Fall 2026 • Vol. 6 / Issue 4

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

GRASP-LGMD CONSORTIUM

Solving Genetic Dilemmas in LGMD and Seeking New Therapies

LGMD AND THE HEART

Part 1: Incidence, Monitoring, Diagnosis by Karim Wahbi, MD, PhD

OUR FAMILY, OUR STORY

Growing Up with a Parent who has Dysferlinopathy

Finding Purpose

Carol Abraham's Journey with LGMD R1/2A

LGMD
Awareness Day
September 30th

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

Advocate • Educate • Celebrate

Raise Awareness with the LGMD Awareness Day Toolkit

Access Toolkit:

- Puzzles
- 30-Day Challenges
- Informational Flyers
- Social Media Posts
- Media Kits + more!

Get a FREE T-SHIRT when you sign up!

GIRDIE 5K WALK-RUN-ROLL FOR LGMD AWARENESS

SEPTEMBER 2026

SPONSORED BY BRIDGEBIO

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

Learn more and register:

Thank you to our sponsors:

PLATINUM SPONSOR
BRIDGEBIO

GOLD SPONSOR
CURELGMD2i
Limb-Girdle Muscular Dystrophy

SILVER SPONSORS
AskBio
The SPEAK Foundation
LGMD2i Fund

LGMD AWARENESS FOUNDATION

lgmd-info.org

LGMD News

Editorial

Editor In Chief
Kathryn Bryant Knudson

Managing Editor
Jessica Evans, PsyD

Senior Editor
Bradley Williams, PhD

Editorial Assistants
Lissa Cone
Barbara Jean Doye
John Faver
Victoria Nedza

French Translation Editor
Mélanie Bordes

Spanish Translation Editor
Noni Uribe

Medical Editor
Peter B. Kang, MD, FAAN, FAAP
University of Minnesota Medical School

Contributors
Carol Abraham
Camille Bouchard, PhD
Peter B. Kang, MD, FAAN, FAAP
Amanda Price
Jonathan Price
Karim Wahbi, MD, PhD

Design + Production

Design Director
Jonathan Price

Advertising

ContactUs@TheSpeakFoundation.com

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.

Contents

Departments

Feature Articles

Connect with Us

Comments, Questions, or Suggestions?
ContactUs@TheSpeakFoundation.com

TheSpeakFoundation.com

Facebook.com/TheSpeakFoundation

Instagram.com/TheSpeakFoundation

Twitter.com/SpeakFoundation
X.com/LGMDFoundation

Straight from the Heart

Dear LGMD Community,

This issue of *LGMD News* Magazine focuses on two things deeply connected to life with limb-girdle muscular dystrophy: **our hearts and our families**.

For some types of LGMD, the heart may be affected. Education, appropriate monitoring, and access to physicians who understand LGMD can make a real difference. Our goal is not to create fear, but to empower patients and families with questions to ask and how to be proactive about cardiac care.

And when we talk about the heart, we are also talking about family.

LGMD may be diagnosed in one person, but rare disease affects an entire family. Spouses, parents, children, siblings, and caregivers adjust, advocate, celebrate victories, and often carry worries others may never see.

There is no one way to be an "LGMD family." What matters is knowing we do not have to navigate this journey alone.

We are also sharing something just as important: the tips and tricks we learn from one another. So much of living with LGMD is figuring out everyday solutions that make life easier — how we travel, conserve energy, adapt our homes, navigate appointments, manage family life, or find new ways to do things that have become more difficult. Many of us have found that some of the most useful advice comes from another person living with LGMD.

As science continues to move forward, we will keep fighting for better care, better treatments, and cures. Along the way, we can also make life a little easier by sharing what we have learned.

Our hearts matter. Our families matter. And the wisdom we share with one another matters. ▪

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

As science continues to move forward, we will keep fighting for better care, better treatments, and cures. Along the way, we can also make life a little easier by sharing what we have learned.

Connect with Us

ContactUs@TheSpeakFoundation.com

Our Mission

The Speak Foundation was based on the principle of "Speak up for those who have no voice." *Speak up for those who cannot speak for themselves.* — Proverbs 31:8

LGMD News is a program of the Speak Foundation. In order to help individuals and families living with limb-girdle muscular dystrophy receive critical and life-saving information, the Speak Foundation began publishing this resource in 2021.

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

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**

Peter B. 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

Meet the Expert

Peter B. Kang, MD, FAAN, FAAP, FCNS is a pediatric neuromuscular neurologist and physician-scientist whose laboratory studies the genetics and disease mechanisms of muscular dystrophy and DNA repair disorders, with a particular interest in LGMDs. He serves as Medical Editor for The Speak Foundation's *LGMD News* magazine and has published extensively on muscular dystrophy. He has also co-edited a textbook of pediatric electromyography and has been awarded multiple grants from the NIH and the Muscular Dystrophy Association to support his investigations. With support from the CDC, he led the successful initiation of the first site in Florida for the MD STARnet consortium on muscular dystrophy and continues to work on this project. His former trainees hold faculty positions in the United States, Canada, and Japan. He is Director of the Greg Marzolf Jr. Muscular Dystrophy Center, Professor and Vice Chair of Research in the Department of Neurology, and a Scholar at the Institute for Translational Neuroscience at the University of Minnesota. He is Editor of *Neurology® Genetics* and previously served as President of the Child Neurology Society, Secretary-Treasurer of the Massachusetts Medical Society, and Associate Editor of *Muscle & Nerve*.

Q

Some people with inherited muscle diseases still don't receive clear answers from standard genetic testing, particularly when very large genes like TTN are involved. Can you explain how newer long-read sequencing may help identify or clarify TTN variants that older testing methods might miss? Also, are there any early lessons from your Minnesota Neurogenetics Repository research that can help families understand when long-read sequencing might be beneficial—especially for individuals with LGMD R10/2J or other TTN-related muscle diseases?

A

Nanopore sequencing is a powerful genetic sequencing tool that enables us to read long strands of DNA. The output from nanopore sequencing is referred to as "long reads" that can stretch for many thousands, sometimes even millions, of base pairs at a time. Most genetic sequencing techniques rely on short reads consisting of several hundreds of base pairs at a time. For both long read and short read techniques, the stretches of DNA reads overlap with each other and can be pieced together using computer software to determine the overall sequence for the patient's DNA. Short read sequencing has become highly accurate in the detection of small variants (mutations) that affect one or a few base pairs. Advances in computer software have enabled us to detect an increasing range of larger variants in short read sequence data, but detection gaps remain for larger variants, especially in stretches of DNA that are highly repetitive. Nanopore long read sequencing by its nature can more easily detect larger variants, including those that are hiding in stretches of highly repetitive DNA. Nanopore sequencing has also become better at detecting smaller variants and is now often on par with short read sequencing in its ability to detect those smaller variants.

TTN is an ideal gene to be examined using nanopore sequencing. *TTN* is a very large gene that has highly repetitive regions and numerous exons (coding segments) that are expressed in different isoforms (combinations). Our laboratory has analyzed DNA samples from individuals enrolled in the Minnesota Neurogenetics Repository and we have begun to identify variants in *TTN* that were missed on clinical genetic testing. We believe that we have only scratched the surface of what nanopore sequencing can do for diagnosing *TTN* variants and would be interested in expanding our analysis of *TTN*-related muscle diseases.

Q

What role is artificial intelligence (AI) currently playing in muscular dystrophies such as LGMD—for example in gene discovery, variant interpretation, drug or gene-therapy design, or predicting disease progression and treatment response?

A

Artificial intelligence (AI) is suddenly all around us, stretching from ordinary online search engines to patient care to research. Patients and families are using online AI tools to ask increasingly complex questions and receive increasingly comprehensive answers. The widespread adoption of electronic medical records is making it feasible to mine medical record systems to automate research activities, increasingly using tools based on large language models (LLMs), a form of AI. Our team examined the ability of LLMs to extract key medical information relevant to muscular dystrophy and found that, at present, they could not detect all the key information that a trained human can (Zhou et al., 2025)[1]. We expect that as LLMs become more sophisticated, they will increasingly be able to match human medical record abstraction capabilities. In muscular dystrophy genetics, AI-based tools have been developed that can mine DNA sequence data and generate lists of candidate genetic variants based on multiple criteria. These new tools promise to discover and classify new genetic variants and new diseases that were previously overlooked. However, those AI-based tools often depend on variant calls that have already been made (i.e., the DNA sequence changes are pre-selected by another computer program) and human phenotype ontology (HPO) terms that are applied inconsistently (i.e., the genes are assigned to clinical disease categories but not always consistently). Thus, there are vulnerabilities in AI-based gene discovery and variant resolution tools, because their conclusions may be based on inaccurate information. We expect that these limitations will also diminish in coming years with rapid advances in AI technology.

Q

Gene therapy for Duchenne muscular dystrophy (DMD) has advanced significantly and is now being used in some patients. Has that progress helped inform or accelerate the development of gene therapies for LGMD R3/2D? If so, what advances or evidence are most encouraging for families affected by LGMD R3?

A

The knowledge we have gained from the commercial use of gene therapy in DMD is beneficial for the entire muscular dystrophy community. We now have a much better idea of what the side effect profile is like for patients with muscular dystrophy who receive adeno-associated virus (AAV)-based gene therapy. Though LGMD is different from DMD, there is enough overlap that our experience with DMD will inform how we approach LGMD gene therapy. The coding region of the *SGCA* (α-sarcoglycan) gene that is associated with LGMD R3/2D fits within an AAV vector. Such AAV-based gene therapy has been tested in mouse models of LGMD R3/2D for about two decades and, based on this experience, human clinical trials of gene therapy for LGMD R3/2D have been conducted. We will learn even more about the potential for gene therapy in LGMD when more information about the latest trials becomes available.

The knowledge we have gained from the commercial use of gene therapy in DMD is beneficial for the entire muscular dystrophy community. We now have a much better idea of what the side effect profile is like for patients with muscular dystrophy who receive adeno-associated virus (AAV)-based gene therapy. Though LGMD is different from DMD, there is enough overlap that our experience with DMD will inform how we approach LGMD gene therapy.

Question

There will come a day when the tables are turned, with therapies for LGMD leading the way to the development of new treatments for other diseases.

Q

Spinal muscular atrophy (SMA) and Duchenne muscular dystrophy (DMD) now have approved disease-modifying therapies, including SMN2 splicing modifiers and agents like givinostat. To what extent could similar approaches—such as RNA-targeting drugs, anti-inflammatory strategies, or other small molecules—be applicable to limb-girdle muscular dystrophy, or are these mechanisms largely disease-specific?

A

The approval of a new compound for muscular dystrophy by the US FDA advances the entire field of muscular dystrophy therapeutics. The potential applicability of an approved therapy for one type of muscular dystrophy to another type hinges on the disease mechanisms being targeted. For example, risdiplam specifically targets and alters the splicing pattern of the *SMN2* gene, making it unlikely that this compound will benefit patients with LGMD. But the lessons learned from compounds such as risdiplam could inspire the development of related compounds in other diseases where gene splicing changes could have a therapeutic effect. In contrast, givinostat is a histone deacetylase inhibitor, with downstream beneficial effects in skeletal muscle that are not necessarily specific to DMD. Thus, in theory, givinostat could be used in LGMD, but human clinical trials will be needed to determine whether givinostat is safe and efficacious for various subtypes of LGMD. There will come a day when the tables are turned, with therapies for LGMD leading the way to the development of new treatments for other diseases.

Q

Are gene therapies typically personalized to individual patients, or can a single therapy be used across all patients within the same subtype, such as LGMD R12/2L?

A

It is important for all of us to use terms consistently with each other to avoid misunderstandings. There is a general term "molecular therapy" that is usually understood to include a variety of categories including antisense oligonucleotides, cell-based therapy, AAV-based gene therapy, and gene editing. Each of these types of molecular therapy is associated with varying degrees of generalizability. Antisense oligonucleotides are short strands of nucleotides that bind to a certain type of RNA called pre-mRNA, often changing the splicing pattern and thus influencing the kinds of proteins that are generated by the gene. Antisense oligonucleotides often target specific types of variants in a gene, which may apply to subsets of patients with a disease such as DMD, or nearly all patients with another disease such as SMA. Cell-based therapies are often, though not always, derived from stem cells. One of the oldest forms of cell therapy is bone marrow transplantation, which is still in widespread use today. More traditional forms of cell therapy often apply to broad categories of patients, with the exception of newer cell therapies that are combined with another technology such as gene editing that yield personalized treatments.AAV-based gene therapies that deliver new copies of a gene to organs that are deficient for that gene often apply to any patient who has a deficiency in that gene, with some exceptions. Gene editing encompasses approaches that fix the DNA sequence so that a harmful sequence is replaced by the

healthy sequence. Gene editing traditionally targets specific genetic variants, but gene editing approaches can be rapidly adapted to different variants depending on the situation.

Q

In preclinical research, is it necessary to develop separate animal models for each muscular dystrophy subtype or can models be shared across related diseases?

A

Traditionally, it would be expected that we would need to generate an animal model for each genetic subtype of muscular dystrophy for which a therapy is to be applied. The main exception is that when a drug is already approved for one type of muscular dystrophy, it may sometimes be possible to start a clinical trial for a related disease without generating as much preclinical data as before. Given the large number of genetic subtypes of LGMD, the task of developing gene-specific animal models for all of them is a daunting prospect. Recently, the FDA began an initiative to promote the use of new approach methodologies (NAMs) in preclinical drug testing, including AI models and human organoids. It is not clear yet what situations NAMs can be applied to, but we anticipate that there will be increasing flexibility over time with a goal of expediting drug discovery and development. ■

Reference

1. Zhou, H., Rajamani, G., Huang, J., et al. (2025). Accelerating medical record data abstraction and analysis in muscular dystrophy: Large language models and International Classification of Diseases codes. *Neurology: Clinical Practice*, 15(6), e200542. https://doi.org/10.1212/CPJ.0000000000200542.

Have a Question for Our Experts?

Send Questions To:
ContactUs@TheSpeakFoundation.com

Solving Genetic Dilemmas in LGMD and Seeking New Therapies

Above: Dr. Peter B. Kang (Left) and his team investigate both genetic dilemmas and therapeutic strategies for LGMD.

As we enrolled more individuals with LGMD who had unsolved and ambiguous genetic diagnoses, we came to appreciate the scope of the problem.

Twenty years ago, I published my first articles on LGMD while training in my mentor's laboratory at Boston Children's Hospital. From those experiences, I learned that many patients with LGMD had trouble securing definitive genetic diagnoses. Several years later, as I started my own laboratory and sought a focus for my research, the topic of genetic dilemmas in LGMD caught my attention. A National Institutes of Health (NIH) grant fortuitously provided the support needed to launch this effort.

As we enrolled more individuals with LGMD who had unsolved and ambiguous genetic diagnoses, we came to appreciate the scope of the problem. These patients faced a knowledge gap that limited genetic counseling and prognostic information, and also was a barrier to participation in natural history studies and clinical trials. Our work gained further momentum when we joined the GRASP-LGMD Consortium in 2019 and later moved to the University of Minnesota in 2021. If anything, these challenges are likely to be magnified once new therapies for LGMDs receive approval, since patients with unclear genetic diagnoses may be unable to access those treatments.

Today, our laboratory investigates both genetic dilemmas and therapeutic strategies for LGMDs. We currently focus on the following laboratory projects:

- ***Genomic Studies of Undiagnosed Individuals with LGMD.***

  As we solve more cases of undiagnosed LGMD, the patients who remain undiagnosed often have genetic variants (mutations) that are increasingly difficult to find. For the past several years, we have been using nanopore long-read sequencing, an advanced DNA sequencing technology, to identify the missing genetic variants. Sometimes we find that those genetic variants are large changes in the DNA sequence, which hide in regions of DNA that were thought to be less important, and sometimes we find unusual combinations of variants.

- ***Modeling and Therapeutic Development for LGMD R19/2T.***
  In partnership with the Cure LGMD2T Foundation, our laboratory has embarked on a new project to develop human cellular models of LGMD R19/2T with known genetic variants in the *GMPPB* gene. We will culture cells that have been genetically modified to replicate patient variants, convert them into muscle, and characterize the effect of those mutations. These cells will be an excellent platform for testing all types of therapeutic candidates.

On the clinical research side, the Greg Marzolf Jr. Muscular Dystrophy Center in Minnesota continues to participate in the GRASP-LGMD Consortium and the Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STAR*net*). We are also working on a newly funded grant from the Muscular Dystrophy Association (MDA) focused on expanding the use of various clinical outcome assessments (COAs) in preparation for future clinical trials in LGMDs. We are proud to be designated a Speak Foundation Center of Excellence (CoE) for LGMD.

Patients and families interested in learning more about our research activities are welcome to contact us. For clinical research studies, please email ***mdcenter@umn.edu***. For laboratory research studies, including projects focused on LGMD genetics and LGMD R19/2T, email ***neurogenetics@umn.edu***. ■

Written by Peter B. Kang, MD, FAAN, FAAP, FCNS
University of Minnesota

Connect with Us

Clinical Research Studies:
MDCenter@umn.edu

Laboratory Research Studies:
Neurogenetics@umn.edu

GRASP-LGMD Consortium Members


Zineb Ammous, MD, FACMG
The Community Health Clinic


Ellie Carrell, PhD
Virginia Commonwealth University


Urvi Desai, MD
Atrium Health Carolinas Medical Center


Jordi Diaz-Manera, MD, PhD
Newcastle University


Stacy Dixon, MD, PhD
University of Colorado


Nicholas Johnson, MD, MSci, FAAN
Virginia Commonwealth University


Peter B. Kang, MD, FAAN, FAAP, FCNS
University of Minnesota


Kathryn Bryant Knudson
The Speak Foundation


Doris Leung, MD, PhD
Kennedy Krieger Institute


Katherine Mathews, MD
University of Iowa


Tahseen Mozaffar, MD, FAAN
University of California, Irvine


Richard Roxburgh, Mb ChB, FRACP, PhD
University of Auckland


Valeria Sansone, MD, PhD
Centro Clinico NeMO


Jeffrey M. Statland, MD
University of Kansas Medical Center Research Institute


Sub Subramony, MD
University of Florida


Conrad "Chris" Weihl, MD, PhD
Washington University in St. Louis


Matthew Wicklund, MD
UT Health San Antonio

Spotlight: Dr. Peter B. Kang | University of Minnesota

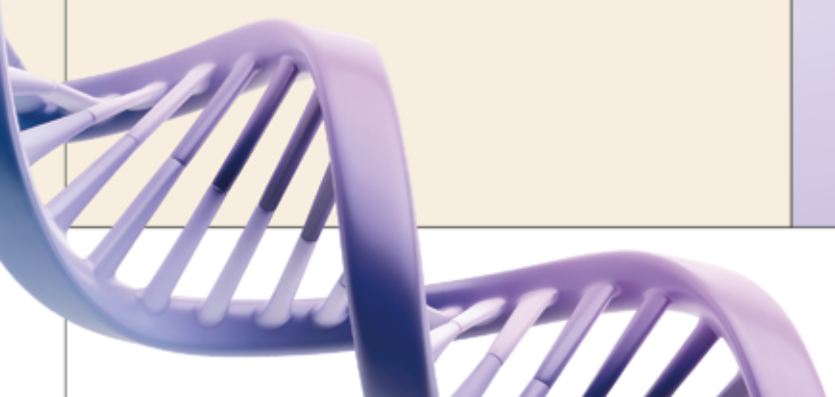
Dr. Peter B. Kang is a pediatric neuromuscular neurologist and physician-scientist whose laboratory studies the genetics and disease mechanisms of muscular dystrophy and DNA repair disorders, with a particular interest in LGMDs. He serves as Medical Editor for The Speak Foundation's *LGMD News* magazine and has published extensively on muscular dystrophy. He has also co-edited a textbook of pediatric electromyography and has been awarded multiple grants from the NIH and the Muscular Dystrophy Association to support his investigations. With support from the CDC, he led the successful initiation of the first site in Florida for the MD STARnet consortium on muscular dystrophy and continues to work on this project. His former trainees hold faculty positions in the United States, Canada, and Japan. He is Director of the Greg Marzolf Jr. Muscular Dystrophy Center, Professor and Vice Chair of Research in the Department of Neurology, and a Scholar at the Institute for Translational Neuroscience at the University of Minnesota. He is Editor of *Neurology® Genetics* and previously served as President of the Child Neurology Society, Secretary-Treasurer of the Massachusetts Medical Society, and Associate Editor of *Muscle & Nerve*.

Confirm. Code. Connect.

Three practical ways to stay informed and connected

Living with LGMD2I/R9 often means navigating a healthcare system that wasn't designed with rare diseases in mind. Along the way, there are decisions about diagnosis, care, support, and research—often with information that continues to evolve.

While no single step removes that complexity, there are practical actions that may help ensure your diagnosis is accurately documented, connect you with available resources, and keep you informed as care and research continue to advance.



Confirm

A confirmed diagnosis of LGMD2I/R9 requires genetic testing—a simple blood or saliva test that identifies changes in the FKRP gene.

Genetic confirmation can make a practical difference. It can support access to specialized care, research opportunities, and community resources. It may also be required for participation in future clinical studies or access to approved treatments, should they become available.

Several programs currently offer genetic testing at low- or no-cost. The LGMD Awareness Foundation maintains an up-to-date list of United States and international testing resources at:

lgmd-info.org/knowledge-base/list-of-international-lgmd-genetic-testing-resources

Some current low- or no-cost options for people in the United States include:

- **The Lantern Project (Sanofi/Revvity)** — A sponsored LGMD gene panel covering all LGMD subtypes. Testing is ordered through your healthcare provider.
- **Detect Muscular Dystrophy (Invitae)** — A sponsored neuromuscular panel of approximately 120 genes, with genetic counseling included, available in the United States and Canada. Testing may be ordered by you or your healthcare provider.

Your neurologist or genetic counselor can help you determine which testing option is most appropriate for your situation.

Code

LGMD2I/R9 now has its own ICD-10 diagnosis code:

G71.036 — Limb-girdle muscular dystrophy due to fukutin-related protein (FKRP) dysfunction

If you haven't already done so, consider asking your healthcare team to confirm that this code is accurately reflected in your medical record.

Accurate coding is more than an administrative detail. It helps ensure your diagnosis is clearly documented and communicated across your healthcare team, and it may influence insurance and reimbursement processes. As new treatment options emerge, accurate medical records may also help healthcare providers identify people who could be eligible for appropriate therapies or programs.

Many people in the LGMD2I/R9 community have found that updating medical records takes persistence. Ensuring your diagnosis is documented accurately can be an important part of receiving coordinated care.

"As a pharmacist and healthcare provider, the ICD-10 code is invaluable for both diagnosis and treatment. It helps identify disease state, guides medical decision-making, and alerts healthcare professionals to important considerations, such as anesthesia precautions or respiratory support needs. It also affects billing and reimbursement. The more accurately our healthcare teams understand our condition, the better equipped they are to provide appropriate care."

Lisa K. Vayda, RPh, MS, MA - Lisa is a pharmacist and health economist living with LGMD2I/R9 and an advocate within the community, including work with the CureLGMD2i Foundation.

Connect

Connection can take many forms—community, your healthcare team, and joining a registry among them.

Community. The Speak Foundation, CureLGMD2i Foundation, and LGMD Awareness Foundation connect people living with FKRP-related LGMD2I/R9 to others with firsthand experience, along with resources and updates as new information becomes available. The Speak Foundation publishes this magazine to share practical information developed with and for the LGMD2I/R9 community.

Your healthcare team. Staying connected with your neurologist or care team can also be valuable. Letting them know you're interested in treatment developments or clinical research can help guide future conversations as new information becomes available.

The Global FKRP Registry. The Global FKRP Registry is an international registry for people living with FKRP gene mutations, including LGMD2I/R9. People choose to participate for many reasons: registry data help researchers better understand how FKRP-related disease changes over time and support the development of future therapies. Participation may also help you stay informed about research studies or clinical opportunities for which you could be eligible.

Participation is voluntary, and your information is kept confidential and password protected.

To register, you'll need your confirmed genetic diagnosis, including the specific variants identified in both copies of your FKRP gene. Your healthcare provider can also contribute detailed medical information to your registry record.

Registration is available at **fkrp-registry.org**.

Confirming your diagnosis, ensuring your medical records are accurate, and staying connected to trusted resources may make it easier to access care, understand new developments, and make informed decisions as opportunities arise.

At a Glance

Confirm — Explore low- or no-cost genetic testing options at **lgmd-info.org/knowledge-base/list-of-international-lgmd-genetic-testing-resources**

Code — Ask your healthcare team to confirm that **ICD-10 code G71.036** is accurately documented in your medical record.

Connect — Reach out to community organizations like the Speak Foundation, CureLGMD2i Foundation, and LGMD Awareness Foundation, stay in touch with your healthcare team, and register with the Global FKRP Registry at **fkrp-registry.org**.

Resources

Community Organizations

The Speak Foundation:
TheSpeakFoundation.com

CureLGMD2i Foundation:
CureLGMD2i.org

LGMD Awareness Foundation:
lgmd-info.org

Genetic Testing and Registry

Low- or no-cost genetic testing options:
lgmd-info.org/knowledge-base/list-of-international-lgmd-genetic-testing-resources

Global FKRP Registry:
fkrp-registry.org

BRIDGEBIO


LGMD
permobil

Written By Carol Abraham
Founder and President, LGMD Awareness Foundation, Inc.

Finding Purpose:
My Journey with LGMD R1/2A

I have lived with LGMD R1/2A for over 60 years, but the diagnosis isn't my identity. Instead, it has shaped my purpose and inspired a mission that has connected me with hundreds of individuals, families, and caregivers around the world.

Before I became involved in LGMD advocacy, I worked as a registered occupational therapist (OTR). Helping people regain independence and adapt to life's challenges was more than a career; it was a calling. Occupational therapy taught me to focus on abilities rather than limitations and to recognize that quality of life is often found in the small victories that allow us to continue doing what matters most.

Growing up, I didn't know anyone else living with LGMD, and I had no friends or family who could truly relate to the daily challenges I faced. As my muscle weakness progressed, continuing to perform everyday tasks required creativity, adaptation, and perseverance. Problem-solving quickly became second nature. I experienced moments of uncertainty and frustration, yet I also discovered I possessed a lot of inner strength.

Over time, I realized that awareness of LGMD was far too limited. Many people, including healthcare professionals, had never heard of these diseases. It was also obvious that patients and families often felt isolated, just as I did growing up. I wanted to change that.

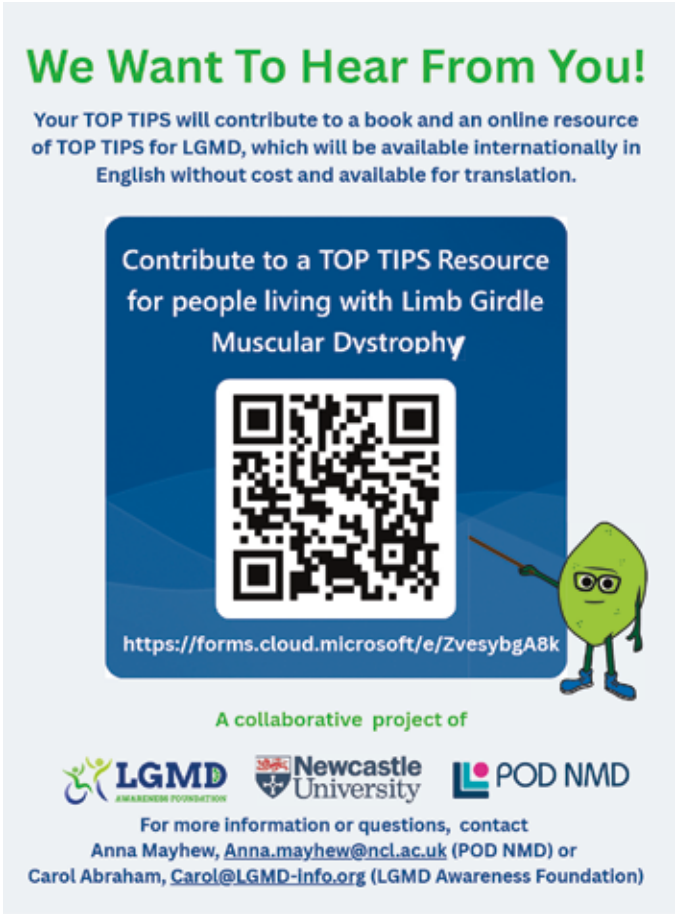
In 2014, I began LGMD Awareness Day, which is observed each year on September 30. What began as a simple effort to raise awareness of a rare disease has grown into an international movement. Today, patients, families, caregivers, clinicians, researchers, organizations, and industry partners around the world unite each September to increase understanding of LGMDs and to bring a sense of community to those affected.

"Over time, I realized that awareness of LGMD was far too limited. Many people, including healthcare professionals, had never heard of these diseases. It was also obvious that patients and families often felt isolated, just as I did growing up. I wanted to change that."

Opposite: Carol and Tim Abraham

As these awareness efforts expanded, I saw the need for an organization dedicated to education, advocacy, and support. That vision led me to found the 501(c)(3) non-profit LGMD Awareness Foundation in 2020, where I currently serve as President. The Foundation works to empower patients, provide curated educational resources, connect families, support research initiatives, and advocate on behalf of the global LGMD community. Through these efforts, we strive to ensure that individuals affected by these progressive, debilitating diseases have improved access to diagnosis, care, and treatment.

One of the greatest gifts this journey has given me has been the opportunity to meet extraordinary people living with LGMDs. Their resilience, courage, and determination continue to inspire me every day. While none of us would have chosen this diagnosis, together we have built a community filled with hope, friendship, and purpose.



My Top 5 Tips for Living with LGMD

1. **Protect your energy.** Think of your energy as a precious resource. Prioritize what matters most to you and don't hesitate to delegate or ask for help. Conserving energy today may help you do more tomorrow.

2. **Embrace adaptive equipment.** As an occupational therapist, I have learned that assistive devices are tools of independence, not signs of defeat. The right equipment can help you continue participating in activities you love while maintaining your mobility, safety, and independence.

3. **Stay connected.** Living with a rare disease can feel lonely, but you are not alone. Connecting with others who understand your journey can provide encouragement, practical advice, and lifelong friendships.

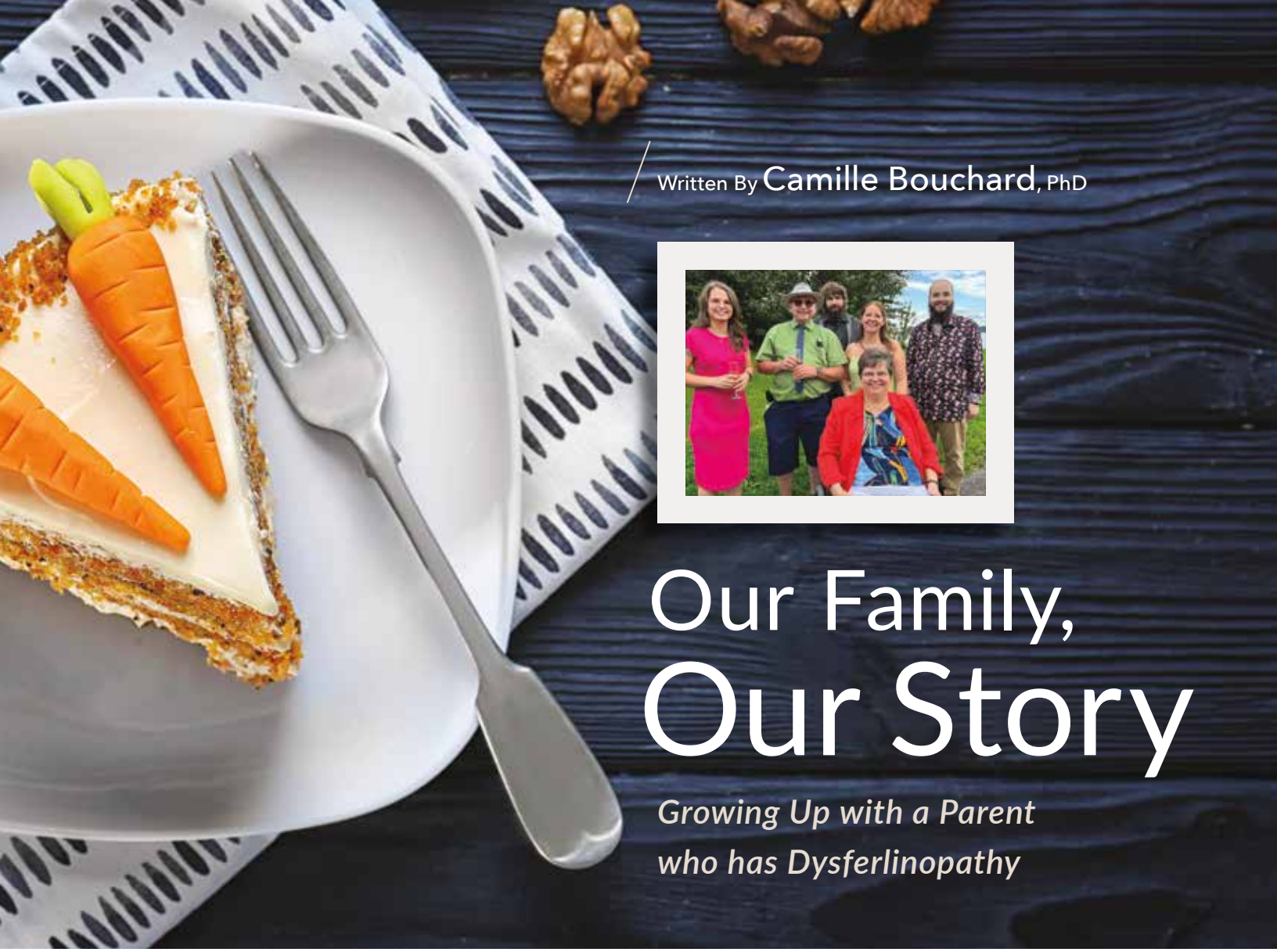
4. **Focus on what you can do.** LGMD may change how we do things, but it doesn't change who we are. Celebrate accomplishments, adapt when necessary, and continue pursuing what brings you joy and purpose.

5. **Become your own advocate.** Learn about your subtype, ask questions, and partner with your healthcare team. Your voice matters, and informed patients help shape the future of care and research.

Looking Forward with Hope

Although living with LGMD presents many daily challenges, I remain hopeful. Scientific advances are accelerating, awareness continues to grow, and our community has never been stronger. I believe the best days are ahead.

My journey with LGMD has taught me that strength is not measured by muscle, but by resilience, faith, determination, and the willingness to lift others along the way. Together, we are stronger! ▪



Written By Camille Bouchard, PhD

Our Family, Our Story

Growing Up with a Parent who has Dysferlinopathy

When my family thinks of our mother, we do not think of limb-girdle muscular dystrophy. Instead, we think of her love, the delicious carrot cake she bakes, the blankets and dish towels she knits, and a family home with laughter and the rhythms of everyday life. Although dysferlinopathy, a form of LGMD, is part of her life and, in turn, part of our family's life, that was never what defined her. She is simply our mom.

This is a story about family, about figuring things out together, and about how life can be beautiful even when it does not look like everyone else's. Every parent faces challenges. For our mom, one of them was LGMD, but it did not stop her from being exactly the parent we needed. A disability does not make you less of a good parent.

Growing up with our mom's condition shaped our lives. It made us curious about the science behind what we were watching, and it led one of us to a career focused on finding a cure. The rest of us work in jobs built around helping people: one in a city parks and recreation department, making sure every detail of a couple's wedding day is cared for; another at a police station, listening to people on their worst days and finding them the help they need. We are people who genuinely love to help others, a value our mother taught us, the same mother who still teaches us great lessons every day.

A Diagnosis that Arrived Quietly, then Revealed Itself

The first clues appeared during one of our mother's pregnancies. A doctor noticed that she was having difficulty walking and that her gait had changed, but no one knew what it meant at the time. Since she was carrying a boy, doctors checked her for Duchenne or Becker muscular dystrophy, both of which are X-linked disorders that primarily affect males, although females can also be affected as manifesting carriers. Testing ruled out both diagnoses. Beyond that, they had no answers for what was causing the changes in her movement, and for years the uncertainty lingered.

Eventually, our mother received a diagnosis of Miyoshi myopathy. Around the same time, her sister received a diagnosis of LGMD R2/2B. Today, we know these are different presentations of the same underlying condition: dysferlinopathy. For the first time, there was an explanation for what she had been experiencing. But for us, growing up, the diagnosis itself was never what mattered most. Our mom used a cane, and that was simply part of everyday life.

Growing up with LGMD: Just How We Lived

It was never strange to us that our mom needed a cane. It was normal to us, as familiar as the kitchen table, just another part of our home. As children, we knew to play different games with her than with our dad, who could run fast. We raced to open doors for her at stores and restaurants, and for anyone else, too, because we thought it was fun. It was never about making her feel different. It was just what we did.

LGMD made us think more carefully about things. We learned to adjust our games so everyone could join in. We did not need to talk about it. We simply knew where mom would sit, how to adapt an activity so she could take part, and which games worked for all of us. It was never a sacrifice. It was simply how our family did things.

At school events, it felt completely normal to make sure she had a good place to sit. Our teachers understood and found creative ways to include her in everything, without treating her as fragile or separate. Looking back, growing up with a mom who has LGMD made our childhood different from those of many others, but in ways we now value and appreciate. It taught us empathy, adaptability, and the importance of making sure everyone feels included.

Then and Now:
Above, Left: My mother just learned she had a muscular dystrophy yet, no clear diagnosis.
Above, Right: Us last Christmas celebrating love with our family.

What LGMD Taught Us about Parenting and Family

As young adults, we are still a close family. We love going home whenever we can to help with whatever our parents need. It is not a burden. It is simply natural because we love our family. We did not just share a house. We shared responsibilities, hopes, and the sense that our family's future belonged to all of us. We learned to come home not only to recharge but to contribute: helping with chores, rallying around a parent who loves to cook, and making sure the home remains a place that everyone can move through with ease and dignity.

LGMD did not derail our dreams. It gave them texture. It turned us into adults who notice when others need help and make sure their needs are met. The lessons from our mother's strength travel with us into every decision we make. They shape how we plan, how we imagine our future, and how we respond when life asks for a little extra thought or energy.

From Watching to Understanding: How Our Mom's Condition Shaped One of Our Paths

Growing up knowing that our mother's muscles worked differently made us curious to understand why, down to the cellular level, and whether there was a way to fix it. That curiosity grew from watching our mom navigate the world with her cane, seeing her strength despite the limitations, and from us wanting to do something about it.

For my part, I turned that curiosity into a career. I now work as a researcher, trying to find a treatment for dysferlinopathy, while also seeking to better understand LGMDs and improve their diagnosis. It is the most meaningful work I can imagine. I'm not just studying any disease; I'm studying my mom's disease.

Every breakthrough, every new discovery, and every improved diagnostic tool feels personal because it could directly change the lives of our mom and all the other people living with these conditions. I don't believe a treatment is good enough for anyone if it is not safe enough to give to my own mother.

"Disability does not define us. It influences how we live and how we take care of one another, and sometimes that can be a beautiful thing."

A Message to those Considering Parenthood with LGMD

Parenting is hard for everyone, but it can be hard in different ways. LGMD adds extra considerations, but it can also bring real strengths: resilience, empathy, teamwork, and a perspective on life that can make a family closer and more thoughtful.

Our mom's advice to anyone living with LGMD who wants to become a parent is simple: find a way to make it work for you. It is not about pretending limits do not exist. It is about knowing that limits are often where creativity and courage are born. You are not alone in this. You deserve to have the family you want with honest support about what care looks like and access to a community that understands.

We are proud of our mom for living life on her own terms, doing the things she loves in the ways that work for her. There is no single right way to parent with LGMD. For everyone, there is a way to parent that honors both your own well-being and your relationship with your child, all while being grounded in love.

For readers who do not have LGMD: disability is not the whole story of someone's life. It is one part of it, mixed in with humor, planning, patience, and the simple fact that we love the people we love. Disability does not define us. It influences how we live and how we take care of one another, and sometimes that can be a beautiful thing.

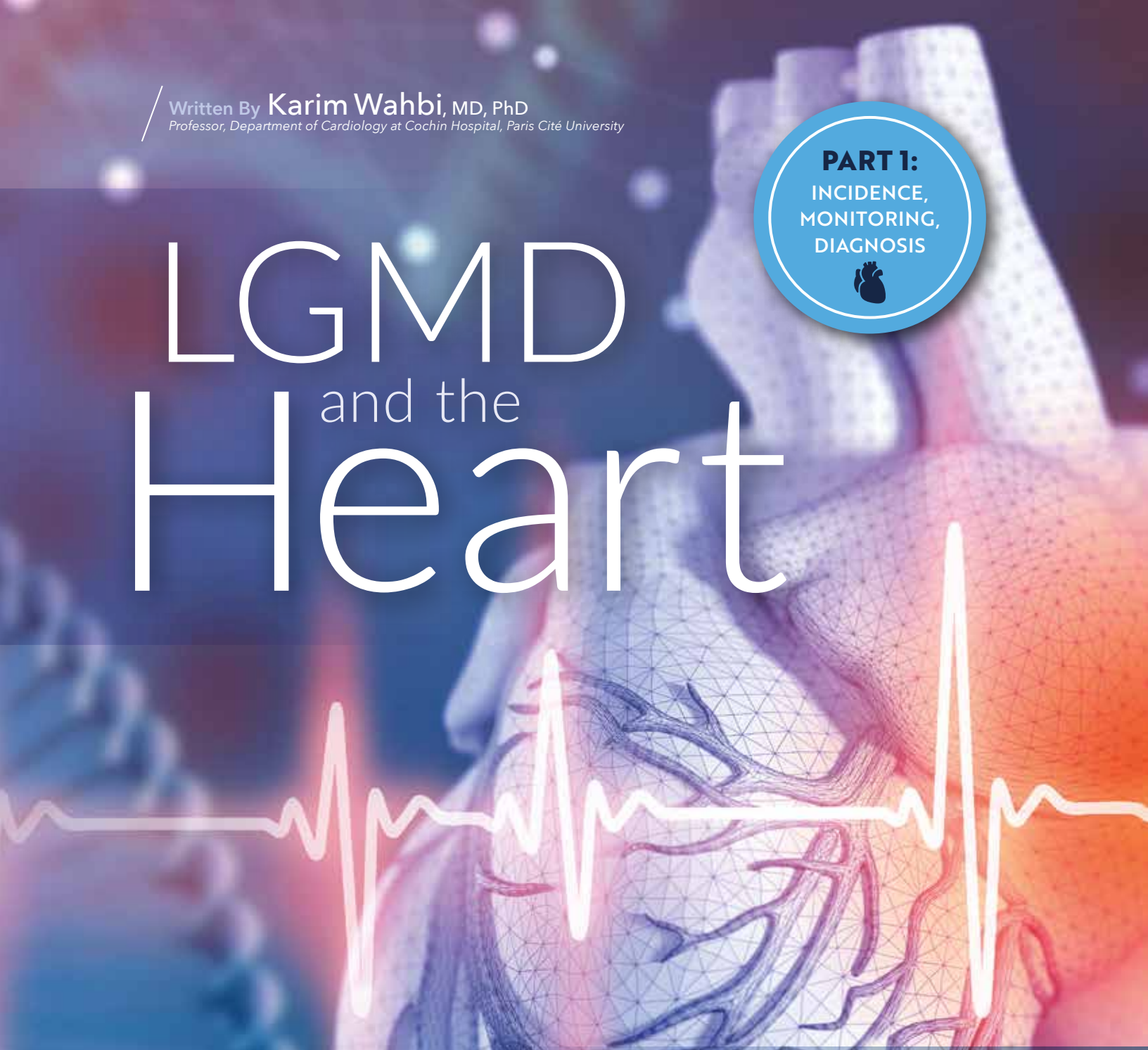
Looking Forward

If you are reading this while considering whether to have children as someone who lives with LGMD, my siblings and I hope you see a path forward. You deserve to build a family in the way that works for you, with the same love, care, and joy that any other family has. ■

Written By Karim Wahbi, MD, PhD
Professor, Department of Cardiology at Cochin Hospital, Paris Cité University

PART 1:
INCIDENCE, MONITORING, DIAGNOSIS

LGMD and the Heart



WHILE LGMD PRIMARILY AFFECTS SKELETAL MUSCLE, SOME FORMS CAN ALSO AFFECT THE HEART, SOMETIMES BEFORE NOTICEABLE SYMPTOMS DEVELOP. IN THIS ARTICLE, DR. KARIM WAHBI, A CARDIOLOGIST AND EXPERT IN CARDIAC INVOLVEMENT IN NEUROMUSCULAR DISEASES, DISCUSSES WHICH LGMD SUBTYPES MAY CARRY GREATER CARDIAC RISK, HOW HEART INVOLVEMENT CAN BE DETECTED, AND THE IMPORTANCE OF ONGOING CARDIAC MONITORING.

Cardiac Involvement in LGMD

LGMD News: **Which LGMD subtypes show the highest risk of cardiac involvement, and how does this differ mechanistically (e.g., sarcolemmal instability vs. fibrosis pathways)?**

Dr. Wahbi: Cardiac involvement (cardiomyopathy) in LGMDs is highly heterogeneous (meaning it varies greatly among different types of LGMD). Among the forms associated with the highest cardiac risk are dystroglycanopathies such as LGMD R9/2I and sarcoglycanopathies (LGMD R3-R6/2C-2F), with cardiac phenotypes similar to those observed in Duchenne and Becker muscular dystrophies. Cardiac involvement can also occur in LGMD R10/2J, depending on the location of the mutation within the TTN gene. Dominant forms of MD historically categorized as LGMD1B (Lamin A/C) and LGMD1E (Desmin), also have cardiac involvement, although the current classification does not include them among LGMDs.

All these diseases share the potential to progress, at some point, towards myocardial fibrosis, which is one of the determinants of heart failure. However, the exact mechanism differs according to the mutated gene. In FKRP-related dystroglycanopathies, deficient glycosylation of alpha-dystroglycan weakens the anchorage of the muscle fiber to the extracellular matrix, causing cellular injury. In sarcoglycanopathies, disruption of the sarcoglycan complex contributes to sarcolemmal instability.

LGMD News: **At what stage of disease does one typically see cardiac changes?**

Dr. Wahbi: Often, significant cardiac involvement can be present before symptoms are noticeable to the patient. This may include reduced contractile function, meaning the ability of the heart muscle to contract and pump blood effectively, as assessed either by ejection fraction or more sensitive parameters such as left ventricular strain. It may also include myocardial fibrosis (scarring of the heart muscle), detectable by cardiac MRI.

Symptoms of heart failure, such as breathlessness or edema (fluid retention and swelling), generally appear at a stage when myocardial contractile function is significantly reduced or when the burden of myocardial fibrosis is relatively substantial. Patients with severe symptoms requiring hospitalization usually have advanced myocardial involvement. Some non-ambulatory patients may remain asymptomatic due to a lack of physical activity, despite severe cardiac involvement. Cardiomyopathy may progress silently, only being discovered after a serious acute complication.

A different aspect of cardiac involvement involves electrical abnormalities, including conduction disease and atrial or ventricular arrhythmias. These are very prominent in laminopathies (formerly LGMD1B) and desminopathies (formerly LGMD1E). In most LGMDs, electrical complications occur at advanced stages of cardiomyopathy, causing symptoms such as palpitations, presyncope (near-fainting), or syncope (fainting). These symptoms are often preceded by abnormalities detectable on electrocardiography (ECG) or Holter monitoring.

Potentially treatable cardiac abnormalities may indeed be present before the onset of symptoms, highlighting the importance of cardiological follow-up from the time of diagnosis.

LGMD News: **How do you distinguish between primary cardiomyopathy versus secondary effects from skeletal muscle disease?**

Dr. Wahbi: Overall, cardiac involvement should not be considered a consequence of skeletal muscle disease. Rather, heart and skeletal muscle involvement are manifestations of the same genetic condition in different tissues. Many genes involved in LGMD affect proteins important in both skeletal and cardiac muscle, meaning both tissues can be affected by their mutations.

While respiratory and cardiac involvement may influence one another, heart involvement in LGMDs is primarily related to the effects of the genetic mutation on cardiac muscle itself.

Cardiac Biomarkers

LGMD News: **What biomarkers on blood tests are informative about cardiac health, and how should they be interpreted by clinicians?**

Dr. Wahbi: BNP or NT-proBNP and troponin are commonly used as indicators of heart damage in the general population. Creatine kinase (CK) is also elevated with heart damage but is generally used to assess skeletal muscle. Some of these biomarkers require care in interpretation in patients with LGMDs, which we'll discuss in more detail.

LGMD News: **In LGMD patients, how should clinicians interpret elevated CK levels in relation to cardiac risk versus skeletal muscle breakdown?**

Dr. Wahbi: CK is an enzyme found in muscle, and serum levels of CK can be elevated when muscle is damaged. Different versions of CK are present in skeletal muscle and cardiac muscle. However, standard lab tests often don't distinguish between the two (referred to as a "total CK" test). An increase in total CK can, in theory, be related to skeletal muscle or cardiac muscle injury. However, in muscular dystrophies, chronic elevation of total CK is almost always related to skeletal muscle involvement, partly because the body contains a much larger amount of skeletal muscle than cardiac muscle.

In LGMD, elevated CK should primarily be considered a marker of skeletal muscle involvement. By itself, it does not indicate cardiac involvement.

LGMD News: **How often do you see troponin elevations in LGMD patients without clear evidence of acute cardiac injury?**

Dr. Wahbi: As with CK, there are different types of troponin. Troponin I is specific to cardiac muscle, so elevated Troponin I generally reflects injury to the heart. By contrast, troponin T is also influenced by skeletal muscle involvement in some neuromuscular diseases. Therefore, elevated troponin T does not necessarily mean that there is a cardiac abnormality.

It is also important to consider how troponin levels change over time. Many questions remain unanswered regarding how these variations should be interpreted and what they mean in terms of cardiac disease progression. Most of the available data on troponin I come from DMD, although some LGMDs (FKRP-related dystroglycanopathies and sarcoglycanopathies) share similarities regarding fragility of the dystrophin-glycoprotein complex and of the sarcolemma.

LGMD News: **Are there specific patterns of troponin levels (persistent vs. transient elevation) that are informative about cardiac involvement?**

Dr. Wahbi: Different patterns of troponin elevation have been reported in patients. Some have normal troponin levels, while others may have temporary elevations that later return to normal. Finally, some patients have persistent, usually moderate, elevations of troponin I, sometimes with additional temporary increases. These patterns may reflect differences in how and when myocardial injury occurs, including periods of increased cardiac stress or injury ("flares"), but the underlying mechanisms are not fully understood.

Beyond the diagnostic step, which involves understanding what is happening in the myocardium at a given time, there is also a prognostic step. This aims to assess the significance of these troponin elevations in terms of the risk of progression of cardiac involvement subsequent to cardiac complications over time. This topic has received study in DMD, but it isn't yet known how it applies in LGMDs.

LGMD News: **How do you clinically interpret BNP or NT-proBNP levels in LGMD patients, especially in those with reduced mobility?**

Dr. Wahbi: BNP and NT-proBNP are the best biological markers for diagnosing heart failure in the general population. They may also be helpful in LGMD, although their utility is less established in this setting.

Two situations should probably be distinguished. The first is the use of NT-proBNP for the early diagnosis of myocardial involvement. Some data suggest that BNP or NT-proBNP levels may remain normal even when changes in the heart are detectable by cardiac MRI or heart ultrasound (echocardiography).

In more advanced stages of cardiac involvement, particularly when left ventricular ejection fraction is reduced, these markers may perform more similarly to how they work in patients without muscle disease.

However, there is an important caveat for non-ambulatory patients. Data from DMD suggests that reduced mobility may affect BNP or NT-proBNP levels, potentially resulting in values lower than expected. This could make these tests less informative in some situations, but we don't yet know how this applies to LGMDs.

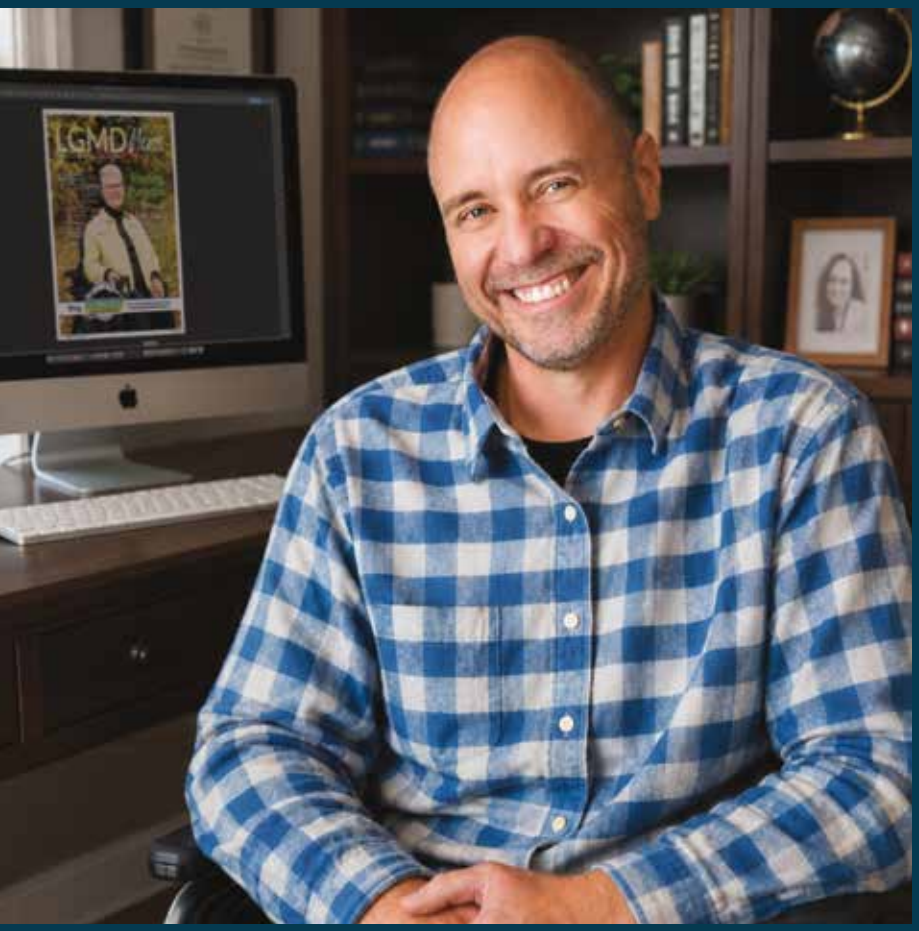
LGMD News: **Do you recommend using high-sensitivity troponin assays, or can they create more ambiguity in neuromuscular populations?**

Dr. Wahbi: High-sensitivity troponin assays are newer blood tests that measure the same marker as conventional troponin tests, but can detect much smaller amounts, allowing identification of subtle changes that might not be detected by a standard test. However, we do not yet know precisely what high-sensitivity troponin adds compared with conventional troponin, for predicting the progression of cardiac disease. Thus, it is unclear whether high-sensitivity troponin testing provides real added value in patients with LGMDs. ▪

Written By Jonathan Price
Creative Director, Jonathan Price Design, LLC

Living and Working with Muscular Dystrophy

One Passion, Two Careers



Above: Jonathan Price founded his design studio on the passion for consistently offering clients award-winning creative solutions, combined with a personal touch and attention to detail that only a hands-on, principal-driven studio can provide.

I loved everything about firefighting. The comradery, the excitement, the adrenaline rush whenever we responded to calls. Most of all I loved the satisfaction of helping folks in the community. But over time, I began noticing subtle physical issues that were difficult to explain. My family noticed the same issues as well. Eventually, my cousin, who happens to be a nurse, encouraged me to see a doctor.

Over the next two years, under the care of three different neurologists, I had an MRI, a CAT scan, an EMG (electromyography), and a stress test. It wasn't until I had a blood draw that I discovered my CK levels were extremely elevated. My doctors originally thought I might have polymyositis or Pompe disease. However, after a muscle biopsy, it was determined that I had LGMD R12/2L.

To be honest, the diagnosis came as a relief. I had an "a-ha" moment where I could look back at embarrassing physical failures — moments when I'd thought, *Oh man, I really need to work out harder* — and feel relieved because everything now made sense. I needed to know what I was dealing with before I could roll up my sleeves and take action.

Left: Jonathan participating in a training exercise while on duty at Columbia College Fire Department.

> **I needed to know what I was dealing with before I could roll up my sleeves and take action.**

Once I realized my firefighting career was no longer a realistic long-term option, I turned my attention to my other childhood passion: graphic design. I grew up doing everything from hand-illustrating my school reports to drawing team logos to catalog my collection of baseball cards. Now it was time to see if I could turn that love into a career.

I went back to school and studied Art History at Biola University. I loved my classes and thought things were going great, until I took an elective illustration class and *failed miserably* (those life-figure renderings I painted still make me laugh to this day). But realizing I was no Rembrandt led me to the university's Graphic Design program, where the right doors finally started to open.

Launching my career more than 30 years ago, I was blessed to have had the opportunity to work as a freelance designer for numerous marketing communication firms and advertising agencies throughout Southern California. I gained valuable experience working on high profile projects for clients such as AAA, Anaheim Ducks, City of Anaheim, Coldwell Banker, Disney, Fatburger, Hunt-Wesson, Los Angeles Angels, Rubys, Taco Bell, and Yamaha, just to name a few. Not long after that experience, I officially launched my own home-based boutique design studio, Jonathan Price Design. Today, my clients come from a wide range of backgrounds, including start-ups, non-profits, municipalities, schools, churches, authors, entertainers, and businesses of all shapes and sizes across the country. I'm especially proud of my collaboration over the years with Kathryn Bryant Knudson and the rest of the Speak Foundation team. I consider *LGMD News* magazine one of my favorite and most meaningful projects.

Above: Before being diagnosed with LGMD R12/2L, Jonathan (Back Row, Second from Left) served the community of Columbia, CA as a full-time firefighter EMT, engineer, and, ultimately, crew leader with the Columbia College Fire Department. On his off-days, Jonathan also served the community of Arnold, CA as a full-time volunteer firefighter with the Ebbetts Pass Fire Department, specializing in low-angle rope rescue.

Jonathan Price Design | Portfolio Snapshot

Logos | Magazines | Ads | Publications | Books

Although I'm no longer able to help people physically, I'm trying to use my eye for design to help raise awareness on behalf of those living with LGMDs.

All in all, I consider myself blessed to live with LGMD R12/2L. Why? Because having this specific adult-onset muscle disease allowed me the chance to have a pretty normal, energetic, and active childhood. I had the opportunity to really discover the things in life that I'm passionate about — including fulfilling my initial childhood dream of being a firefighter — without the confines of the many symptoms we typically encounter later in the progression of the disease. But more than that, it's given me a chance to share my story and hopefully be an inspiration to others. Although I'm no longer able to help people physically, I'm trying to use my eye for design to help raise awareness on behalf of those living with LGMDs.

Today, I approach each day choosing to be positive, optimistic, and focused entirely on what I *can* do rather than what I *can't*. My faith in Christ has given me the strength to carry on, especially on the days when I'm feeling especially fatigued.

I'm also fortunate to have a great support system I know I can count on. I can rely on my amazing wife, Amanda, to help me out each day with the seemingly endless daily tasks, including helping me get ready in the mornings, cooking meals, assisting me getting in and out of vehicles safely and always looking out for my well-being. I also have a caring family who have installed wheelchair ramps and have made their homes wheelchair accessible so I can visit safely and comfortably. For all of this, I am truly grateful.

When people ask me how long I intend to work, I tell them, "As long as I can continue to push myself creatively, have eyes that can see the screen, and a hand that can still move a mouse… I'm still in business!"

For my friends in the LGMD community, I'd like to share that there is in fact a place for you. Find your calling — the work, hobbies, and/or causes you love and are passionate about. Then put yourself out there. Figure out a way to move forward and make things happen. Focus on the positive, not the negative. Push yourself and build up an accountability support group around you. It's also important to give yourself permission once in a while to have an off day. We all need time to recharge. After all, LGMD is what it is. But always keep sight of your purpose, and watch those doors start to open! ▪

Jonathan started his graphic design studio in 1996. He designs the LGMD News magazine, which has received a Gold Davey Award and a Communicator Award of Distinction for design.

Connect with Jonathan Price Design

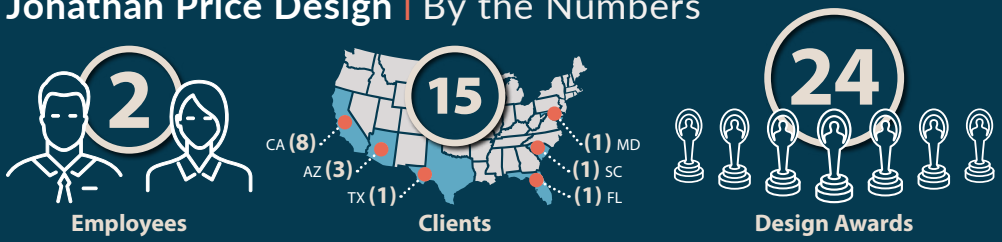
Info@JonathanPriceDesign.com

JonathanPriceDesign.com

Facebook.com/JonathanPriceDesign

But he said to me, "My grace is sufficient for you, for my power is made perfect in weakness." Therefore I will boast all the more gladly about my weaknesses, so that Christ's power may rest on me. –2 Corinthians 12:9

Jonathan Price Design | By the Numbers



Committed to advancing the science of gene therapy

AskBio™
A Bayer Cell & Gene Therapy Platform Company

FOCUS ON PATIENTS | PIONEERING SCIENCE | END-TO-END PLATFORM APPROACH | DETERMINED AND COMPASSIONATE EMPLOYEES | POWERFUL PARTNERSHIPS

Research and clinical development programs focused on areas of unmet medical need

MORE COMMON DISEASES

Heart failure
>64M globally[1]

DISCOVERY → PRE-CLINICAL → PHASE 2

Parkinson's disease
~6M globally[2]

DISCOVERY → PRE-CLINICAL → PHASE 2

RARE DISEASES

Multiple system atrophy
~100,000-500,000 globally[3]

DISCOVERY → PRE-CLINICAL → PHASE 1

Pompe disease
~1 in every 40,000 births globally[4]

DISCOVERY → PRE-CLINICAL → PHASE 1/2

Limb-girdle muscular dystrophy 2I/R9
<5,000 in the U.S[5]

DISCOVERY → PRE-CLINICAL → PHASE 1/2

All programs shown are investigational. Safety and efficacy have not been established. Development stages, timelines, and outcomes are subject to change.

[1]Severese G, et al. Cardiovasc Res 2023;118:3272–3287
[2]GBD 2016 Parkinson's Disease Collaborators, et al. Lancet Neurol. 2018;17(11):939-953; 3. Post B, et al. J Parkinsons Dis. 2020;10(s1):S29-S36
[3]MSA (Poewe et al. Multiple system atrophy. Nat Rev Dis Primers. 2022;8:56; Wenning et al. Lancet Neurol. 2004;3:93-103)
[4]Extracted from: Stevens, D., Milani-Nejad, S. & Mozaffar, T. Pompe Disease: a Clinical, Diagnostic, and Therapeutic Overview. Curr Treat Options Neurol 24, 573–588 (2022). https://doi.org/10.1007/s11940-022-00736-1
[5]NIH.https://rarediseases.info.nih.gov/diseases/12533/autosomal-recessive-limb-girdle-muscular-dystrophy-type-2i Accessed 06/09/2025

Working to help shape the future of gene therapy research

END-TO-END PLATFORM

- Global clinical development capabilities
- Integrated manufacturing platform development

BROAD THERAPEUTIC RESEARCH FOCUS

- Cardiovascular
- Central Nervous System
- Neuromuscular

SUPPORTING THE GOAL OF GLOBAL ACCESS

- Manufacturing capabilities designed to support late-stage and potential commercial supply
- Global reach, in partnership with Bayer
- Continued innovation to address the industry's biggest challenges

AskBio™
A Bayer Cell & Gene Therapy Platform Company

AskBio is a wholly owned and independently operated subsidiary of Bayer AG

Have a question or want to learn more about AskBio's research and clinical development programs?

Contact AskFirst@askbio.com or scan the QR code.

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

Announcing: The 2027 International LGMD Conference is returning to Orlando, Florida

SAVE THE DATE!

July ▪ 23-25 ▪ 2027

REGISTRATION OPENS: SEPTEMBER 18, 2026

2027 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE

JULY 23–25, 2027 • HYATT REGENCY ORLANDO
9801 INTERNATIONAL DRIVE • ORLANDO, FL 32819

Presented By The SPEAK Foundation

www.InternationalLGMDConference.com

The 2027 International LGMD Conference is a program of the Speak Foundation.