Summer 2026 • Vol. 6 / Issue 3

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

2026 LGMD SCIENTIFIC SUMMIT

Elevating the Patient Voice, Advancing Research, and Shaping the Future of LGMD Care

A NEW MODEL FOR DRUG DEVELOPMENT

Patients Going where Pharma Won't

ASK THE EXPERT

Urvi Desai, MBBS, MD

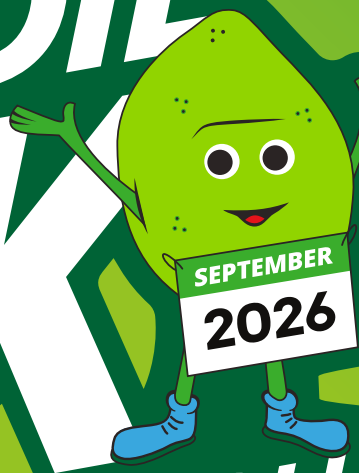
GRASP-LGMD CONSORTIUM

Accelerating the Development of Gene Therapies for LGMD

Approaches to Better Sleep

for Individuals Living with LGMD

GIRDIE 5K WALK-RUN-ROLL FOR LGMD AWARENESS



SPONSORED BY BRIDGEBIO

Visit lgmd-info.org/5k to learn more and register by August 15th!

LGMD
Awareness Day
September 30th

Join the global movement for Limb-Girdle Muscular Dystrophy at the inaugural **Girdie 5K Walk-Run-Roll for LGMD Awareness** this September 2026.

Wherever you are in the world, you can walk, run, or roll to raise awareness and support organizations advancing care, research, and hope for the LGMD community.

Together, we can make a difference. Be part of the movement!

Mark your calendar for the 12th Annual LGMD Awareness Day!

LGMD Awareness Day Toolkit:

Scan here to access flyers, social media posts, media kits, merch, and more!

LGMD AWARENESS FOUNDATION

lgmd-info.org

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How To Donate

Visit **TheSpeakFoundation.com** and select "Donate" to support our mission and programs. The Speak Foundation is a 501(c)(3) nonprofit charity. All donations are secure and tax deductible.

Thank you for your support!

The Speak Foundation
Uniting the entire LGMD community to make a difference together in future treatments for this rare disease.

The origin of The Speak Foundation's name comes from Proverbs 31:8. It is: "Speak up for those who have no voice." Living with a rare disease means many of us wait years to have a voice in areas that impact our daily lives personally. The Speak Foundation helps our voices to be heard.

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X.com/LGMDFoundation

Uniting Science, Care, and Community

Dear LGMD Community,

The upcoming LGMD Scientific Summit hosted by The Speak Foundation on July 31, 2026, marks an important milestone for the limb-girdle muscular dystrophy community. This Summit brings together patients, advocates, regulators, clinicians, researchers, and industry leaders with a shared mission of accelerating progress toward effective treatments and improved care for individuals living with LGMD.

The Summit highlights that, despite setbacks over the past year, there is growing momentum in LGMD therapy development, including both progress in clinical trials and development of novel treatment approaches.

The Summit highlights that, despite setbacks over the past year, there is growing momentum in LGMD therapy development, including both progress in clinical trials and development of novel treatment approaches. There is an increasing emphasis on patient-centered research and multidisciplinary collaboration between stakeholders. Events like this are critical for strengthening partnerships between the scientific community and patients and families whose voices continue to shape the future of research and care.

A particularly exciting advancement is the launch of the LGMD Centers of Excellence in the US, three of whose clinical leads will be presenting at the Summit. These Centers provide coordinated, multidisciplinary care for US patients while expanding clinical trial readiness and therapeutic development. This infrastructure is needed to support the transition of future treatments to the clinic.

We also recognize the tremendous contributions of Dr. Urvi Desai, an active member of the GRASP-LGMD Consortium and this issue's featured Q&A expert. The GRASP-LGMD network plays a vital role in advancing research, developing biomarkers, improving clinical understanding of LGMD, and preparing the field for precision-based therapies. This issue also features a practical article by Dr. Desai on improving sleep for individuals living with LGMD — a helpful resource we are proud to share.

Finally, as part of a new series on Work with LGMD, Hank Eichner shares his story of how he redefined his career by creating a business centered on flexibility, accessibility, and independence. His story reflects challenges many individuals with muscle weakness face in traditional employment while highlighting adaptability, resilience, and innovation.

If you have a story to share that highlights adaptability in work, daily life, caregiving, or overcoming challenges, we invite you to submit a brief proposal for consideration to ***ContactUs@TheSpeakFoundation.com***. ■

Kat Bryant Knudson

Kathryn Bryant Knudson
Editor In Chief
Founder & CEO, The Speak Foundation

Connect with Us

ContactUs@TheSpeakFoundation.com

International Consortium of LGMD Organizations

United States

The Speak Foundation
Uniting the entire LGMD community
TheSpeakFoundation.com

Beyond Labels & Limitations
Funding research for LGMD R1/2A and educating on its disease course
BeyondLabelsLimitations.com

Breathe with MD
Educating and raising awareness about breathing muscle weakness in neuromuscular disease
BreatheWithMD.org

CamronsCure
Funding research for LGMD R18/2S
Facebook.com/LGMDCC

Coalition to Cure Calpain 3
Funding research for LGMD R1/2A
CureCalpain3.org

CureLGMD2D Research Foundation
Funding research for LGMD R3/2D
sujivasu01@gmail.com

CureLGMD2i
Funding research for LGMD R9/2I
CureLGMD2i.org

CureLGMD2T
Raising awareness for LGMD R19/2T and GMPPB-related Congenital Muscular Dystrophies
CureLGMD2T.org

Dion Foundation
Funding research for LGMD R5/2C
TheDionFund.org

Kurt + Peter Foundation
Funding research for LGMD R5/2C
KurtPeterFoundation.org

LGMD Awareness Foundation
Raising awareness of and advocating for the LGMD community
LGMD-Info.org

LGMD-1D DNAJB6 Foundation
Representing LGMD D1/1D and DNAJB6 subgroup
LGMD1D.org

LGMD2D Foundation
Funding research for LGMD R3/2D and educating patients and physicians
LGMD2D.org

LGMD2i Research Fund
Funding research for LGMD R9/2I and educating the patient community
LGMD2iFund.org

LGMD2L Foundation
Representing the LGMD R12/2L Anoctamin5-related community
LGMD2L-Foundation.org

Team Titin
Strengthening the titin community: LGMD R10/2J
TitinMyopathy.com

The Jain Foundation
Funding research for LGMD R2/2B and educating the patient community
Jain-Foundation.org

Argentina

ADM Argentina Muscular Dystrophy LGMD Group
Funding research for neuromuscular diseases
ADM.org.ar

Australia

Daniel Ferguson LGMD2A Foundation
Funding research for LGMD R1/2A and educating the patient community
DFFoundation.com.au

France

"GI LGMD"/LGMD Patient Group of AFM-Telethon
Focusing on all subtypes of LGMD, supporting research and educating the patient community
LGMD.AFM-Telethon.fr

Italy

Conquistando Escalones Association
Funding research for LGMD D2/1F
ConquistandoEscalones.org

"GFB ONLUS"/Family Group of Beta-Sarcoglycanopathy
Representing the LGMD R5/2C Gamma Sarcoglycan-related, LGMD R3/2D Alpha Sarcoglycan-related, LGMD R4/2E Bèta-Sarcoglycan-related, and LGMD R6/2F Delta-Sarcoglycan-related communities
LGMD2e.org

UILDM – Unione Italiana Lotta alla Distrofia Muscolare
Focusing on all subtypes of LGMD, raising awareness and providing support for the entire Italian community
UILDM.org

Italian Association Calpain 3
Funding research for the LGMD R1/2A Calpain 3-related community
AICA3.org

Japan

Patients' Association for Dysferlinopathy Japan
Representing the Japanese and International LGMD R2/2B Dysferlin-related and Miyoshi Muscular Dystrophy 1 (MMD) communities
PADJ.jp/index.html

Netherlands

Stichting Spierkracht
Raising awareness and supporting the LGMD R3/2D Alpha Sarcoglycan-related community
StichtingSpierkracht.com

Pakistan

Muscular Dystrophy Pakistan
Advancing care and awareness for Muscular Dystrophy, including LGMDs, in Pakistan
MuscularDystrophy Pakistan.com.Free

South Korea

Korean Dysferlinopathy Patients Association
Providing patients with LGMD R2/2B information and research updates
Cafe.Naver.com/ UniteDsyferlinopathy

Spain

Conquistando Escalones Association
Funding research for LGMD D2/1F
ConquistandoEscalones.org

Proyecto Alpha
Funding research for LGMD R5/2C Gamma Sarcoglycan-related, LGMD R3/2D Alpha Sarcoglycan-related, LGMD R4/2E Bèta-Sarcoglycan-related, and LGMD R6/2F Delta-Sarcoglycan-related communities
ProyectoAlpha.org

United Arab Emirates

CureSCG
Dedicated to accelerating research and treatment for Sarcoglycanopathies
CureSCG.org

For additional information about the International Consortium of LGMD Organizations: **ContactUs@TheSpeakFoundation.com**

Urvi Desai, MBBS, MD

Director of Levine Children's Comprehensive Neuromuscular Program, Director of Adult and Pediatric MDA Care Center at Atrium Health and Professor of Neurology at Wake Forest University of School of Medicine

Meet the Expert

Urvi Desai, MBBS, MD, is Director of Levine Children's Comprehensive Neuromuscular Program, Director of Adult and Pediatric MDA Care Center at Atrium Health and Professor of Neurology at Wake Forest University of School of Medicine. She obtained her graduate medical degree at MS University, India, followed by residency in Neurology and a fellowship in Neuromuscular Disorders, both at UNC Chapel Hill. She has established cross-sub-specialty physician and allied health team collaborations in multidisciplinary Adult and Pediatric MDA clinics. She has received MGFA and CMT/HNF Center for Excellence awards as well as the "Researcher of the Year" award for Atrium Health Neuroscience 2020. Her research interest includes researching the overlap of sleep and neuromuscular disorders to help deploy non-invasive ventilation earlier than suggested by diurnal measures in patients with muscular dystrophies and ALS. She serves as the Principal Investigator in multiple interventional and observational trials in MG, CIDP, DMD, SMA, hereditary TTR Amyloidosis, CMT, and LGMDs including R2/2B, R9/2I, and R4/2E. She is a member of the GRASP-LGMD consortium. She serves on the Advisory Board for the North Carolina site for MD Star Net, a CDC-funded network that conducts public health surveillance of muscular dystrophy.

Q

I have been diagnosed with LGMD R2/2B and would like to ask about the current status of treatment development. What is a realistic outlook for patients in terms of when effective therapies might become available?

A

There are currently no FDA-approved treatments for LGMD R2/2B (dysferlinopathy), but progress in research is underway across multiple therapeutic strategies. Gene therapy is more complicated for dysferlinopathy than for sarcoglycanopathies or LGMD R9/2I because the size of the dysferlin gene exceeds the carrying capacity of AAVs, which are used to deliver DNA in most current gene therapies. Thus, work on R2/2B gene therapy has concentrated on "dual vector" approaches, in which the gene is divided into two pieces, each delivered by an AAV. The two pieces then combine (as the DNA/RNA or the dysferlin protein), resulting in production of the full-length protein.

The dual-vector approach is inherently more difficult and less efficient than gene therapy that delivers a gene in one piece, which is one reason why gene therapies for sarcoglycans and LGMD R9/2I have advanced more rapidly. However, the first drug using dual vector gene therapy (for otoferlin, a protein closely related to dysferlin, which is necessary for hearing) received FDA approval in April 2026. At the American Society of Gene and Cell Therapy in May 2026, new dysferlin studies highlighted ways to improve delivery efficiency, including better muscle targeting and improved reassembly of split gene fragments inside cells.

In addition to gene therapy, a number of approaches are being investigated in dysferlinopathy. Some of these are targeted to specific mutations, while others are drugs approved for other types of muscular dystrophy or other diseases. The dysferlinopathy field is at a pivotal point — natural history data, outcome measures, and preclinical therapeutic candidates are now largely in place, and the transition to interventional clinical trials is anticipated in the coming years.

Q

My follow up question is: if a treatment does become available in the future, will it be accessible to patients from financially limited backgrounds? Are there likely to be any support programs, financial assistance options, or affordable access pathways for patients who may not be able to afford high-cost therapies?

A

A diagnosis often brings legitimate concerns regarding the eventual cost and accessibility of these first-in-class medicines. It is important for patients to know that the medical and advocacy communities are working on access long before a drug ever hits the pharmacy shelf. For rare diseases, several structural safeguards typically come into play. Most pharmaceutical companies developing "orphan drugs" establish robust Patient Assistance Programs (PAPs) to cover co-pays or provide the drug at no cost to those who are uninsured or underinsured. Furthermore, international and domestic advocacy organizations often provide travel grants for clinical trial participation and financial navigation services.

The most impactful action you can take today is to ensure you are connected with the larger LGMD advocacy community and are enrolled in your subtype-specific registry (the Dysferlin Registry). This ensures that when therapies become available or clinical trials are initiated, you are visible to the researchers and the systems designed to provide them, regardless of your financial background.

Q

Is there any known connection between menopause and progression or symptom changes in muscle-wasting conditions like LGMD?

A

There are currently no studies that have specifically looked at how menopause affects the progression of LGMD, but research does show that the hormonal changes of menopause can have an impact on muscle health. Estrogen, a hormone that drops sharply during menopause, plays an important role in keeping muscles strong by protecting muscle cells from damage, helping muscles repair themselves, and reducing inflammation.

After menopause, women can lose a noticeable amount of muscle mass — studies show about a 6% decrease compared to before menopause — and muscle strength also tends to decline faster during this time. While more research is needed to confirm these connections in LGMD patients specifically, it may be helpful for women with LGMD to work closely with their doctors during the menopausal transition to monitor muscle function more carefully.

Staying physically active within individual limitations and maintaining good nutrition, including adequate protein intake, are practical steps that may help preserve muscle strength during this time. Hormone replacement therapy is another option worth discussing with a doctor on an individual basis, though it has not been specifically studied in people with muscular dystrophy.

The most impactful action you can take today is to ensure you are connected with the larger LGMD advocacy community and are enrolled in your subtype-specific registry (the Dysferlin Registry). This ensures that when therapies become available or clinical trials are initiated, you are visible to the researchers and the systems designed to provide them, regardless of your financial background.

Question

Gentle, low-impact aerobic exercise such as swimming and stationary bicycling improves cardiovascular performance, increases muscle efficiency, and lessens fatigue. Patients should hydrate adequately, avoid exercising to exhaustion, and avoid supramaximal, high-intensity exercise.

Q

Are there specific types of exercise you would recommend for someone with LGMD R12/2L to maintain strength and function safely?

A

The American Academy of Neurology (AAN) recommends that clinicians advise patients with muscular dystrophy that aerobic exercise combined with supervised submaximal strength training is probably safe. Gentle, low-impact aerobic exercise such as swimming and stationary bicycling improves cardiovascular performance, increases muscle efficiency, and lessens fatigue. Patients should hydrate adequately, avoid exercising to exhaustion, and avoid supramaximal, high-intensity exercise.

Clinicians should educate patients about warning signs of overwork-related weakness and myoglobinuria, including feeling weaker rather than stronger after exercise, excessive muscle soreness 24–48 hours later, severe muscle cramping, heaviness in the extremities, and prolonged shortness of breath as well as dark-colored urine. Referral to a multidisciplinary clinic with access to physical therapy, occupational therapy, and other specialties is recommended for long-term complex care strategies.

An exercise trial specifically in LGMD R12/2L patients was conducted by Vissing et al.[1] It demonstrated that moderate-intensity aerobic cycling (30 minutes, 3×/week for 10 weeks) produced a 27% improvement in VO_2 max (maximum oxygen intake) and a 35% improvement in sit-to-stand time, with creatine kinase (CK) levels remaining stable and no adverse effects noted. This is particularly reassuring given the pronounced hyperCKemia characteristic of ANO5 myopathy and confirms that moderate aerobic exercise does not exacerbate membrane damage.

A more recent randomized controlled trial[2] of personalized home-based aerobic exercise with coaching in patients with various neuromuscular diseases (6-month intervention) similarly showed improved peak oxygen uptake (mean difference 2.2 mL/min/kg) with stable CK levels and no increase in adverse events compared to usual care.

- **Avoid:**
  Avoid heavy resistance training of already weakened muscles. High-impact activities that involve strong "lengthening under load" of muscles (such as jumping, running downhill, or fast lowering movements) should also be avoided. Exercising to exhaustion or using very high-effort ("all-out") effort is not recommended.
- **Monitor:**
  CK can be considered to establish baseline before starting an exercise program. Stable CK during training is reassuring, as demonstrated in the Vissing study. Any significant CK spikes, myoglobinuria, or worsening weakness after exercise should prompt program modification or cessation.
- **Individualize:**
  Generic exercise protocols may lead to fatigue, pain, and poor compliance in LGMD patients. Programs should be tailored to the individual's specific pattern of weakness and supervised by a physical therapist familiar with neuromuscular disease.

References

1. C.R. Vissing, N. Preisler, E. Husu, K.P. Prahm, and J. Vissing, Aerobic training in patients with Anoctamin 5 Myopathy and HyperCKemia, *Muscle Nerve*. 2014 Jul;50(1):119-23. doi: 10.1002/mus.24112.
2. S. Oorschot, M.-A. Brehm, A.C. van Groenestijn, T. Veneman, J. Twisk, et al., Efficacy of Combined Aerobic Exercise and Coaching on Physical Fitness in People with Neuromuscular Diseases: A Randomized Clinical Trial, *Neurology*. 2025 Jul 8;105(1):e213781. doi: 10.1212/WNL.0000000000213781.

Q

I have been living with LGMD R1/2A for 20 years. Who are the leading doctors in India specializing in neuromuscular genetic diseases? When can we expect research or clinical trials for LGMD in India?

A

A large-scale clinical exome sequencing study of 207 genetic myopathy patients from the Indian subcontinent found that LGMD R1/2A accounted for 19% of confirmed diagnoses. This highlights its relatively high frequency in this population and the importance of continued clinical research and expertise in neuromuscular diseases in India, particularly LGMD R1/2A. Fortunately, several institutions throughout India have established expertise in LGMDs. It may be helpful to contact clinicians at one of the following centers, which have established expertise in LGMDs:

- **NIMHANS** (National Institute of Mental Health and Neurosciences), Bangalore
- **Bombay Hospital**, Mumbai
- **AIIMS** (All India Institute of Medical Sciences), New Delhi
- **PGIMER** (Post Graduate Institute of Medical Education and Research), Chandigarh

EDITOR'S NOTE: The Spring 2025 Issue of *LGMD Magazine* contains an article describing visits by members of the John Walton Muscular Dystrophy Research Centre (JWMDRC) in Newcastle, UK, and the Jain Foundation with clinicians and patients at Bombay Hospital and AIIMS.

Have a Question for Our Experts?

Send Questions To:
ContactUs@TheSpeakFoundation.com

From ML Bio to BridgeBio Neuromuscular: A Story of Urgency, Partnership, and Evolution

For many in the neuromuscular disease community, the name ML Bio Solutions carries deep meaning. It represents persistence in the face of uncertainty, a family's determination to find answers, and a scientific journey that refused to stall. Today, that same spirit lives on—expanded and accelerated—within BridgeBio Neuromuscular.

This is a story of evolution.

ML Bio Solutions
a bridgebio company

BRIDGEBIO NEUROMUSCULAR

A Beginning Rooted in Urgency

The origins of ML Bio Solutions (also known as "ML Bio") are as personal as they are scientific. More than two decades ago, a family set out to confront a diagnosis that offered few options: limb-girdle muscular dystrophy type 2I/R9 (LGMD2I/R9), affecting their own child.

At the time, the scientific understanding of the disease was still nascent, and therapeutic strategies to treat it were only theoretical. What followed was a sustained, deeply focused effort to change that reality. In 2001, a gift from the McColl & Lockwood families enabled the creation of the McColl-Lockwood Laboratory for Muscular Dystrophy Research. Since its founding, a team of experts has diligently advanced research and contributed to a growing body of knowledge aimed at addressing the root causes of—and potential treatments for—LGMD2I/R9.

In 2018, this research delivered promising preclinical evidence for a potential new therapy for LGMD2I/R9: BBP-418. This same year, ML Bio Solutions was founded to carry the baton forward into clinical trials, by forming a team to develop BBP-418 as quickly and safely as possible.

For patients and care partners, ML Bio became synonymous with hope, grounded in action. For researchers and clinicians, it represented a rare combination of scientific rigor and lived experience.

2000

2001
A generous gift from the McColl & Lockwood families supports creation of research laboratory to look for a potential therapy for LGMD2I/R9

2018
Published preclinical work establishing impact of BBP-418 in LGMD2I/R9 model from McColl-Lockwood Laboratory at Atrium Health

Late 2018
ML Bio team formed

2019
ML Bio partners with BridgeBio Pharma, Inc. to advance program and reach patients as quickly as possible

A Pivotal Transition

As the science matured, the ability to increase speed and scale became both possible and increasingly urgent, marking the beginning of a new chapter. In 2019, ML Bio joined forces with BridgeBio, a biotechnology company built to accelerate the development of therapies for genetic diseases.

The partnership between ML Bio and BridgeBio did not erase ML Bio's identity. Instead, it amplified it.

For nearly a decade, ML Bio and BridgeBio have worked relentlessly to develop BBP-418 for LGMD2I/R9.

Accelerating Toward Impact

One of the most tangible outcomes of this partnership is the development of BBP-418, a molecule targeting the underlying biology of LGMD2I/R9.

In a field where timelines often stretch across decades, BBP-418 was advanced from Investigational New Drug application to Phase 3 clinical trial results and a New Drug Application in just 6 years. This pace reflects more than efficiency—it reflects a deliberate alignment of science, patient need, and operational execution. For clinicians, it signals a shift toward more responsive drug development. For researchers, it demonstrates what is possible when infrastructure supports innovation. And for patients and care partners, it brings the prospect of a therapy closer than ever before.

Importantly, this progress builds directly on the foundation laid by ML Bio. Partnership with BridgeBio did not restart the clock; it accelerated it.

The Emergence of BridgeBio Neuromuscular

With BridgeBio and ML Bio's continued commitment to developing therapies for neuromuscular diseases comes the creation of BridgeBio Neuromuscular—a dedicated group of scientists, patient advocates, and trailblazers united by a clear mandate: to improve the lives of people living with neuromuscular disease, as quickly and effectively as possible.

The team carries forward the founding vision of ML Bio while expanding its reach. Where the early work focused intensely on LGMD2I/R9, the scope now includes a broader commitment to neuromuscular disease communities.

Continuity Within Change

ML Bio continues to exist—its legacy, expertise, and foundational work remain intact. What changed is the context: its mission is now supported by a larger ecosystem, purpose-built to translate scientific insight into approved therapies more efficiently.

The name may have changed in certain contexts, but the driving force has not. The same urgency that began with a single family's search for answers now powers a broader effort to deliver solutions for entire communities.

ML Bio's legacy is not behind us; it is embedded within everything BridgeBio Neuromuscular does.

The same urgency, the same commitment, and the same focus on patients continue—now supported by more resources and a broader effort to reach more people.

BBP-418 is an investigational product and has not been approved by the FDA or any other Regulatory Health Authority for any indication.

GRASP-LGMD Consortium:

Accelerating the Development of Gene Therapies for LGMD

Established in 2018, the GRASP-LGMD Consortium (Genetic Resolution and Assessments Solving Phenotypes in Limb-Girdle Muscular Dystrophy) is an international clinical research network dedicated to accelerating the development of therapies, including gene therapies, for LGMD. The consortium currently includes sites across the United States, Europe, and New Zealand that collaborate closely with one another.

This work is carried out through a variety of natural history studies involving multiple LGMD subtypes, with the goal of developing standardized outcome measures to track disease progression across patient populations. ■

Have a Question for Our Experts?

Send Questions To:
ContactUs@TheSpeakFoundation.com

1. **Zineb Ammous**, MD, FACMG
   The Community Health Clinic, Indiana
2. **Ellie Carrell**, PhD
   Virginia Commonwealth University
3. **Urvi Desai**, MD
   Atrium Health Carolinas Medical Center
4. **Jordi Diaz-Manera**, MD, PhD
   Newcastle University, UK
5. **Stacy Dixon**, MD, PhD
   University of Colorado
6. **Nicholas Johnson**, MD, MSCI, FAAN
   Virginia Commonwealth University
7. **Peter Kang**, MD
   University of Minnesota
8. **Kathryn Bryant Knudson**
   The Speak Foundation
9. **Doris Leung**, MD, PhD
   Kennedy Krieger Institute, Baltimore
10. **Katherine Mathews**, MD
    University of Iowa
11. **Tahseen Mozaffar**, MD, FAAN
    University of California, Irvine
12. **Richard Roxburgh**, Mb ChB, FRACP, PhD
    University of Auckland
13. **Valeria Sansone**, MD, PhD
    Centro Clinico NeMO, Milan
14. **Jeffrey M. Statland**, MD
    University of Kansas Medical Center Research Institute
15. **Sub Subramony**, MD
    University of Florida
16. **Conrad "Chris" Weihl**, MD, PhD
    Washington University in St. Louis
17. **Matthew Wicklund**, MD
    UT Health San Antonio

ELEVATE THE PATIENT VOICE, ADVANCE RESEARCH, AND SHAPE THE FUTURE OF LGMD CARE

2026 LGMD SCIENTIFIC SUMMIT

LIMB GIRDLE MUSCULAR DYSTROPHY

The **2026 LGMD SCIENTIFIC SUMMIT** will bring together an exceptional lineup of leading researchers, expert clinicians, and innovative biotech companies who are at the forefront of limb-girdle muscular dystrophy (LGMD) research and care. These respected voices from across the scientific and medical community will share the latest breakthroughs in emerging treatments, provide updates on active and upcoming clinical trials, and offer insight into the future of therapeutic development for LGMD. ***Be part of this conversation of hope, innovation, and what's ahead for the LGMD community!***

A HUGE THANK YOU
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AskBio

A PROJECT OF The SPEAK Foundation

TheSpeakFoundation.com/lgmd-summit

2026 LGMD SCIENTIFIC SUMMIT
LIMB GIRDLE MUSCULAR DYSTROPHY

Schedule
July 31, 2026

11:00 AM — **WELCOME**
- Kathryn Bryant Knudson | Founder & CEO, The Speak Foundation

11:05 AM — **SESSION 1: CLINICAL TRIAL UPDATES**

11:05 – 11:15 AM — **LION-CS-101-AskBio's LGMD R9/2I Investigational Gene Therapy Program**
- Susan Sifers, MD | Vice President, Global Clinical Development for LGMD and Pompe Programs, AskBio

11:15 – 11:35 AM — **Where We Are Today: Sarepta's LGMD Gene Therapy Program**
- Louise Rodino-Klapac, PhD | President, R&D and Technical Operations, Sarepta Therapeutics, Inc.

Session Chair:
- Kathryn Bryant Knudson | Founder & CEO, The Speak Foundation

11:35 – 11:50 AM — **Program Update from BridgeBio Neuromuscular**
- Douglas Sproule, MD, M.Sc | Chief Medical Officer, BridgeBio Neuromuscular

11:50 AM – 12:05 PM — **Gene Therapy for LGMD R5/2C: Preliminary Safety and Efficacy Data from a Phase 1 Study of ATA-200**
- Barry Byrne, MD, PhD | Genethon/Atamyo | Professor and Associate Chair, Department of Pediatrics Director, Powell Gene Therapy Center and UF Heath Advanced Therapeutics, University of Florida, College of Medicine

12:05 – 12:15 PM — **ATA-100, an AAV9 Gene Therapy Treatment for LGMD R9/2I FKRP-related: Results from a Dose Escalation Study in Patients**
- John Vissing, MD, DMSCI | Genethon/Atamyo | Professor of Neurology at the University of Copenhagen, Denmark; Director of the Copenhagen Neuromuscular Center at the National Hospital, Rigshospitalet (Copenhagen)

12:15 PM — **BREAK**

12:30 PM — **SESSION 2: POLICY DISCUSSIONS**

12:30 – 1:00 PM — **Fireside Chat: Advancing Rare Disease Legislation through Community Collaboration**
- Neha Gandhi | Head of Global Patient Advocacy, BridgeBio Neuromuscular
- Amanda Malakoff, MPA | Executive Director, Government Affairs, BridgeBio Pharma, Inc.

1:00 – 1:40 PM — **FDA Roundtable/Patient Panel: Patient-Centered Trial Design & Regulatory Harmonization**
- Robyn Bent, MS, RN | Center for Drug Evaluation and Research (CDER); Patient-Focused Drug Development (PFDD) Program; U.S. Food and Drug Administration (FDA)
- Michelle Campbell, PhD | Associate Director for Stakeholder Engagement and Clinical Outcomes, Office of Neuroscience, Office of New Drugs, Center for Drug Evaluation and Research (CDER); U.S. Food and Drug Administration (FDA)

Co-Moderators:
- Annie Kennedy | Chief Mission Officer, EveryLife Foundation for Rare Diseases
- Kathryn Bryant Knudson | Founder & CEO, The Speak Foundation

Patient Panel:
- Carol Abraham | President & Founder, LGMD Awareness Foundation
- Kelly Brazzo | Co-Founder & CEO, CureLGMD2i Foundation
- Joshua Thayer | Advisor and former General Counsel, Jain Foundation
- Bradley Williams, PhD | Director of Research & Diagnostic Innovation, Jain Foundation

ALL TIMES ARE IN EASTERN DAYLIGHT TIME (EDT), UTC-4 | Schedule is subject to change.

2026 LGMD SCIENTIFIC SUMMIT

LIMB GIRDLE MUSCULAR DYSTROPHY

Schedule

July 31, 2026

1:40 PM **BREAK**

1:55 PM **SESSION 3: EMERGING TREATMENTS**

1:55–2:15 PM **Fast Skeletal Myosin Inhibition for Becker Muscular Dystrophy: Potential Implications for LGMD**

- Joanne Donovan, MD, PhD | Chief Medical Officer, Edgewise Therapeutics

2:15–2:35 PM **DELI550 and DualR3: Gene-Corrected Primary Human Muscle Stem Cells for Therapy in LGMD R1/2A and R3/2D**

- Simone Spuler, MD | Charite, University Medicine (Berlin, Germany)

2:35–2:50 PM **Dual AAV Gene Therapy for LGMD R2/2B**

- Casey Childers, DO, PhD | Chief Executive Officer, AskBio, Kinea Bio, Inc.

2:50–3:20 PM **Development of Allogeneic Off-the-Shelf Myogenic Progenitors for the Treatment of Muscular Dystrophies**

- Rita Perlingeiro, PhD | Lillehei Endowed Professor in Stem Cell and Regenerative Cardiovascular Medicine and Deputy Director of Scientific Engagement and Commercialization for the LHI at the University of Minnesota; Co-founder of Myogenica Inc.
- Peter Kang, MD, FAAN, FAAP, FCNS, | Director, Greg Marzolf Jr. Muscular Dystrophy Center; Professor and Vice Chair of Research in the Department of Neurology, Scholar at the Institute for Translational Neuroscience at the University of Minnesota Medicine

3:20 PM **BREAK**

3:35–3:55 PM **Developing a New Dual Gene AAV Therapy for LGMD R9/2I**

- Paul Martin, PhD | Principal Investigator, Jerry R. Mendell Center for Gene Therapy at Nationwide Children's Hospital; Associate Director, Paul D. Wellstone Muscular Dystrophy Specialized Research Center at Nationwide Children's Hospital

3:55–4:10 PM **A Novel Therapeutic Approach to Combat Degeneration and Fatty Replacement of Muscles in Dysferlinopathy**

- Monique Floer, PhD | Co-Founder, President & CEO, Advertent Biotherapeutics, Inc.

4:10–4:25 PM **Second Generation AAV Therapies for the LGMDs**

- Nicholas Johnson, MD, MSCI, FAAN | Director of the Center for Inherited Myology Research (CIMR), Armas Family Professor, Vice Chair of Research in Neurology at Virginia Commonwealth University

4:25–4:55 PM **Using Genetic Modifier Signals to Develop New Muscular Dystrophy Drugs**

- Elizabeth McNally, MD, PhD | Ikaika Therapeutics | Director, Center for Genetic Medicine, Northwestern University's Feinberg School of Medicine

Development of Anti-LTBP4 Treatment for Muscular Dystrophy

- Alexis Demonbreun, PhD | Ikaika Therapeutics | Associate Professor, Center for Genetic Medicine, Department of Pharmacology, Northwestern University's Feinberg School of Medicine

4:55–5:10 PM **Cure Rare Disease and the LGMD Pipelines: LGMD R9/2I Second Generation Gene Therapy is Under Development**

- Tahseen Mozaffar, MD, FAAN | Professor of Neurology and Pathology and Laboratory Medicine; Former Founding Director of the Division of Neuromuscular Disorders, UCI Health, Orange County, CA

5:10–5:30 PM **Closing: Addressing Structural Barriers to Treatment Development**

- Bradley Williams, PhD | Director of Research & Diagnostic Innovation, Jain Foundation

ALL TIMES ARE IN EASTERN DAYLIGHT TIME (EDT), UTC-4 | Schedule is subject to change.

2026 LGMD SCIENTIFIC SUMMIT

LIMB GIRDLE MUSCULAR DYSTROPHY

Speakers

Susan Sifers, MD
Vice President, Global Clinical Development for LGMD and Pompe Programs, AskBio

Louise Rodino-Klapac, PhD
President, R&D and Technical Operations, Sarepta Therapeutics, Inc.

Douglas Sproule, MD, M.Sc
Chief Medical Officer, BridgeBio Neuromuscular

Barry Byrne, MD, PhD
Professor and Associate Chair, Department of Pediatrics Director, Powell Gene Therapy Center and UF Heath Advanced Therapeutics, University of Florida, College of Medicine

John Vissing, MD, DMSCI
Professor of Neurology at the University of Copenhagen, Denmark; Director of the Copenhagen Neuromuscular Center at the National Hospital, Rigshospitalet (Copenhagen)

Neha Gandhi
Head of Global Patient Advocacy, BridgeBio Neuromuscular

Amanda Malakoff, MPA
Executive Director, Government Affairs, BridgeBio Pharma, Inc.

Robyn Bent, RN, MS
Center for Drug Evaluation and Research (CDER); Patient-Focused Drug Development (PFDD) Program; U.S. Food and Drug Administration (FDA)

Michelle Campbell, PhD
Associate Director for Stakeholder Engagement and Clinical Outcomes, Office of Neuroscience, Office of New Drugs, Center for Drug Evaluation and Research (CDER); U.S. Food and Drug Administration (FDA)

Annie Kennedy
Co-Moderator
Chief Mission Officer, EveryLife Foundation for Rare Diseases

Kathryn Bryant Knudson
Co-Moderator
Founder & CEO, The Speak Foundation

Carol Abraham
Patient Panelist
President & Founder, LGMD Awareness Foundation

Kelly Brazzo
Patient Panelist
Co-Founder & CEO, CureLGMD2i Foundation

Joshua Thayer
Patient Panelist
Advisor and former General Counsel, Jain Foundation

Bradley Williams, PhD
Patient Panelist
Director of Research & Diagnostic Innovation, Jain Foundation

Joanne Donovan, MD, PhD
Chief Medical Officer, Edgewise Therapeutics

Simone Spuler, MD
Charite, University Medicine (Berlin, Germany)

Casey Childers, DO, PhD
Chief Executive Officer, AskBio, Kinea Bio, Inc.

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Monique Floer, PhD
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Elizabeth McNally, MD, PhD
Director, Center for Genetic Medicine, Northwestern University's Feinberg School of Medicine

Alexis Demonbreun, PhD
Associate Professor, Center for Genetic Medicine, Department of Pharmacology, Northwestern University's Feinberg School of Medicine

Tahseen Mozaffar, MD, FAAN
Professor of Neurology and Pathology and Laboratory Medicine; Former Founding Director of the Division of Neuromuscular Disorders, UCI Health, Orange County, CA

Accepting Applications Beginning July 1, 2026

We Offer HOPE.

Are you an individual living with limb-girdle muscular dystrophy who needs assistance covering the costs of mobility and durable medical equipment (DME)? The SPEAK Foundation is here to help.

Through **The HOPE Project**, we are proud to launch the second round of our 2026 HOPE Grants program. A limited number of $350 one-time stipends will be awarded to qualified applicants living in the United States who have a diagnosis of limb-girdle muscular dystrophy and need assistance affording mobility-related DME expenses.

For additional information or to submit an application between July 1, 2026, and July 31, 2026, please visit **TheSpeakFoundation.com/grant-programs**.

The HOPE Project

A Program of The SPEAK Foundation

TheSpeakFoundation.com/grant-programs

Accepting applications from July 1, 2026 through July 31, 2026.
Available to US residents only.

An Update on the Treatment Landscape: Interim Analysis of Phase 3 FORTIFY Study of BBP-418 in LGMD2I/R9 Complete

By Andrew Huy Cao Nguyen, MD, BridgeBio Neuromuscular Medical Writing, and Jan M. Logan, BridgeBio Neuromuscular Patient Advocacy

Living with limb-girdle muscular dystrophy type 2I/R9 (LGMD2I/R9) can affect movement, strength, and daily activities over time. At present, care focuses on managing symptoms and maintaining function, as there are no **approved therapies** that target the underlying biological processes of the condition. Several investigational approaches are in development, aiming to slow progression or improve aspects of muscle function.

One investigational product in development is **BBP-418**, sponsored by ML Bio Solutions Inc., a BridgeBio Pharma, Inc. company. Clinical development of BBP-418 includes **FORTIFY**, an international, ongoing, randomized, placebo-controlled Phase 3 study.

The FORTIFY study is planned to run for 36 months. **A planned interim analysis was conducted at 12 months** and evaluated efficacy and safety outcomes that were prespecified in the study protocol.

What was evaluated in the FORTIFY study?

The FORTIFY study includes both **efficacy endpoints** and **safety assessments** to evaluate the effects of BBP-418 compared with placebo. These include laboratory measures, assessments of muscle and physical function, and safety monitoring.

Primary areas of evaluation also known as **Alpha Controlled Endpoints** include:

Biochemical markers

- Levels of **glycosylated-alpha dystroglycan (αDG)** in muscle biopsy samples, a marker related to the molecular pathway affected in LGMD2I/R9
- **Creatine kinase (CK)** levels in the blood, a marker commonly used to assess muscle damage

Functional and physiologic outcomes

- **100-meter timed test (100MTT)** which assesses walking speed
- **Forced vital capacity (FVC)**, a measure of respiratory function

Secondary outcome measures also known as **Non-Alpha Controlled Endpoints** include:

Functional and physiologic outcomes

- Walking assessment, the **10-minute walk test (10MWT)**
- **North Star Assessment for Limb-Girdle Muscular Dystrophies (NSAD)**, which evaluates muscle function across a range of daily activities
- **Performance of Upper Limb scale 2.0 (PUL 2.0)** assessment of upper limb function.

Patient-reported outcomes

- Patient-reported outcomes are being collected using **PROMIS-57** and **PROMIS-37**, a validated system of questionnaires that assess physical, mental, and social health. Data from PROMIS assessments are still being evaluated and have not been publicly reported at this time.

Throughout the study, participant safety is carefully monitored by assessing adverse events, clinical laboratory values, and other safety parameters.

Study Measures Evaluated in FORTIFY

Alpha Controlled Endpoints	What it Assesses	Why it Is Included
Glycosylated αDG	A form of alpha-dystroglycan measured in muscle tissue	To assess activity within a molecular pathway known to be altered in LGMD2I/R9
Serum CK	Creatine kinase levels in blood	A commonly used indicator of muscle injury
100MTT	100-meter timed test	To measure walking speed
FVC	Forced vital capacity	To measure lung function
Non-Alpha Controlled Endpoints	**What it Assesses**	**Why it Is Included**
NSAD	North Star Assessment for Limb-Girdle Muscular Dystrophies	To evaluate muscle function across a range of movement-related tasks
10MWT	10-minute walk test	To assess walking endurance
PUL 2.0	Performance of Upper Limbs	To assess upper limb function
PROMIS 57	Patient-reported outcome questionnaires	To assess patient-reported physical, mental, and social health (data pending)

What do these results mean for people with LGMD2I/R9?

The interim results from FORTIFY met prespecified endpoints supporting submission of a New Drug Application (NDA).

While these data are encouraging, it is important to note that:

- BBP-418 is an **investigational product and has not been approved by any regulatory agency**
- The FORTIFY study is ongoing
- Final conclusions about long-term efficacy and safety will require completion of the full trial and regulatory review

Researchers will continue to collect and analyze data as the study progresses, with the goal of informing future treatment development for people living with LGMD2I/R9. Continued participation in clinical research and careful evaluation of study results are essential steps toward improving future treatment options for the LGMD2I/R9 community.

Understanding NSAD Scores

The North Star Assessment for Limb Girdle Muscular Dystrophies (NSAD) is a clinician-administered tool that evaluates a person's ability to complete a structured set of movement related tasks. These tasks assess different aspects of muscle function, including balance, strength, and transitions between positions.

A 1-point difference in NSAD can mean the difference between:

This article is made available by our medical expert for educational purposes only to give general information and a general understanding, not to provide specific medical advice. By using this advice, you understand that there is no physician-patient relationship between you and the expert, and this advice should not be used as a substitute for a consultation with your attending medical professional.

Written By Urvi Desai, MBBS, MD
Director of Adult and Pediatric MDA Care Center at Atrium Health

Approaches to Better Sleep

for Individuals Living with LGMD

Sleep disturbances are highly prevalent in neuromuscular disorders, including LGMDs.

There can be various causes for sleep disorders in patients, in addition to those present in the general population. These include breathing difficulties which often occur at night before they become apparent in the daytime, as well as difficulty turning over or finding a comfortable position in bed. Therefore, a comprehensive approach to improving sleep in patients is needed.

Sleep Disordered Breathing (SDB) is the most clinically consequential sleep problem in neuromuscular disease and may be the earliest manifestation of respiratory muscle weakness presenting as nocturnal oxygen desaturations before daytime symptoms appear. The American Academy of Neurology (AAN) recommends referral for pulmonary or sleep medicine consultation in muscular dystrophy patients with excessive daytime sleepiness, nonrestorative sleep (frequent nocturnal arousals, morning headaches, excessive daytime fatigue), or respiratory insufficiency on pulmonary function tests (PFTs). The American College of Chest Physicians (CHEST) recommends PFTs at minimum every 6 months for NMD patients at risk of respiratory failure and sleep study/polysomnography for symptomatic patients with normal PFTs and overnight oximetry. Noninvasive ventilation (NIV) is the standard-of-care treatment for SDB in NMD and improves survival, sleep quality, sleep architecture, and quality of life.

Addressing Insomnia

Insomnia is common amongst patients with neuromuscular disorders due to a number of reasons, including but not limited to: sleep disordered breathing, inability to find comfortable positioning, and other comorbidities. In the LGMD-R9/2I cohort (currently the only LGMD subtype with dedicated insomnia data), 32% of patients met the criteria for insomnia, with prevalence rising to 54% among fatigued patients.[1]

An important first step in addressing insomnia is to practice good sleep hygiene, meaning emphasizing factors that make it easy to sleep, while avoiding those which interfere with it.

Good Sleep Hygiene Tips

- **Maintain a consistent sleep-wake schedule**
- **Maintain a cool, dark, and quiet sleep environment**
- **Avoid caffeine and alcohol later in the day and before bedtime**
- **Limit screen exposure before bedtime**
- **Time exercise appropriately** (moderate aerobic exercise can improve sleep quality, but it should not be performed close to bedtime)

"An important first step in addressing insomnia is to practice good sleep hygiene, meaning emphasizing factors that make it easy to sleep, while avoiding those which interfere with it."

Cognitive behavioral therapy for insomnia (CBT-I) is the first-line treatment for chronic insomnia recommended by multiple professional society guidelines, and it produces durable improvements in sleep architecture. A 2025 meta-analysis in JAMA Internal Medicine confirmed the efficacy of CBT-I in chronic disease populations, with adaptations for mobility-limited patients.

Core CBT-I Components Applicable to Neuromuscular Patients

- **Sleep restriction:** The most effective single component – limiting time in bed to actual sleep time, then gradually expanding
- **Stimulus control:** Associating the bed with sleep only; modified for mobility limitations by sitting upright rather than leaving bed
- **Cognitive restructuring:** Addressing catastrophic beliefs about sleep consequences
- **Relaxation techniques:** Progressive muscle relaxation (adapted to functional capacity), abdominal breathing, and mindfulness
- **Digital and telehealth CBT-I platforms** can improve access for patients with mobility limitations

When CBT-I is insufficient or unavailable, pharmacotherapy may be considered, but caution in drug selection is required in patients with neuromuscular diseases.

Applicability of Specific Drug Classes

- **Avoid benzodiazepines:** The U.S. Department of Veterans Affairs (VA) and the U.S. Department of Defense (DoD) guidelines specifically cite patients with neuromuscular diseases as a population where the risks of benzodiazepines (hypoventilation in particular) substantially outweigh benefits
- **Dual orexin receptor antagonists** (suvorexant, lemborexant, daridorexant) have a favorable profile with lower risk of respiratory depression and cognitive/psychomotor impairment compared to benzodiazepine receptor agonists, making them a reasonable option in NMDs
- **Low-dose doxepin** (3–6 mg) is effective for sleep maintenance with minimal adverse effects and no respiratory depression risk
- **Melatonin/ramelteon** may help with sleep onset but have limited efficacy for sleep maintenance. ■

Reference

1. S. Jensen, K. Abeler, O. Friborg, A. Rosner, C. Olsborg, et al., *Insomnia and sleep-disordered breathing in FKRP-related limb-girdle muscular dystrophy R9. The Norwegian LGMDR9 cohort study (2020),* J Neurol. 2024 Jan; 271(1):274–288. doi: 10.1007/s00415-023-11978-7.

Written By Hal Tily, PhD

Patients Going where Pharma Won't:

A New Model for Drug Development

For families living with rare diseases overlooked by the pharmaceutical industry, patients themselves are becoming the driving force behind drug development.

Traditional US drug development has struggled to meet the needs of the rare disease community. Commercial drug development companies must make profits and recoup the costs of developing drugs and running clinical trials. Rare diseases (defined as those affecting fewer than about 1 in 2,000 people) don't have a large market, and until 1983, suffered from almost complete "pharmacologic neglect." It was impossible for pharma companies to earn a reasonable return on their investment for such small patient populations. At that time, only 38 drugs had been approved (for use in the US) by the FDA across all rare diseases.

In the USA, the 1983 Orphan Drug Act changed some of this situation. The law introduced tax credits, grants, and fee waivers to make neglected diseases more attractive commercial targets. However, these incentives only go so far, and most rare diseases, including all LGMD subtypes are considered ultra-rare conditions — sometimes defined as those affecting fewer than 1 in 50,000 people. Even today, most ultra-rare diseases still draw very little commercial attention.

Now, patient-led organizations have developed an alternative model. A new gene therapy program targeting LGMD R12/2L (ANO5-related) illustrates this shift.

A Disease that Got Left Behind

Like other LGMDs, R12/2L causes progressive muscle weakness that eventually affects mobility and leads to loss of independence and reduced quality of life.

For several years, the R12/2L community followed developments within the broader LGMD therapeutic landscape, including gene therapy efforts underway at Sarepta Therapeutics. While Sarepta advanced programs across several LGMD subtypes, no specific updates were announced for R12/2L.

In 2025, Sarepta restructured portions of its neuromuscular pipeline, resulting in the discontinuation of multiple LGMD programs. Although this represented a setback for many within the LGMD community, including people living with R12/2L, the need for effective therapies for R12/2L remained unchanged. Accordingly, the need for a new strategy became apparent.

Today, progress continues through the commitment of patients, families, researchers, clinicians, nonprofit organizations, and industry partners working together to advance potential treatments. Recognizing this need, the 2L Foundation and Cure Rare Disease have joined forces to help accelerate research and therapeutic development for the R12/2L community.

CRD Grows from Personal Urgency to Serve Many

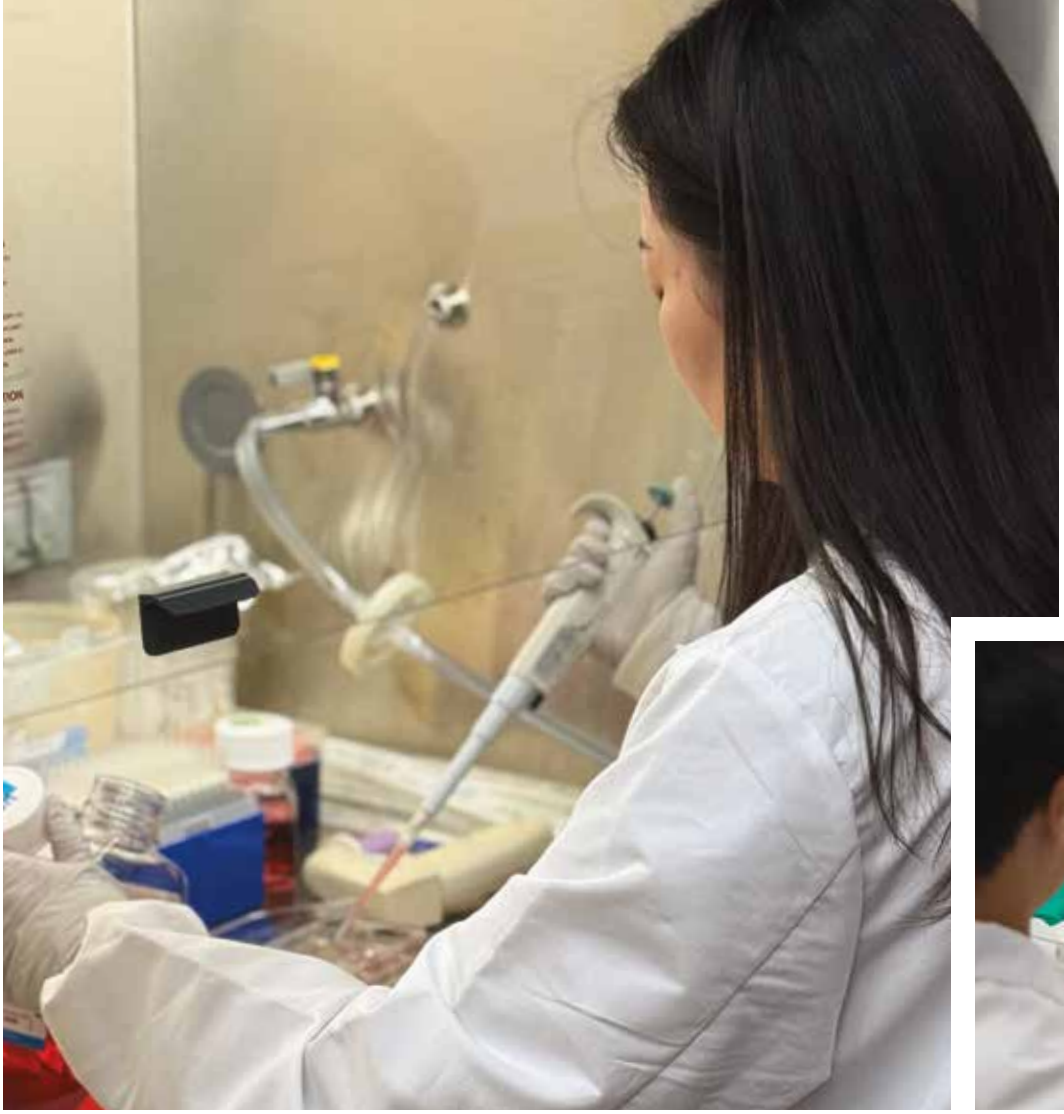
CEO Rich Horgan founded what would become Cure Rare Disease (CRD) in 2017 to develop a gene therapy for his brother, Terry, who had been battling a rare mutation of DMD since childhood. Horgan assembled a coalition of experts and created a novel, patient-oriented biotech company that was efficient enough to enable the development of therapies for even ultra-rare genetic diseases. The model was different: a nonprofit structure could take risks and set priorities that a for-profit company could not.

"I wanted to make sure the needs of patients were our ultimate outcome, not return on investment (ROI) or the amount of money raised or invested," Horgan has said. "Our goal was to help patients with rare diseases, and to prove there was a potential pathway to develop therapeutics for ultra-rare diseases."

Nonprofit biotechnology groups can also help make the process of commercial drug development more transparent. "For patients and their families, drug development has often been a black box that is difficult to see into,"

"I wanted to make sure the needs of patients were our ultimate outcome, not return on investment (ROI) or the amount of money raised or invested. Our goal was to help patients with rare diseases, and to prove there was a potential pathway to develop therapeutics for ultra-rare diseases."

— Rich Horgan • *CEO, Cure Rare Disease*



Left: Research conducted during the early stage of development is critical to understanding the safety and efficacy of treatments before human clinical trials.

Below: Cure Rare Disease focuses on research aimed at treating the genetic root causes of rare and ultra-rare diseases using ASO, gene replacement and gene editing strategies, depending on the disease indication.

Horgan observes, echoing the concerns of rare disease patients broadly and R12/2L patients in particular, who received no updates on Sarepta's development programs until they were terminated. CRD strives to be transparent about progress, while commercial entities may have conflicting incentives leading them to deprioritize patient communication and engagement.

The organization is now developing therapies covering gene replacement, gene editing (CRISPR), and antisense oligonucleotides (ASOs) for four neuromuscular and neurodegenerative rare disease subtypes, including LGMD R12/2L.

Nonprofit Groups Step Up

There is a broader movement of community groups who, knowing that industry is incentivized by differing metrics, have decided to act themselves. Though controversial, one of the earliest and most widely known examples is Augusto and Michaela Odone, who in 1984 taught themselves the biochemistry of their son Lorenzo's disease (adrenoleukodystrophy, or ALD), helped organize research efforts, and ultimately contributed to the development of a treatment celebrated in the 1992 film *Lorenzo's Oil*.

This playbook has been refined since those early days. In 2000, the Cystic Fibrosis Foundation pioneered what it called "venture philanthropy," investing $40 million into a biotech company to develop therapies targeting the defective protein in people with cystic fibrosis (CF). At the time, the move was considered radical. The bet paid off beyond anyone's expectations, and in 2012, the FDA approved Ivacaftor (Kalydeco), the first drug to address the underlying cause of CF. The Michael J. Fox Foundation, the Leukemia & Lymphoma Society, and the Juvenile Diabetes Research Foundation have all since modeled themselves after the CF Foundation's approach. The lesson across these cases is the same: when the math of commercial profit doesn't work, affected communities must step in to drive progress themselves.

Below: Cure Rare Disease works with scientists and researchers at academic institutions, like Yale University (pictured here), to develop genetic treatments for neuromuscular and neurodegenerative diseases.

Gene Therapy, and Why the Vector Matters

Gene replacement therapy is a simple idea: if a disease is caused by a faulty or missing gene, deliver a working copy of that gene directly into the patient's cells. Unlike drugs that modify symptoms, a successful gene replacement addresses the root cause.

For many conditions, results have been transformative. Successes include FDA-approved gene therapies for inherited retinal dystrophy (Luxturna), spinal muscular atrophy (Zolgensma), and hemophilia B (Hemgenix). Zolgensma, approved in 2019 for a devastating childhood neurological disorder, is a dramatic demonstration: delivering a functional copy of the SMN1 gene leads to a remarkable change in outcomes. Children who would previously have required mechanical ventilation within two years of birth are now walking in some cases. Zolgensma's development was enabled by the SMA Foundation, a nonprofit which funded the scientific foundations for the treatment. Without the nonprofit's work, it is unlikely that Novartis would have invested in a treatment for a disease affecting only 1 in 10,000 births.

The key challenge for gene therapy is delivery. DNA cannot simply be injected into the bloodstream and expected to reach the right cells on its own. It requires a delivery vehicle, and for decades, the most effective option has been modified viruses.

Adeno-associated viruses (AAVs) are the newest stars of modern systemic gene therapy. Their relative safety, tissue versatility, and ability to facilitate long-term gene expression makes them the vehicle of choice. AAV vectors are naturally occurring viruses that have been engineered so that their own genetic material is removed and replaced with a therapeutic gene.

However, there have been significant challenges, the most important being liver toxicity. First-generation AAV capsids used in most approved therapies — the viral delivery shells that transport gene therapies into cells —

distribute broadly throughout the body after infusion. While they can effectively reach target tissues such as muscle, they also accumulate in the liver. Dose-dependent liver toxicity, observed in approximately 20% to 90% of patients across trials, can range from asymptomatic biomarker elevations to acute liver failure. This toxicity has been associated with serious adverse events in gene therapy trials, including the deaths in Sarepta's trials.

The last five years have seen the development of second-generation AAV capsids which are better targeted to the desired tissues (e.g. muscle) than the capsids previously used. In pursuit of safer and more efficacious delivery mechanisms, CRD's programs are using a next-generation AAV vector engineered to preferentially target muscle tissue and avoid the liver, which has been validated in preclinical studies.

CRD's Growing LGMD Pipeline

The LGMD R12/2L program is the newest addition to CRD's pipeline of LGMD therapies, all built on the same next generation vector. The most advanced is an investigational gene therapy for LGMD R9/2I. CRD has already completed a pre-IND meeting with the FDA and secured Orphan Drug Designation for this program, establishing a strong precedent for other candidates in the pipeline. A program targeting LGMD R7/2G is also underway.

The shared backbone of these programs is critical. Data generated from the R9/2I trials will support the R12/2L and R7/2G programs, and vice versa. Each program makes the others stronger, de-risking and accelerating the broader pipeline.

About the LGMD2L Foundation

The 2L Foundation was established in 2018 and expanded its mission in 2024 to begin actively funding research toward treatments. The Foundation has raised funds to support several research projects, including the development of a novel LGMD R12/2L mouse model (a mouse carrying the affected gene) in collaboration with Virginia Commonwealth University (VCU). That model is under active development and is expected to be used to evaluate the new gene therapy.

The partnership with Cure Rare Disease represents the LGMD2L Foundation's largest funding commitment to date. The Foundation's scientific advisors believe that gene replacement therapy represents the most promising near-term therapeutic strategy for LGMD R12/2L.

The Foundation and its work are funded entirely by donations. Drug development is expensive, and for rare diseases like ours, there are limited prospects for investors to recoup investment. It's the grassroots participation of community members like all of us that makes this revolutionary model possible. ▪

Want to Be Part of this Movement?

Both organizations welcome donations, volunteers, and advocates.

Written By Henry P. Eichner
Senior Drone Pilot, HHLT Marketing & Drone Services

Living and Working with Muscular Dystrophy

Why Flying Drones Became My Perfect Career

Above: Henry Eichner steped away from the traditional workforce and became his own boss — a life-changing decision that gave him flexibility, independence, and the ability to continue working and contributing in a meaningful way.

Before I became a full-time wheelchair user, I worked extensively in the information technology (IT) field. Much of that work required physical activity such as climbing ladders, crawling under desks, running cables, and handling equipment installations. As my muscular dystrophy progressed, those types of physical tasks became increasingly difficult and eventually were no longer possible for me to perform safely.

After that, I transitioned into work where I helped people with disabilities find employment opportunities. I assisted with writing resumes, teaching interview techniques, and helping individuals prepare themselves for entering or re-entering the workforce. While I truly enjoyed helping others in this way, I was still working for someone else and operating on someone else's schedule.

One of the biggest challenges of living with a disability is that everyday tasks simply take longer. Many people do not realize how long it can take just to get up, get dressed, get situated in a wheelchair, and prepare for the day. Traditional work schedules can be very difficult to maintain consistently when dealing with those realities.

Beyond just getting ready and commuting, there are also everyday challenges within a traditional workplace environment that many people may not think about — even simple things like restroom accessibility, navigating office spaces, and having the flexibility to manage physical needs throughout the day. In many cases, work environments simply are not designed to be truly conducive or realistic for someone living with a progressive physical disability like LGMD.

On top of that, there were many days where I would be in my wheelchair for extremely long periods of time — sometimes up to 12 hours a day between getting ready, commuting, working, and returning home. Over time, that became incredibly physically and mentally taxing. Eventually, I realized that continuing in a traditional workforce environment outside of the home simply was not sustainable for my long-term health and well-being.

That realization led me to step away from the traditional workforce and become my own boss, which turned out to be a life-changing decision for me. It gave me the flexibility to work in a way that accommodated my physical limitations and my good and bad days. It has given me flexibility, independence, and the ability to continue working and contributing in a meaningful way while still managing my disability.

Owning my own business (HHLT Marketing & Drone Services) has allowed me to transition away from relying on disability assistance and build something for myself, which is something that happens all too rarely in the disability community.

Flying drones, which had been just a hobby, became my career. Today, I co-lead the company, which provides high-quality aerial photography, video, orthomosaic mapping, and even thermal inspections. Our clients include construction companies, developers, and property owners across the Southeastern U.S.

Eventually, I realized that continuing in a traditional workforce environment outside of the home simply was not sustainable for my long-term health and well-being.

HHLT Marketing & Drone Services

Above: Henry and Tammie Eichner

Connect with HHLT Marketing & Drone Services

Sales@HHLTMarketing.com

(727) 484-0013

Facebook.com/HHLTMarketing

I think many people are genuinely surprised when they learn that I am a full-time wheelchair user, especially after seeing the type of work we produce and the places we travel to. One thing I always tell people is, "I may not be able to physically get everywhere on the job site, but my drone has no problem getting there." In many ways, that's what makes this career so exciting for me. The drone removes so many of the physical barriers that would normally limit someone with a disability, and because of that, I really don't feel limited when it comes to doing this kind of work.

For me, a lot of it comes down to outlook. There are real physical limitations that come with LGMD, but one's mindset can either expand or narrow what feels possible within those limits. Of course, none of this would be possible without an incredible support system behind me. My family has been incredibly supportive and has encouraged me every step of the way. Having people who believe in you and support your vision makes all the difference, and I'm incredibly grateful for that.

In closing, my advice to anyone reading this is to explore what you're passionate about. Three years ago, I barely even knew what a drone was. As I started learning more about the technology and what it could do, I realized it was the perfect fit for my skills and circumstances. That one discovery completely changed the direction of my life.

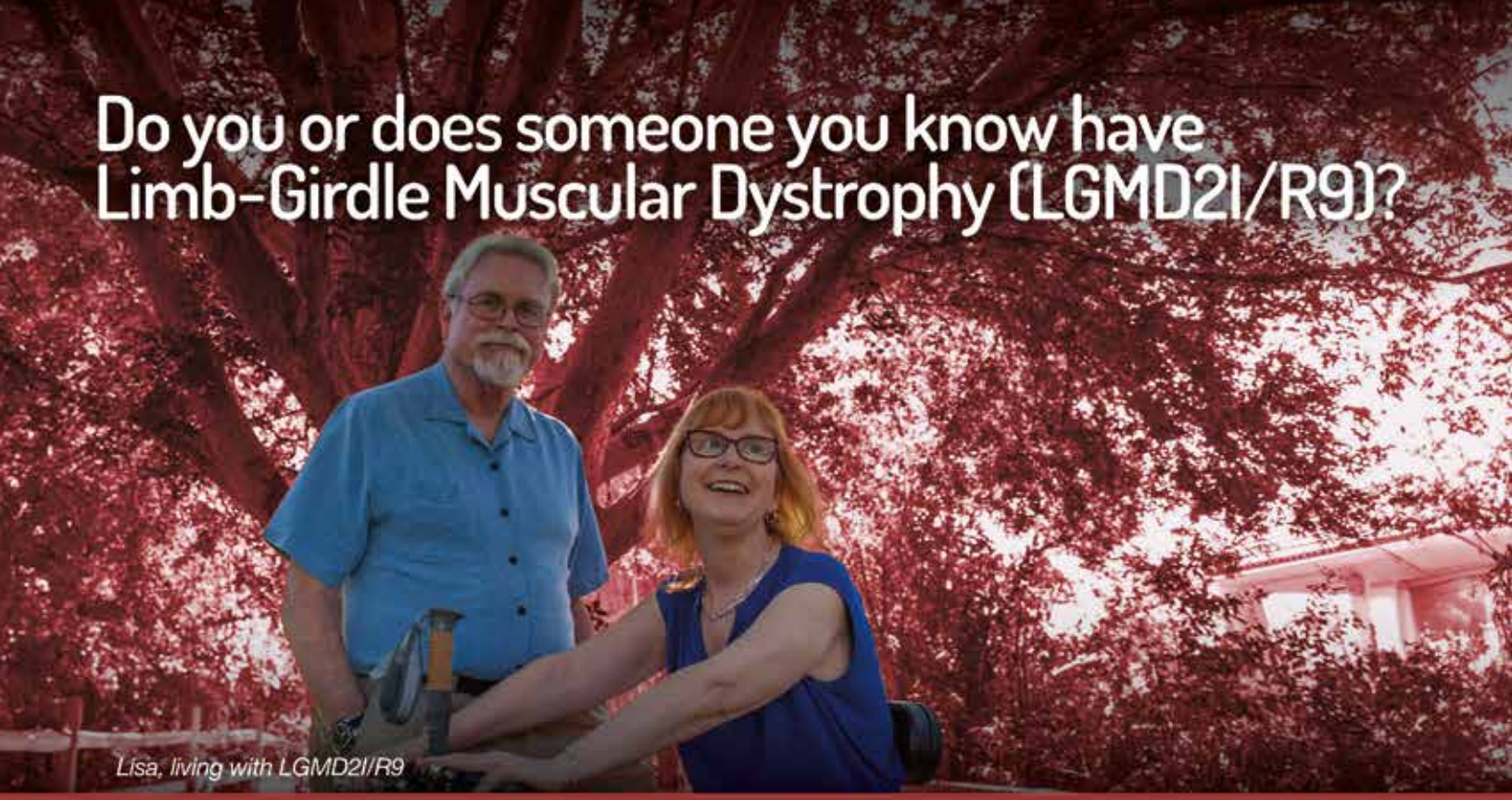
Equally important is being willing to seek out help and resources that are available to you. Organizations like vocational rehabilitation made a huge difference in my life. They helped me obtain a wheelchair-accessible van, assisted with modifications to my bathroom, and supported the installation of a ramp at my house. There are services and people out there who genuinely want to help you succeed, but many people are either not aware of them or choose not to reach out.

I believe there are good opportunities in work or creative paths for many people out there, though it often takes time, persistence, and a willingness to keep exploring different options to find them. Don't give up on yourself. Keep pushing forward, even when progress feels slow or uncertain. You may be surprised where life can take you when you stop focusing on what you can't do and start focusing on what you can.

My hope is that sharing my story will encourage others living with disabilities to realize there are still opportunities to create meaningful careers and successful businesses despite the challenges we face. ▪

"For me, a lot of it comes down to outlook. There are real physical limitations that come with LGMD, but one's mindset can either expand or narrow what feels possible within those limits."

Do you or does someone you know have Limb-Girdle Muscular Dystrophy (LGMD2I/R9)?



Lisa, living with LGMD2I/R9

AskBio is actively recruiting clinical study[1] participants with a confirmed genetic diagnosis of LGMD2I/R9 for an investigational[2] gene therapy treatment.

Find out if you qualify today

- This study is designed to evaluate the safety and tolerability of investigational gene therapy AB-1003 to treat LGMD2I/R9.
- Participants will receive a one-time intravenous infusion (injected directly into your vein) of the investigational gene therapy called AB-1003. AB-1003 is designed to produce fukutin-related protein (FKRP) in the body, primarily in muscle, heart, and diaphragm (the muscle that helps you inhale and exhale).
- There are two cohorts in the study. Cohort 1 is complete. We are currently recruiting for cohort 2.
- Some visits will take place at the study site, while others may occur at home. Study visit support including transportation, lodging and meals may be available.

We invite you to learn more

Email us
askfirst@askbio.com
Visit us online
Askbio.com
Clinicaltrials.gov
clinicaltrials.gov/study/NCT05230459
Scan QR code

We celebrate LGMD patients and advocacy groups for their tireless efforts and dedication to the community!

AskBio
A Bayer Cell & Gene Therapy Platform Company

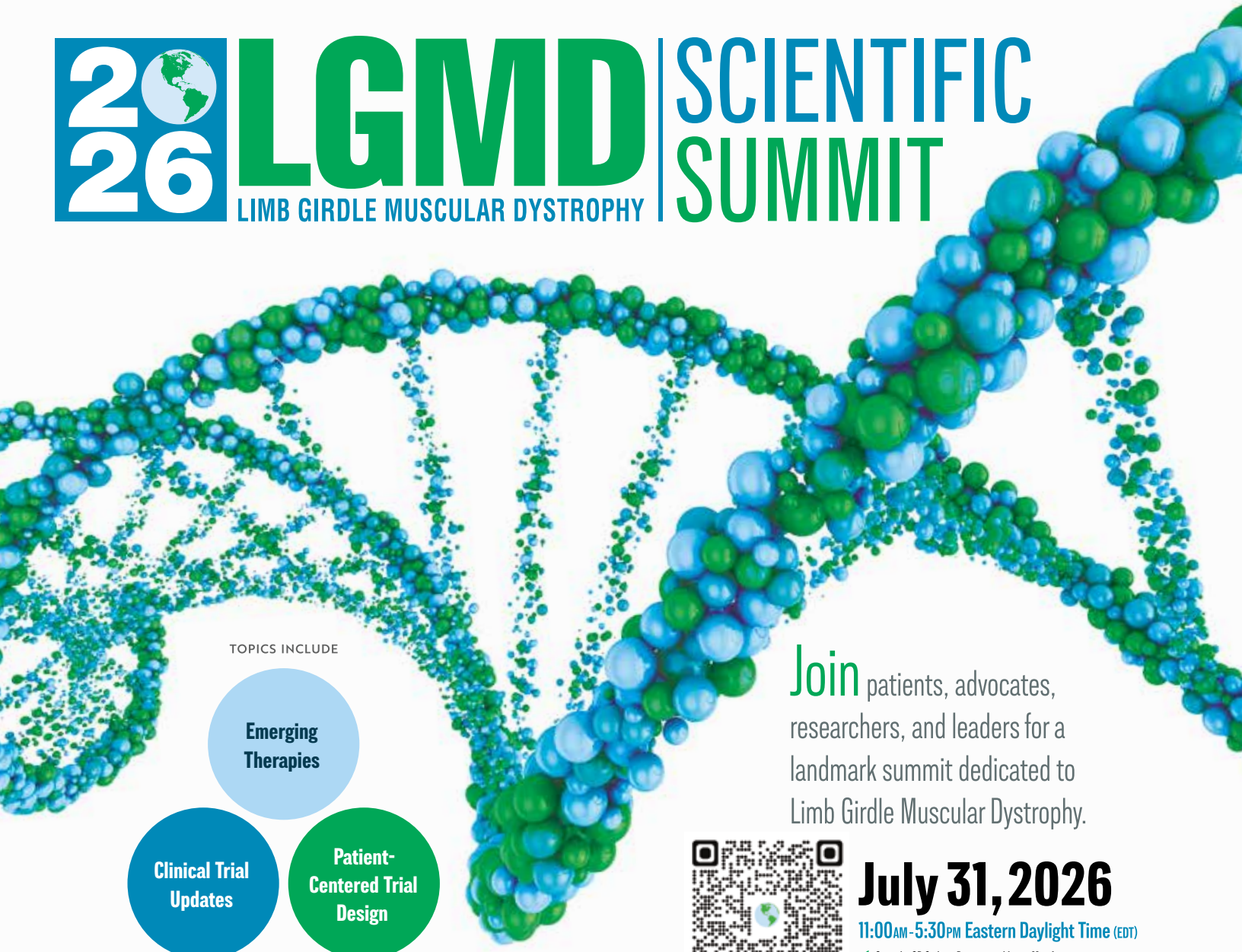
[1]Clinical Study: A clinical study is a research study for which people volunteer to help scientists find answers to certain health questions
[2]Investigational Treatment: An investigational treatment is a treatment being studied to see if your disease or medical condition improves while taking it. It has not yet been approved by regulatory authorities, such as the U.S. Food and Drug Administration (FDA).

The SPEAK Foundation
3551 Blairstone Road #105-62
Tallahassee FL 32301

ELEVATE THE PATIENT VOICE, ADVANCE RESEARCH, AND SHAPE THE FUTURE OF LGMD CARE

2026 LGMD SCIENTIFIC SUMMIT

LIMB GIRDLE MUSCULAR DYSTROPHY



TOPICS INCLUDE

- Emerging Therapies
- Clinical Trial Updates
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Join patients, advocates, researchers, and leaders for a landmark summit dedicated to Limb Girdle Muscular Dystrophy.

July 31, 2026

11:00AM-5:30PM Eastern Daylight Time (EDT)

Scan the QR Code to Register and Learn More!

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