MDA INNOVATIONS IN SCIENCE

RESEARCH REPORT

MDA® | Muscular Dystrophy Association

> “MDA has always encouraged the funding of **out-of-the-box** ideas that look at neuromuscular diseases from novel perspectives.”
>
> —Robert Baloh, MD, PhD, Cedars-Sinai

Charging Toward Cures

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

A single breakthrough can lead to a cure, and in 2018, MDA supported 224 research projects worldwide. Our umbrella model of funding research across many neuromuscular diseases means findings from one disease often enable progress in others, maximizing the speed at which we can make progress.

MDA is the largest non-governmental funder of neuromuscular disease research in the country, supporting more than 40 neuromuscular diseases including muscular dystrophy, ALS, and many others

SINCE ITS INCEPTION IN 1950, MDA HAS INVESTED MORE THAN

$1 BILLION

IN NEUROMUSCULAR DISEASE RESEARCH TO UNCOVER NEW TREATMENTS AND CURES

IN 2018 MDA FUNDED

224

GRANTS IN 31 STATES AND 11 COUNTRIES REPRESENTING A COMMITMENT OF MORE THAN

$58M

IN 2018 MDA INVESTED MORE THAN

$16M

IN RESEARCH PROJECTS AND AWARDED 88 NEW GRANTS

How Yesterday's Investments Have Created Today's Breakthroughs: Approved Therapies

Support for MDA's research enables MDA to fund leading research teams working toward breakthrough therapies, which can have a life-changing impact on patients. MDA-funded breakthroughs include drugs for amyotrophic lateral sclerosis (ALS), Duchenne muscular dystrophy (DMD), periodic paralysis, Pompe disease, and spinal muscular atrophy (SMA). Work MDA has funded in the past has helped enable drug development and new therapies, some of which are described on the following pages.

MDA'S RESEARCH INVESTMENTS HELPED LEAD TO THE DEVELOPMENT OF

SIX (6)

OF THE TEN (10) FDA-APPROVED DRUGS TO TREAT NEUROMUSCULAR DISEASE

> "The basic science research that is annually supported by the MDA is like a fountain, springing forth new discoveries that drive future treatments and cures."
>
> —Joe Metzger, PhD, University of Minnesota

SPINRAZA FOR SMA

In December 2016, the US Food and Drug Administration (FDA) approved nusinersen (brand name Spinraza) for treating SMA, the leading genetic cause of death in infants. Spinraza is the first FDA-approved treatment for SMA and works by increasing levels of the missing survival motor neuron protein (SMN) to address the underlying cause of the disease.

MDA Role in Funding
MDA-backed research laid the foundation for what would one day become Spinraza. MDA invested nearly $750,000 in awards to Dr. Adrian Krainer at Cold Spring Harbor Laboratory in New York for early-stage development of this therapy. Dr. Krainer went on to win the 2019 Breakthrough Prize in Life Sciences for his work in developing Spinraza.

EXONDYS 51 FOR DMD

In September 2016, the FDA approved eteplirsen (brand name Exondys 51) for treating DMD, a deadly genetic disorder characterized by progressive loss of voluntary muscle movement. Exondys 51 is the first drug approved to treat DMD and the first disease-modifying drug for Duchenne. It is designed to make a partially functional version of the critical missing protein in DMD.

MDA Role in Funding
In 1986, MDA-supported researchers identified a gene that, when flawed, leads to DMD. In 1987, the protein associated with this gene was identified and named dystrophin. MDA has funded many DMD projects over the years. In 2000, MDA began funding work by Dr. Steve Wilton at Murdoch University in Australia. It was Dr. Wilton's work in exon skipping that specifically led to the development of Exondys 51.

THE HUMAN IMPACT

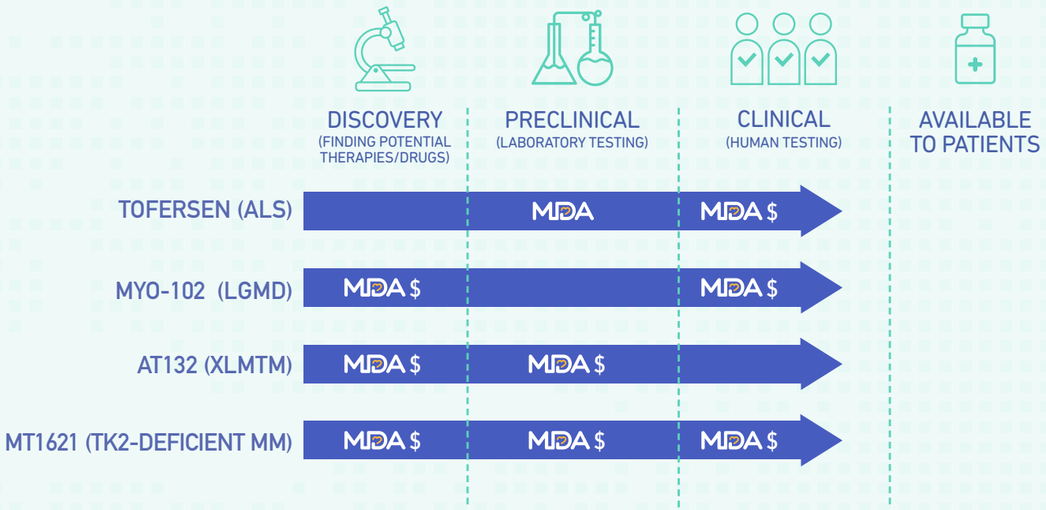
MDA National Ambassador Faith Fortenberry, a second grader who lives with SMA type 2, is just one patient benefitting from treatment with Spinraza. The drug has had a dramatic impact on Faith's life, improving her upper body strength and respiratory function.

"Spinraza has been a game-changer," says her mother, Leeann. "Faith can lift things, take the lids off of markers, hold a glass, roll on her own, brush her own teeth. The biggest thing, though, is that she's able to breathe better."

Spinraza has helped Faith stay out of the hospital, which has not only enabled her to take on a busy travel schedule as a National Ambassador but also might very well have saved her life.

"Her lungs were so weak before that whenever she had sniffles she had to go to the ICU," Leeann says. "Now she's breathing deeper and longer. She's coughing now — she never was able to cough, or even sneeze. It's been huge for her."

How Yesterday's Investments Have Created Today's Breakthroughs: Therapies in Development



"MDA has supported my research into gene therapy for muscular dystrophy for more than 25 years. In the early years few people believed that gene therapy would ever work. However, MDA believed in my ideas, and without their support the AAV/micro-dystrophins that are currently being tested in clinical trials for DMD would have never been developed."

— Jeff Chamberlain, PhD, University of Washington

TOFERSEN IN CLINICAL TRIALS FOR ALS

Tofersen is an investigational therapy for ALS caused by mutations in the superoxide dismutase 1 gene (*SOD1*), which is one type of familial ALS that makes up 2% of all ALS cases. Tofersen is designed to prevent or decrease the SOD1 protein, which is thought to be toxic when mutated in ALS. If successful, this treatment would be the first ALS therapy targeting the root genetic cause of the disease.

MDA Role in Funding
MDA granted Dr. Timothy Miller at Washington University School of Medicine more than $1 million from 2007 to 2012 to conduct toxicology studies and a phase 1 clinical trial on a first-generation therapy against SOD, which was developed by Ionis Pharmaceuticals. While this first therapy was determined to need adjustments to improve its potency, this was the first trial testing this type of drug in neuromuscular disease through injection into the spinal canal, and it paved the way for the trials currently in progress.

MDA also funded natural history studies run by Drs. Miller and Michael Benatar that were essential to the design of the clinical trials. MDA granted Dr. Benatar about $1 million from 2007 to 2013, and Dr. Miller was awarded more than $227,000 in MDA funding from 2012 to 2015.

MYO-102 IN TRIALS FOR LGMD

In January 2019, the FDA awarded Orphan Drug Designation to MYO-102, an investigational gene-replacement therapy for limb-girdle muscular dystrophy type 2D (LGMD2D).

In LGMD2D, mutations in the *SCGA* gene cause decreased levels of the gene product alpha-sarcoglycan. MYO-102 uses an adeno-associated virus (AAV) to deliver a functional copy of the *SCGA* gene to the muscles of individuals with LGMD2D, targeting the root genetic cause of the disease. This technology, initially licensed from Nationwide Children's Hospital (NCH) Myonexus Therapeutics, is now in clinical trials by Sarepta Therapeutics.

MDA Role in Funding
MDA awarded grants to Drs. Steven Roberds and Kevin Campbell supporting the work that led to the discovery of the original gene mutations for LGMD2D in 1994. MDA also supported the early development of this gene therapy in LGMD2D and other work done by Dr. Jerry Mendell of NCH by way of multiple grants totaling more than $600,000. These grants allowed for the earliest LGMD2D gene therapy trials in humans, work that has directly led to the current trials of MYO-102.

AT132 IN TRIALS FOR XLMTM

Audentes Therapeutics is currently conducting a phase 1/2 clinical trial (ASPIRO) of AT132, an *MTM1* gene-replacement therapy for people with X-linked myotubular myopathy (XLMTM). Interim results presented at the World Muscle Society conference in October 2018 showed that all treated patients continued to exhibit meaningful improvements in neuromuscular and respiratory function.

MDA Role in Funding
Multiple MDA grants helped Dr. Jocelyn Laporte then at the University of Strasbourg, France, discover and characterize the gene for myotubularin, *MTM1*, in the late 1990s. Later, Dr. Martin (Casey) Childers received MDA support in the amount of $369,365 from 2011 to 2014 to test AAV-MTM1 in animal models. This preclinical work has formed the basis for Audentes' testing AAV-MTM1 therapy in humans.

MT1621 IN DEVELOPMENT FOR TK2-DEFICIENT MITOCHONDRIAL MYOPATHY

In February 2019, the FDA awarded Breakthrough Therapy Designation to MT1621, Modis Therapeutics' investigational therapy for thymidine kinase 2 (TK2) deficiency. TK2 deficiency is a type of myopathy caused by a reduction of mitochondrial DNA. These types of diseases are also referred to as mitochondrial DNA depletion syndromes.

MDA Role in Funding
In 2005, Dr. Michio Hirano of Columbia University was awarded an MDA grant totaling $198,800 to create a TK2-deficient mouse model. In 2012, with an additional $380,000 in MDA funding, he demonstrated that by supplementing those animals with building blocks to make more mitochondrial DNA, the health of the animals was restored.

In 2012, Dr. Hirano obtained approval from the FDA for "compassionate use" of his experimental therapy and treated a child with severe, infantile-onset TK2 deficiency. The child responded favorably, and to date, more than 30 other patients have been treated. In August 2018, MDA awarded Dr. Hirano $300,000 to help support an ongoing open-label, expanded-access clinical trial of the TK2 therapy, while Modis Therapeutics is further developing the therapy for FDA approval.

How MDA Supports Research

Today's funding shapes tomorrow's breakthroughs

Our grants represent a continued commitment to fund groundbreaking research that will one day lead to treatments and cures. Each grant impacts neuromuscular disease research in a different way, from better understanding the underlying mechanisms of a particular disease to uncovering therapeutic targets to building clinical research infrastructure that will expedite clinical trials.

OUR FUNDING PROCESS

OUR MISSION
Accelerate therapies and cures

NEUROMUSCULAR DISEASE
MDA funds research in more than 43 different diseases that affect muscle function

APPLICATION SUBMITTED
Researchers with creative ideas for impacting neuromuscular disease submit research projects

APPLICATION REVIEW
Research projects are reviewed by our panels of expert scientists and clinicians

PROJECT FUNDED
The most meritorious and impactful projects are funded

AWARD MANAGEMENT
MDA regularly evaluates scientific progress of projects

RESEARCH OUTCOMES
MDA-funded projects have been impacting lives for more than 65 years

Grant Types

MDA's grant programs span the entire drug development spectrum, from the earliest discovery stages through clinical trials. The strategy of our research programs is to evaluate for every disease where the biggest gaps and opportunities lie, aiming to fill them with impactful efforts.

DISCOVERY RESEARCH

- **Research Grant (RG)** — Accelerates a progress toward understanding and treating neuromuscular disease
- **Development Grant (DG)** — Promotes training of promising new neuromuscular disease researchers

TRANSLATIONAL RESEARCH

- **MDA Venture Philanthropy (MVP)** — Funds discovery and clinical application of treatments and cures for neuromuscular diseases in order to de-risk drug development efforts in academia and industry

CLINICAL RESEARCH

- **Human Clinical Trial Grant/Clinical Research Grant (HCTG/CRG)** — Focuses on clinical studies such as natural history and biomarker development, as well as clinical trials of compounds already on the market
- **Clinical Research Network Grant (CRNG)** — Accelerates "clinical trial-readiness" for specific diseases by helping a distributed network of clinics enhance communications, coordinate activities, and standardize procedures, with the goals of collecting robust natural history data and defining the outcome measures and infrastructure to facilitate future clinical trials
- **Clinical Research Training Scholarship (CRTS)** — Promotes the training of early-stage investigators entering the field of academic clinical neuromuscular research

INFRASTRUCTURE

- **Research Infrastructure Grant (RIG)** — Supports the development of tools, techniques, and services needed by the neuromuscular research community for therapy development

CONFERENCES

- **Conference Grant (CG)** — Supports conferences, meetings, and workshops to facilitate the exchange of scientific ideas and data

> "We are grateful to donors for supporting us and to the MDA for the careful stewardship of these funds."
>
> —Christine Vande Velde, PhD, University of Montreal

2018 Grant Highlights

IN 2018, MDA CONTINUED FUNDING PROMISING DISCOVERY, PRECLINICAL, AND CLINICAL RESEARCH TO HELP ACCELERATE THERAPIES AND CURES

These are highlights of some projects from MDA's 2018 funding portfolio, grouped according to MDA's areas of strategic focus.

MDA STRATEGIC PRIORITIES

DISCOVERY	PRECLINICAL	CLINICAL		
Understanding cause of disease • Gene/protein function and dysfunction mechanisms • Creation of disease models	**Developing or improving treatments** • Identify new therapeutic approaches • Test and optimize new experimental drugs	**Screening & diagnosis** • New gene discovery • Genetic testing programs • Technology/ resources for diagnostic testing	**Preparing for clinical trials** • Biomarkers • Natural history of disease progression • Identify and standardize measures to use in future trials	**Clinical trials** • Early-stage tests of therapeutic interventions in humans
$23.8M COMMITMENT	**$20.2M** COMMITMENT	**$13.6M** COMMITMENT		

DISCOVERY

Understanding cause of disease

Todd J. Cohen, PhD, at the University of North Carolina at Chapel Hill was awarded $300,000 over three years to create and study a novel ALS mouse model. Drug development for ALS has been hindered by the lack of a mouse model that recapitulates the most common form of the disease, sporadic ALS. This new mouse model develops many hallmarks seen in humans with sporadic ALS and may be useful for drug testing in the future.

64 ACTIVE ALS GRANTS IN 2018

Rashmi Kothary, PhD, at Ottawa Hospital Research Institute was awarded $300,000 over three years to study abnormal fatty acid metabolism in SMA. Spinraza can increase levels of the missing SMN protein in the brain and spinal cord, but other disease-related abnormalities may arise in peripheral tissues. With this funding, Dr. Kothary will gain a better understanding of the multi-organ nature of the disorder.

MDA HAS INVESTED MORE THAN **$48 MILLION** IN SMA RESEARCH TO DATE

Lori Wallrath, **PhD**, at the University of Iowa was awarded $285,254 over three years to use a fruit fly model to examine ways to restore muscle function in Emery-Dreifuss muscular dystrophy (EDMD). Dr. Wallrath will test whether mutations in lamins — proteins that line the inner membrane of the nucleus — contribute to muscle disease and whether experimental drugs might reverse muscle loss.

Anita Corbett, PhD, at Emory University was awarded $299,997 over three years to study the molecular basis of oculopharyngeal muscular dystrophy (OPMD). The underlying genetic cause of OPMD is a mutation in a single gene, *PABPN1*. Dr. Corbett plans to test whether it is sufficient to inhibit the disease-causing mutant PABPN1 protein, or if, in addition, normal PABPN1 protein will need to be replaced.

2018 Grant Highlights

FUNDING EXAMPLES

PRECLINICAL

Developing or improving treatments

Mark Rich, MD, PhD, at Wright State University was awarded $251,565 over three years to study the blocking of transient receptor potential ion channel, TRPV4, as an approach to treating myotonia congenita (MC).

22 PROJECTS WITH COMPANIES OR ACADEMIC-INDUSTRY PARTNERSHIPS IN 2018

AcuraStem Inc. was awarded MDA Venture Philanthrophy (MVP) funding totaling $300,000 over two years to support preclinical development of a small molecule therapeutic for ALS. AcuraStem will perform proof-of-concept studies in a mouse model of ALS and develop biomarkers for its orally delivered, blood-brain-penetrating drug candidate.

MDA HAS INVESTED MORE THAN **$56 MILLION** IN MG RESEARCH TO DATE

Henry Kaminski, MD, at George Washington University was awarded $367,187 over three years to test a therapeutic strategy (based on a complement inhibitor drug) in cell and rat models for treating myasthenia gravis (MG), with the intent to demonstrate the feasibility of the approach and then move to human clinical trials.

Melissa Spencer, PhD, at the University of California at Los Angeles was awarded $300,000 over three years to develop nanoparticles for the delivery of gene therapy for DMD and other neuromuscular disorders. The current delivery platform for gene-editing and gene-replacement therapies, adeno-associated virus, has a number of limitations. In this work, Dr. Spencer will develop nanoparticles that are designed to more efficiently carry drugs or nucleic acids to specific tissues in the body.

Andrew Lieberman, MD, **PhD**, at University of Michigan was awarded $300,000 over three years to test a modified antisense oligonucleotide therapy to treat spinal-bulbar muscular atrophy (SBMA). Working with Ionis Pharmaceuticals, Dr. Lieberman will complete preclinical studies in a mouse model to establish the safety and efficacy of the therapeutic molecule.

CLINICAL

Screening and diagnosis

Alan Beggs, PhD, at Boston Children's Hospital was awarded $300,000 over three years to discover new genes that cause nemaline myopathy in patients and families who don't have any of the previously known mutations. To better understand the biological pathways that lead to disease and to aid in the search for effective therapies, Dr. Beggs will then study animal models containing the newly uncovered mutations.

James Lupski, MD, PhD, at Baylor College of Medicine, was awarded $300,000 over three years to use next-generation sequencing methods for patients and families whose genetic diagnosis has been elusive, leading not only to more diagnoses for individuals but also to discovery of new disease-associated genes for a host of neuromuscular diseases.

52 GRANTS HAVE APPLICABILITY ACROSS MULTIPLE DISEASES COVERED BY MDA

Daniel MacArthur, PhD, at the Broad Institute of MIT and Harvard was awarded $110,000 to create a neuromuscular disease-specific platform for the Rare Genomes Project. The LGMD Rare Genomes Project will offer free whole-genome testing and genetic variation analysis to individuals whose prior LGMD gene panel testing did not provide a definitive genetic explanation of their disease.

14 ACTIVE LGMD GRANTS IN 2018

"MDA's support of our research program was critical in characterizing the underlying mechanisms of a novel genetic cause of ALS."

—Giovanni Manfredi, MD, PhD, Weill Cornell Medical College

"MDA's support of our research program was critical in the discovery of a new epigenetic defect in the *FXN* gene that causes Friedreich's ataxia. With MDA's continued support, this defect is now being investigated as a therapeutic target, which is a source of much hope for people and families affected by Friedreich's ataxia."

—Sanjay I. Bidichandani, MBBS, PhD, University of Oklahoma College of Medicine

2018 Grant Highlights

FUNDING EXAMPLES (CONTINUED)

Preparing for clinical trials

Mary M. Reilly, MD, FRCP, FRCPI, at University College London Institute of Neurology was awarded $1 million over three years to evaluate a new magnetic resonance imaging (MRI) protocol designed to detect disease-related changes in muscles over time in Charcot-Marie-Tooth disease (CMT).

13 ACTIVE CMT GRANTS IN 2018

Nicholas Johnson, MD, at Virginia Commonwealth University was awarded $598,348 over three years to conduct a natural history study in children with congenital myotonic dystrophy (DM) to define endpoints that could be used in future clinical trials.

MDA HAS INVESTED MORE THAN **$50 MILLION** IN DM RESEARCH TO DATE

Chad Heatwole, MD, at the University of Rochester was awarded $200,000 over two years to develop and validate a health index to be used as a patient-reported outcome tool in clinical trials for DMD.

Clinical trials

Stanley Appel, MD, at Houston Methodist was awarded $558,800 over 2.5 years to test in a phase 2a clinical trial whether infusion of a type of expanded immune cell called regulatory T-lymphocytes is safe and could be used as a potential therapy for ALS. This grant was co-funded by the ALS Association and ALS Finding a Cure as part of the ALS ACT program.

19 MDA GRANTS WERE CO-FUNDED WITH OTHER ORGANIZATIONS

Kevin Flanigan, MD, at Nationwide Children's Hospital and Ohio State University was awarded $528,798 over two years to conduct a pilot clinical trial in boys age 4 to 7 years old with DMD to study whether treatment with spironolactone can ameliorate the negative side effects of prednisolone.

MDA SPENT MORE THAN **$26 MILLION** ON DMD GRANTS IN THE LAST FIVE YEARS

CLINICAL RESEARCH NETWORK GRANTS

In addition to the other grants to help prepare disease fields for clinical trials, MDA also aims to promote and accelerate clinical-trial-readiness via Clinical Research Network Grants. These strategic awards allow a distributed network of clinical sites to enhance communications, coordinate activities, and standardize procedures with the goals of collecting robust natural history data and developing the outcome measures and infrastructure to facilitate clinical trials.

Four clinical research networks were active in 2018 with MDA support:

Charles Thornton, MD, at the University of Rochester leads the Myotonic Dystrophy Clinical Research Network, which currently includes nine medical centers. MDA, along with other funders, helped establish the network with funding in 2013. A second MDA award of $918,000 over three years has continued to support this important resource.

Michael Benatar, MD, PhD, at the University of Miami Miller School of Medicine and **Jonathan Katz, MD,** at California Pacific Medical Center were awarded $300,000 over two years to support work conducted through the Clinical Research in ALS and Related Disorders for Therapy Development (CReATe) Consortium.

Jeffrey Statland, MD, at the University of Kansas Medical School was granted an award of $1.2 million over three years to support seven medical centers that specialize in facioscapulohumeral muscular dystrophy (FSHD) research and clinical care.

Nicholas Johnson, MD, of Virginia Commonwealth University was awarded $705,003 over three years to establish and lead the Limb-Girdle Muscular Dystrophy Clinical Research Network consisting of seven medical centers.

> "We are extremely grateful for support from MDA in getting the network launched and keeping it going. When we started we hoped that we could encourage more companies and scientists to work on developing treatments for myotonic dystrophy because they would see that all the tools are in place to conduct good clinical trials. We can see that this is happening."
>
> —Charles Thornton, MD, University of Rochester

2018
Grant Highlights

PIPELINE OF SCIENTISTS

SINCE 1952, MDA HAS INVESTED NEARLY **$120 MILLION** IN PROVIDING EARLY CAREER SUPPORT TO MORE THAN **2,200 EARLY-STAGE SCIENTISTS AND CLINICIANS**

IN 2018, MDA FUNDED **48 GRANTS** GEARED TOWARD THE TRAINING AND DEVELOPMENT OF EARLY-STAGE SCIENTISTS AND CLINICIANS

Similar to fostering and maintaining a robust pipeline of new drugs, it is also important to encourage the next generation of scientists and clinicians to work in the neuromuscular disease field and make the important discoveries of tomorrow. MDA has a long-held commitment to early-in career professionals, through a number of grant mechanisms that help the best and brightest postdoctoral scientists and clinical fellows establish independent research programs and secure their first faculty positions.

Katharine Nicholson, MD, at Massachusetts General Hospital was awarded $86,666 over two years to study early markers of disease in C9ORF72 ALS, the most common genetic form of ALS. This grant is being co-funded by the American Academy of Neurology and the American Brain Foundation and originated as part of a partnership with the ALS Association.

Johanna Hamel, MD, at the University of Rochester in New York was awarded a fellowship to compare RNA toxicity in DM type 1 versus type 2. The two-year award — co-sponsored by MDA with the American Academy of Neurology and the American Brain Foundation — will provide a total of $130,000 to support Dr. Hamel's work to shed light on the molecular processes that drive DM.

Dwi Kemaladewi, PhD, was awarded $180,000 over three years to explore the potential of CRISPR/Cas9 gene-editing technology as a potential therapy in merosin-deficient congenital muscular dystrophy (MDC1A). Since the award was made, Dr. Kemaladewi started an independent faculty position at Children's Hospital of Pittsburgh.

Lukasz Sznajder, PhD, at the University of Florida was awarded $180,000 over three years to use next-generation sequencing to discover blood biomarkers for ALS, frontotemporal dementia, and DM2.

"I really wouldn't be able to do what I am doing now without the initial support from MDA."

— Jinsy A. Andrews, MD, MSc,
Columbia University

MDA Clinical Research Training
Grant recipient 2007-2009

Building for the Future

GENE CATALYST INITIATIVE (GCI)

At MDA we are focusing intensely on understanding the genetic and molecular origins of neuromuscular diseases to advance genetic diagnoses and to support the development of disease therapies — including antisense oligonucleotides and gene-replacement and gene-editing therapies — that target the underlying genetic cause. Our GeneCatalyst Initiative is a multi-faceted effort to drive support for continued gene-targeted research and therapy development.

This initiative builds upon the long history that MDA has had in supporting genetic medicine, appreciating that the best way to move toward treatments and cures is to understand the root cause of the disease and target that as closely as possible. Thus, from inception, MDA has contributed greatly to the field of muscle disease and toward landmark research advances, including the identification of the first human disease-causing gene as well as the first human trial of gene transfer for a muscle disease. For several decades, MDA-supported researchers have discovered the gene-causing mutations for many other neuromuscular disorders, developed and refined gene-delivery tools and methods, and funded innovative research looking for other creative ways to circumvent the problems created by genetic defects.

Indeed, recent FDA approvals for gene-targeted therapies for neuromuscular diseases, as well as even more experimental therapies currently in advanced clinical trials, are the result of decades of effort from a large community of partners, including families, donors, researchers, and clinicians. With the GeneCatalyst Initiative, MDA will continue to work together with these partners to keep the momentum going toward more treatments and cures.

IN 2018 MDA FUNDED
31 RESEARCH PROJECTS UNDER
THE GENE CATALYST INITIATIVE

MOVR DATA HUB

NEUROMUSCULAR OBSERVATIONAL RESEARCH

In 2018, MDA launched the MOVR (neuroMuscular ObserVational Research) Data Hub, an expanded disease registry platform leveraging MDA's Care Center network to capture more real-world data in a number of different diseases. Building upon the pilot neuromuscular disease registry MDA has had in place since 2013 in four diseases — ALS, SMA, DMD, and Becker muscular dystrophy (BMD) — in 2019 MOVR also began including clinician-entered data in FSHD, LGMD, and Pompe disease at Care Centers in the MDA network.

The goal of MOVR, as a centralized data hub for multiple neuromuscular diseases with an eye toward integrating the clinician-entered data with other datasets (such as genomics and patient-reported outcomes), is to provide a robust repository of real-world information that will be available to a broad community of healthcare providers, researchers, and industry partners in order to improve health outcomes as well as clinical trial design and recruitment.

Thus, as MOVR accumulates more longitudinal data and integrates with other datasets, the data hub could serve as an unparalleled one-stop resource, an asset to researchers probing for a richer understanding of neuromuscular disease and improved coordination of care for individuals.

Sonia Skarlatos Public Service Award

We are proud to announce that the American Society for Gene and Cell Therapy awarded MDA the Sonia Skarlatos Public Service Award for 2019. The award recognizes a person or group that has consistently fostered and enhanced the field of gene and cell therapy through governmental agencies, public policy groups, public education, or non-governmental charitable organizations.

Appendix

2018 ADVISORY COMMITTEES

MDA RESEARCH ADVISORY COMMITTEE MEMBERS

Stanley Appel, MD - Chair	Houston Methodist, TX
Louis Kunkel, PhD - Chair	Boston Children's Hospital, MA
Robert Baloh, MD, PhD	Cedars-Sinai Medical Center, CA
Elisabeth Barton, PhD	University of Florida, FL
Aaron Beedle, PhD	SUNY-Binghamton, NY
Alan Beggs, PhD	Boston Children's Hospital, MA
Sanjay Bidichandani, MBBS, PhD	University of Oklahoma, OK
Dean Burkin, PhD	University of Nevada, NV
Jeffrey Chamberlain, PhD	University of Washington, WA
M. Casey Childers, DO, PhD	University of Washington, WA
Thomas Cooper, MD	Baylor College of Medicine, TX
Thomas Crawford, MD	Johns Hopkins University, MD
Merit Cudkowicz, MD, MSc	Massachusetts General Hospital, MA
John Day, MD, PhD	Stanford University, CA
James Dowling, MD, PhD	Hospital for Sick Children, CAN
Fen-Biao Gao, PhD	University of Massachusetts, MA
Matthew Harms, MD	Columbia University, NY
Lawrence Hayward, MD, PhD	University of Massachusetts, MA
Michio Hirano, MD	Columbia University, NY
Joe Kornegay, DVM, PhD	Texas A & M University, TX
Clothilde Lagier-Tourenne, MD, PhD	Massachusetts General Hospital, MA
Christian Lorson, PhD	University of Missouri, MO
Giovanni Manfredi, MD, PhD	Weill Cornell Medical College, NY
Nicholas Maragakis, MD	Johns Hopkins University, MD
Katherine Mathews, MD	University of Iowa, IA
Elizabeth McNally, MD, PhD	Northwestern University, IL
Lynn Megeney, PhD	Ottawa Health Research Institute, CAN
Matthew Meriggioli, MD	Rush University, IL
Daniel Miller, MD, PhD	University of Washington, WA
Timothy Miller, MD, PhD	Washington University, MO
Carlos Moraes, PhD	University of Miami, FL
Brett Morrison, MD, PhD	Johns Hopkins University, MD
Kanneboyina Nagaraju, DVM, PhD	SUNY – Binghamton, NY
Bradley Olwin, PhD	University of Colorado, CO
Udai Pandey, PhD	University of Pittsburgh, PA
John Ravits, MD	University of California, San Diego, CA
Louise R. Rodino-Klapac, PhD	Ohio State University, OH
Alessandra Sacco, PhD	Sanford Burnham Institute, CA
Rita Sattler, PhD	Barrow Neurological Institute, AZ
Michael Shy, MD	University of Iowa, IA
Melissa Spencer, PhD	University of California, Los Angeles, CA
Charlotte Sumner, MD	Johns Hopkins University, MDw
Christine Vande Velde, PhD	University of Montreal, CAN
Eric Wang, PhD	University of Florida, FL
C. Chris Weihl, MD, PhD	Washington University, MO
Noah Weisleder, PhD	Ohio State University, OH
*R. Rodney Howell, MD	University of Miami, FL

*Ex Officio Member

> "I truly enjoyed being part of the Research Advisory Committee. It has always been one of the best-run review boards. It was a privilege to be part of the great MDA team."
>
> —Merit Cudkowicz, MD, Massachusetts General Hospital

MDA VENTURE PHILANTHROPY ADVISORY COMMITTEE MEMBERS

Stanley Appel, MD	Houston Methodist, TX
Andrew Billin, PhD	Gilead Sciences, CA
Barry J. Byrne, MD, PhD	University of Florida, FL
Thomas Cheever, PhD	NIAMS/NIH, MD
Cristina Csimma, PharmD, MHP	Csimma, LLC, MA
Kurt Fischbeck, MD	NINDS/NIH, MD
Amelie Gubitz, PhD	NINDS/NIH, MD
John E. Howell	Prometheus Partners Holdings, LLC, VA
Louis Kunkel, PhD	Boston Children's Hospital, MA
Melanie Leitner, PhD	Accelerating NeuroVentures, LLC, MA
Elizabeth McNally, MD, PhD	Northwestern University, IL
Jeffrey Rothstein, MD, PhD	Johns Hopkins University, MD
Lee Wrubel, MD	EgonZehnder, NY
Bruce Wentworth, PhD	BMW Research Consulting, MA
Dione T. Kobayashi, PhD	AkeOla, MA

JUNE 2019