Vol 3 / Issue 4

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

2023 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE

WASHINGTON, D.C.

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

3RD INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY

CONFERENCE PROGRAM INSIDE

2023 International LGMD Conference

Do you have LGMD2I/R9?

FOCUS GROUP

We Want to Hear From You...

Join Us for a Focus Group Discussion on what individuals with LGMD2I/R9 and their caregivers want from potential therapies.

Sunday
October 29
12:10PM – 1:10PM ET

Grand Hyatt Washington
Renwick/Bulfinch
1000 H St NW
Washington, DC 20001

To register email:
2ipatientfocusgroup@gmail.com

ML BioSolutions
a bridgebio company

Welcome

To Our 2023 International LGMD Conference!

We all know that the world is changing and treatments are in development now for many forms of LGMD. We now are facing a potential future where many forms of LGMD could be treated. This is why our conference theme is Believe there is Hope for a Cure.

You will notice that we have devoted this entire issue to our upcoming conference! Did you know that even if you cannot attend in person, you can attend virtually and watch the conference live? If you are interested in this option, you can purchase a virtual ticket through EventBrite by visiting **InternationalLGMDConference.com**. We are also live streaming (for **FREE**) the Saturday night banquet with Dr. Peter Marks, Director of the Center for Biologics Evaluation and Research (CBER) for the Food and Drug Administration (FDA). The link to watch this special banquet **FREE** is included in our program.

It is very important for our community to stay informed about the latest in progress towards treatments. We are now seeing multiple companies developing therapies for several LGMD subtypes. You do not want to miss any clinical trial that is available to you. You can stay updated through our *LGMD News* magazine and by visiting **ClinicalTrials.gov** for the latest trials published. I also highly recommend the GRASP LGMD consortium and finding a neurologist within this network, as they are on the cutting edge of all potential treatments.

Also, stay tuned for an important announcement from The Speak Foundation regarding a new program for health equity grants coming in early 2024 in conjunction with high-performing LGMD Centers.

If you have any questions, please reach out to us at **ContactUs@TheSpeakFoundation.com**.

Kathryn Bryant Knudson ★ Conference Administrator

WE WANT TO THANK OUR INCREDIBLE SPONSORS!

2023 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE
WASHINGTON, D.C.

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

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International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

CONNECTING FOR A CURE

CURELGMD2i
Limb Girdle Muscular Dystrophy

Join us Sept. 30 at one of our **Connecting for a Cure** events in FL, PA, CA, or host your own. Learn more:

CURELGMD2i.org

and follow us on

LGMD News

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Thank you for your support!

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

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

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CONFERENCE SCHEDULE

2023 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE
WASHINGTON, D.C.

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

FRIDAY ★ OCTOBER 27, 2023

4:00 – 6:30 PM	**General Registration — Check In**		Constitution Ballroom Foyer
5:00 – 6:00 PM	**Exhibitor Setup**		Constitution Ballroom Foyer
6:30 – 8:00 PM	**Welcome Reception & Mixer**		Constitution Ballroom Foyer

SATURDAY ★ OCTOBER 28, 2023

8:00 – 11:00 AM	**General Registration — Late Check In**		Constitution Ballroom Foyer
9:00 – 10:30 AM	**Plenary Session I:**	**Overview of LGMDs and Natural History Studies:** **Conference Welcome** • Kathryn Bryant Knudson, President, The Speak Foundation **LGMDs: Overview and Progress in Developing Treatments** • Nicholas E. Johnson, MD, Virginia Commonwealth University **How Natural History Studies Enable Clinical Trials** • Meredith James, PhD, University of Newcastle, UK **Current LGMD Natural History Studies** • Linda Lowes, PhD, Nationwide Children's Hospital	Constitution Ballroom
10:30 – 10:45 AM	**Break**		Constitution Ballroom Foyer
10:45 – 11:45 AM	**Plenary Session II:**	**LGMD Diagnosis and Clinical Care:** **The NIH Diagnostic Program for Neuromuscular Diseases** • Carsten Bonnemann, MD, National Institutes of Health **Clarifying the Genetics of LGMDs** • Conrad (Chris) Weihl, MD, PhD, Washington University School of Medicine **Developing Standards of Care for LGMDs** • Volker Straub, MD, PhD, University of Newcastle, UK	Constitution Ballroom
11:45 AM – 12:55 PM	**Lunch & Learn**	**Fortify – Launch of a Phase 3 Study and What We've Learned So Far about BBP-418 for LGMD2i/R9** Presented by Platinum Sponsor ML BioSolutions	Constitution Ballroom

Grand Hyatt — Washington, D.C. ★ All Conference Session Times are in Eastern Daylight Time (EDT), UTC-4 ★ Conference schedule is subject to change

The International Limb Girdle Muscular Dystrophy Conference is a project of the Speak Foundation, a 501 (c)(3) tax-exempt public charity since 2008. Learn more at: TheSpeakFoundation.com

CONFERENCE SCHEDULE

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

SATURDAY ★ OCTOBER 28, 2023 — *Continued*

12:55 – 1:55 PM	**Breakout Session 1:**	**Managing Respiratory & Cardiac Involvement in LGMDs** • Noah Lechtzin, MD, Johns Hopkins • Amy Fulford, RRT, Millennium Respiratory Services • Kan Hor, MD, Nationwide Children's Hospital	**Constitution Ballroom**
		Ask the Expert: Q&A • All Speakers from Plenary Sessions I & II	**Cabin John / Arlington**
		Family and Parenting as a Person with LGMD • Chris Carroll, Jessica Evans, PsyD, Yumi Shim	**Wilson / Roosevelt**
		Building Community Connections: Children and Teens • Hosts: Sammi Brazzo, Brooklyn Garza	**Renwick / Bulfinch**
1:55 – 2:05 PM	**Break**		**Constitution Ballroom Foyer**
2:05 – 3:35 PM	**Plenary Session III:**	**Advocacy Initiatives in LGMD:** **Overview of Advocacy for Rare Diseases** • Annie Kennedy, Everylife Foundation **Collaborating for Advocacy** • Paul Melmeyer, Muscular Dystrophy Association **Muscular Dystrophy Coordinating Committee & NIH Support for Research** • Glen Nuckolls, PhD, National Institutes of Health **Community Interactions with FDA** • Jennifer Levy, PhD, Coalition to Cure Calpain 3 • Brad Williams, PhD, Jain Foundation	**Constitution Ballroom**
3:35 – 3:45 PM	**Break**		**Constitution Ballroom Foyer**
3:45 – 4:45 PM	**Breakout Session 2:**	**Building Community Connections: 20's - 30's - 40's** • Hosts: Andrea Lane, Julianna Rodrigues	**Cabin John / Arlington**
		Living a Fulfilling Life with LGMD • Melissa Grove MS, LPC	**Wilson / Roosevelt**
		Building Community Connections: Adults 50's+ • Hosts: Carol Abraham, Pat Moeschen	**Renwick / Bulfinch**
4:45 – 6:00 PM	**Break**		

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2023 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE
WASHINGTON, D.C.

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

SATURDAY ★ OCTOBER 28, 2023 — *Continued*

6:00 – 8:30 PM	**Banquet Dinner**	**Welcome Back:** • Kathryn Bryant Knudson, The Speak Foundation **Videos: Youth Living with LGMD** **Panel Discussion: Individuals Living with LGMD** **Keynote Address:** **FDA Fireside Chat with Peter Marks, MD, PhD, CBER, FDA** • Hosts: Kathryn Bryant Knudson & Brad Williams, PhD **LGMD Documentary from Proyecto Alpha: *Nunca Dejes de Moverte (Never Stop Moving)***	**Constitution Ballroom**

FREE Saturday Stream
Keynote Address: "FDA Fireside Chat with Peter Marks, MD, PhD, CBER, FDA"
VirtualEventPortal.com/Videos/LGMDKeynoteSpeaker

SUNDAY ★ OCTOBER 29, 2023

10:00 – 10:50 AM	**Breakout Session 3:**	**Making Gene Therapy More Versatile:** **Applying Gene Therapy to LGMDs** • Matthew P. Wicklund, MD, University of Texas in San Antonio **Bespoke Gene Therapy Initiative** • Phillip (PJ) Brooks, PhD, National Institutes of Health **What Will Gene Therapy 2.0 Look Like?** • Sharon Hesterlee, PhD, Muscular Dystrophy Association	**Constitution Ballroom**
		Building Community Connections: Parents of Children with LGMD • Hosts: Faran Day, Rachel DeConti	**Cabin John / Arlington**
		Enhancing your Clinical Care Experience • Vovanti Jones, MD, University of Missouri	**Wilson / Roosevelt**
10:50 – 11:00 AM	**Break**		**Constitution Ballroom Foyer**

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CONFERENCE SCHEDULE

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

SUNDAY ★ OCTOBER 29, 2023 — *Continued*

11:00 AM – 12:10 PM	**Plenary Session IV:**	**Clinical Trial Updates (Part 1):** **ML BioSolutions: Update on BBP-418 clinical studies for LGMD 2i/R9** • Doug Sproule, MD, ML BioSolutions **Sarepta Therapeutics: LGMD Program Update** • Louise Rodino-Klapac, PhD, Sarepta Therapeutics **Preliminary Experiences with Atamyo's FKRP Gene Therapy** • John Vissing, MD, University of Copenhagen	**Constitution Ballroom**
12:10 – 1:10 PM	**Lunch & Learn**	**Relax, Recharge, and Reconnect Patient Networking Lunch** **Presented by Platinum Sponsor Sarepta Therapeutics**	**Constitution Ballroom**
1:10 – 1:50 PM	**Plenary Session V:**	**Clinical Trial Updates (Part 2):** **An Update on AskBio's LION-101 Study for the Treatment of LGMD 2i/R9** • Anh Nguyen, MD, AskBio **EDG-5506: A Novel Approach to Protecting Muscle in DMD, BMD, and LGMDs** • Joanne Donovan, MD, PhD, Edgewise Therapeutics	**Constitution Ballroom**
1:50 – 2:00 PM	**Break**		**Constitution Ballroom Foyer**
2:00 – 3:00 PM	**Plenary Session VI:**	**Development of New Treatments and New Assessments** **Developing Cell Therapy for Muscular Dystrophy** • Peter Kang, MD, University of Minnesota **Chaperone Compounds for Rescue of Missense Sarcoglycan Mutations** • Dorianna Sandonà, PhD, University of Padova **Regenerative Medicine in LGMD2A/R1. VTA-100: A Novel Therapeutic Cell-based Approach** • Michael Molyneaux, MD, Vita Therapeutics **Therapeutic Approaches for Dominantly Inherited LGMDs** • Andrew Findlay, MD, Washington University	**Constitution Ballroom**
3:00 PM	**Closing Remarks**	• Kathryn Bryant Knudson, The Speak Foundation	**Constitution Ballroom**
3:05 PM	**Adjourn**		

Grand Hyatt — Washington, D.C. ★ All Conference Session Times are in Eastern Daylight Time (EDT), UTC-4 ★ Conference schedule is subject to change

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SPONSORED CONTENT

Gene Therapy 101

Produced by
Sarepta Therapeutics

Sarepta Therapeutics is a biotech company headquartered in Cambridge, Massachusetts. We have over 20 gene therapy programs in development, including for limb-girdle muscular dystrophy types 2E/R4, 2D/R3, 2C/R5, 2B/R2, 2L/R12, and 2A/R1.

What is assessed in gene therapy clinical trials?

3 primary categories may include :

Safety

First is safety. During a clinical trial, participants are closely monitored. Doctors look for and record side effects – which may also be called adverse events. This helps protect the overall health and well-being of the participants and may also help researchers potentially adjust treatment plans for future clinical trials.

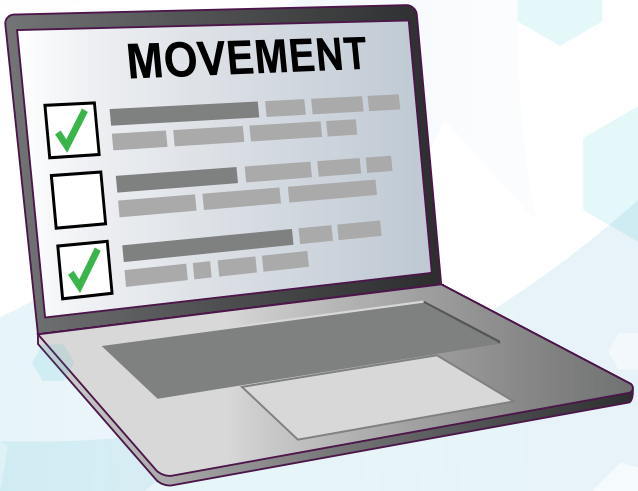
Participants in gene therapy clinical trials are also monitored long-term—often for years. This provides information on the long-term effects of gene therapy.

If you are considering gene therapy for yourself or a loved one, speaking with your doctor is the best way to learn more.

Intended for US audiences

CHAPTER 4

OF A FOUR-PART SERIES



Protein Expression

Next is protein expression. Gene therapy is designed to produce a new protein inside disease-impacted cells. Expression is the measurement of how much protein has been produced in a collection of cells. Knowing whether the disease-impacted cells are producing the intended protein helps researchers understand if the gene therapy is working as intended.

Impact on Disease

Finally, remember that the goal of gene therapy is to slow or stabilize the disease. Therefore, researchers may ask clinical trial participants to perform tests before and after the treatment to help determine any impact on their disease. For gene therapies that aim to address muscular dystrophy, this could include tests examining a person's muscle movement and function.

CHAPTER 1 CHAPTER 2 CHAPTER 3

Sarepta is committed to providing gene therapy educational resources to rare disease communities

Check out more GT 101 in the previous 2023 issues of the *LGMD News Magazine* or email Advocacy@Sarepta.com

SAREPTA THERAPEUTICS

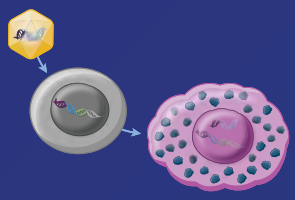
SPONSORED CONTENT

Gene Therapy 101

is a four-part educational series featured in the 2023 issues of the *LGMD News Magazine*.

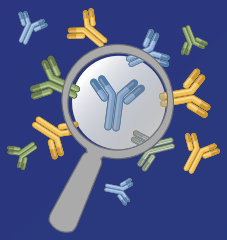
Throughout this series, we have explored common questions and areas of interest regarding investigational gene therapy research and development.

What have we covered in Chapters 1-3?



Chapter 1

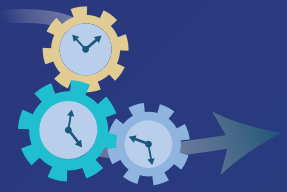
Investigational gene therapy's goal is to slow or stabilize disease by delivering the right instructions to cells to make a new protein.



Chapter 2

Gene therapy has unique features that may affect treatment eligibility. Two tests to determine eligibility are:

- Genetic diagnosis of the disease, including subtype
- Vector antibody testing to confirm that the body doesn't have elevated antibodies that could prevent the gene therapy from working as intended



Chapter 3

Gene therapy development takes place over the course of years, with significant investment required to manufacture the product once it's been designed.

If you are considering gene therapy for yourself or a loved one, speaking with your doctor is the best way to learn more.

Intended for US audiences

How can the LGMD community help further research?

There are many ways to be involved. A few suggestions are detailed below.

Connect with a Neuromuscular Specialist

Understand your LGMD subtype with a Genetic Test

and get your genetic test report which may help inform your care plan

Participate in Registries, Natural History Studies, and Surveys

which can provide information that may help research in rare diseases

Ask your Doctor about Potential Clinical Trials

for your subtype. You can also search for trials on clinicaltrials.gov

Engage with Patient Advocacy Organizations

to access community news, support, education, and events

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SAREPTA THERAPEUTICS

TOGETHER WE CAN DO EVERYTHING:

We Are One!

Dear members with LGMD: know that you are never alone. We are here for you. We listen, advise, and stand up for those without a voice. Every day we get closer to a cure. Until then, we are one family. **Please send us your experiences, worries, and hopes!**

Hosting the LGMD CAB and representing all LGMD subtypes

StichtingSpierKracht.com

Stichtingspierkracht@ziggo.nl

Facebook.com/stichtingspierkracht

Instagram.com/stichting_spierkracht

Representing the LGMD R5/2C, LGMD R3/2D, LGMD R4/2E, and LGMD R6/2F communities

ProyectoAlpha.org

info@ProyectoAlpha.org

Facebook.com/asociacion.proyectoalpha

Instagram.com/proyectoalpha_

Charity for those living with muscle-weakening and wasting conditions in the UK

MuscularDystrophyUK.org

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Representing all types of muscular dystrophy

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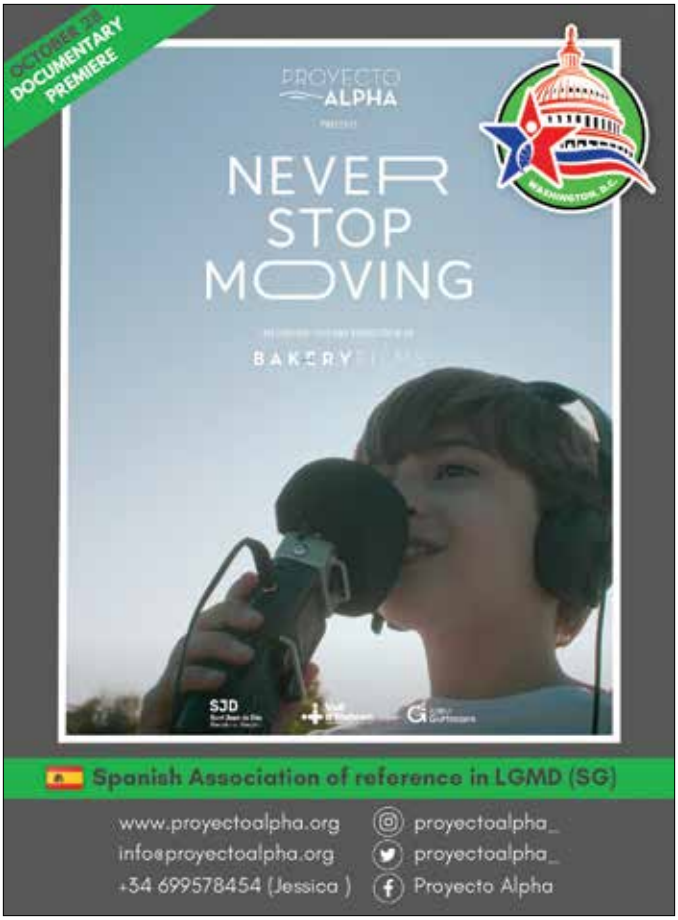

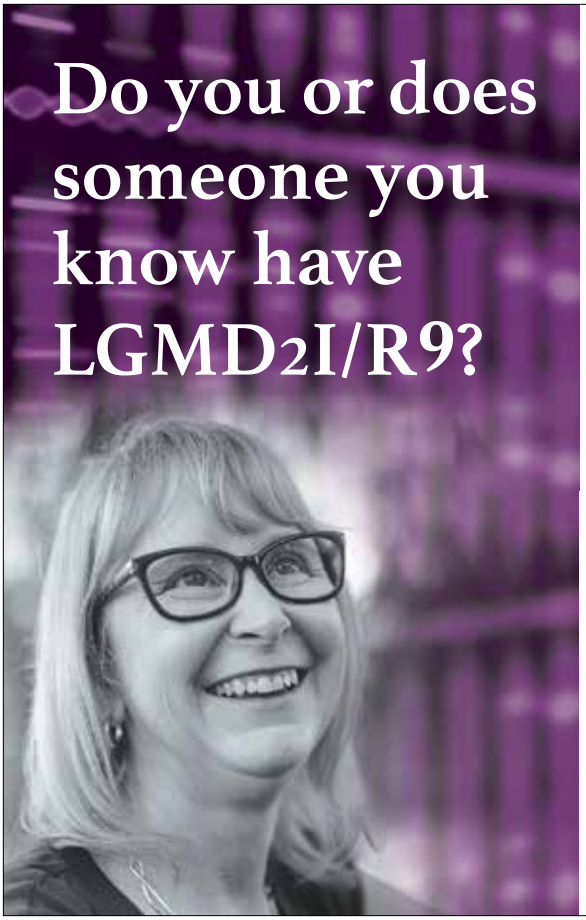

AskBio will be conducting a clinical study of an investigational gene therapy for individuals with a confirmed genetic diagnosis of LGMD2I/R9.

- This is a one-time intravenous infusion of gene therapy designed to produce fukutin-related protein (FKRP) in the body, primarily in muscle.
- Part 1 of the study will assess the safety of LION-101 only in adults (ages 18 and 65 years).
- This is a randomized, placebo controlled, double-blind study.
- The study is designed to investigate at least two different doses of LION-101 versus placebo.
- The initial phase of this first-in-human dose-finding study will be conducted in the US.
- Travel to study sites may be reimbursed; local and home-based testing will be used when possible.
- Information on the clinical trial can be found on clinicaltrials.gov.

To learn more, please visit AskBio.com, email AskFirst@AskBio.com or go to clinicaltrials.gov (NCT05230459)

AskBio™

SPONSORED CONTENT

Connecting People. Providing Answers.

The only way to bring about change is to unify those who are driven for a cure.

At the LGMD2L Foundation, our goal is to unite people afflicted by this rare disease. We provide an open forum to connect, discuss, and ultimately build a community of LGMD2L members. Although our disease is rare and poorly understood, we have established our foundation to build a database of LGMD2L patients for future scientific research and clinical trials.

LGMD2L Foundation
Connecting People. Providing Answers.

LGMD2LFoundation.org

A History of Hope

Celebrating some of the Achievements of the LGMD Community* in Advancing Science and Quality of Life

Launch of The Speak Foundation

The SPEAK Foundation

2008 – 2018 | The Speak Foundation sponsored yearly MD & LGMD conferences in Atlanta

Launch of CureLGMD2i Foundation (originally known as the Samantha J. Brazzo Foundation)

2011

2008

LGMD2iFund
Limb-Girdle Muscular Dystrophy 2I Research Fund

2013

GLOBAL FKRP REGISTRY

LGMD AWARENESS FOUNDATION

*ML Bio Solutions is grateful for the partnership of the featured advocacy organizations, which include The Speak Foundation, CureLGMD2i Foundation, LGMD Awareness Foundation and the LGMD2i Research Fund.

ML Bio Solutions

LIMB GIRDLE MUSCULAR DYSTROPHY

LGMD Awareness Day September 30th

LGMD Awareness Foundation (LAF) website launched the "Information Hub for the LGMD community" (LGMD-Info.org)

2014

2015

Lime Green for LGMD Awareness campaign (LAF)

2016

The Speak Foundation Personal Care Attendant (PCA) stipend program

For more information visit **MLBioSolutions.com**

Congratulations on these and many more accomplishments...

First Patient Listening Session for LGMD with the FDA (LGMD Coalition)

First International LGMD Conference in Chicago (Speak Foundation)

First Annual "Pioneer in LGMD2i" awarded to Lacey Woods

2020

Connecting and Reaching Everyone
C.A.R.E.
A Program of The SPEAK Foundation

The C.A.R.E. Program
Provides newly diagnosed patients with LGMD with a giftbox full of helpful tools donated by the International Consortium of LGMD Organizations

2018

Girdie, the LAF LGMD Ambassador was introduced to the LGMD community

2021

The HOPE Project
A Program of The SPEAK Foundation

The HOPE Project
Awards a stipend to qualified applicants living in the United States who have a diagnosis of LGMD and financial need for durable medical equipment (DME)

ML Bio Solutions

...and **cheers to all those to come!**

Girdie debuted at the Wellstone Dystroglycanopathies Patient & Family conference in Iowa (pictured: Rich Yates, the designer of Girdie)

CONNECTING FOR A CURE

CURELGMD2i

2022 LGMD GLOBAL ADVOCACY SUMMIT
LIMB GIRDLE MUSCULAR DYSTROPHY

CureLGMD2i surpasses $1,000,000 in funding awareness, advocacy, scientific research and drug development programs

2023

2022

LGMD EL-PFDD (LGMD Coalition)

LAF LGMD Advocacy Bundle, information packets for newly diagnosed LGMD patients, pilot program launched in 6 clinics

New 'Voice of the Patient' Report Highlights Unmet Medical Needs of Six Subtypes of Limb-Girdle Muscular Dystrophy (LGMD Coalition)

For more information visit **MLBioSolutions.com**

The Top 3 Reasons to Join the New

LGMD2A/CALPAINOPATHY REGISTRY
A C3 INITIATIVE

1. To attract the attention of scientists and share with them how this disease impacts your life
2. To learn about opportunities to participate in research, including clinical trials
3. To help C3 inform and drive medical research towards a cure

The Coalition to Cure Calpain 3 team looks forward to meeting you at the International LGMD Conference in DC

Website: LGMD2A.iamrare.org
Email: Registry@CureCalpain3.org

---

www.lgmd2e.org

GRUPPO FAMILIARI BETA-SARCOGLICANOPATIE
GFB ONLUS

2013-2023
10 YEARS OF RESEARCH

In 2023 GFB celebrates its first 10 years of scientific research activities, with two major ongoing research projects funded entirely by GFB:

IPSCs generation, characterization and differentiation for LGMDR4 patients.

Yvan Torrente, University of Milan, Italy, Giulio Pompilio, Centro cardiologico Monzino, Italy, Cerletti Massimiliano, UCL UK, Carles Sanchez Riera, University La Sapienza Roma, François Gros-Louis, Centre de Recherche CHU de Quebec, Université Laval, Canada.

Observational study: the quality of life in patients with Alpha-sarcoglycan, Beta-sarcoglycan and Gamma-sarcoglycan gene mutations.

Prof. Yvan Torrente, University of Milan, Italy, Dr. Carles Sanchez Riera, University La Sapienza Roma.

Recruitment is still open, contact segreteria@beta-sarcoglicanopatie.it

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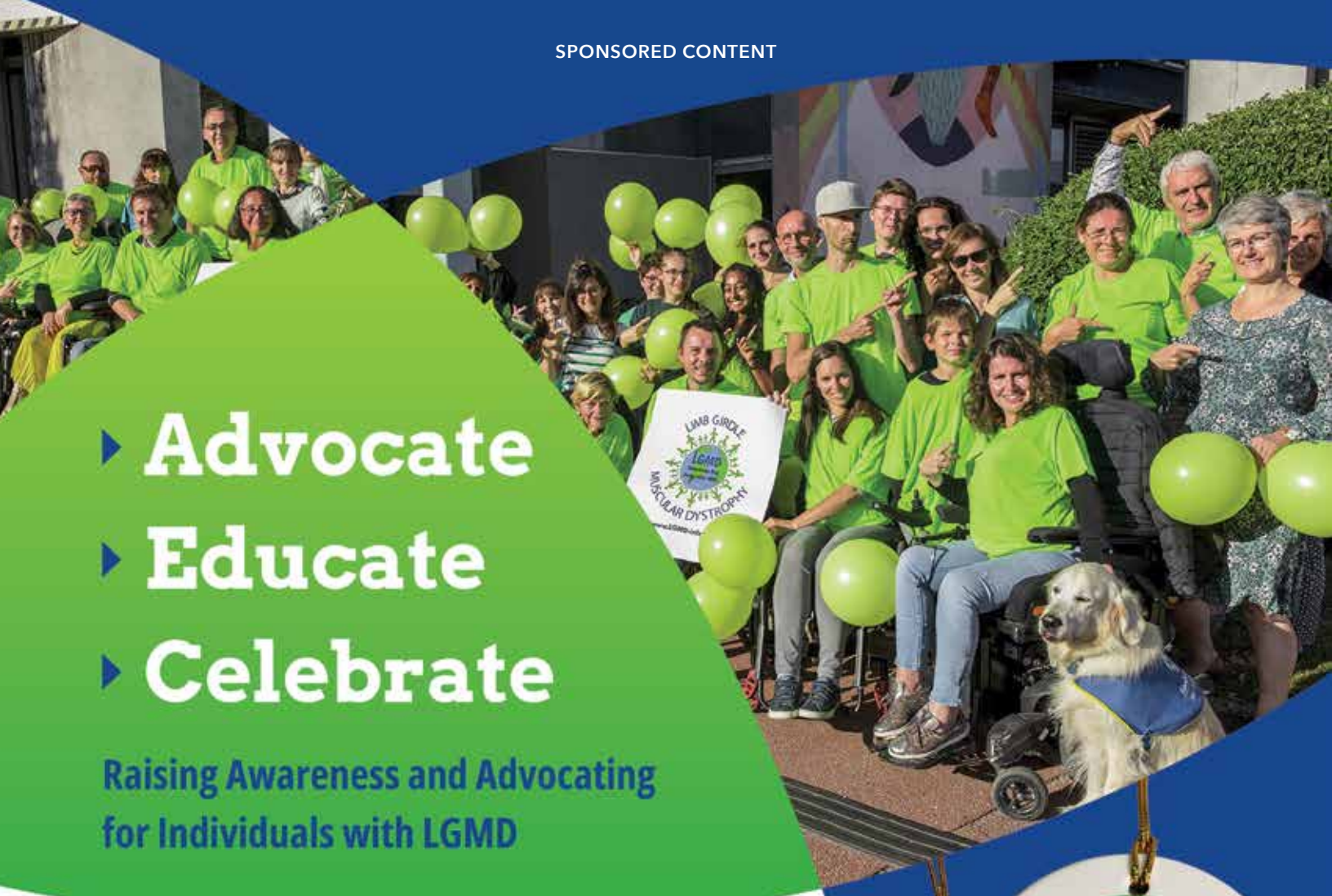
LGMD-1D DNAJB6 Foundation

Foundation Assisted Genetic Testing: **lgmd1d.org**



Advocate
Educate
Celebrate

Raising Awareness and Advocating for Individuals with LGMD

Whether you are a long-time community member, or new to the scene, you are not alone on your LGMD journey. We are the informational hub for the LGMD community and we truly believe that ***Together We Are Stronger.***

Visit lgmd-info.org today to connect with the LGMD community and access resources and support.

2023 Girdie Limelight Collectible

Come see us at the

International LGMD Conference
Washington, DC
October 27-29

Collectibles will be available at our booth!

LGMD AWARENESS FOUNDATION

lgmd-info.org

Sarepta is the proud sponsor of

LimbGirdle.com

a U.S. educational website.

Members of the U.S. community can sign up at **limbgirdle.com/stay-connected** to receive information on community resources, news, and research on limb-girdle muscular dystrophy.

U.S. community members may also choose to follow Sarepta on our social media platforms (Facebook, LinkedIn, Instagram, Twitter).

If you wish to speak directly to a member of the Sarepta Patient Affairs team and share about yourself and hear about community resources, we encourage members of the U.S. and international communities to connect with us by emailing **Advocacy@Sarepta.com**.

Sarepta is advancing the science behind limb-girdle muscular dystrophy research.

Sarepta Therapeutics is a global biotechnology company on an urgent mission to engineer precision genetic medicine to reclaim futures otherwise impacted or cut short by rare diseases, including limb-girdle muscular dystrophy (LGMD).

Sarepta currently has 6 LGMD development programs: sarcoglycanopathies (LGMD2C/R5, LGMD2D/R3, LGMD2E/R4), dysferlin (LGMD2B/R2), anoctamin-5 (LGMD2L/R12), and calpain-3 (LGMD2A/R1).

Sarepta will be conducting non-interventional research studies over the coming months

Overview

Non-interventional (non-treatment) research studies, such as interviews and surveys, provide an opportunity for people living with LGMD to share their unique experiences and perspectives.

When you participate in non-interventional studies, like interviews and surveys, you can help propel understanding of the impact of LGMD on daily living and help researchers understand what matters to people living with LGMD.

Your input can help healthcare professionals and researchers:

Gain a deeper understanding of LGMD
from your perspective as someone living with LGMD. This can help researchers develop potential therapies to address unmet medical needs

Advance drug development:
Researchers and regulators often rely on patient-reported data to make decisions regarding drug development

Evaluate the effectiveness of current disease management
and assess the impact of interventions. This information can help enable better patient-centric care

Visit **limbgirdle.com/stay-connected** to receive updates

Please note: *Participation in these types of studies will not impact your ability to enroll in future clinical trials for potential new treatments.*

If you are eligible and participate in a non-interventional research study, you will be compensated for your time.

SAREPTA THERAPEUTICS

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SPONSORED CONTENT

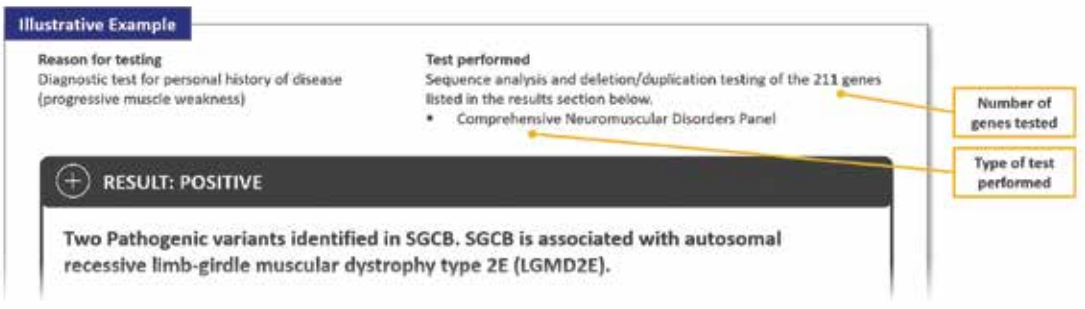
Do you have your genetic test report?

You are an important member of your care team.

While all subtypes of LGMD share some common features, each has unique characteristics that may impact your care team composition and clinical management decisions.

Some clinical trials may require a genetic diagnosis, or may only be available for people with specific subtypes of LGMD caused by a variant(s) in a certain gene. Therefore, if you have not had a genetic test, or your genetic test was done years ago, it may be time to pursue genetic testing.

What information can you learn from a genetic test report?



Possible results from a genetic test:

Positive	Uncertain	Negative
2 Pathogenic or Likely Pathogenic variants in the same gene associated with an autosomal recessive LGMD subtype	1 Pathogenic or Likely Pathogenic Variant and 1 Variant of Uncertain Significance (VUS) identified in the same gene, OR >1 VUS detected in the same gene*	No Pathogenic, Likely Pathogenic, or VUS detected in any of the tested genes
▸ This is a definitive diagnosis of LGMD and your LGMD subtype can be determined	▸ This is not a definitive diagnosis. Discuss next steps with a healthcare provider, such as variant reclassification or family testing, and ask questions to clarify results	▸ The test did not identify any of the subtypes of LGMD caused by genes that were screened for in the specific genetic test. To see which genes were screened for, visit the laboratory website and search for the name of the genetic test on the report

- Uncertain results do not offer a clear and final diagnosis, and additional efforts may be undertaken to clarify the diagnosis
- Negative results do not rule out the possibility of LGMD, if not all LGMD genes were tested or if the subtype has not been discovered yet
- Always discuss questions and results with a healthcare provider

*Some LGMD subtypes are characterized by a dominant inheritance pattern and require only one Pathogenic or Likely Pathogenic variant to cause disease. However, these subtypes are more rare.

What are My Next Steps?

If you do not have a **copy of your genetic test report**, contact your doctor and ask for a copy.

If your result is negative or uncertain, ask your healthcare provider (neurologist or genetic counselor) if getting a new test or reclassification could be an option for you.

SPONSORED CONTENT

Did you know? New ICD-10 codes for limb-girdle muscular dystrophy (LGMD) became available for use by your doctor in October 2022.

What are ICD-10 codes?

ICD-10 refers to the 10th edition of the International Classification of Diseases (ICD), a medical classification list that is used worldwide. The ICD provides a standardized method for classifying and coding health conditions and diseases, with codes representing diagnoses, symptoms, and procedures

ICD codes are generally highly specific. There are over 68,000 codes!

Why is using the accurate ICD-10 code important for LGMD?

Accurately recording a patient's LGMD subtype is important to the development of new potential therapies and provides meaningful de-identified data to drug developers. Use of specific ICD-10 codes will support clinical and research communities in ongoing efforts to:

- Understand LGMD epidemiology
- Assess natural history/disease progression of the condition
- Understand economic burden (i.e., healthcare costs) of LGMD
- Help manage care of patients
- Facilitate reimbursement and patient access when targeted therapies potentially become available in the future

What can you do with this information?

At your next visit, speak with your physician about LGMD subtype-specific ICD-10 codes to help ensure that your diagnosis is coded properly.

To Learn More

Visit Sarepta's booth at the conference to pick up an ICD-10 code wallet card

Visit limbgirdle.com/ICD-10

Team Titin
Strengthening the Titin Community

Variants in the TTN (titin) gene can cause a spectrum of muscle and heart disorders including LGMD R10 Titin-related, formerly known as LGMD2J.

Team Titin, Inc.'s mission is to serve those living with, caring for, or researching titin (TTN) related muscle and heart disorders.

visit TitinMyopathy.com to learn more

@teamtitin

facebook.com /TeamTitin

DYSFERLIN REGISTRY

by Jain Foundation

GENETICALLY CONFIRMED REGISTRY MEMBERS WORLDWIDE **1145**

61 **COUNTRIES REPRESENTED IN THE DYSFERLIN REGISTRY**

INDIVIDUALS WITH DYSFERLINOPATHY APPLY ONLINE

VITA Therapeutics

Harnessing the Power of Genetics

Vita Therapeutics is a cell engineering company harnessing the power of genetics to replace defective muscle cells. The company utilizes induced pluripotent stem cell (iPSC) technology to engineer specific cell types designed to replace those that are defective in patients.

The first target is limb-girdle muscular dystrophy 2A/RI, caused by a defective gene for the protein Calpain-3. The lead asset, VTA-100, is an autologous cell therapy using gene editing technology to replace the defective gene which is drawn from the blood of a patient. The goal is to repair and replace defective muscle tissue in affected patients, regenerating healthy muscle. The second target is FSHD with the development of VTA-120 aiming to replace cells producing destructive DUX4 protein.

VTA-100 is currently in the pre-clinical stages and working towards initiating our First-in-Human IND trial. The trial will enroll 9 patients with each patient serving as their own control, and endpoints including cellular, imaging and functional testing. We anticipate prescreening of patients in early 2024.

For more information on Vita's clinical programs, please inquire to clinicaltrials@vita-therapeutics.com

Vitatx.com

clinicaltrials@vita-therapeutics.com

SPONSORED CONTENT

LGMD2D Foundation

Limb-Girdle Muscular Dystrophy

CELEBRATING 10 YEARS OF ADVOCATING FOR LGMD2D/R3.

Founded in 2013, the LGMD2D Foundation was built for families living with LGMD type 2D/R3, by families with the same diagnosis. Ten years later, we are still driven by our mission to expedite the development of treatments or a cure for LGMD2D.

The LGMD2D Foundation continues to:

- **Maintain a global patient registry.**
- **Monitor progress of natural history studies & clinical trials.**
- **Provide financial support to accelerate research for treatments.**
- **Educate patients, researchers & physicians on LGMD2D/R3.**
- **Encourage scientific collaboration & partnerships.**

FOR MORE INFORMATION VISIT:

Follow us:

The LGMD2D Foundation is a proud Advocacy Sponsor of the 2023 International LGMD Conference.

fortify

Fortify is a Phase 3 clinical trial evaluating if an investigational oral therapy (BBP-418) is safe and effective for treating Limb Girdle Muscular Dystrophy type 2I / R9, FKRP-related (LGMD2I/R9)

About Fortify

Fortify is a 36 month randomized, double-blind, placebo-controlled trial for individuals with genetically confirmed LGMD2I/R9 measuring patient response to treatment by measuring both biomarkers and clinical assessments. For every three study participants, two will receive BBP-418 and one will receive placebo.

About the Therapy

In patients with LGMD2I, the enzyme FKRP does not work properly. FKRP is responsible for a critical step in a process called "glycosylation", whereby a crucial string of sugars are added to alpha dystroglycan (α-DG). Without this string, α-DG does not work correctly in its role as a "shock absorber" for muscle fibers. BBP-418's theoretical mechanism of action supplements the FKRP enzyme by adding more of the molecule that FKRP normally reacts with to drive residual activity of FKRP and helping it to stabilize muscle cells and act as a shock absorber. BBP-418 is an investigational therapy and is not yet approved by any health authorities for the treatment of LGMD2I/R9.

Who Can Participate

You may be eligible to participate in Fortify if you:

- Have a genetically confirmed diagnosis of LGMD2I/R9
- Are 12 to 60 years of age
- Have not used ribose or systemic corticosteroids prescribed for the treatment of LGMD or other investigational therapies for the treatment of LGMD within 90 days of screening

There are other requirements to participate in Fortify. A physician or study team member will help determine if you are eligible to participate and if this study is a good fit for you. Speak with your physician about your ability to participate in Fortify.

Fortify Locations:

The trial will be conducted at clinical sites in the United States and Europe.

Additional information about our BBP-418 study is available at **www.clinicaltrials.gov** and at **www.mlbsolutions.com**.

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Scan to visit our website

ML Bio Solutions

ClinicalTrials.gov Identifier:
NCT05775848

Fortify
Trial Locations

Scan here for more information:

United States

1. University of California Irvine — *California*
2. University of Colorado Anschutz Medical Campus — *Colorado*
3. University of Florida — *Florida*
4. University of Iowa — *Iowa*
5. University of Kansas Medical Center — *Kansas*
6. Kennedy Krieger Institute — *Maryland*
7. University of Minnesota, Twin Cities — *Minnesota*
8. Washington University School of Medicine — *Missouri*
9. Oregon Health & Science University — *Oregon*
10. Penn State — *Pennsylvania*
11. The Children's Hospital of Philadelphia — *Pennsylvania*
12. University of Pennsylvania — *Pennsylvania*

Europe

1. Rigshospitalet, Neuromuscular Clinic and Research Unit — *Denmark*
2. University Hospital Essen — *Germany*
3. Ospedale Maggiore Policlinico Milano; U.O. Neurologia — *Italy*
4. Leiden University Medical Center — *Netherlands*
5. Universitetssykehuset Nord-Norge, Department of Neurology — *Norway*
6. Great Ormond Street Hospital for Children — *United Kingdom*
7. International Centre for Life — *United Kingdom*

Fortify Trial locations are accurate as of August 1, 2023, however locations may be subject to change. For the most up to date list of clinical trial locations and for more information about the Fortify trial, please visit clinicaltrials.gov.

fortify

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 R19/2S
CamronsCure.com

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

Cure LGMD2I
Funding research for LGMD R9/2I
CureLGMD2I.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
Beta-Sarcoglicanopathy.org

Gruppo Cingoli of 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

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
ProyectoAlpha.org

ML Bio Solutions
a bridgebio company

we want to know!

Do you or your loved one have a confirmed diagnosis of limb-girdle muscular dystrophy, subtype 2I, also known as LGMD2I/R9, FKRP related? ML Bio Solutions would like to ask people living with LGMD2I/R9 and their caregivers to participate in a survey to:

- Learn what symptoms have the most impact on their lives and to better understand the day-to-day experiences of people living with LGMD2I/R9
- Learn what changes might have the biggest impact on your daily life or quality of life and to better understand what people with LGMD2I/R9 (and families) want to see from new treatments or therapies.
- Learn from you about your experience living with LGMD2I/R9.

Let's get started!
Scan QR Code

This survey should take approximately 15 minutes to complete. Survey participation is completely voluntary, anonymous, and will have no impact on current or future clinical trial eligibility.

We sincerely appreciate your time! Thank you in advance for your participation and contribution.

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

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WASHINGTON, D.C.

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The Speak Foundation thanks all participants of the 2023 International Limb Girdle Muscular Dystrophy Conference, including our corporate and advocacy sponsors.