Spring 2026 • Vol. 6 / Issue 2

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

LGMD CENTERS OF EXCELLENCE

A Patient-Led, Academic-Aligned Model for the Future of Rare Disease Care

ASK THE EXPERT

Valeria A. Sansone, MD, PhD

A GLOBAL CONVERSATION FOR THE LGMD COMMUNITY

International LGMD Community Leaders Assess the Global Landscape of Care, Research Infrastructure, and Patient Access

LGMD and Medical Test Misinterpretation

Avoid a Misdiagnosis or a Missed Diagnosis by Being Aware of the Limitations of Test Outcomes

TREAT-NMD®
Neuromuscular Network

Limb Girdle Muscular Dystrophy Family Guide

LGMD Guide for Patients, Families and HCPs

An easy to follow guide, designed to provide you with clear, accessible information on disease progression, care and treatments.

Scan the QR code to view our guide

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LGMD News

Spring 2026 • Vol. 6 / Issue 2

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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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Vol. 6/Issue 1
Correction

In the last issue, The GFB Foundation article incorrectly stated that "29 clinical histories have been studied, spanning from the first symptoms of disease to loss of ambulation." — but should have stated that "229 diagnoses have been studied, linked to the first symptoms of disease and to the loss of ambulation."

LGMD Centers of Excellence

Building the Infrastructure Patients Have Been Waiting For

Dear LGMD Community,

As the therapeutic pipeline for LGMDs continues to expand, one reality is clear: without the right clinical infrastructure, even the most promising therapies will struggle to reach patients.

With the official launch of the LGMD Centers of Excellence on May 4, 2026, our community reaches a pivotal milestone. For more information on this initiative, visit **TheSpeakFoundation.com**.

Individuals living with LGMDs often face fragmented care, long wait times, and limited access to neuromuscular specialists. The Centers of Excellence model addresses these challenges by delivering a more coordinated care experience, including a dedicated LGMD Care Coordinator to streamline scheduling. Participating clinics commit to providing appointments within an 8–10-week timeframe, creating a more timely and predictable system of care. *Please note that clinics will continue to charge for healthcare appointments as usual. Patients are responsible for any associated copays and fees.*

This model also unites leading neuromuscular experts and standardizes care across institutions such as the University of Florida (Dr. Barry Byrne), Virginia Commonwealth University (Dr. Nicholas Johnson), University of Minnesota (Dr. Peter Kang), the University of Iowa (Dr. Katherine Mathews), and UT Health San Antonio (Dr. Matthew Wicklund).

These centers are not only improving care — they are creating readiness. Readiness for emerging therapies, for innovative trial designs, and for ensuring that patients are not left behind as science advances.

Importantly, this is a patient-led initiative. The Speak Foundation has worked to build the connective infrastructure across the LGMD landscape — aligning clinicians, researchers, industry, and policymakers around a shared goal: accelerating access to treatments while ensuring the patient voice is central to every stage of drug development for LGMDs. We are also pleased to announce our care partners on this initiative: LGMD Awareness Foundation and CureLGMD2i.

As multiple therapeutic approaches — from small molecules to gene therapies — move closer to reality, success will depend on our ability to match innovation with execution. The LGMD Centers of Excellence are a critical step in making that possible. ▪

These centers are not only improving care — they are creating readiness. Readiness for emerging therapies, for innovative trial designs, and for ensuring that patients are not left behind as science advances.

Connect with Us

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

For additional information about the LGMD Centers of Excellence and to connect with a coordinator, visit **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**

Valeria A. Sansone, MD, PhD

Centro Clinico NeMO Milano

Meet the Expert

Valeria A. Sansone, MD, PhD is a Professor of Neurology at the University of Milan and a senior neurologist at Centro Clinico NeMO Milano, a leading Italian center for neuromuscular diseases. She is internationally recognized for her clinical and research expertise in muscular dystrophies, with a particular focus on LGMD, myotonic dystrophy, and other inherited neuromuscular disorders. Prof. Sansone has played a key role in advancing multidisciplinary care models for neuromuscular patients and is actively involved in clinical trials, natural history studies, and translational research. She serves as a member of the European Reference Network for Rare Neuromuscular Diseases (ERN Euro-NMD) and has authored over 200 peer-reviewed publications. Her work bridges academic research and patient-centered clinical care, contributing significantly to improved diagnosis, management, and outcomes for individuals living with rare neuromuscular conditions.

Q

I have been diagnosed with a probable pathogenic homozygous variant in the TRIM32 gene, which is associated with limb-girdle muscular dystrophy type 2H (LGMD R8/2H). I would like to know if there is any research on this subtype of LGMD, including any potential treatments being investigated.

A

To my knowledge, there are no clinical trials specifically targeting TRIM32 related LGMD at present. This may be due in part to the large size of the TRIM32 cDNA, its complex regulatory domains (meaning the gene's activity and function are tightly and intricately controlled), and the clinical heterogeneity observed among affected individuals, ranging from mild forms to more severe forms.

However, cellular and animal models are being developed to study disease mechanisms, and on the clinical side, observational studies are collecting data from patients over time. These efforts are necessary to better understand the disease, stratify patients based on severity and clinical features, and ultimately pave the way for future clinical trials.

Q

How much daily protein intake is generally recommended for someone with LGMD?

A

There is currently no official, disease-specific dietary recommendations for individuals with LGMD. However, I normally recommend that individuals with LGMD follow the same age-appropriate nutritional guidance that applies to the general population, except for the fact that it is as though the "spare" energy, the "stores" are reduced to begin with in people living with neuromuscular conditions.

The goal is to maintain an adequate caloric intake while avoiding obesity, support muscle maintenance and repair through sufficient protein intake, and reduce the risk of cardiovascular and metabolic problems (connected to a dysmetabolic syndrome due to excessive carbohydrate and lipid intake). Physical activity or adaptive sports and exercise should be considered and tailored to the individual's residual functional capacity (the amount of muscle strength and mobility a person still has). Taken together, these factors are helpful to implement a balanced nutritional approach.

Question

Q

I have heard that GLP-1 medications may not be recommended for people with LGMDs due to concerns about muscle loss. If this is accurate, are there safer alternatives for individuals with LGMDs who, like myself, may have diabetes and also need support with weight loss but have limitations in their ability to exercise?

A

There is currently insufficient data on the use of GLP-1 medications in individuals living with neuromuscular conditions, including LGMDs. Obesity is a significant global health issue, and reductions in lean muscle mass with increases in fat mass is of course greater in people living with neuromotor disabilities. Therefore, a thorough analysis is needed to weigh the health risks and limitations associated with obesity against the potential effects of GLP-1 medications on muscle loss.

I would recommend a multidisciplinary approach to evaluate treatment options on a case-by-case basis. Broad generalizations are not possible in this case. I would not exclude GLP-1 medications *a priori*, but instead suggest working with your team of medical professionals to consider all the potential risks and benefits over a limited trial period. In some cases, initial weight loss may facilitate increased activity and excerise, which could then support longer-term nutritional and lifestyle interventions without medications.

Q

Is it possible for LGMD R9/2I to affect gastrointestinal function? Over the past few years, I have noticed significant changes related to my stomach and eating, including a feeling of fullness after only a few bites. I have also discussed this with other individuals affected by LGMD R9/2I in a patient support group, and several reported experiencing similar symptoms.

A

Our bodies contain three major types of muscle. Skeletal muscle, which allows us to move, is the type primarily affected in LGMDs. Some LGMD subtypes can also affect cardiac muscle, the muscle of the heart. The third type, smooth muscle, is found in internal organs such as the digestive system. Smooth muscle is not typically involved in muscular dystrophies, and I am not aware of gastrointestinal (GI) symptoms being considered a defining feature of LGMD R9/2I. However, GI symptoms are common across many neuromuscular conditions. These may include constipation, delayed gastric emptying, dysphagia (difficulty swallowing), and bloating. Such symptoms can have a number of causes and may be related to reduced activity, nutritional factors, and body weight rather than direct muscle involvement. ■

Obesity is a significant global health issue, and reductions in lean muscle mass with increases in fat mass is of course greater in people living with neuromotor disabilities. Therefore, a thorough analysis is needed to weigh the health risks and limitations associated with obesity against the potential effects of GLP-1 medications on muscle loss.

Have a Question for Our Experts?

Send Questions To:
ContactUs@TheSpeakFoundation.com

Introducing LGMD Centers of Excellence

A Patient-Led, Academic-Aligned Model for the Future of Rare Disease Care

While scientific progress is advancing, the systems designed to deliver care and conduct trials have not kept pace. The LGMD Centers of Excellence aim to change that.

The Speak Foundation is proud to announce the launch of the LGMD Centers of Excellence, a national, patient-led initiative designed to transform care, accelerate research readiness, and modernize the clinical trial landscape for individuals living with LGMD.

For decades, LGMD — an umbrella term encompassing more than 30 genetic subtypes — has faced fragmented care pathways, limited coordination, and no FDA-approved therapies. While scientific progress is advancing, the systems designed to deliver care and conduct trials have not kept pace. The LGMD Centers of Excellence aim to change that.

Unlike traditional models that are institution-driven alone, the LGMD Centers of Excellence are stewarded by The Speak Foundation and are based on formal partnerships with leading academic neuromuscular programs. This structure ensures that patient priorities and long-term sustainability are incorporated from the start, while real-world barriers are proactively addressed. We are also pleased to announce our care partners on this initiative: LGMD Awareness Foundation and CureLGMD2i, who will be providing funding along with the Speak Foundation and sponsors to launch the program.

The inaugural Centers include nationally recognized leaders in neuromuscular medicine such as: Dr. Barry Byrne, Dr. Nicholas Johnson, Dr. Peter Kang, Dr. Katherine Mathews, and Dr. Matthew Wicklund. These physicians have decades of expertise in neuromuscular disease, clinical trials, and natural history research. Their participation signals a strong alignment between academic rigor and patient-centered infrastructure.

The LGMD Care Coordinator: A Transformational Patient-Centered Feature

A defining and essential component of every LGMD Center of Excellence is the dedicated LGMD Care Coordinator. This role is foundational to how the Centers function for patients.

Individuals living with LGMD often require appointments with multiple specialists including neurology, cardiology, pulmonology, physical therapy, and genetic counseling. Historically, patients and families have been left to coordinate these appointments themselves — making repeated phone calls, navigating separate scheduling systems, and spreading visits across multiple days or even months.

Under the Centers of Excellence model, patients will no longer be expected to manage this complexity alone.

Each Center will provide a single, direct point of contact: the LGMD Care Coordinator. This individual will take responsibility for organizing and aligning all specialty appointments, working internally across departments so patients do not have to. *Please note that clinics will continue to charge for healthcare appointments as usual. Patients are responsible for any associated copays and fees.* ▸▸▸

> *Each Center will provide a single, direct point of contact: the LGMD Care Coordinator.*

LGMD Centers of Excellence Endorsements

The opportunity to create meaningful therapies for the LGMD community are finally within reach thanks to many years of foundational basic and clinical research. At the University of Florida Powell Center and UF Health Center for Advanced Therapeutics, we are excited to bring these new discoveries to all individuals who are living with LGMD.

– Dr. Barry Byrne • University of Florida Powell Gene Therapy Center

The launch of the LGMD Center of Excellence at VCU represents a meaningful step forward for patients and families living with limb-girdle muscular dystrophy. Our goal is to deliver coordinated, multidisciplinary care while strengthening our readiness for clinical trials and access to emerging therapies. By partnering with The Speak Foundation, we are aligning clinical excellence with patient-centered infrastructure in a way that truly meets the needs of this community.

–Dr. Nick Johnson • Virginia Commonwealth University

The LGMD Centers of Excellence create a collaborative framework that allows academic centers to work more efficiently to keep patients at the center of research and clinical care. This model positions us to advance therapies responsibly as they emerge.

– Dr. Peter Kang • University of Minnesota Medical School and M Health Fairview

As clinicians, we see firsthand the burden that fragmented care places on families. The LGMD Centers of Excellence model prioritizes coordination, genetic diagnosis where possible, and long-term follow-up – ensuring that patients receive comprehensive care that evolves alongside scientific discovery.

– Dr. Katherine Mathews • University of Iowa Hospitals & Clinics

This initiative brings together clinical expertise and structured infrastructure in a way that strengthens both patient care and research readiness. By standardizing multidisciplinary coordination, we can facilitate data synchrony for research trials and improve the overall patient experience.

– Dr. Matthew Wicklund • UT Health San Antonio

The goal is clear and measurable: whenever clinically feasible, all specialist appointments will be coordinated within one day — and at most two consecutive days.

The goal is clear and measurable: whenever clinically feasible, all specialist appointments will be coordinated within one day — and at most two consecutive days.

For patients who must travel long distances, require accessible transportation, manage fatigue, or depend on caregivers, this coordination is life changing. It reduces financial strain, physical exhaustion, missed work, and emotional stress. It transforms a fragmented experience into a structured, supportive one.

This is more than scheduling efficiency. It is an intentional redesign of care delivery around the realities of living with LGMD.

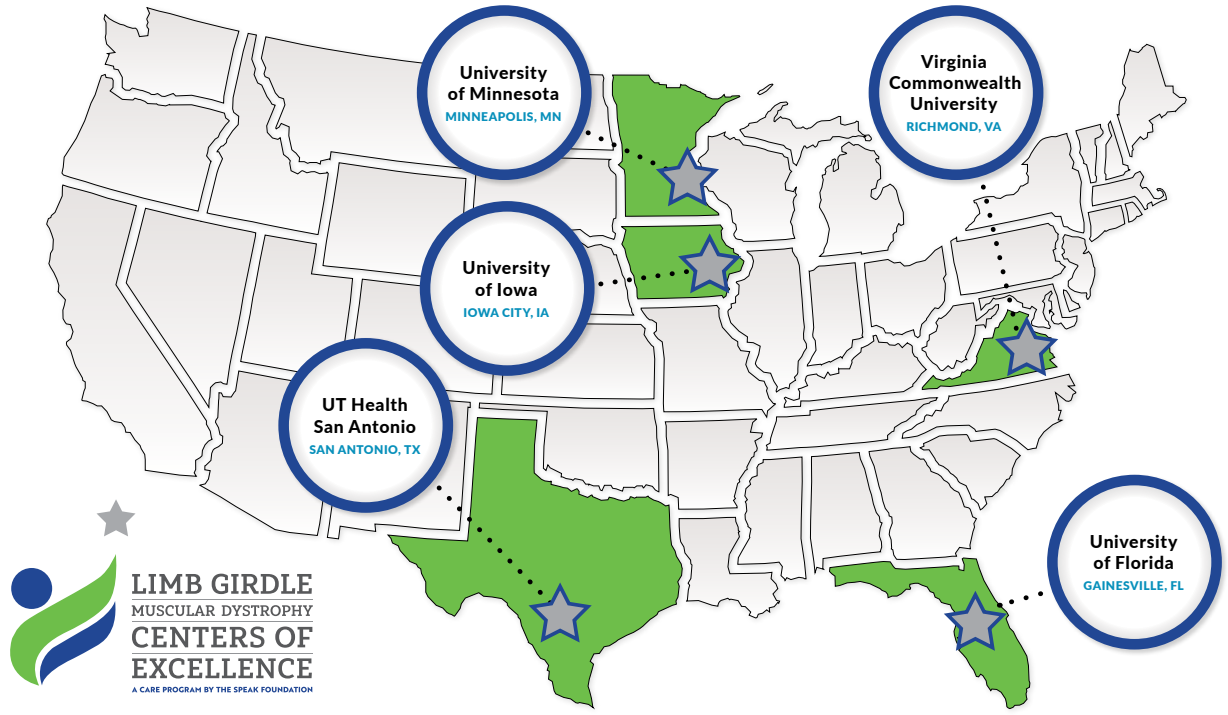
The LGMD Centers of Excellence are built around multidisciplinary care, dedicated coordination, clinical trial readiness, thoughtful expansion to additional partner institutions, and oversight led by patients and families.

As gene therapies and emerging treatments advance, readiness is no longer optional. Centers must be prepared not only to conduct trials, but to provide durable care, manage long-term follow-up, and support patients before and after therapeutic milestones.

For the LGMD community, this is more than a designation. It is a commitment — to coordination, to excellence, and to building a care system that is cognizant of how LGMD patients actually live. ▪

Written by
Kathryn Bryant Knudson, *The Speak Foundation*
Nicholas Johnson, *MD, MSci, FAAN*

LGMD Centers of Excellence Locations ▪ Officially Launches May 4, 2026



For additional information about the LGMD Centers of Excellence and to connect with a coordinator, visit

TheSpeakFoundation.com

UPDATED: MAY 1, 2026

Active GRASP-LGMD Natural History Study

Recruiting:

Trial Readiness and Endpoint Assessment in LGMD R1/2A (GRASP-01-003)*

Inclusion Criteria

- Age between 12 – 50 at enrollment
- Clinically affected (defined as weakness on bedside evaluation in a pattern consistent with LGMD R1/2A)
- Genetic confirmation of LGMD R1/2A (presence of homozygous or compound heterozygous pathogenic mutations in CAPN3)
- Must be able to provide written informed consent and be willing and able to comply with all study requirements

Exclusion Criteria

- Have contraindications to MRI or MRS (e.g., non-MR compatible implanted medical devices or severe claustrophobia)
- Non-ambulatory, defined as unable to safely walk 10 meters without assistive devices (ankle foot orthotics allowed)
- Positive pregnancy test at any timepoint during trial
- Have dominantly inherited CAPN3 mutations (i.e. LGMD D4)
- Any other illness that would interfere with the ability to undergo safe testing or would interfere with interpretation of the results in the opinion of the site investigator

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*ENROLLMENT FOR THIS STUDY CLOSES: APRIL 30, 2026

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Virginia Commonwealth University


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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: Kathryn Bryant Knudson | The Speak Foundation

I am honored to join GRASP-LGMD and represent the voice of individuals who live daily with limb-girdle muscular dystrophy. I am deeply encouraged that our distinguished medical community recognizes the essential role of the patient voice in shaping research, care, and policy. The Speak Foundation has partnered with leading academic institutions to establish LGMD Centers of Excellence. Meaningful patient engagement informs trial design, care coordination, outcome measures, and responsible data sharing. When lived experience is integrated early and structurally, we build smarter research and stronger systems together. I look forward to working alongside these incredible clinicians to ensure that every person living with LGMD is not only represented, but actively shaping the future of our field.

DAY ON THE HILL EVENT

LIMB GIRDLE MUSCULAR DYSTROPHY

LGMD

Would You Like To Become a Delegate for **LGMD Day on the Hill?**

LGMD Day on the Hill is an advocacy event and program of the Speak Foundation. This event is for delegates living with limb girdle in the USA to meet with their legislators on **September 15, 2026**.

Let Your Voice Be Heard.

Selected delegates will need to travel to Washington, D.C., the day before to participate in mandatory advocacy training on September 14th , 2:30 – 5:30 P.M., in preparation for LGMD Day on the Hill. Limited travel stipends are available, provided by **The Speak Foundation** and the **LGMD Awareness Foundation**.

Delegate Applications Accepted through June 15, 2026.

APPLY TO BECOME

A DELEGATE TODAY!

Written By **Bradley Williams**, PhD
Jain Foundation; Senior Editor, LGMD News

and **Marianela Schiava**, MD, PhD
John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle upon Tyne, UK

LGMD and Medical Test Misinterpretation

As if living with LGMD weren't challenging enough, the disease can cause unusual and misleading results in certain common blood and urine tests. The misinterpretation of these test results can, in turn, cause clinicians to think that a person with LGMD has a medical condition they don't actually have or to miss one that they do have. In this article, we highlight some common tests in which LGMD can cause anomalous results that can be misinterpreted by clinicians unfamiliar with muscular dystrophies (MD). All of the misinterpretations mentioned in this article have been experienced and reported by members of the LGMD patient community. To avoid a misdiagnosis or a missed diagnosis, people living with LGMD, as well as their family members, caregivers, and clinicians, should be aware of the limitations of these tests.

AST, ALT, and Liver Function

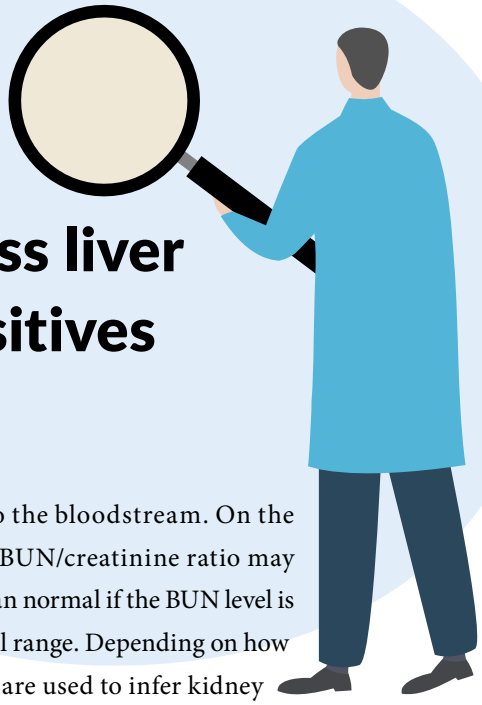
Aspartate Aminotransferase (AST) and Alanine Aminotransferase (ALT) are enzymes commonly included in standard blood test panels. Because both enzymes are found in the liver, and are released into the bloodstream when liver cells are damaged, AST and ALT levels are often referred to by clinicians as "liver function tests."

However, both AST and ALT are also present in skeletal muscle. And because both enzymes can be leaked into the bloodstream from damaged muscle, people with LGMDs, and other types of MD, often have elevated levels of AST and ALT whether or not their liver is damaged.[1-3] This has led to many instances of people with LGMD undergoing unnecessary liver biopsies, either because their clinicians didn't consider damaged muscle as a possible source of the heightened enzyme levels or because the LGMD hadn't been diagnosed, which is more likely if the affected individuals are not experiencing obvious muscle weakness. Testing for creatine kinase (CK, also called creatine phosphokinase) is a useful way to pinpoint whether elevated AST and ALT blood levels originate from muscles rather than the liver, because CK isn't present in the liver.[4]

"All of the misinterpretations mentioned in this article have been experienced and reported by members of the LGMD patient community."

Why should liver health be monitored in a person with MD? People with MD can develop liver disease from causes such as obesity or alcohol consumption just like the general population. Also, some types of MD (though not most LGMD subtypes) and certain metabolic myopathies are also known to adversely affect the liver. Liver toxicity has also emerged as a safety concern in gene therapy, making ongoing liver monitoring necessary for many MD patients.

Fortunately, there are alternative blood markers which can be used to assess liver function without false positives from muscle disease. Two of these, gamma-glutamyl transferase (GGT) — discussed in Ref. [4]) and bilirubin, are being used to monitor liver function in Duchenne muscular dystrophy (DMD) patients following gene therapy.[5] Another more recently identified liver biomarker is glutamate dehydrogenase (GLDH). GLDH is an enzyme that is found primarily within liver cells. When the liver is damaged, GLDH can leak into the blood, where it can be measured with a blood test. Because GLDH in blood comes almost entirely from the liver, its presence in the blood helps doctors and researchers determine whether abnormal blood test results reflect true liver damage rather than other causes, such as muscle



"Fortunately, there are alternative blood markers which can be used to assess liver function without false positives from muscle disease."

injury or strenuous exercise. For this reason, GLDH is often used in clinical trials as a helpful marker to monitor liver health and detect potential liver problems early.[6]

Creatinine and Kidney Function

Creatinine (not to be confused with creatine kinase) is a waste product of muscle metabolism that is released into the blood at a relatively constant rate, largely independent of physical activity. Creatinine is then filtered from the blood by the kidneys and excreted in urine. Because of this, blood and/or urine creatinine levels can be used as a measure of kidney function — indicating how efficiently the kidneys are removing creatinine from the bloodstream.

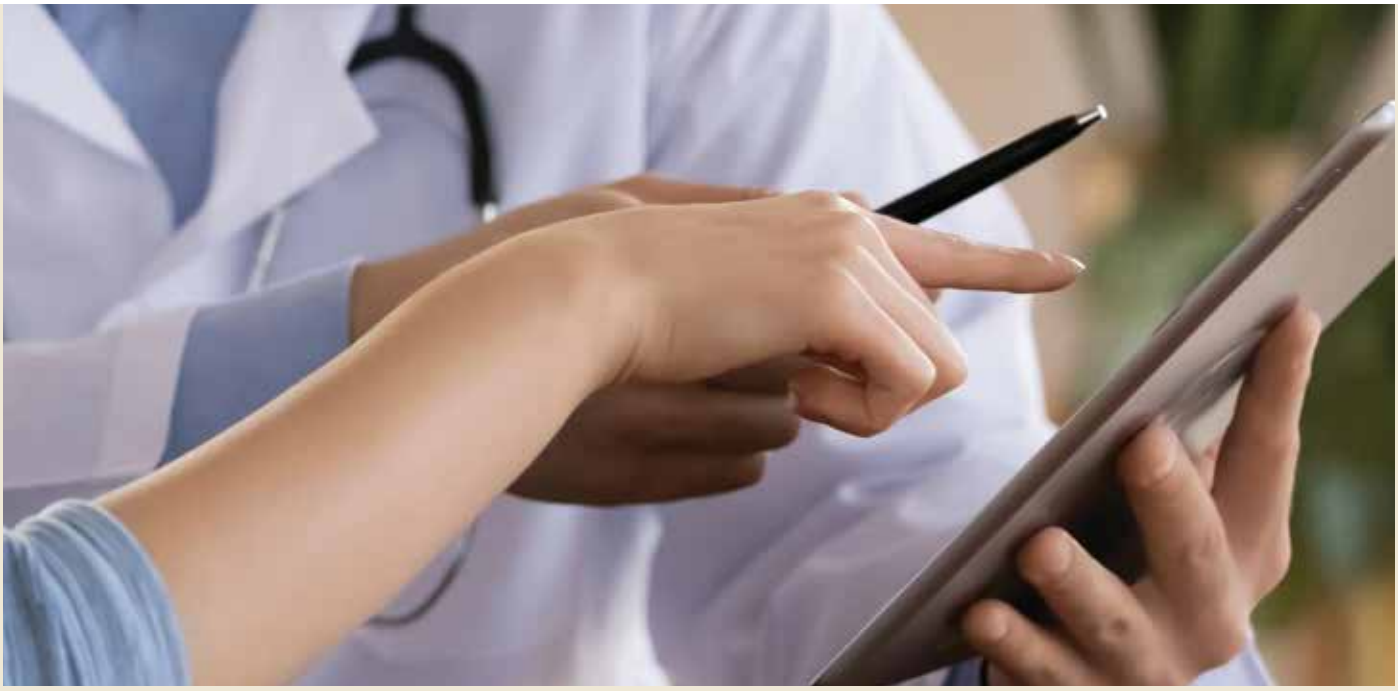
The most commonly used quantitative measure of kidney function is called the glomerular filtration rate (GFR). GFR can be estimated using formulas that take into account a person's age, body size, gender, and other factors, based on the serum (blood) level of creatinine.[7] An abnormally high blood creatinine level is often interpreted as an indication of a kidney problem. Other biomarkers, such as blood urea nitrogen (BUN, produced by protein breakdown and filtered by the kidneys), are also used to assess kidney function. The BUN/creatinine ratio is commonly reported on blood test panels, with an elevated ratio indicating there may be a problem with kidney function.

But what happens if a person doesn't have normal muscle mass, for instance, if they have LGMD? Lower muscle mass can lead to abnormally low blood and urine levels of creatinine, because less creatinine is being released from the muscles into the bloodstream. On the other hand, the BUN/creatinine ratio may appear higher than normal if the BUN level is within the typical range. Depending on how creatinine levels are used to infer kidney function, the test results can be misleading: the elevated BUN/creatinine ratio might suggest a kidney problem that isn't actually present, or the low creatinine level could make it appear that the kidneys are functioning normally even when there is an underlying issue.

The Jain Foundation-sponsored Clinical Outcome Study for Dysferlinopathy,[8] conducted at 16 clinical centers, found that about 70% of 200 study participants with LGMD R2/2B had abnormally low blood levels of creatinine. The clinicians conducting the study concluded that "creatinine-dependent methods will be uninformative" for monitoring kidney function in people with dysferlinopathy. Since all types of MD cause a loss of muscle mass, this finding isn't specific to LGMD R2/2B and has been reported in other types of MD as well.[9]

An alternative biomarker for assessing kidney function is cystatin C, which unlike creatinine, is excreted by tissues other than muscle. As with creatinine, formulas have been developed to estimate GFR from the blood levels of cystatin C. Some studies investigating the use of cystatin C to determine kidney function in patients with muscular dystrophy have concluded that it is more reliable than creatinine.[10,11]

People living with LGMD (or any neuromuscular disease) should also be aware that creatinine levels are often used as a "validation" measure in urine testing, for



instance, in drug screening for employment. Some testing protocols require that the creatinine concentration be above a certain level (typically 20 mg/dl). If it is below this level, the test result may be considered inconclusive, as the sample could be suspected of having been deliberately diluted.[12]

CK Tests

Many LGMD patients are familiar with creatine phosphokinase (CPK), also known as creatine kinase (CK), from their diagnosis and, in some cases, from its use as a biomarker used in clinical studies and trials. CK is a protein that exists in three different forms ("isoenzymes"), which are found in different tissues in the body. CK-MM is the primary form in skeletal muscle, while CK-MB is found primarily in cardiac muscle, and CK-BB is found in the central nervous system. A CK test may be specific to one type of CK or detect multiple isoenzymes.

CK test results may cause confusion if the person hasn't yet been diagnosed with MD. Therefore CK values, whether from a single test or as trends over time, should be interpreted with caution, taking into consideration the individual's overall clinical picture. If possible, unexpected results should be discussed with specialists, because normal levels can vary widely between people. CK is known to be affected by factors such as age, sex, muscle mass, physical activity, and race.[13] Levels can also rise temporarily due to internal or external factors that stress or affect the muscles, such as exercise, illness, or trauma such as a car accident. In people with MD, this may lead physicians to mistakenly attribute elevated CK to an injury or, conversely, to ascribe the CK levels to a known injury when they are actually being caused primarily by the MD.

In one fortunate case, a patient with LGMD was diagnosed after a car accident when his high CK was discovered in tests meant to determine the severity of his injuries. While the emergency room doctors assumed the CK levels showed accident-related muscle damage, follow-up testing ruled this out and ultimately led to his LGMD R2/2B diagnosis.

Because CK-MB is elevated after heart muscle damage, clinicians may misinterpret a general CK elevation (particularly if the test doesn't distinguish between CK isoenzymes) as indicative of cardiac injury. However, other heart biomarkers, such as troponins, are better indicators of heart damage[14] and are therefore preferred when evaluating suspected heart problems.

Share Knowledge with Your Clinicians

A person diagnosed with LGMD will see a variety of clinicians, not just neurologists, because the individual can have a range of medical issues just as anyone else might experience. Because LGMD is so rare, it may be unfamiliar to general practitioners and other medical specialists, which can sometimes lead to misinterpretation of test results, as the examples above illustrate. It is therefore important for patients to be aware of these issues and to communicate openly with their clinicians, encouraging them to consult with neuromuscular specialists when interpreting laboratory findings. ■

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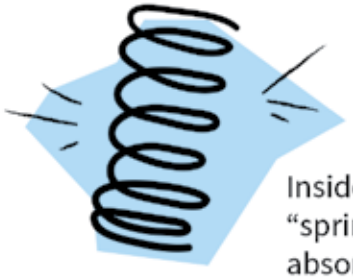
Did you know your muscles have a built-in shock absorber?

Here's the rare rundown on LGMD2I/R9, a subtype of limb-girdle muscular dystrophy



Inside healthy muscles are tiny "springs" that act as shock absorbers—protecting muscle fibers with every contraction.

An enzyme called **FKRP helps build these springs onto a key protein**, alpha-dystroglycan. When FKRP is mutated, those protective springs aren't properly formed.

Without these springs, **muscles absorb damage instead of cushioning it**—leading to repeated **microtears**, **scar tissue**, and **gradual muscle weakness**.

Over time, everyday actions like **walking**, **standing**, or even **breathing** can become more difficult.

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BRIDGEBIO

Written By Kathryn Bryant Knudson

A Global Conversation for the LGMD Community

In October 2025, leaders from the LGMD community across key countries convened for a Global Advocacy Meeting organized by the Speak Foundation to assess the global landscape of limb-girdle muscular dystrophy care, research infrastructure, and patient access. The discussion documented both measurable progress and persistent gaps.

Across continents, clinicians are building multidisciplinary programs, researchers are deepening understanding of genetic subtypes, and advocacy organizations are strengthening patient networks and policy engagement. The dedication is extraordinary. Yet in many parts of the world, patients still struggle to obtain timely genetic diagnoses, consistent standards of care, and opportunities to participate in clinical trials.

In this special feature, *LGMD News* is honored to present perspectives from leading organizations around the world. Their updates offer insight into where innovation is accelerating, where systems are beginning to take shape, and where urgent patient needs remain.

This gathering went beyond discussion, marking a step toward deeper collaboration: breaking down barriers, sharing expertise across borders, and working toward a future where geography does not determine care, research access, or representation for those living with LGMD.

LGMD does not recognize borders. Neither should progress.

PARTICIPATING ORGANIZATIONS:

INTERESTED? If your LGMD organization would like to participate in future international meetings — where we share a global "State of the Union" for LGMDs — please contact *Jessica@TheSpeakFoundation.com* to stay informed.

Proyecto Alpha

Written By
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Irune García
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Advancing Together in Research, Rehabilitation, and Global Visibility for LGMD

Proyecto Alpha is a non-profit organization founded in 2018 in Barcelona, Spain, by families and individuals affected by LGMDs caused by sarcoglycan deficiencies (LGMD R3–R6/2C–2F). Our mission is to improve the quality of life of people living with these conditions through biomedical research, comprehensive rehabilitation, scientific dissemination, social awareness, and emotional support. Our organization participates in international networks for research and patient advocacy, including the LGMD CAB (Community Advisory Board), in which our president serves as an executive member, as well as TREAT-NMD, the World Muscle Society (WMS), and Orphanet.

Living with LGMD in Spain

Living with LGMD in Spain often involves a complex diagnostic process due to the heterogeneity and severity of the disease. Despite the country's strong biomedical research infrastructure, access to genetic testing remains uneven, and disease-specific clinical trials are still limited.

There remains significant regional differences within Spain in terms of access to specialized neuromuscular care, rehabilitation services, and psychological support. These factors further highlight the need for standardized clinical guidelines and shared protocols to ensure equitable access to high-quality care across the country.

In response to this need, Proyecto Alpha collaborated with the European Neuromuscular Centre (ENMC) in the development of international standards of care for sarcoglycanopathies, contributing patient perspectives to help define best clinical practices. Complementing these efforts at the community level, our organization has also co-hosted the Ibero-American LGMD Conference in collaboration with advocacy organizations across Latin America, helping to bridge LGMD communities in Spain and Latin America and to promote stronger Spanish-speaking representation within the global LGMD community.

Patient Needs in Spain

People living with LGMD in Spain face significant physical, psychological, and social challenges that require a comprehensive and coordinated approach. Proyecto Alpha has conducted the Psychosocial and Quality of Life Study since 2024 in collaboration with the University of Deusto (Neuro-e-Motion Research Team). The study has found a marked reduction in health-related quality of life (HRQoL) among both pediatric and adult patients, particularly in physical and psychosocial domains. In children, higher levels of emotional symptoms and social difficulties have been observed relative to the equivalent age- and sex-matched control group drawn from the general population without the condition, underscoring the importance of integrating psychological and emotional care into standard clinical management.

In addition to psychosocial needs, individuals with LGMD require consistent access to specialized physiotherapy to preserve mobility, prevent contractures, optimize respiratory and postural function, and maintain functional independence. However, many of these services are not fully covered by the public healthcare system, creating disparities in access to essential supportive care.

Ways in Which Proyecto Alpha is Addressing these Needs

In response to identified psychosocial needs, Proyecto Alpha will launch group psychotherapy sessions this year aimed at strengthening emotional well-being and community connection. Led by a licensed professional psychologist, these sessions will provide a safe space for individuals affected by LGMD to connect, share achievements and concerns, and foster mutual support through guided group dynamics.

To address the physical care needs, Proyecto Alpha has developed several patient-centered support initiatives. Since 2019, the Rehabilitation Program has ensured access to specialized physiotherapy for individuals with LGMD, covering services that are not fully financed by the public healthcare system. The program focuses on enhancing both physical and emotional well-being, with the goal of maintaining independence and quality of life.

Natural History Studies and Research Landscape in Spain

Proyecto Alpha promotes research projects focused on the clinical, genetic, and psychosocial aspects of sarcoglycan-related LGMDs, with the aim of generating clinically relevant knowledge and advancing toward future clinical trials.

- Since 2022, the organization has supported a clinical and pathological Natural History Study at Hospital Sant Joan de Déu (Barcelona-Spain) involving 14 pediatric patients with LGMD R3/2D, R4/2E, and R5/2C. This study analyzes clinical and genetic evolution through muscle biopsies, MRI, and molecular analysis. Preliminary findings indicate a more stable clinical course in SGCG variants and more variable progression in SGCA and SGCB variants. Characteristic muscle MRI patterns with potential value as progression biomarkers have also been identified. This work represents the first comprehensive pediatric characterization of sarcoglycanopathies in Spain.
- This study was expanded in 2025 to include the adult population via a cross-sectional project: Adult Natural History Study at Hospital Clínic de Barcelona, aiming to compare pediatric and adult phenotypes and to analyze functional evolution across the lifespan.
- Proyecto Alpha is continuing its structured Psychosocial Research Project (previously mentioned) for both pediatric and adult populations living with LGMD. This initiative complements clinical natural history efforts by incorporating patient-reported outcomes and psychosocial dimensions into the understanding of LGMD across a range of ages, helping to inform more holistic standards of care.
- In collaboration with the University of Almería, which hosts a dedicated University Chair on Rare Diseases, Proyecto Alpha has carried out a Quality of Life Study highlighting the impacts of LGMD on autonomy, emotional well-being, and the burden assumed by families and caregivers. These findings reinforce the need for an integrated, multidisciplinary approach to care that addresses not only physical aspects, but also psychosocial and family-related dimensions of the disease.
- Proyecto Alpha also maintains a National Patient Registry, active since 2018, which connects families, clinical centers, and research networks. This strengthens collaboration and readiness for future natural history studies and clinical trials.

Conclusion

In Spain, the LGMD community continues to transform challenges into collective action. While significant gaps remain in equitable access to specialized care, rehabilitation, psychological support, and clinical trials, the growing collaboration among patients and families, clinicians, researchers, and patient organizations signals meaningful progress. Research initiatives, expanded registries, and international partnerships will improve understanding and management of LGMDs. ■

▶ **ProyectoAlpha.org**

EDITOR'S NOTE The story of how **Proyecto Alpha** developed was highlighted in our Winter 2026 Issue.

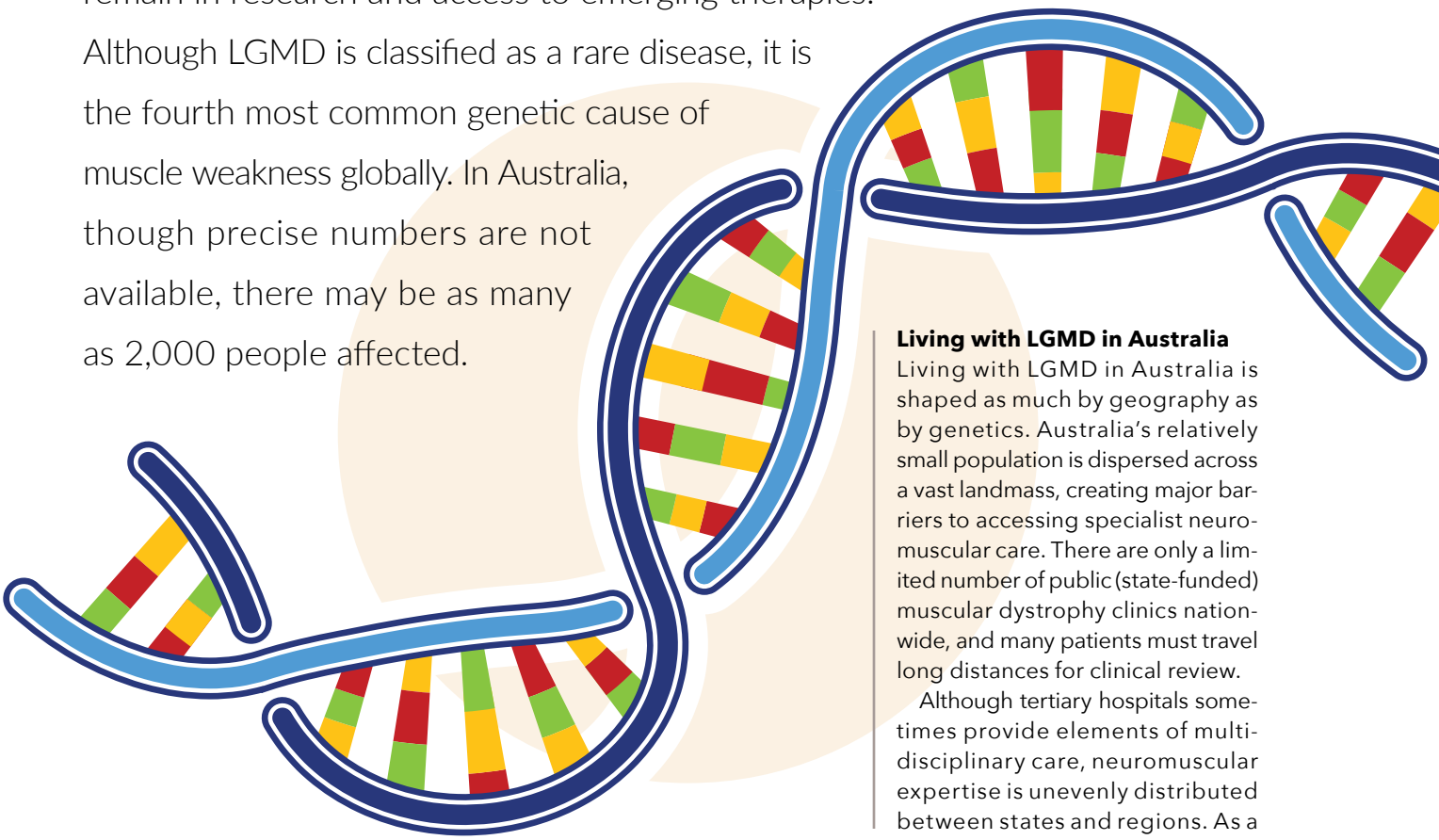
Daniel Ferguson LGMD Foundation

Written By Jo Offord, FRACP MBBS BSc (Hons 1)
and Michelle Lorentzos, PhD FRACP MBBS BA (Comms)

LGMD: An Australian Perspective

On behalf of the Daniel Ferguson LGMD Foundation, this article provides an overview of LGMD in Australia—the realities of living with this condition, the supports currently available, and the significant gaps that remain in research and access to emerging therapies.

Although LGMD is classified as a rare disease, it is the fourth most common genetic cause of muscle weakness globally. In Australia, though precise numbers are not available, there may be as many as 2,000 people affected.



Living with LGMD in Australia

Living with LGMD in Australia is shaped as much by geography as by genetics. Australia's relatively small population is dispersed across a vast landmass, creating major barriers to accessing specialist neuromuscular care. There are only a limited number of public (state-funded) muscular dystrophy clinics nationwide, and many patients must travel long distances for clinical review.

Although tertiary hospitals sometimes provide elements of multidisciplinary care, neuromuscular expertise is unevenly distributed between states and regions. As a

result, the experience of care can vary significantly depending on where a person lives. Often, care coordination falls largely on the patient and their family.

Patient Needs

Australian management of LGMD focuses primarily on supportive, rehabilitative, and preventative care. Core patient needs include:

- **Physiotherapy and tailored exercise programs to maintain mobility and prevent secondary complications**
- **Cardiac surveillance, including access to cardiac MRI, due to the risk of cardiomyopathy**
- **Respiratory monitoring and specialist care**
- **Gastroenterology and orthopaedic surgery input**
- **Timely access to mobility aids, home modifications, and personal care support**

While these elements represent the intended framework of care, access and delivery are often inconsistent, and many patients experience delays or gaps in services.

Beyond medical care, patients need equitable access to emerging therapies, clinical trials, and coordinated multidisciplinary services. The lack of active LGMD-specific therapeutic options in Australia remains a major unmet need.

Patient Supports Available

Disability-related supports are primarily accessed through the National Disability Insurance Scheme (NDIS), which funds mobility equipment, home modifications, and personal support workers for eligible individuals.

Community and advocacy support is provided by organisations including:

- **Muscular Dystrophy Australia**
- **Daniel Ferguson LGMD Foundation**

These organisations provide education, peer support, advocacy, and assistance navigating complex health and disability systems.

Status of Natural History Studies

Australia has had limited participation in LGMD-specific natural history studies, with one example being the Dysferlinopathy Clinical Outcome Study (2013-2018) focused on LGMD R2/2B, which included a study site in Sydney. The Daniel Ferguson LGMD Foundation is set to support a natural history study based in Melbourne. While international research efforts continue to characterize genotype–phenotype relationships and disease progression patterns, Australian involvement is often dependent on individual site capacity.

The lack of coordinated national registries and structured natural history programs limits readiness for future clinical trials and reduces opportunities for Australian patients to contribute to – and benefit from – global research advances. Many LGMD registries operate at an international level, so participation of Australian patients in these global databases may improve opportunities for inclusion in future clinical trials and access to emerging research developments.

Clinical Trials

Brisbane, Queensland is a trial site for the FORTIFY trial of ribitol in LGMD R9/2I. But according to the Australian New Zealand Clinical Trials Registry, there are no other active LGMD-specific interventional clinical trials recruiting in Australia.

Novel therapies which have been discussed for LGMDs and in some cases approved for other conditions include gene replacement, antisense oligonucleotides (ASOs), RNA interference (RNAi), stem cell transplantation, and gene editing. The current situation in Australia regarding treatment approval and access to therapies is as follows:

- **Stem cell transplantation and gene therapies have been established clinically in other conditions, but not for LGMDs.**
- **ASO therapy is funded for other neuromuscular conditions, such as nusinersen for spinal muscular atrophy, but not for LGMDs.**
- **RNA interference therapies (applicable to diseases with dominant inheritance) are government-funded in Australia through the Pharmaceutical Benefits Scheme (PBS) for other conditions, including vutrisiran for hereditary transthyretin amyloidosis, but none are approved for LGMD**
- **Gene editing approaches remain confined to research or clinical trials for other conditions.**

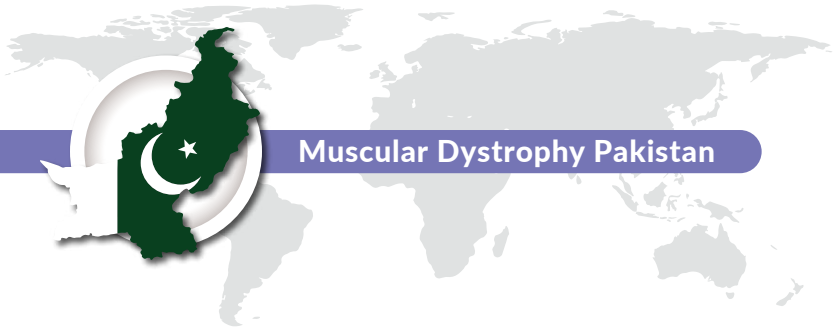
Conclusion

LGMD in Australia is characterised by resilience – from patients, families, clinicians and advocacy organisations – in the context of geographic challenges and limited therapeutic options. While supportive care is well established, access to coordinated national research infrastructure and advanced therapies remains limited.

Bridging this gap will require investments and participation in natural history studies. registries, trial readiness, and stronger alignment between global scientific advances and Australian regulatory and funding pathways. Only then can patients with LGMD fully participate in – and benefit from – the next generation of therapies. ■

▶ **DFFoundation.com.au**

EDITOR'S NOTE **The story of how the Daniel Ferguson LGMD Foundation developed was highlighted in our Winter 2026 Issue.**



Muscular Dystrophy Pakistan

Written By Ghulam Ali

The State of LGMD in Pakistan

In Pakistan, LGMD remains under-diagnosed and poorly supported. Limited diagnostic infrastructure, lack of specialist care, and low awareness at community and policy levels create significant medical, social, and economic challenges.

Muscular Dystrophy Pakistan (MDP) was established in August 2021 to address these gaps by advocating for patients, raising awareness, facilitating research collaboration, and working toward equitable access to diagnosis, care, and future therapies. As a patient-led organization operating in a low-resource setting, MDP serves as a critical voice for LGMD families across the country.

What Is It Like to Have LGMD in Pakistan?

Living with LGMD in Pakistan is complex and often isolating. Many patients experience symptoms in childhood or adolescence, but diagnosis is frequently delayed due to limited awareness among primary healthcare providers and restricted access to genetic testing. Families often consult multiple doctors before receiving an accurate diagnosis—if they receive one at all.

Mobility challenges are compounded by inaccessible infrastructure, limited availability of assistive devices, and social stigma surrounding disability. Public spaces, transportation, schools, and workplaces are rarely designed to accommodate people with physical disabilities. As a result, individuals with LGMD often face educational disruption, unemployment, and social exclusion.

Cultural misconceptions about genetic disease deepen isolation. Families may experience blame or shame, particularly in communities where genetic conditions are poorly understood. Patients frequently experience anxiety and depression, but these are rarely addressed due to stigma and limited psychosocial services.

Patient Needs

The unmet needs of LGMD patients in Pakistan are extensive and multifaceted:

1. **Early and Accurate Diagnosis**
   Access to affordable genetic testing remains the most urgent need. Only a handful of laboratories in the country can perform genetic testing for neuromuscular disorders, often at costs beyond the reach of most families.

2. **Multidisciplinary Care**
Management of LGMD requires coordinated care involving neurologists, physiotherapists, cardiologists, pulmonologists, orthopedic specialists, and rehabilitation experts. Such multidisciplinary clinics are largely absent in Pakistan.

3. **Rehabilitation and Assistive Support**
Regular physiotherapy, mobility aids, orthotics, and respiratory support devices are essential but inconsistently available and rarely covered by insurance or government programs.

4. **Psychosocial and Peer Support**
Patients and caregivers need counseling services, peer networks, and community-based support systems to cope with the emotional and social impact of LGMD. These resources are scarce.

5. **Financial Protection**
Most families bear healthcare costs out of pocket. Loss of income due to disability further exacerbates poverty and limits access to care.

Patient Support Systems Available

Patient support for LGMD in Pakistan is limited and largely driven by non-governmental efforts. MDP plays a central role by:

- Providing patient guidance and referrals
- Conducting awareness campaigns in communities and online
- Organizing rare disease and LGMD awareness events
- Supporting families in navigating diagnosis and care
- Advocating with hospitals, policymakers, and international partners

While some tertiary hospitals offer neurology services, these are concentrated in major cities, leaving rural populations underserved. There is no national LGMD registry, structured home-based care system, or government-funded LGMD support program.

Despite these limitations, patient communities are growing through digital platforms, allowing families to connect and access information—often for the first time.

Status of Natural History Studies

Pakistan currently lacks formal, nationwide natural history studies for LGMD. This absence represents a significant gap in clinical understanding and research readiness.

However, there is strong potential. Pakistan's large population, high prevalence of consanguinity, and growing diagnostic capacity create an opportunity to contribute valuable data to global LGMD research. MDP has begun informal patient mapping and data collection and is working toward establishing patient registries in collaboration with academic and clinical partners.

Partnerships with institutions such as Aga Khan University Hospital and international research networks are being explored to support ethically conducted, locally relevant natural history studies. Building this foundation is essential for future clinical trials and ensuring Pakistani patients are not excluded from emerging therapies.

Clinical Trials Landscape

At present, there are no active LGMD clinical trials in Pakistan. Patients who wish to participate in trials abroad face major barriers, including visa restrictions, travel costs, documentation challenges, and physical limitations.

Interest among patients is high, and clinician awareness is gradually increasing. With investment in diagnostic confirmation, registry development, and research partnerships, Pakistan could become a valuable site for observational studies and future interventional trials.

MDP advocates for inclusion of low- and middle-income countries in global drug development efforts. Expanding trial access beyond high-income regions is essential to achieving equity in rare disease research and treatment availability.

Looking Ahead: Opportunities for Impact

Despite these challenges, the LGMD community in Pakistan is resilient and motivated. With strategic collaboration, meaningful progress is possible. Key opportunities include:

- Establishing a national LGMD patient registry
- Expanding access to genetic testing through partnerships
- Developing multidisciplinary care models
- Launching natural history studies
- Engaging pharmaceutical companies and researchers early in the research process

Conclusion

LGMD in Pakistan exists at the intersection of rare disease, disability, and health inequality. While current support systems are limited, patient voices are growing stronger. By investing in awareness, research infrastructure, and inclusive drug development, the global LGMD community has an opportunity to transform lives in underserved regions.

Muscular Dystrophy Pakistan is committed to serving as a bridge between patients, clinicians, researchers, and global advocacy networks. International recognition and collaboration are critical to ensuring LGMD patients in Pakistan are seen and included. Our organization stands ready to lead this effort nationally and collaborate globally – so that no LGMD patient, regardless of geography or nationality, is left behind. ▪

▶ **Muscular Dystrophy Pakistan.com.Free**

UILDM

Written By Cristina Sancricca

LGMD in Italy: The UILDM Perspective

About UILDM

Unione Italiana Lotta alla Distrofia Muscolare (UILDM) — the Italian Muscular Dystrophy Association — has a unique dual role in Italy, serving both as a national patient association and as a network of healthcare and rehabilitation providers. Since 1961, UILDM has supported people living with muscular dystrophies and other neuromuscular diseases through social inclusion, advocacy, clinical services, and research.

What Is It Like to Live with LGMD in Italy?

Living with LGMD in Italy means navigating a healthcare landscape that achieves high levels of clinical expertise but also has significant regional variability. Italy has a long-standing tradition of specialized neuromuscular care, supported by experienced clinicians, dedicated centers, and strong links between patient organizations and the medical community. At the same time, access to care and services can differ markedly depending on geographic location, creating unequal experiences for individuals and families affected by LGMD.

Within this context, UILDM plays a unique role. Founded in 1961, it operates both as a national patient association and as a network of healthcare and rehabilitation providers. This dual identity enables UILDM to support individuals with LGMDs through advocacy, social inclusion, and direct involvement in clinical care and long-term disease management.

Patient Needs

Despite the presence of specialized centers and experienced professionals, people living with LGMD in Italy continue to face important unmet needs. Through patient testimonials and ongoing dialogue within the UILDM patient chat group, several recurring challenges have been identified.

Patients report unequal access to care between regions, with some areas offering comprehensive services while others lack coordinated follow-up. Psychological support is often insufficient, and care pathways are sometimes fragmented, placing additional strain on individuals and families. Administrative barriers can also pose significant difficulties, particularly in relation to prescriptions, assistive devices, and disability benefits.

Beyond the healthcare system, accessibility remains a major issue. Public transportation and urban infrastructure frequently limit independence and participation. Financial burdens – including the costs of rehabilitation, transportation, and assistive technologies – further compound these challenges. Many individuals also face challenges to independent living due to shortages in personal assistance services and accessible housing options.

These realities highlight that clinical management alone is not enough. Living successfully with LGMD requires integrated, long-term approaches that address medical, social, psychological, and economic needs together.

Patient Support Available

UILDM provides a broad and structured network of support for people living with LGMD across Italy. With 65 local branches and more than 9,000 volunteers, the organization works daily to promote autonomy, inclusion, and access to specialized care.

In many regions, UILDM branches offer direct healthcare and rehabilitation services, including physiotherapy, respiratory care, speech and occupational therapy, psychological support, and clinical follow-up. A key example is UILDM Lazio, which includes the Fondazione UILDM Lazio, a highly specialized rehabilitation and clinical follow-up center serving hundreds of individuals with neuromuscular diseases each year. Through multidisciplinary programs, instrumental assessments, respiratory and cardiological monitoring, and long-term management, this center illustrates how UILDM integrates community support with high-standard clinical care.

In addition, the UILDM LGMD Group brings together individuals affected by all forms of LGMD nationwide. The group fosters peer connection, shares scientific and medical updates, and serves as an important channel for understanding real-world patient needs. It collaborates closely with other Italian LGMD associations and with international organizations, strengthening both national and global networks of support.

Status of Natural History Studies

Significant progress has been made in understanding the natural history of LGMD in Italy. Data from 599 patients collected across 12 Italian neuromuscular centers provides insight into the clinical and genetic diversity of the disease. Autosomal recessive forms account for the majority of cases (84%), with calpainopathy (LGMD R1/2A), dysferlinopathy (LGMD R2/2B), and the sarcoglycanopathies (LGMD R3–R6/2C–2F) among the most common subtypes.

These results highlight the wide variability in age of onset, disease progression, severity, and cardiac and respiratory involvement in the Italian LGMD patient population, reinforcing the need for coordinated, multidisciplinary care throughout the disease course.

UILDM is actively supporting the creation of the Telethon–UILDM national LGMD Registry. Currently in development, this registry aims to harmonize clinical, genetic, and functional data across centers, improving understanding of disease progression and strengthening readiness for future therapeutic developments.

Clinical Trials and Research

UILDM is deeply committed to advancing scientific research. Through its long-standing partnership with the Telethon Foundation, more than 67 research projects have been funded, and over 6,000 individuals have participated in clinical studies related to neuromuscular diseases. This collaboration has also contributed to the development of specialized neuromuscular centers, such as the NEMO Clinical Centers, which now form a cornerstone of Italy's clinical expertise.

These efforts to develop research infrastructure, encourage patient participation, and develop registries lay the groundwork for clinical trials by improving natural history knowledge, standardizing data collection, and strengthening collaboration between centers.

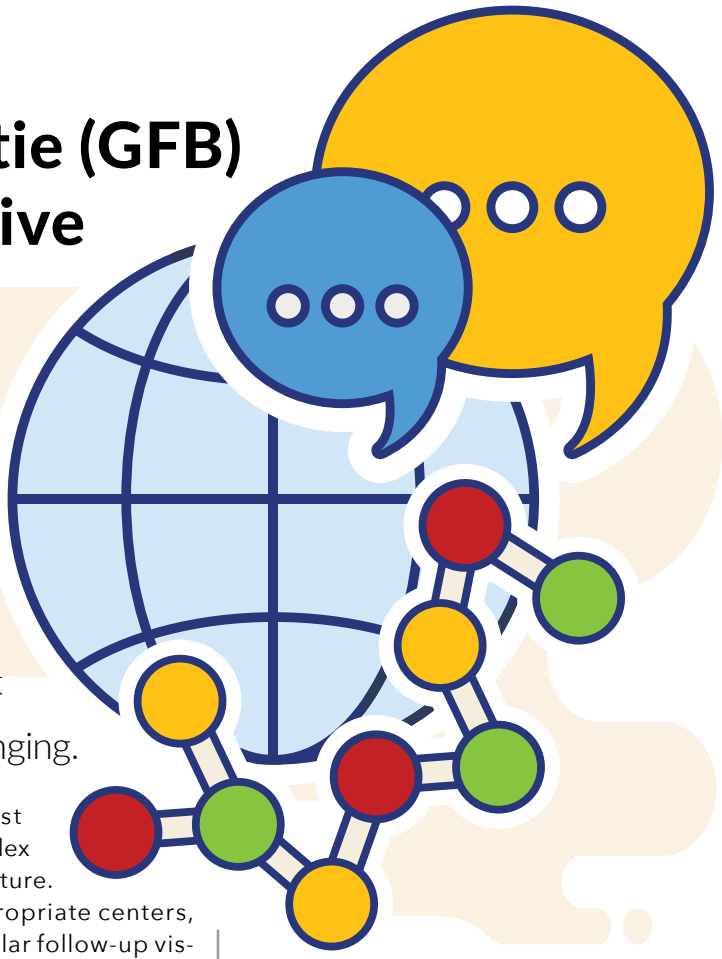
Looking Ahead

By strengthening national and international networks and continuing to address unmet patient needs, UILDM aims to ensure that every person living with LGMD in Italy has access to equitable care, updated knowledge, and meaningful opportunities for full participation in society. ■

▶ UILDM.org

Written By Beatrice Vola

LGMD in Italy: The Gruppo Familiari Beta-sarcoglicanopatie (GFB) Foundation Perspective



What Is It Like to Have LGMD in Italy?

Over the last two decades, Italy has built a well-organized and increasingly visible LGMD community, much like efforts underway in other countries. Patient advocacy, clinical expertise, and research efforts have grown, but living with LGMD in Italy remains challenging.

Specialized clinical centers for LGMD are primarily located in central and northern Italy, in cities such as Milan, Turin, Padua, Genoa, Pisa, and Rome. The establishment of the NEMO Clinical Centers in 2008 created a national network of seven rehabilitation centers focused on neuromuscular diseases. However, because these centers are concentrated in the north and center of the country, individuals living in southern Italy often must travel long distances to access specialized care.

Although medical treatments and specialist visits are covered by the Italian national health system, families still must navigate a complex healthcare structure. Identifying appropriate centers, scheduling regular follow-up visits, and coordinating local services can be difficult and time-consuming, making daily life with LGMD demanding despite there being universal healthcare coverage.

Patient Needs

People living with LGMD in Italy often face ongoing unmet needs, particularly related to geographic disparities in care. As mentioned above, families in southern regions are often disadvantaged due to travel requirements and limited local expertise.

Assistive devices and supportive equipment are another critical area of need. While these "aids" are provided free of charge through the national health service, many devices required by people living with LGMD fall outside standard coverage models. In such cases, families must pay the difference out of

pocket, which can create significant financial strain, especially for those with limited resources.

Beyond specialist medical care, individuals and families must also personally navigate and coordinate community-based health and care services in their local area, adding another layer of complexity to long-term disease management.

Patient Support Available

Italy has a strong and active LGMD patient advocacy community. Three main disease-specific organizations and one broader "umbrella" organization support people living with LGMDs:

- **AICa3 (Italian Association Calpainopathy)**, founded in 2012, focuses on calpainopathy (LGMD R1/2A) and has organized multiple international conferences.
- Our foundation, **GFB Foundation**, founded in 2013 by parents of two children with sarcoglycanopathy, supports individuals with sarcoglycanopathies (LGMD R3–R6/2C–2F).
- **Conquistando Escalones**, established in 2015, supports individuals with LGMD D2/1F and includes more than 100 patients.
- **UILDM Limb-Girdle Group**, founded in 2020, works within the broader Italian Muscular Dystrophy Association to focus specifically on LGMDs, promote therapeutic awareness, and facilitate dialogue among patients, families, and clinicians.

Together, these organizations provide education, peer support, advocacy, and connections to research and clinical expertise at both national and international levels.

Research Efforts

In Italy, relatively few institutions provide funding for scientific research, and public research budgets are limited, resulting in support for only a small number of investigators. Private organizations periodically fund research through competitive grants; among these, the Telethon Foundation is the most active and represents one of the largest sources of neuromuscular research funding in Italy. To advance their work, Italian researchers often seek financial support outside the country through international research networks, including grants from organizations such as AFM-Téléthon in France and the Muscular Dystrophy Association (MDA) in the United States.

Patient Registries

Patient registries have become important tools for advancing research. The Italian Register of People with Muscular Dystrophies and Myopathies (RegistroNMD), promoted by Telethon Foundation and UILDM, collects clinical and genetic data to support research and trial readiness. In addition, the GFB Sarcoglycan Registry, launched in 2018, specifically focuses on LGMD R3–R5 (2C–2E) and currently includes more than 350 registered participants from Italy and abroad.

Status of Natural History Studies and Clinical Trials

Italy has contributed to several natural history studies on LGMD over the past two decades, primarily through specialized neuromuscular centers. For sarcoglycanopathies, the JOURNEY study sponsored by Sarepta Therapeutics was initiated in 2021 for LGMD R3–R5 and R1 (2C, 2D, 2E, and 2A) at sites in Milan, Genoa, and Padua.

Italy also participates in international clinical research efforts, although access to clinical trials has been limited. The Policlinico of Milan was selected as a reference center for both the EMERGENE trial for LGMD R4/2E and the Atamyo Therapeutics trial for LGMD R5/2C. However, to date, no patients in Italy have received treatment within these trials.

Initiation of clinical trials in Italy requires authorization from both the European Medicines Agency (EMA) and the Italian Medicines Agency (AIFA). In the past, there have been some drugs that were not approved by the EMA despite having already been approved by the U.S. Food and Drug Administration (FDA). In the United States, regulatory pathways may allow for drug approval based primarily on safety and early evidence of benefit, with functional outcomes further evaluated after approval. In contrast, European regulatory authorities typically require more extensive evidence of clinical benefit prior to approval.

Authorization for the EMERGENE trial in Europe was granted only after patient recruitment elsewhere was nearly complete. Consequently, only one patient from Belgium was enrolled in the European arm of the study; notably, this individual is also a member of the GFB Foundation.

Looking Forward

The LGMD community in Italy has been very active in establishing clinical networks, conducting natural history studies, developing patient registries, and supporting drug development and clinical trials. However, much remains to be done, particularly in supporting patient care throughout the country and achieving approved therapies for LGMDs. ▪

▶ **LGMD2e.org**

EDITOR'S NOTE **The story of how GFB developed was highlighted in our Winter 2026 Issue.**

CureSCG

Written By Rana Abu Khadra

LGMD in the UAE: The CureSCG Perspective

About CureSCG

CureSCG is a United States-registered 501(c)(3) nonprofit organization founded by our family. We approach this work first and foremost as parents of a child living with LGMD. Although we currently reside in the United Arab Emirates (UAE), we do not speak on behalf of the country, its healthcare system, or any national programs. The perspectives shared here reflect solely our lived experience as a family navigating LGMD while residing in the UAE, informed by our engagement in international patient advocacy and research communities.

Living with LGMD while Residing in the UAE

From our family's experience, living in the UAE has afforded us access to advanced medical facilities and specialized services critical for managing a progressive neuromuscular condition. Our daughter has been able to receive multidisciplinary care including regular neuromuscular follow-up visits, cardiac monitoring, pulmonary assessments, physiotherapy, and hydrotherapy. These services have been essential in maintaining our daughter's health and quality of life. This reflects the high level of care available in certain tertiary healthcare facilities in the country.

Patient Needs and Available Support

Our diagnostic journey proceeded quickly once we recognized that our daughter was having difficulty with movement, after only showing subtle signs such as toe-walking previously. We consulted a pediatrician who ordered blood tests, which showed elevated creatine kinase (CK) levels, suggesting a neuromuscular disorder. We were referred the following day to a neurologist who, based on the clinical findings and laboratory results, suspected LGMD even before genetic confirmation. Genetic testing was subsequently requested to confirm the diagnosis and determine the specific subtype. Within approximately one month, the genetic results confirmed the diagnosis of LGMD.

Despite access to high-quality clinical services, many of the challenges we have faced will be familiar to families affected by rare diseases globally. LGMD remains a condition with limited public awareness, and families often bear the responsibility of raising awareness, coordinating care, seeking disease-specific information, and connecting with experts. Local patient support structures specific to LGMD are limited, which means families must frequently turn to international patient organizations, online communities, and overseas specialists for education, emotional support, and guidance.

Our experience highlights the need for stronger patient-centric resources, improved awareness of LGMD subtypes, and greater integration of patient voices into research and care discussions. These observations are based on our personal journey and should not be interpreted as a comprehensive assessment of all patient experiences in the UAE.

A very positive recent development has been the opening of a dedicated LGMD clinic in Abu Dhabi in collaboration with local

medical groups. Our exposure to this initiative has been limited to our personal interactions, but its existence represents a meaningful step toward more coordinated, disease-specific care for families in the UAE affected by LGMDs.

The UAE Genome Program is a national project aimed at sequencing the genomes of Emirati citizens to support precision medicine and improve the diagnosis of genetic diseases. By expanding access to genetic testing and building a national genomic database, the program may help identify individuals with rare genetic conditions such as LGMD. This could facilitate earlier diagnosis, support natural history studies, and improve patient identification for clinical research and trials.

Research, Natural History, and Clinical Trials

From our perspective as a family, access to natural history studies and clinical research opportunities within the region appears limited. We are not aware of large-scale natural history studies for LGMD being conducted locally. This has meant that our engagement with research has largely occurred through international channels, particularly in the United States and Europe.

Similarly, access to disease-modifying therapies and clinical trials, especially gene therapy, remains constrained worldwide. Only a small number of LGMD gene therapy programs are currently in development, and participation in these studies often requires families to travel internationally. This reality has shaped our advocacy efforts and reinforces the global nature of the challenges faced by the LGMD community.

Conclusion: A Patient- and Advocacy-Led Narrative

This narrative reflects our family's lived experience rather than a policy analysis or comprehensive assessment of the UAE healthcare system. By sharing our perspective, we hope to contribute meaningfully to broader conversations among clinicians, researchers, and patient advocates, and to highlight the real-world challenges families may face when navigating care and research pathways for LGMD. ▪

▶ **CureSCG.org**

EDITOR'S NOTE **The story of how CureSCG developed was highlighted in our Winter 2026 Issue.**

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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!***

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The LGMD Awareness Foundation, Inc. is a 501(c)(3) non-profit advocacy organization committed to raising global awareness about limb-girdle muscular dystrophy (LGMD), a group of rare and often debilitating neuromuscular diseases. Help support our efforts at **lgmd-info.org/donate** to raise awareness of LGMD and advocate for individuals living with these rare conditions — so they can more easily access diagnosis, care, and treatment.

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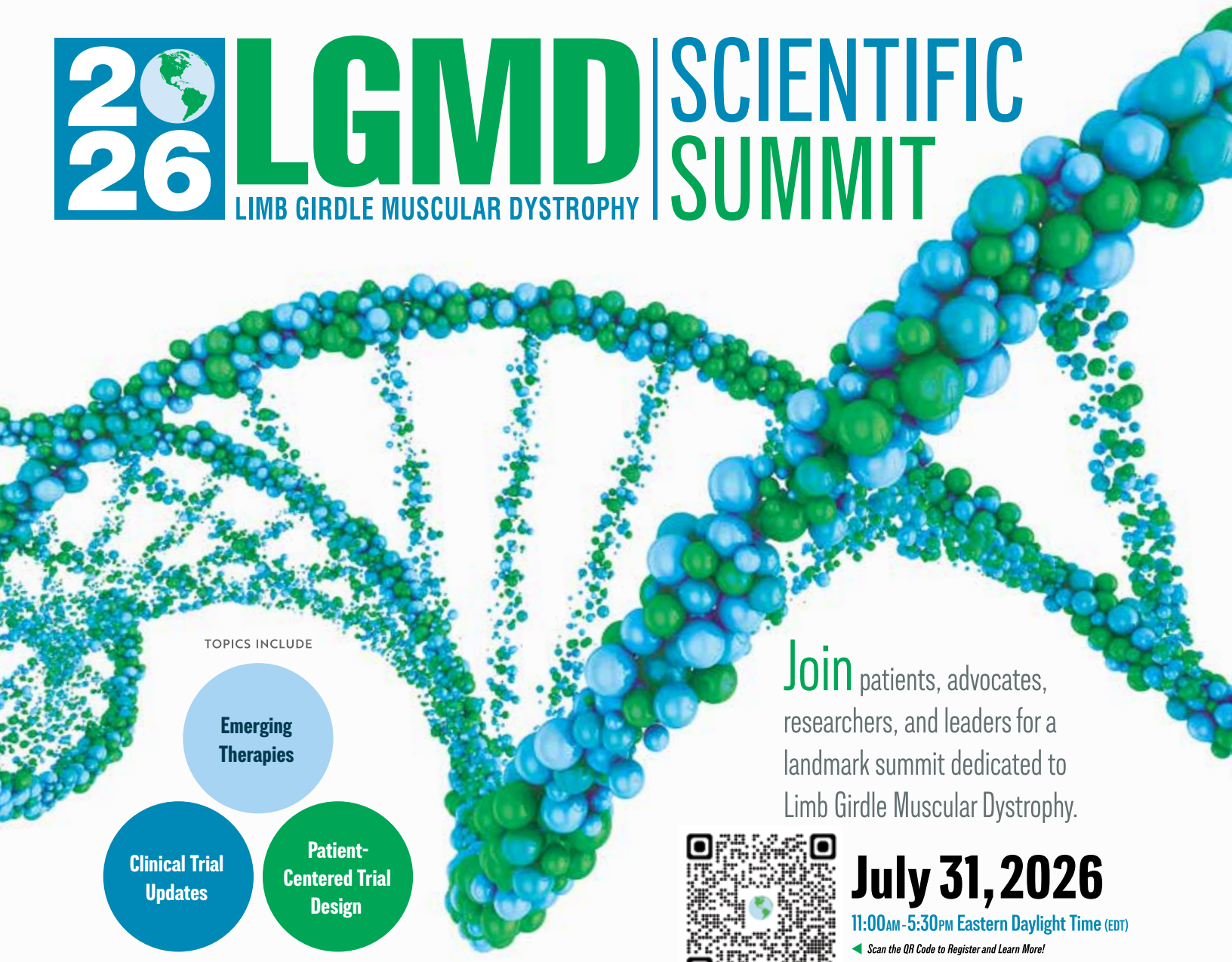

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