration of AVXS-101 for SMA type 2. The order stops enrollment of patients in the high-dose group of AveXis' phase 1/2 STRONG trial.

This partial hold impacts only the IT (injected into the spinal canal) formulation and does not impact the currently FDA-approved and available Zolgensma or AVXS-101 intravenous clinical trials. The hold was issued in response to a finding in a preclinical study in 12 animals that showed dorsal root ganglia (DRG) mononuclear cell inflammation, which can be associated with sensory effects such as tingling, prickling, chilling, burning, or numbness.

The low- and mid-dose cohorts in STRONG already completed enrollment, and AveXis reported encouraging interim results from these groups. AveXis will work with the FDA to identify any additional actions necessary to resume the trial.



The STRONG trial is designed to evaluate the efficacy, safety, and tolerability of one-time IT administration of AVXS-101 in patients with SMA type 2, ages 6 months to 5 years.

Read more about AVXS-101 at avexis.com. For questions, please contact AveXis Medical Information at medinfo@avexis.com.

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

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

*Actual patient

SANOFI GENZYME

IT COULD BE POMPE DISEASE

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

Genetic Testing is Vital.

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

Know the Pompe Spectrum
at pompe.com

OUTSIDE the lab

MDA families can get involved in research and help advance treatments for neuromuscular diseases

BY CHERYL ALKON

Mikey (left) is in a clinical trial for a DMD therapy. The medication could benefit his brother, Reid (right), and many other boys with DMD.

Michael Lo Sapio, father of Mikey, 7, and Reid, 5, proudly admits that he's pushy. Both his boys live with Duchenne muscular dystrophy (DMD), which causes progressive muscle degeneration and weakness beginning in early childhood. People with DMD typically live to their late teens or early 20s.

These facts are grim, but being pushy means not accepting the statistics without a fight. For Michael — and many people living with neuromuscular diseases — one way to push back on the numbers is by participating in research.

Biomedical researchers are constantly seeking information about neuromuscular diseases, testing therapies that could pave the way for potential cures and improve quality of life for people living with these diseases. There are many ways that individuals with neuromuscular disease can be involved in research, whether enrolling in a clinical trial or natural history study, electing to participate in a disease registry, or giving to a tissue bank. All have the potential to advance treatments for neuromuscular diseases.

That's why Michael got pushy when he learned Mikey qualified for a clinical trial of a gene-targeted therapy that has the potential to slow the progression of DMD.

"I called every hospital listed in the trial and pestered them until I finally got answers on when it was starting and made sure Mikey was on the list," he says.

While Mikey has been in the trial for two years, his younger brother did not make the age cutoff to enroll. Michael continues to push to see if Reid could qualify for compassionate use of the medication Mikey is taking. "As a parent, you have to really take control and advocate for your kid," he says.

WHY PARTICIPATE IN RESEARCH?

Getting access to the newest medications available is one potential benefit of participating in clinical trials. But being involved in research has other far-reaching effects.

"Many patients tell me that their participation helps define the future research opportunities in clinical care when there are no approved therapies," says Barry Byrne, MD, PhD, a professor of Pediatrics and director of the Child Health Research Institute at the University of Florida in Gainesville. "It helps advance the science and improve outcomes in the long-term."

In other words, you don't have to have an MD or PhD to make a difference in the neuromuscular disease community.

"For most patients, on a basic level, participating is a way to give back to the community," says Nicholas Maragakis, MD, a professor of Neurology at Johns Hopkins Medicine, specializing in amyotrophic lateral sclerosis (ALS). "A lot of people with ALS say, 'This may not help me, but it may help others who come after me.'"

GETTING INVOLVED

There are many ways that people with neuromuscular diseases can get involved with research. Finding the right way for you depends on many factors, including your diagnosis, your location, and your available time.

Clinical trials: Clinical trials are experiments done in humans to see if a potential treatment is safe and effective. Clinical trials are run at institutions and hospitals by research teams that often include doctors, nurses, study coordinators, social workers, and other healthcare professionals. But trials are very different from routine medical care, because researchers don't know exactly how the drug or device being tested will affect people.

MDA'S MOVR DATA HUB

Currently, people who see their physicians through MDA Care Centers may be able to elect to have their clinical information shared in a safe and confidential way through the MOVR (neuroMuscular Observational Research) Data Hub for future research opportunities.

The MOVR Data Hub collects information from people with amyotrophic lateral sclerosis (ALS), some forms of muscular dystrophy, and other neuromuscular diseases to help scientists better understand how the conditions affect people and what treatments are effective. MOVR also helps researchers identify individuals who may qualify for clinical trials.

MDA plans to expand MOVR to include more neuromuscular diseases and increase the number of locations that compile this information. Stay tuned for updates on this ambitious project at **mda.org/movr**.

IMAGE: ISTOCK.COM/FINGERMEDIUM

Michael Lo Sapio at a clinic visit with his sons, Reid and Mikey, who live with DMD

Each trial has its own inclusion and exclusion criteria to determine who can participate. Many clinical trials require genetic confirmation of a disease so researchers know that they are testing their treatment on the intended target. Genetic testing can pinpoint an individual's disease-causing mutation and help individuals qualify for clinical trials (see "The Importance of Genetic Testing" on page 22).

Research studies: Some clinical studies are not used to test a treatment or device but instead follow patients over time in order to better understand how a disease develops or how to treat it. These observational or natural history studies are especially important for rare diseases, which tend to be poorly understood.

The knowledge gained through observational studies may be used to determine how to detect or diagnose a disease, or to optimize testing procedures for future trials.

Research studies also have specific criteria for enrollment, based on the condition they are studying.

Disease registries: Disease registries collect health information from people with certain diseases. Registries make that information (without names or other personal details) available to researchers who can look at it to see how a disease affects various populations or how people are responding to certain treatments. The more people involved in a disease registry and the more data available, the more useful it is to researchers.

Registry data can also help improve future clinical trial design and help identify potential participants for upcoming studies.

Different types of registries collect different information, but most focus on one or a few diseases.

Tissue banks: A number of tissue banks store samples of blood, muscle, skin, spinal fluid, and other clinical specimens from people with neuromuscular diseases. These samples can be collected with consent during scheduled office visits or surgeries, or after death with the family's consent. Donated tissue samples can be used by qualified scientists for research purposes.

Tissue banks can be extremely valuable to researchers when it is difficult to procure samples from a large number of patients, such as in the case of rare diseases.

For example, the Stanford Neuromuscular Biobank "has supported many different neuromuscular research programs around the world, helping scientists better understand the biology behind these conditions in order to improve the lives of patients," says Katharine A. Hagerman, PhD, a research scientist who manages the bank along with neuroscientist John W. Day, MD, PhD.

WHAT TO EXPECT

Clinical trials and research studies generally are the most involved forms of research, often requiring office visits, medical tests, or extensive surveys.

Participating in disease registries might involve providing health information on a regular basis. Or, as in the case of MDA's MOVR Data Hub, you may simply opt in to have information from

RESEARCH RESOURCES

Figuring out how you can be involved in research takes some, well, research. Here are resources to get you started.

Clinical trials and research studies

MDA Clinical Trials Finder Tool: **mda.org/research/clinical-trials**

US National Library of Medicine database of clinical studies around the world: **clinicaltrials.gov**

Tissue banks

Congenital Muscle Disease Tissue Repository at the Medical College of Wisconsin: **mcw.edu/departments/congenital-muscle-disease-cmd-tissue-repository**

Stanford Neuromuscular Biobank: **med.stanford.edu/day-lab/biobank.html** or call **650-497-9807**

Disease registries

MDA MOVR Data Hub: **mda.org/science/movr-data-hub-neuromuscular-observational-research**

National ALS Registry: **cdc.gov/als**

While it's important to do your own research, the amount of information available on the internet can be overwhelming. Working through your MDA Care Center and care providers often is the best way to learn about the opportunities to participate in research that are available to you.

The MDA Resource Center can also answer questions and guide you to resources. Contact us at **833-ASK-MDA1**.

IMAGE: ISTOCK.COM/FINGERMEDIUM

your clinical visits combined, in a safe and confidential way, with data from others living with the same disease.

Your care team might ask if you're interested in donating samples to a tissue bank during a regular office visit or before a procedure. If they don't, you can ask your doctor about sharing samples with a tissue bank. Some tissue banks ask you to contact them and fill out a questionnaire, as they are looking for specific types of samples. They may then contact you with instructions for collecting and donating your sample.

A REWARDING EXPERIENCE

Chris Anselmo, a 33-year-old Westbrook, Conn., resident lives with limb-girdle muscular dystrophy type 2B, also called Miyoshi myopathy or dysferlinopathy. From 2013 to 2016, he took part in a natural history study called the Clinical Outcome Study for Dysferlinopathy, sponsored by the Jain Foundation. Researchers tracked his disease progression over time.

Participating was intense. Over three years, Chris made a series of clinical visits where he underwent physical tests, such as bloodwork, a skin biopsy, two MRIs, and several strength tests, and answered questions about his disease progression, such as whether he had fallen since his last visit.

"It was very taxing, and I was exhausted by the end," he says. "It was also difficult because I could see myself getting weaker from visit to visit, but they needed to track this to see how my disease progressed year to year."

The goal of collecting this information from Chris and others with the same condition was to determine the best biomarkers and strength tests to use for potential future clinical trials. "This way, they know what to test and how to measure if a therapy is working," he says.

Despite the hardships, Chris says the experience was rewarding. "There aren't many of us with this disease, so I knew my efforts were needed," he says. "Without this data, it is much harder for a company to find a therapy for my condition. It felt good to contribute in some positive way toward research that may one day lead to a therapy." Q

Cheryl Alkon is a freelance writer based in Massachusetts.

Chris Anselmo

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INDICATION & IMPORTANT SAFETY INFORMATION FOR EMFLAZA® (deflazacort)

INDICATION

EMFLAZA® is a corticosteroid indicated for the treatment of Duchenne muscular dystrophy (DMD) in patients 2 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, ocular infections and glaucoma and you should be monitored if corticosteroid therapy is continued for more than 6 weeks.
- **Vaccination:** The administration of live or live attenuated vaccines is not recommended in patients on EMFLAZA. Live-attenuated or live vaccines can be administered at least 4 to 6 weeks prior to starting EMFLAZA.
- **Serious Skin Rashes:** Seek medical attention at the first sign of a rash.
- **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 www.emflaza.com for the 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 report adverse events to FDA at 1-800-FDA-1088 or www.fda.gov/medwatch.

PTC THERAPEUTICS™

THE IMPORTANCE OF GENETIC

Kelly Berger, 31, of Cincinnati, spent most of her life chasing a diagnosis. When she was 3, her parents noticed that, although she reached physical milestones for her age, she did them in unusual ways. For example, to step up, she pushed off her thighs with her hands, and she preferred crawling on stairs to walking them. Her parents took her to a neurologist and, after bloodwork, an electromyography (EMG), and a muscle biopsy, she received a diagnosis of spinal muscular atrophy (SMA) type 3. That was in the early 1990s, when fewer types of neuromuscular disease were understood.

"Fast-forward to my mid-teens," Kelly recounts. "After more bloodwork, another neurologist concluded that my tests showed I didn't have the SMA gene mutation. They settled on congenital muscular dystrophy [CMD], type undefined. I was my own category. Kind of cool when you think about it."

But Kelly and her neurologist weren't ready to give up on getting an accurate diagnosis. Finally, in 2017, genetic tests run on blood samples and a skin biopsy showed she has collagen VI A1 intron 11, a rare type of CMD.

SEARCHING FOR ANSWERS

Struggling to get an accurate diagnosis is a common experience among people living with neuromuscular diseases. In a survey of more than 3,000 individuals living with a neuromuscular disease or a family member who has one, MDA found 25% have not had their diagnosis confirmed through genetic testing.

Having a genetically confirmed diagnosis is important because it can lead to better treatment and open doors to participating in clinical trials and other research aimed at treatments and cures.

Genetic testing not only is used to confirm a diagnosis but also can help predict how a disease will progress and give physicians vital information for targeting therapies to an individual's disease.

"Recently, we had a case of an individual who had been visiting his neurologist for many years without a clear diagnosis," says molecular geneticist Madhuri Hegde, PhD, FACMG, a professor at Emory University School of Medicine and vice president

GOOD QUESTION

My doctor gave me a diagnosis. Why should I get a genetic test?

Genetic testing can improve the accuracy of a clinical diagnosis by pinpointing an individual's disease-causing mutation. This can lead to eligibility for certain therapies or medications and clinical trials, as well as a better understanding of how the disease will progress.

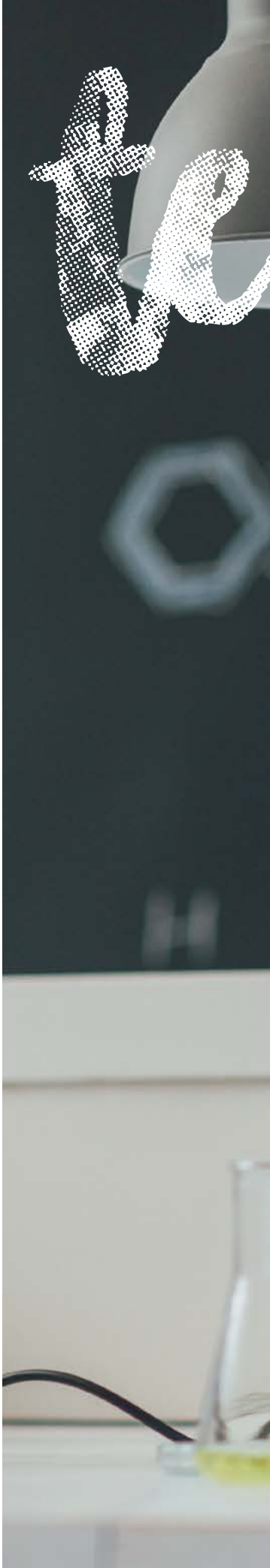

IMAGE: ISTOCK.COM/VGAJIC

Testing

Better testing allows more people to get a diagnosis that opens doors to treatments and research

BY DONNA ALBRECHT


Kelly Berger

GOOD QUESTION

Can't I order a genetic testing kit on the internet? There are many DNA testing kits on the market that claim to reveal your ancestry or provide health insights. But over-the-counter genetic testing kits can't match the specificity of the tests ordered in a clinical setting and don't offer the clinical help and counseling your care team delivers with test results.

and chief scientific officer of PerkinElmer Genomics. "When the sample was sent to us for genetic testing, the neurologist was planning a muscle biopsy, which can be painful. We were able to find a disease-causing change in the *GAA* gene, which can cause adult-onset Pompe disease, and thereby did not need the invasive procedure." That genetic diagnosis also allowed the individual to start enzyme-replacement therapy, currently the most effective treatment for Pompe disease.

ADVANCES IN GENETIC TESTING

Genetic testing becomes more accessible and accurate every year. This has as much to do with scientific discoveries in the neuromuscular disease field as it does with advances in testing technology.

Genetic tests look for specific gene mutations that are known to be associated with particular diseases. Genetic mutations have been identified for a fraction of the many neuromuscular diseases we know. But scientists are continuing to search for and identify more disease-causing gene mutations. With each discovery, the possibilities for genetic testing expand.

At the same time, testing technology is improving. "Years ago, we could only test for one gene at a time," says Kristin Engelstad, MS, CGC, a genetic counselor at Columbia University. "As the technology changed, we had tests that could cover more. Now, if I have a patient with muscle weakness, I can check 70–100 genes for sequencing."

In addition to finding gene mutations, some test panels can evaluate how a patient will respond to a particular treatment.

WHERE TO START

If you're considering genetic testing, begin by talking with your doctor and a genetic counselor (see "What's a Genetic Counselor?"). They will consider your symptoms and family history to determine the appropriate tests to order.

Most genetic tests involve taking a blood sample or cheek swab, which is sent to a lab for testing. When the results are in, the physician and genetic counselor will share the results and advise you on appropriate treatments or clinical trials.

Some people are hesitant to undergo genetic tests when there is no cure for their disease. But at a time when science is advancing quickly in the field of gene-targeted therapies, it makes sense to have a genetically confirmed diagnosis in order to be prepared as new therapies emerge.

Having an accurate diagnosis can have positive impacts on other areas of your life. For example, you and your family may be able to plan for the future knowing

GOOD QUESTION

I already had a genetic test. Do I need another? Genetic testing is advancing quickly. If your previous genetic test was negative or inconclusive, keep asking about new testing. Anyone considering a gene-targeted therapy also should ask if their testing needs to be repeated.

The First Step to Testing

If you're interested in genetic testing, talk with the care team at your MDA Care Center. To find your MDA Care Center, visit **mda.org/care/mda-care-centers**.

WHAT'S A GENETIC COUNSELOR?

Genetic counselors are medical professionals who have advanced training in medical genetics and counseling.

Jennifer Roggenbuck

Their role is to guide and support patients and families who are seeking information about genetic conditions.

At MDA Care Centers, genetic counselors are often part of multidisciplinary care teams. Your doctor may refer you to a genetic counselor before or after genetic testing.

Jennifer Roggenbuck, MS, CGC, a genetic counselor and associate professor at the Ohio State University Wexner Medical Center, explains that when she meets with a patient, she begins by asking about their family and medical history. With this history, she can address concerns about diseases that might run in the family, and she can recommend specific genetic tests.

"It's important that patients and clinicians understand the possible outcomes before embarking on the testing journey," Jennifer says.

After genetic testing, she can help patients and their families understand test results and provide them with emotional support as they continue on that journey.

the possible progression of your disease. You may also find social networks of people who share the same diagnosis.

GETTING ANSWERS

Since receiving her genetically confirmed diagnosis, Kelly has joined a clinical trial. She hopes she can help advance research and discover treatments for her condition.

She's found another benefit that is less tangible but no less important. "I finally have the answer to that question that's been lingering my whole life," Kelly says. "Yes, it's just a name and some numbers; it ultimately doesn't define me. But it feels like I have a bit of closure after not knowing for so long."

Donna Albrecht is a health writer based in San Francisco.

GOOD QUESTION

Is genetic testing expensive? Genetic testing is becoming more affordable. MDA's Resource Center may be able to help you find resources for genetic testing. Call us at **833-ASK-MDA1**.

GOOD QUESTION

If I have a diagnosis, should my family be tested? Talk with your genetic counselor about who in your family should be tested based on the type of disease and who else is showing symptoms. Relatives may be interested in genetic testing to find out if they are carriers. Carriers have a chance of having children with the same genetic disease.

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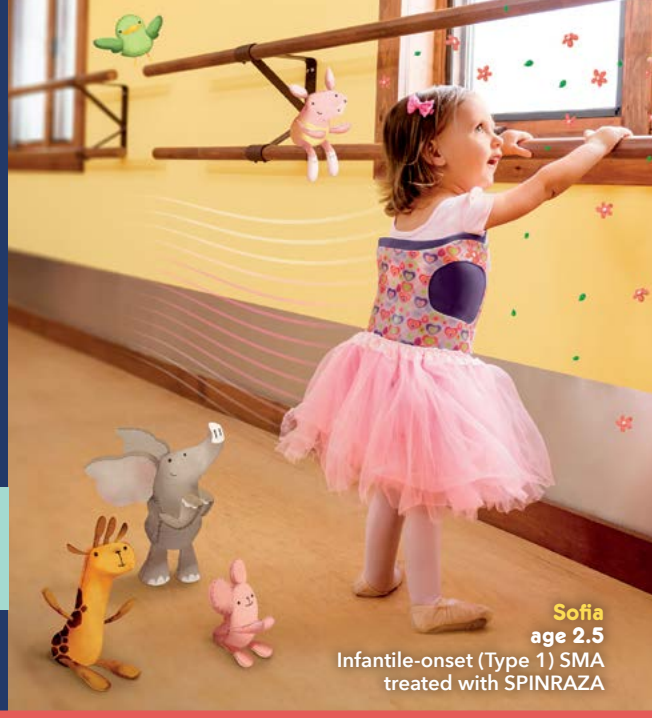

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*Based on commercial patients in the US (including Puerto Rico) as of September 2019.
†Based on commercial patients in the US (including Puerto Rico) as of August 2019.

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.

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

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.

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.

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

QUESTIONS?

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

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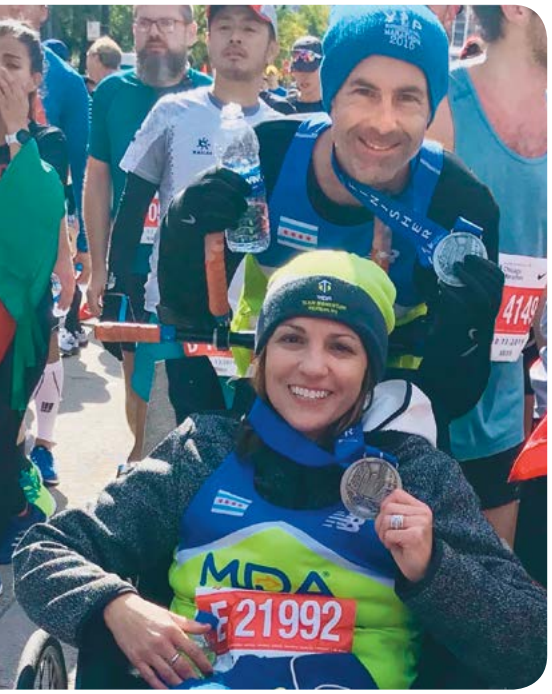
Your guide to the MDA community, from news briefs to inspiring profiles



Race of a Lifetime

"Running as a bike duo in the Chicago Marathon was life-changing for me. My favorite part was the roaring of the crowd as we went by. Hearing them cheer us on by name was validation that the 45 years I have lived with muscular dystrophy haven't been for nothing — they've been for encouraging people along the way without even realizing it."

—Amy Shinneman and her husband, Jamie, finished the 2019 Bank of America Chicago Marathon with Team Momentum

Off to the Races

Visit **mdateam.org** and save your spot on MDA's Team Momentum for one of these upcoming races:

- United Airlines NYC Half Marathon, March 15, 2020
- Trans New Hampshire Bike Ride, June 26-28, 2020
- San Francisco Giant Race, Sept. 6, 2020
- Bank of America Chicago Marathon, Oct. 12, 2020
- 50th TCS New York City Marathon, Nov. 1, 2020

The Shamrocks Are Coming

There's more to celebrate in March than just St. Patrick's Day — it's also a time to make a difference for those living with neuromuscular diseases. For almost 40 years, MDA has been partnering with businesses and institutions to raise money for MDA services with its Shamrocks campaign. From restaurants to gas stations and grocery stores, there are many opportunities to turn an errand into an act of good.

Find participating stores or donate online at mda.org/shamrocks.

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 ClinicalTrials.gov (NCT02814019) or SiderosDMD.com or by emailing us at sideros@santhera.com.

santhera

Living, Not Suffering

An MDA Ambassador learns to live life to the fullest

BY CHRISTOPHER CARROLL

Christopher Carroll, fifth from left in the second row, was team captain of the Carroll Crusade at a recent MDA Muscle Walk event in the Philadelphia area.

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**.

I often hear people say that I suffer from muscular dystrophy. I don't like this use of the word "suffer." I am living my life to the fullest, and don't feel like I am suffering at all.

I may need to ask for help sometimes or adapt to my surroundings, but I can always be myself. Coming to that realization took years of hard work and some tough life lessons.

KEEPING UP APPEARANCES

Growing up, I was always an active kid. I would spend hours outdoors playing basketball, football, and going to the pool with my friends.

As I grew older, I realized that I moved a bit differently than other kids. I am the second-youngest of eight children, and my oldest brother was already diagnosed with Becker muscular dystrophy (BMD). Being familiar with his condition, I realized I must have muscular dystrophy, too. Soon, my doctors diagnosed me with BMD.

Instead of embracing it, I tried to hide the fact that I had a neuromuscular disease. I started to avoid physical activities in front of strangers, and I made up ridiculous reasons why I couldn't run as fast as the other kids. I would stand and walk abnormally straight to try to hide my condition.

I was so determined not to let my condition define me that I completely turned my back on it. In retrospect, this caused more harm than good, as I stopped seeing my doctors and didn't seek out help when I was having trouble coping with the effects of the disease.

For years, I struggled with a tug of war between my feelings of self pity and the need to keep a happy outward appearance.

FATE STEPS IN

In 2012, I was struck by a car while crossing the street in Sea Isle City, N.J., and I had to stay off my feet as I healed. Being off my feet for so long caused my muscles to weaken to the point that I never regained the ability to walk. I have been in a manual wheelchair ever since. This made it harder to hide that I have a disability, but it took me a few more years to finally embrace my condition and start to take control of my disease. I could not have done this without the support of my family; my amazing fiancé, Joy; and my wonderful doctors.

Around this time, I began to see my neurologist at the University of Pennsylvania again. It wasn't long before my doctors began to suspect that I did not have BMD.

At my doctor's suggestion, I met with a genetic counselor. We talked at length about

how muscular dystrophies are inherited and the importance of understanding the gene sequencing of each type. She also explained how the genetic testing process works and urged me to get it done immediately. That day, I spat into a tube, and about two weeks later I received my official diagnosis: limb-girdle muscular dystrophy type 2D.

This was a game changer for me. I finally had a definitive diagnosis that I could research and do something about.

Since finding my diagnosis, I have become an MDA Ambassador and joined my local Muscle Walk Committee.

At a recent appointment with my doctor, I noticed that she always talks about "when" a cure is found, never "if." This gives me the drive to continue to do my part to make that "when" sooner rather than later.

NO MORE HIDING

Throughout the year, you can find my fiancé and me at a ton of concerts or my nieces' and nephews' sports events. We love staying active, and we never let my condition stand in the way.

Looking back, one of my biggest regrets is that I kept my neuromuscular disease in the shadows for so long. I am now in a position to live my life to the fullest and help find a cure. I would not be in this position if I didn't decide to find out my official diagnosis and get involved with the MDA community. Q

Christopher (right) met Philadelphia Phillies player Rhys Hoskins and gave a speech at an MDA Muscle Walk event.

Christopher Carroll, 36, is a sales representative for a remodeling company. He lives in East Norriton, Penn., and looks forward to getting married in July 2020.

Making Waves

Our photo contest winner has a taste for adventure

2019 PHOTO CONTEST WINNER

Congratulations to Brent Gillespie of Benton, Ark., our Lasting Impression Photo Contest winner. This photo, taken in the summer of 2019, captures Brent, 43, trying adaptive wakeboarding for the first time on Lake Ouachita, a popular spot for water sports in Arkansas. The men wakeboarding with him are volunteers from Wake the World, a nonprofit offering water sport experiences for people with disabilities, wounded veterans, and others.

Brent was among a group of people who took turns on the wakeboard and cheered each other on from the shore. "It was like a family; everyone kind of had something in common," says Brent, who has a clinical diagnosis of limb-girdle muscular dystrophy (LGMD) and uses a power wheelchair.

Being on the water gave him a taste for adventure. "I was having a blast, just trying to lick the water," Brent says. He hopes to try skydiving or hang gliding next.

More Reader Photos

Congratulations to Sarah Krieher of Mesa, Ariz., and Whitney Powell of Caney, Kan., the runners-up in our contest. See their photos at **mda.org/quest**.

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