Vol 1 / Issue 4

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

SPECIAL CONFERENCE EDITION

CONFERENCE PROGRAM INSIDE

The SPEAK Foundation

The 2021 International LGMD Conference is a program and project of The Speak Foundation – *the voice for limb-girdle muscular dystrophy.*

Transforming Medicine. Changing Lives.

The desire to improve the quality of life for patients who are fighting genetic diseases is all the inspiration we've ever needed to find the curative answers that may be close at hand.

For questions or information on our gene therapy technology and clinical programs, email us at **askfirst@askbio.com**.

Proud to Support the Limb-Girdle Muscular Dystrophy Community

AskBio™

askbio.com

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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AFMTÉLÉTHON
CURE THROUGH INNOVATION

AFM-Telethon federates patients who are affected by neuromuscular diseases and their relatives. In order to fight those diseases, AFM-Telethon chose to initiate innovative actions and a strategy of general interest that benefits all rare diseases. Thanks to donations from the Telethon, AFM-Telethon has become **a major player in biomedical research** for rare diseases.

- **3 leading laboratories** in innovative biotherapies gathered within **Rare Diseases Biotherapies Institute** (500 experts): Genethon, Myology Institute, I-Stem
- **40 trials** in man supported for 31 different diseases
- More than **200 programmes** and young researchers funded every year
- An **innovative biotherapies and rare diseases' fund** created with Bpifrance

For more information : www.afm-telethon.com

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

Welcome

To Our 2021 International LGMD Conference!

Conference Administrator
Kathryn Bryant Knudson

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

We are now seeing multiple companies with gene therapy programs in development for LGMD. You do not want to miss any clinical trial that is available to you. Make sure to stay informed through our *LGMD News* magazine and by going to **clinicaltrials.gov** for the latest trials published.

I also recommend the GRASP LGMD consortium and finding a neurologist within that network. They are the best and are on the cutting edge of all potential treatments. If you have any questions, please reach out to us at **ContactUs@TheSpeakFoundation.com**.

Thank You to Our Sponsors:

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MDA Muscular Dystrophy Association

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Conference Schedule

Featured Speakers

Volker Straub, MD
Director, John Walton Muscular Dystrophy Research Centre, Newcastle University, Newcastle upon Tyne, England, UK.

Peter B. Kang, MD
Pediatric Neuromuscular Neurologist and Physician-Scientist, University of Minnesota Medical School, USA.

Tahseen Mozaffar, MD
Professor of Neurology and Laboratory Medicine and Vice Chair for Research, Department of Neurology at University of California, Irvine, USA.

Katherine D. Mathews, MD
Director, Iowa Neuromuscular Program. Professor of Pediatrics-General Neurology, University of Iowa, USA.

Nicholas E. Johnson, MD
Associate Professor and Vice Chair of Research, Virginia Commonwealth University, USA.

Friday | September 17, 2021

All conference session times are in Eastern Daylight Time (EDT), UTC -4. | *Conference schedule is subject to change.*

12:00 – 12:15 PM — **Welcome**
- Kathryn Bryant Knudson | *Conference Administrator*
- Brad Williams, PhD | *Conference Chairman*
- Carol Abraham | *Conference Vice Chairwoman*

12:15 – 12:45 PM — **Overview of Limb-Girdle Muscular Dystrophies**
An overview of what LGMDs are, their genetics, and their symptoms. Included will be an update on what has happened in the limb-girdle community since our last conference in 2019.
- Volker Straub, MD | *Newcastle University, UK*

12:45 – 12:50 PM — **My LGMD Story: Aric Hershley | USA**

12:50 – 1:00 PM — **Break**

1:00 – 2:20 PM — **Updates on Clinical Trials and Emerging Treatments**
An overview of treatments being developed for various LGMD subtypes and clinical trials which are being organized to test them.
- **Moderator:** Jenn Levy, PhD | *Coalition to Cure Calpain 3, USA*

GRASP LGMD Panel Discussion
- Peter B. Kang, MD | *University of Minnesota, USA*
- Tahseen Mozaffar, MD | *University of California, Irvine, USA*
- Katherine D. Mathews, MD | *University of Iowa, USA*
- Nicholas E. Johnson, MD | *Virginia Commonwealth University, USA*

2:20 – 2:30 PM — **Break**

2:30 – 3:10 PM — **Patient-Focused Session: Planning for the Future**
How can a person with LGMD live their best life? People living with LGMD and caregivers share their strategies for navigating different aspects of life.
- **Moderator:** Melissa Grove, MS, LPC | *Legacy Counseling Center, USA*
- Aleksandra Leijenhorst | *Netherlands*
- Jasmine Robinson | *USA*
- Joshua Thayer | *USA*
- Kae Tran | *Canada*

3:10 – 3:15 PM — **Break**

3:15 – 4:00 PM — **Patient-Focused Session: Caregivers – Who, What and How?**
Individuals living with LGMD share their experiences of hiring caregivers to assist with their activities of daily living.
- **Moderator:** Carol Abraham | *Coalition to Cure Calpain 3, USA*
- Mark Barnett | *USA*
- Anne Brastow | *USA*
- Andrew Robertson | *UK*
- Bliss Welch | *USA*

4:00 – 5:00 PM — **Community Reception**

Join a Conference Community Reception Room

Zoom link:
https://us02web.zoom.us/j/86934608410?pwd=bW9Zb3NmU0NsUnNYZnNLZzdGcjJUZz09
Meeting ID: **869 3460 8410**
Passcode: **770112**

Questions? Visit: **www.TheSpeakFoundation.com**

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Conference Schedule

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

Saturday | September 18, 2021

All conference session times are in Eastern Daylight Time (EDT), UTC -4. | *Conference schedule is subject to change.*

11:30 AM – 12:00 PM
Introduction to Gene Therapy (Led by Sarepta Therapeutics)
In this 30-minute presentation, Sarepta will be presenting introductory gene therapy concepts. This will be an interactive presentation with the opportunity for the patient community to assess their current knowledge of gene therapy and expand their knowledge base.
- Allison Kreuzer, PhD | *Director, Patient Affairs, Sarepta Therapeutics, Inc., USA*

12:00 – 1:00 PM
Sarepta Therapeutics LGMD Pipeline Update
In this 60-minute presentation, Sarepta will be sharing an update on their pre-clinical and clinical LGMD pipeline and facilitating a short Q&A with the patient community following their presentation.
- Louise Rodino-Klapac, PhD | *Executive Vice President, Chief Scientific Officer, Sarepta Therapeutics, Inc., USA*

1:00 – 1:30 PM — Break

1:30 – 1:35 PM — My LGMD Story: Tatyana Vyatkina | Russia

1:35 – 1:45 PM — LiftSeat® Demonstration

1:45 – 1:50 PM — "Neurologist of the Year" Award

1:50 – 2:00 PM — Break

Keynote Session
2:00 – 3:00 PM
Regulatory Aspects of Drug Development and the Patient's Role
Former FDA Commissioner (2017-2019) discusses challenges in developing therapies for rare diseases involving new, cutting-edge technologies such as gene and cell therapies, and gene editing. Dr. Gottlieb will also discuss how people living with LGMD can contribute towards the development of treatments.
- **Keynote Speaker:** Scott Gottlieb, MD | *Former FDA Commissioner, USA*

3:00 – 3:45 PM
Patient Interaction with the FDA
Learn how individuals living with rare conditions such as LGMD can communicate their experiences and priorities with the FDA.
- James Valentine, JD, MHS | *Associate, Hyman, Phelps & McNamara, P.C., USA*

3:45 – 3:50 PM
Patient-Focused Drug Development (PFDD) Introduction for Patient Engagement
A Patient-Focused Drug Development (PFDD) meeting with the FDA is being organized for several LGMD subtypes. Discover how the LGMD community can participate.

3:50 – 4:00 PM — Break

4:00 – 5:00 PM
Patient-Focused Session: Maintaining Bone Health, Flexibility & Function
Loss of flexibility and decreased bone density are common complications impacting mobility in LGMD. Clinicians in these areas of practice share strategies for managing these challenges.
- **Moderator:** Carol Abraham | *Coalition to Cure Calpain 3, USA*
- Linda Lowes, PT, PhD | *Nationwide Children's Hospital, USA*
- Meredith James, PT | *Physiotherapist, University of Newcastle, UK*
- Philip Zeitler, MD | *Children's Hospital Colorado, USA*

Featured Speakers

Louise Rodino-Klapac, PhD
Executive Vice President, Chief Scientific Officer, Sarepta Therapeutics, Inc., USA.

Keynote Speaker
Scott Gottlieb, MD
Scott Gottlieb is a physician and served as the 23rd Commissioner of the U.S. Food and Drug Administration. He is a resident fellow at the American Enterprise Institute and a partner at the venture capital firm New Enterprise Associates. Dr. Gottlieb is an aggressive advocate for advancing public health through developing and implementing innovative approaches to improving medical outcomes, reshaping healthcare delivery, and expanding consumer choice and safety. Dr. Gottlieb is widely published in leading medical journals, and is a regular contributor to CNBC *and* Face the Nation. Fortune Magazine *recognized him as one of the "World's 50 Greatest Leaders" in 2018 and 2019.*

James Valentine, JD, MHS
Hyman, Phelps & McNamara, USA.

Linda Lowes, PT, PhD
Principal Investigator, Center for Gene Therapy, Nationwide Children's Hospital, USA.

Meredith James, PT
Clinical Physiotherapist, John Walton Muscular Dystrophy Research Centre, Newcastle, UK.

Philip Zeitler, MD
Professor of Pediatrics, University of Colorado Anschutz Medical Campus, USA.

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International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

Conference Schedule

Featured Speakers

Anh Nguyen, MD
VP, Sector Lead, Musculoskeletal, AskBio, USA.

Sharon Hesterlee, PhD
Chief Research Officer, Muscular Dystrophy Association, USA.

Jorge Bevilacqua, MD, PhD
Professor, Department of Neurology and Neurosurgery, Hospital Clínico Universidad de Chile, Chile.

Conrad (Chris) Weihl, MD, PhD
Professor of Neurology, Washington University School of Medicine, St. Louis, Missouri, USA.

Monkol Lek, PhD
Assistant Professor of Genetics, Yale University School of Medicine, USA.

Sunday | September 19, 2021

All conference session times are in Eastern Daylight Time (EDT), UTC -4. | *Conference schedule is subject to change.*

12:00 – 1:30 PM

Sponsored Session: AskBio

In this 90-minute presentation, AskBio will introduce and give an overview of their patient advocacy program and present the pre-clinical data supporting the clinical advancement of LION-101. They will also offer an inside look from a GRASP LGMD Investigator into LGMD gene therapy trials, and into the perspectives and challenges of living with LGMD2i from a patient family.

- Anh Nguyen, MD | *AskBio, USA*
- Nicholas E. Johnson, MD | *Virginia Commonwealth University, USA*
- Craig Norton | *USA*
- CJ Norton | *USA*
- Sharon Hesterlee, PhD | *Muscular Dystrophy Association, USA*

1:30 – 1:35 PM — **My LGMD Story: Szymczak Family | USA**

1:35 – 1:55 PM — **Break**

1:55 – 2:00 PM — **My LGMD Story: Andrew Robertson | UK**

2:00 – 4:00 PM

Diagnostic Landscape for LGMDs

Correctly diagnosing LGMDs and other conditions causing similar symptoms is often challenging. Clinicians and geneticists describe the diagnostic process, with patients sharing their journeys.

- **Moderator:** Laura Rufibach, PhD | *Jain Foundation, USA*
- Jorge Bevilacqua, MD, PhD | *University of Chile, Chile*
- Conrad (Chris) Weihl, MD, PhD | *Washington University School of Medicine, USA*
- Monkol Lek, PhD | *Yale University School of Medicine, USA*

Patient Panel:

- Mark Barnett | *USA*
- Kyle Harrington | *USA*
- Jenna Keindel | *Canada*
- Andrea Klein | *USA*
- Noni Uribe | *USA*

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Conference Schedule

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

Monday | September 20, 2021

All conference session times are in Eastern Daylight Time (EDT), UTC -4. | *Conference schedule is subject to change.*

11:15 AM – 12:00 PM — **Global LGMD Updates: India, Egypt, and Latin America**

LGMD is a global disease affecting all parts of the world. Leading clinicians from different regions discuss diagnosis, research, and opportunities for advocacy.

- **Moderator:** Sarah Emmons | *Jain Foundation, USA*
- Satish Khadilkar, MD | *Bombay Hospital Institutes of Medicine, India*
- Rasha el Sherif, MD, PhD | *Myocare National Foundation, Egypt*
- Jorge Bevilacqua, MD, PhD | *University of Chile, Chile*

12:00 – 1:30 PM — **Sponsored Session: GFB ONLUS**

In this 90-minute presentation, GFB Onlus will share information about the clinical determinants of disease progression in patients with beta-sarcoglycan gene mutations, a quality of life study for patients with alpha-sarcoglycan, beta-sarcoglycan, and gamma-sarcoglycan mutations, CFTR correctors in sarcoglycanopathies, and unraveling muscle differences to approach sarcoglycanopathies.

- Yvan Torrente, MD, PhD | *University of Milan, Italy*
- Claudia Paniga | *GFB Onlus, Italy*
- Dorianna Sandonà, MD, PhD | *University of Padua, Italy*
- Carles Sánchez Riera, PhD | *Spain*

1:30 – 1:35 PM — **My LGMD Story: Brandi Benton | USA**

1:35 – 1:45 PM — **LiftSeat® Demonstration**

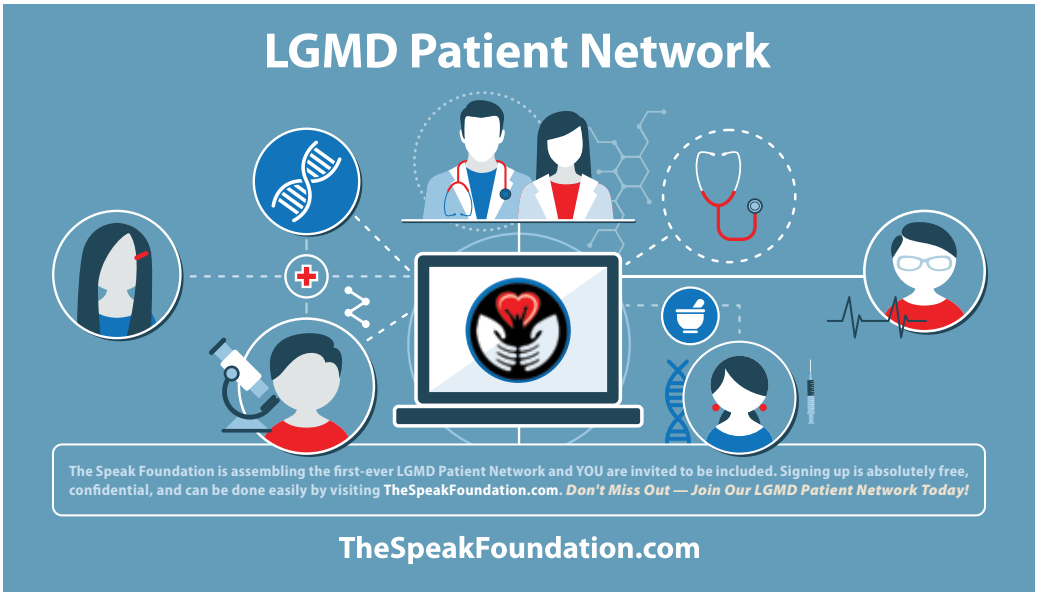
1:45 – 2:00 PM — **Break**

2:00 – 4:00 PM — **Respiratory and Cardiac Complications and the Importance of Screening**

Respiratory and cardiac complications are associated with many forms of LGMD. It is vitally important for individuals with LGMD to receive proper screening and symptom management.

- **Moderator:** Kathryn Bryant Knudson | *Conference Administrator*
- Matthew P. Wicklund, MD | *University of Colorado, USA*
- Oren Kupfer, MD | *University of Colorado, USA*
- Elizabeth McNally, MD, PhD | *Northwestern University, USA*

4:00 – 4:30 PM — **Conference Wrap-Up**



Featured Speakers

Satish Khadilkar, MD
Dean, Medical Faculty of Bombay Hospital Institute of Medical Sciences. Head of the Department of Neurology, BHIMS, Mumbai, India.

Rasha el Sherif, MD, PhD
Founder of Myocare Neuromuscular Foundation. TGDOC LGMD Subgroup Lead in TREAT-NMD, Egypt.

Yvan Torrente, MD, PhD
Professor, Università degli Studi, Milan. Co-Founder, UNISTEM Center, Università degli Studi, Milan, Italy.

Claudia Paniga
Interpreter, GFB Onlus, Italy.

Dorianna Sandonà, MD, PhD
Professor, Biomedical Sciences, University of Padua, Italy.

Carles Sánchez Riera, PhD
Postdoctoral Researcher, University of La Sapienza, Rome, Italy.

Matthew P. Wicklund, MD
Professor of Neurology, University of Colorado, USA.

Oren Kupfer, MD
Pediatric pulmonologist, University of Colorado School of Medicine and Children's Hospital, Colorado, USA.

Elizabeth McNally, MD, PhD
Cardiologist, Center for Genetic Medicine at Northwestern, Chicago, Illinois, USA.

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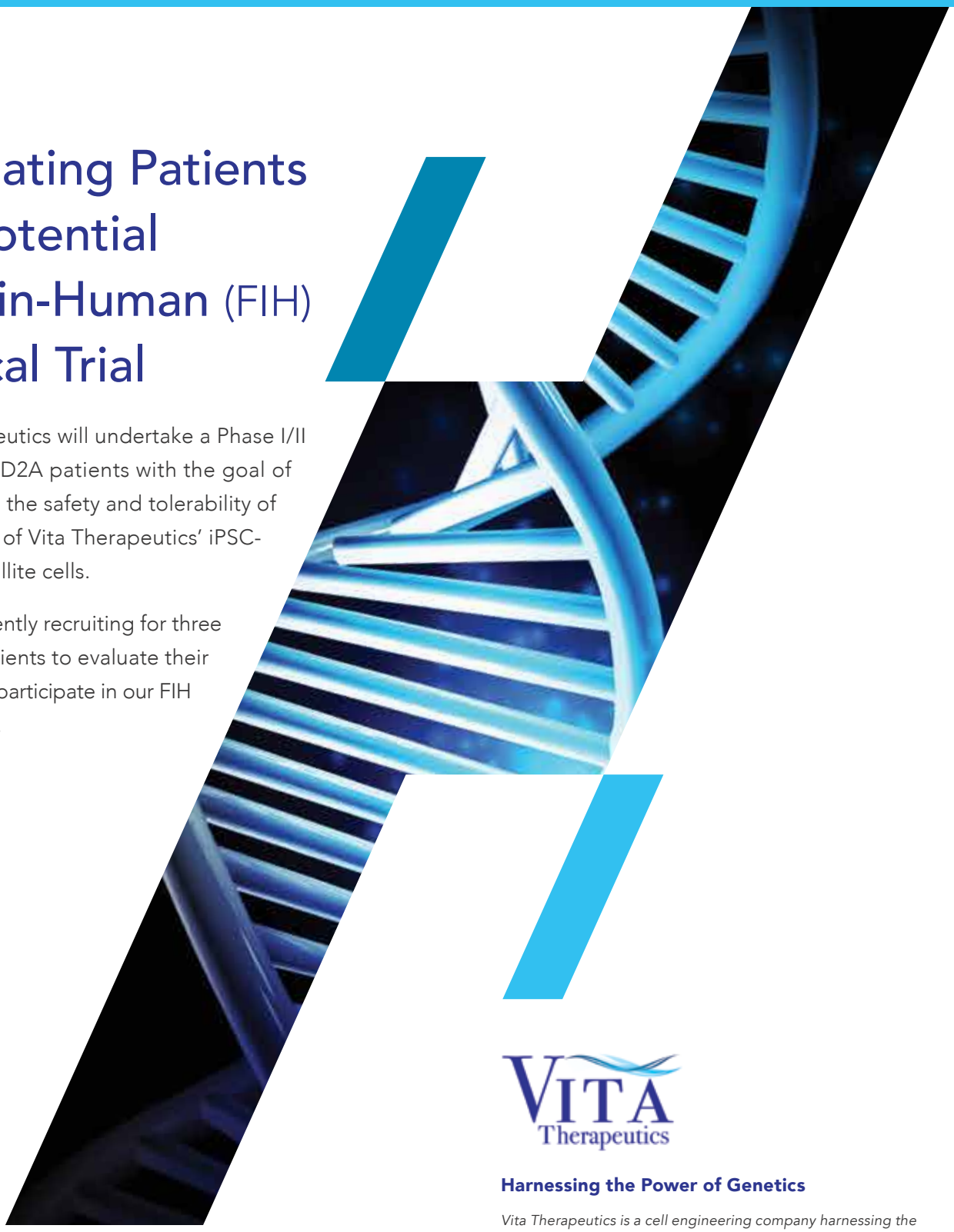
Evaluating Patients for Potential First-in-Human (FIH) Clinical Trial

Vita Therapeutics will undertake a Phase I/II trial of LGMD2A patients with the goal of determining the safety and tolerability of IM injection of Vita Therapeutics' iPSC-derived satellite cells.

We are currently recruiting for three (3) initial patients to evaluate their potential to participate in our FIH Clinical Trial.



Learn More About Our Planned LGMD2A/RI Clinical Trial:

Vitatx.com

clinicaltrials@vita-therapeutics.com

VITA Therapeutics

Harnessing the Power of Genetics

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

The lead asset, VTA-100, is an autologous cell therapy designed to repair and regenerate healthy muscle in patients with limb-girdle muscular dystrophy 2A/R1. VTA-100 is currently in the pre-clinical stages and working towards initiating IND-enabling studies with the goal to file an IND in the second half of 2022.

AMRA
PRECISION FOR DECISION

Shed Light Through MRI

Whole body, whole muscles - precisely the answer

All bodies are unique. Limb-girdle muscular dystrophy (LGMD) presents differently among individuals living with this disorder. As you know, this genetic disease is characterized by progressive pelvic and scapular girdle weakening, where muscle changes can be quite small at times. Therefore, LGMD muscle measurements must be precise and reliable.

What does this mean for those living with LGMD and researchers in this space? As long as someone is having an MRI – get the full picture. The disease presents heterogeneously thus requiring a whole-body MRI to follow where LGMD is actually impacting a body. AMRA can deliver single muscles, muscle groups, and single MRI slice measurements. But why stop there? A person is not a single muscle or a single slice. A person is a whole body. So, scan and measure muscles and fatty infiltration throughout the entire body. Use AMRA's MRI measurements to potentially move drug treatment approval forward faster.

AMRA's regulatory compliant processes paired with our precision and accuracy, are the safe and reliable way to see the small changes potentially sooner than functional tests.

Courtesy: Fulcrum Therapeutics

FOR MORE INFORMATION

www.amramedical.com
info@amramedical.com
@AMRAMedical

Working urgently with the hope to bring therapies to the LGMD community

Sarepta is on an urgent mission: engineer precision genetic medicine for rare diseases that devastate lives and cut futures short.

As part of this mission, Sarepta's current gene therapy pipeline addresses six LGMD subtypes (see sidebar), which together represent more than 70% of all known LGMDs. Learn about our long-standing commitment to LGMD and the progress we've made to date below. We look forward to sharing more at our live presentation during the 2021 International Limb-girdle Muscular Dystrophy Conference weekend.

A Pioneer in LGMD Research

Louise Rodino-Klapac, Ph.D., Sarepta's Chief Scientific Officer, is a gene therapy pioneer dedicated to advancing medical research to treat genetically based diseases. She is renowned for contributions to molecular genetics and gene therapy research that have advanced the field, particularly in LGMD. She is currently based in Columbus, Ohio, home to our Gene Therapy Center of Excellence, where she and her team are working to further advance Sarepta's investigational gene therapy pipeline.

Based on the research and foundational work performed by Dr. Rodino-Klapac and her team, Sarepta's industry-leading LGMD pipeline aspires to treat this complex group of diseases which represent significant unmet need.

Learn more about our progress in LGMD:

1. Advancing Development of our Lead Clinical Program, SRP-9003

Our lead clinical program, SRP-9003, is designed to treat LGMD2E (R4). The first clinical trial is ongoing – participants have received their one-time infusion and long-term participant follow-up is continuing as planned.

"We recently shared clinical data from all participants in the SRP-9003 study, including up to 2 years post-SRP-9003 infusion. We are excited about these early data and will continue this study while also developing and preparing to launch new SRP-9003 studies. We look forward to discussing our plans with regulators, such as the FDA in 2021."

ERICA KOENIG, Ph.D., Senior Director, Clinical Development

2. Sarepta's LGMD Gene Therapy Engine Gains Steam

Our LGMD pipeline programs have a number of commonalities in their biological design, and in how they're constructed. Sarepta will leverage this knowledge, with hopes of enabling research to develop potential new therapies for a variety of subtypes.

"We are excited with the tremendous progress made in advancing our lead LGMD program, SRP-9003, as well as our broader LGMD pipeline. Some near-term milestones for our other programs include completing our toxicology work for LGMD2D (R3) and LGMD2C (R5), which is an integral part of pre-clinical development. We are also progressing toward our first clinical trial for LGMD2B (R2)."

ERIC POZSGAI, Ph.D, Senior Director, Gene Therapy Research

3. Advancing our Understanding of LGMD Disease Progression

One critical aspect in propelling our LGMD work forward is our ongoing natural history study, Journey. Learnings from this study will have the potential to help enable deeper understanding of LGMD types 2E, 2D, and 2C and may inform how we approach and design future clinical trials across our LGMD pipeline.

"Natural history studies provide foundational understanding of how the disease impacts people to facilitate development, approval, and ultimate availability of meaningful treatments."

DONNA-LEE DESTOUCHE, M.Sc., CCRP, Director, Clinical Operations

4. Sarepta is Consistently Seeking Patient Perspectives

We are continually seeking information about the experience and perspective of people living with limb-girdle muscular dystrophy and mapping our programs toward the needs expressed by the community. We do this through advisory boards, focus groups, one-to-one interactions when possible, and through our collaboration with Patient Advocacy Organizations.

"We are always seeking opportunities to engage, partner, support and learn important perspective from the communities we serve. We have already built meaningful relationships with many talented and thoughtful people in the LGMD community, and these engagements help to inform critical decisions."

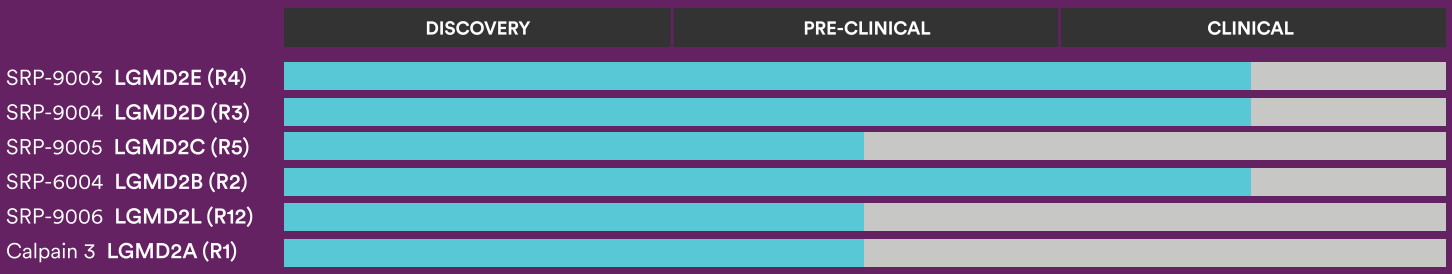
SIOBHAN FITZGERALD, Executive Director, Patient Affairs

Learn more at Sarepta.com

SAREPTA THERAPEUTICS

Sarepta's LGMD Pipeline in Development

Information is current as of 7/1/2021, updates are made on a quarterly basis



On an urgent mission, in partnership with the LGMD community

Your partnership and engagement play an integral role in our mission as we seek to incorporate the patient and caregiver perspective in all aspects of our work. The LGMD community often asks what they can do to help speed research and development. Here are some general suggestions:

Connect with a Neurologist

Connect with a neuromuscular specialist who is an expert in LGMD research and clinical care. Connect with a network of medical centers with expertise in LGMD: **lgmd-info.org/resources/lgmd-specialists**.

Get Genetically Tested to Learn Your LGMD Subtype

Genetic testing is the only approach that may conclusively diagnose a specific subtype. Knowing your specific subtype provides valuable information to help you discuss with your doctor which LGMD care plan may be right for you, and your genetic diagnosis may be required for entry into future clinical trials. Several programs in the U.S. offer free testing for LGMD and other muscle diseases. Learn more at: **limbgirdle.com/genetic-testing**, and talk to your doctor or genetic counselor about which test might be right for you.

Learn More About Natural History Studies

Natural history studies play an important role in developing a thorough understanding of the progression of LGMD, which is a critical component to the research and development of therapies. Ask your doctor to learn more.

Engage with the Community

Connecting and engaging with Patient Advocacy Organization(s) is a great way to access community news, education resources, and a network of support. Find an organization: **lgmd-info.org/resources/lgmd-organizations**.

Join a Patient Registry

Patient registries are essential to rare disease research and the ability to develop a foundational understanding of the number of people who are living with LGMD. Connect with Patient Advocacy Organizations/your doctor to learn about registries and find a list of registries here: **lgmd-info.org/resources/international-lgmd-patient-registries**.

Connect with Us

Sarepta's Patient Affairs team understands the value of direct conversation and engagement with advocates and members of rare disease communities around the world. Each day, the team strives to represent the many voices of the patient communities we serve.

Please do not hesitate to reach out to us at **Advocacy@Sarepta.com**.
We would love to speak with you!

Sarepta 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 LGMD.

KEISHA

A Guide to Genetic Testing for **LGMD**

WHO

Genetic testing is **recommended for individuals with symptoms** of limb-girdle weakness that suggest LGMD, such as difficulties walking, rising to stand, raising the arms, falling and others.

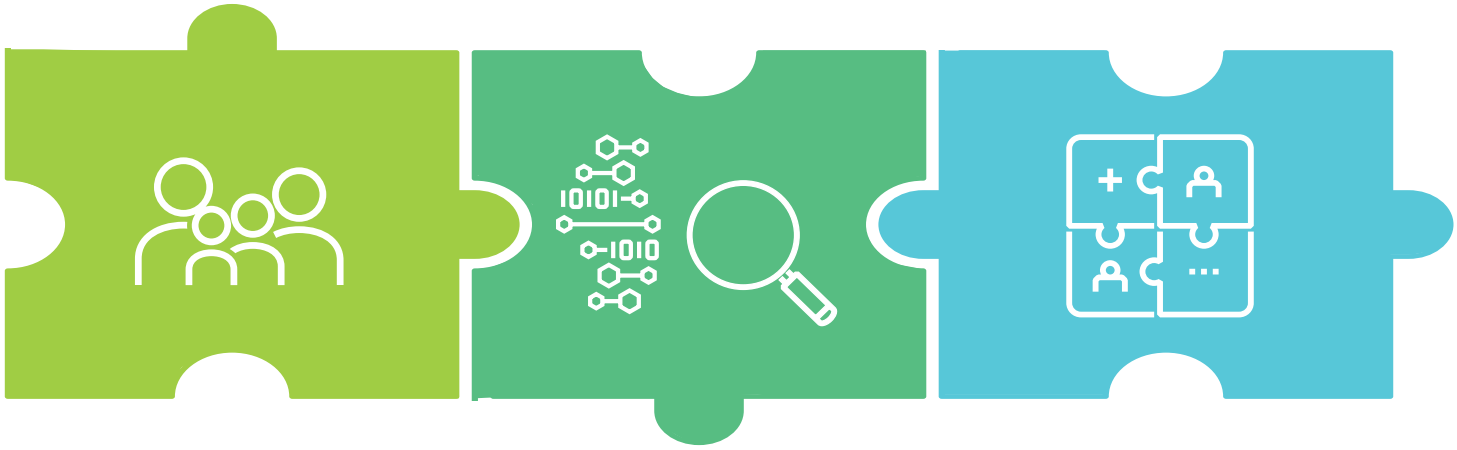
WHAT

Testing analyzes the **30+ genes** associated with **LGMD subtypes** (plus sometimes genes for other muscle diseases) to look for **gene variants** (changes) that may be disease-causing.

WHY

A genetically confirmed subtype diagnosis opens up new options to:

- Work with doctor to create **personalized patient care plan** based on subtype



A genetic test may confirm a **clinical diagnosis**—that is, one based solely on patient symptoms and medical history. Advances in testing technology means even those with prior, inconclusive genetic test results **may wish to consider getting re-tested now**.

What Does "Diagnosis" Really Mean?
"LGMD" alone isn't a diagnosis—it's a broad disease category of 30+ separate subtypes. Genetic testing is the **only approach that may conclusively diagnose a specific subtype**, by identifying a known disease-causing gene variant.

- Discuss with a doctor the possibility of participating in **LGMD clinical trials**, which generally require a genetic subtype diagnosis as a first step in possible patient eligibility
- Understand wider **family risk**, testing, and planning
- Connect with others in **subtype-specific LGMD communities** and advocacy organizations

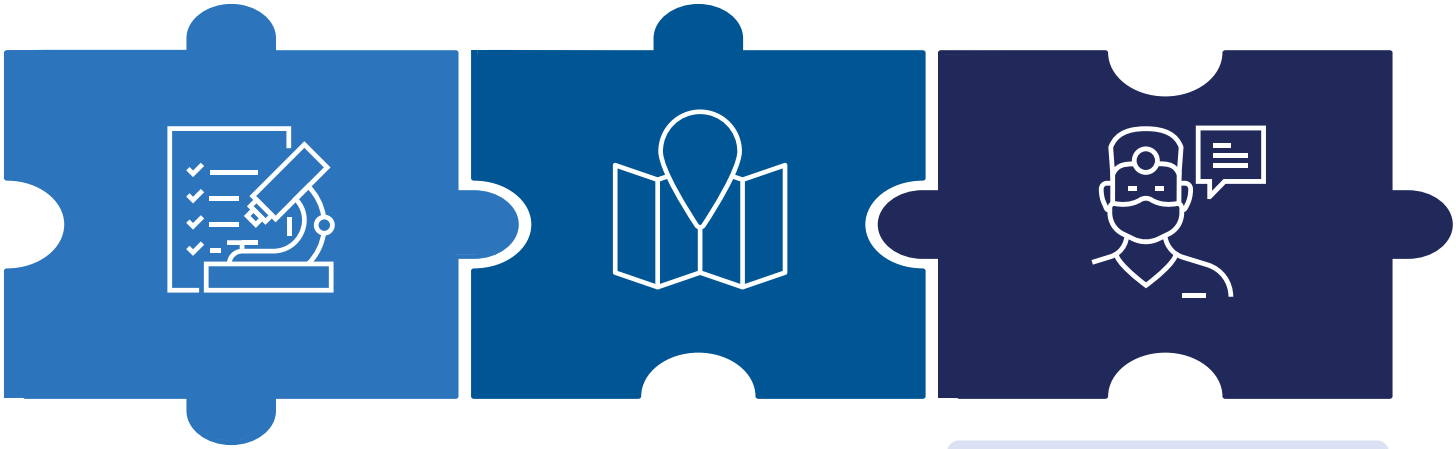
LimbGirdle.com

Sign up for updates on LGMD news, research, and community resources at limbgirdle.com/stay-connected

Today, genetic testing is accessible to many people and considered a first-line approach to diagnosing limb-girdle muscular dystrophy (LGMD) or other similar muscle diseases.

HOW

It's best to order a genetic test through a doctor, but some programs, such Invitae's Detect Muscular Dystrophy, allow patient-ordered testing supported by lab-staffed genetic counselors. **The testing process is typically straightforward:**



- Work with a doctor or genetic counselor to determine the appropriate test and coordinate collecting a **DNA sample (typically blood or saliva)** and sending it to the lab
- Lab analyzes DNA for **gene variants** that may be disease-causing
- Lab provides **test report** within **~2-5 weeks** in U.S.

Afterwards, it's important to **review results** with **a doctor and/or genetic counselor**.

WHERE

Several programs in the U.S. offer **free testing** for LGMD and other muscle diseases. Learn more at **limbgirdle.com/genetic-testing**, and talk to your doctor or genetic counselor about which might be right for you.

U.S. Free Programs:

Detect Muscular Dystrophy—Invitae
Phone: (800) 436-3037
Website: invitae.com/en/detect-muscular-dystrophy

The Lantern Project—PerkinElmer Genomics
Phone: (866) 354-2910
Website: lanternprojectdx.com

Rare Genomes Project—The Broad Institute of MIT and Harvard
Phone: (617) 714-7395
Website: raregenomes.org/limb-girdle-muscular-dystrophy

POSSIBLE TEST RESULTS

Conclusive
Close to half of those tested for suspected LGMD get a **definitive subtype diagnosis**.

Uncertain
Roughly half do not get a diagnosis because testing finds **variants of uncertain significance (VUS)**: not enough data to determine if a variant is disease-causing or not.

Take Action
Uncertain results are not the end of the diagnostic process. Patients and doctors can take follow-up actions collaboratively with the lab to collect more data, which over time may help clarify the variant.

Seek Support
Genetic counselors are a valuable resource to **help interpret test results, plan next steps, and provide support** throughout the testing process.

Helpful Resources:

- **Find a genetic counselor** at findageneticcounselor.nsgc.org
- **Search for LGMD clinical trials** at clinicaltrials.gov
- **Consider genetic data-sharing** at genomeconnect.org

Sarepta 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 LGMD.

PAT

Are you an individual or a parent of an individual living with Limb-girdle muscular dystrophy (LGMD) Type 2E (LGMD2E/R4), Type 2D (LGMD2D/R3), and Type 2C (LGMD2C/R5)? You may be eligible to join Sarepta's natural history study.

JOURNEY (NCT04475926)

What is a natural history study?

In a natural history study, individuals living with a disease are followed closely by researchers to better understand how that disease progresses over time. No study drug is administered, and individuals continue their current care plan as designed by their regular doctor.

Why is Sarepta conducting a natural history study for LGMD 2E, 2D, and 2C?

Sarepta is conducting Journey to better understand the natural progression of LGMD2E, LGMD2D, and LGMD2C. Information from Journey may help Sarepta better design potential future clinical trials using investigational medicines.

Why consider joining Sarepta's natural history study?

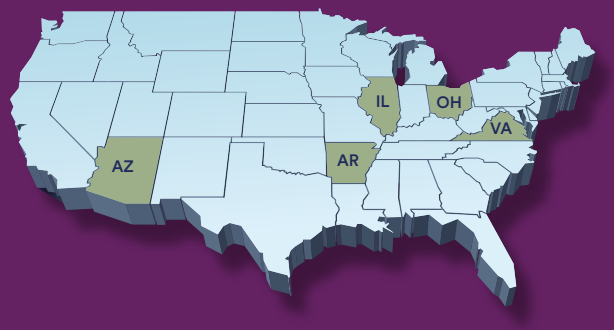
In addition to supporting research and development overall, this is a unique opportunity to be clinically followed by a care team focused on limb-girdle muscular dystrophy. Individuals will also be assessed for eligibility for potential future gene therapy clinical trials. Please note that participation in an interventional trial is not guaranteed.

Who may be eligible to enroll in Sarepta's natural history study?*

- Male or female individuals ≥4 years of age with a confirmed genetic diagnosis of LGMD2E, LGMD2D, or LGMD2C.
- Willing to travel to an active clinical trial site (travel and some associated expenses are reimbursed as part of the study)

**Other inclusion/exclusion criteria may apply. Please contact a study site for more details.*

Check out the locations of our currently activated sites in the United States:



• Arizona • Arkansas • Illinois • Ohio • Virginia

Additional sites in the United States, and parts of Asia, Europe, and South America are planned.

Learn more at Sarepta.com

SAREPTA THERAPEUTICS

Sarepta is a 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.

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 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 LGMD.

KAREN

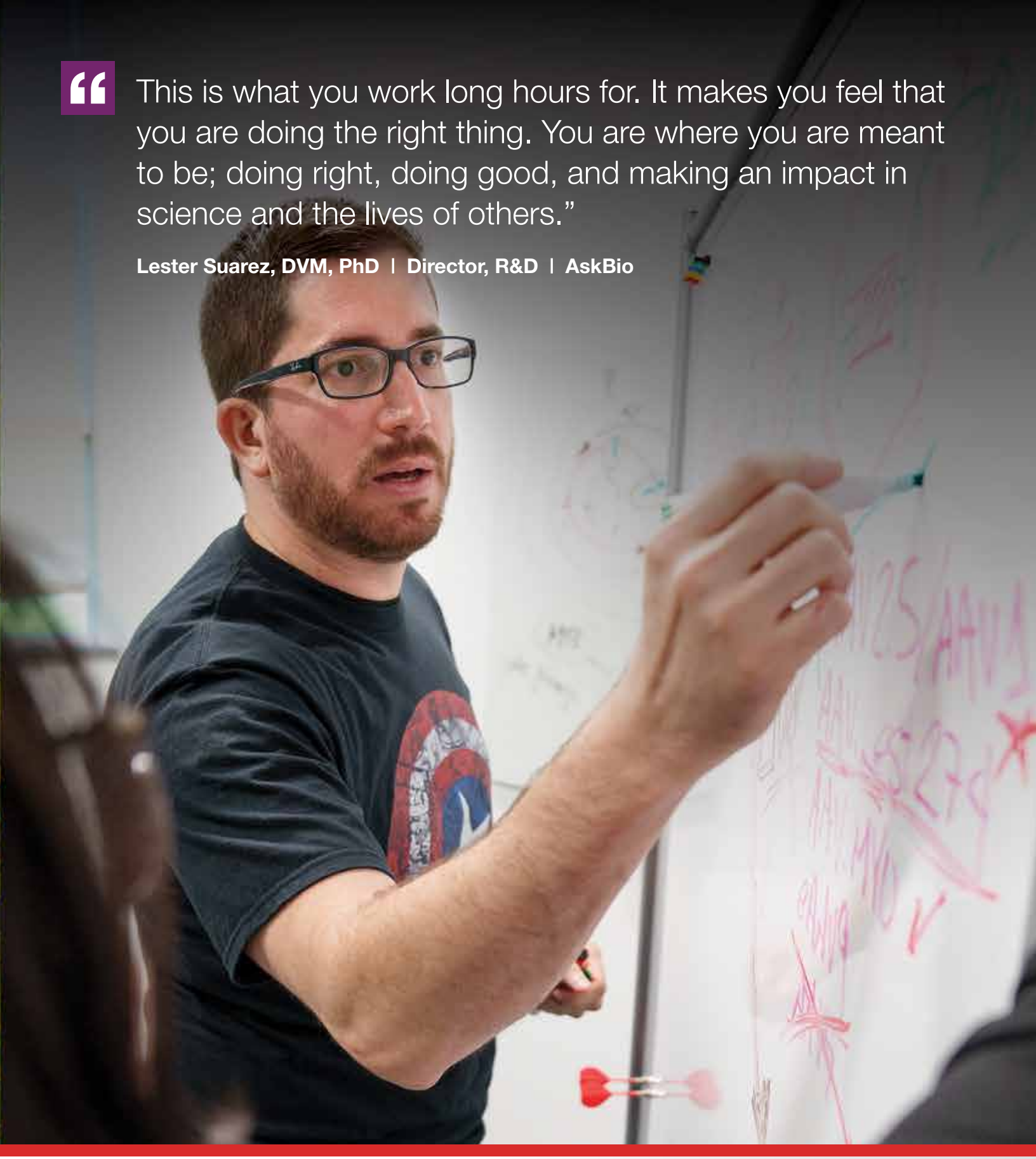
“I’m not afraid of much. Sometimes I’m afraid of limb-girdle, but most of the time I’m not.”

Sammy

AskBio™
askbio.com

“The first time I was told I had a rare form of muscular dystrophy, it was a profound moment. I wanted to know more, but they said they had no more information than the diagnosis.”

Lisa



AskBio™
askbio.com

Coming soon!
AskBio's Gene Therapy Study in LGMD2I/R9

A study of 2 doses of LION-101 in LGMD2I/R9

- LION-101 is one-time intravenous infusion of gene therapy designed to express fukutin-related protein (FKRP), primarily in muscle
- Safety and efficacy of LION-101 will be studied first in adults, and eventually adolescents, in a placebo-controlled, crossover study (where all enrolled participants will receive gene therapy)
- At least 2 doses of LION-101 will be studied
- This first-in-human dose-finding study will be run in the US
- Travel to study sites will be reimbursed; home-based assessments will be used when possible

Visit AskBio.com for updates, or email AskFirst@AskBio.com

AskBio Supports the Patient and Family Heroes Across the LGMD Community

AskBio™

askbio.com

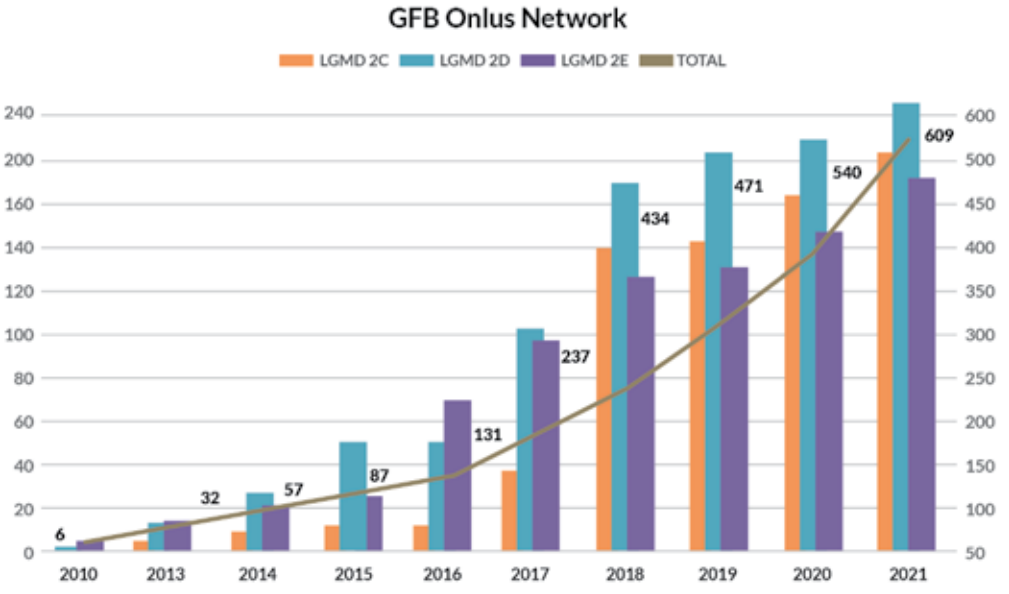
GFB | GRUPPO FAMILIARI BETA-SARCOGLICANOPATIE

GFB

GRUPPO FAMILIARI BETA-SARCOGLICANOPATIE

We founded GFB because we couldn't find any information on our disease anywhere.
For nearly seven years we searched, looking into different associations, at conferences, with doctors, on the web, and on social media. Everybody talked about different neuromuscular diseases, but never about ours, LGMD2E/R4. There were no foundations that were specifically concerned with LGMD2E/R4. It seemed that nobody was interested in this disease at all. In 2010, the GFB internet site was born and in 2013, our first statute was registered. In the beginning, GFB was focused only on LGMD2E/R4, but since then, we have also added LGMD2C/R5 and LGMD2D/R3 to our program.

Because we were alone, we started to look for other families affected by this disease.
In 2003, we started looking for other families, and finally, after six years of searching, we found one. Since then, our patient research strategy has been improved year after year, especially by way of the internet and social media.



Increase in GFB patients from 2010 to 2021

Over
600 patients
in the world

We have funded the preclinical part of the LGMD2E/R4 gene therapy project

Because there was no scientific research on this disease, we started to support research efforts.

We have always believed in the importance of research because only the systematic and targeted study of the disease can lead to a better quality of life for patients. Since 2012, GFB has funded the preclinical part of the gene therapy project for LGMD2E, which is now already in the clinical phase.

We started these clinical studies because very little data existed for patients affected by this pathology.

GFB didn't stop at their first project, and in 2018, a natural history study on Italian patients commenced, with the support of Dr. Yvan Torrente at the Milan Polyclinic in Italy.

Because the number of Italian patients are few, this study also needs data from patients outside of Italy. To help with this effort, in 2021, the Quality Project was born, a study on the quality of life, conducted by GFB, with the support of the Milan Polyclinic. This project has led to many collaborations with foundations and clinical centers from around the world.

A Natural History Study on Italian LGMDR4/2E Patients

Dr. Yvan Torrente, Professor of Neurology at the University of Milan and Medical Director of the Neurology Unit of the IRCCS Ca'Granda Foundation, Ospedale Maggiore Policlinico of Milan, Italy.

This study was developed by:

- G.B. Marchetti: Investigation, writing original drafts;
- L. Valenti: supervision, statistical analysis, writing, review, and editing;
- Y. Torrente: conceptualization, supervision, writing, review, and editing.

This research was funded by GFB Onlus. The funder of the study had no role in study design, data analysis, data interpretation, or writing of the report.

We conducted a retrospective review of the records of 26 Italian patients with LGMDR4. Our primary objective was to compare the rates of decline among creatine phosphokinase (CPK) values, pulmonary function test (PFT) measures, and echocardiographic estimates, and to relate them to patients' ages.

Twenty-six patients, with a defined genetic diagnosis of LGMDR4 were enrolled, ranging in age from 8 to 55 years, and with data collected on at least three different visits. Patients were enrolled within the "Gruppo Familiari Beta-sarcoglicanopatie" Italian network and evaluated in different Italian centers once or twice per year.

The study was approved by the Ethics Committee of the Fondazione IRCCS Ca' Granda Ospedale Policlinico of Milan (EC approval 386_2020, 19 May 2020) and registered on ClinicalTrials.gov with the following number: NCT04509609.

The rates of decline/year of CPK, PFTs and LV function estimates are significatively bound to age, with the LV ejection fraction (EF) being the strongest independent variable describing disease progression. Moreover, the rate of decline of CPK, PFTs, and LV differed in patients grouped according to their genetic mutations, demonstrating a possible genotype-phenotype correlation. The parallel trend of decline in CPK, PFTs, and EF values demonstrates the presence in LGMDR4 of a simultaneous and progressive deterioration in muscular, respiratory, and cardiac function.

This study expands the current knowledge regarding the trend of CPK values and cardiac and respiratory impairment in patients with LGMDR4, to optimize the monitoring of these patients, to improve their quality of life and provide clinical indices capable of quantifying the effects of any new gene or drug therapy.

Yvan Torrente

GFB Quality Project

A GFB study on the quality of life of patients affected by LGMD2E-2C-2D from all over the world.

The imminent arrival of clinical trials for LGMD has spurred GFB to launch a worldwide study on the quality of life of patients with LGMD2C-2D-2E dystrophy (or LGMDR3-R4-R5). We think that associations and patients must engage and give researchers, doctors, and industries what they need to complete their studies of these diseases. At present, GFB is the only existing organization dealing with LGMD2E/R4. For the last eleven years, GFB have been concerned with looking for other families affected by this disease and two others (LGMD2C/R5 and LGMD2D/R3). Nowadays, we are in contact with more than 600 patients of different subtypes from all over the world. We strongly believe that together we can do more! GFB has started the Quality Project to better define the data collected in the past years. The project is fully funded by GFB.

Participate in the Quality Project so that you can help advance important research!

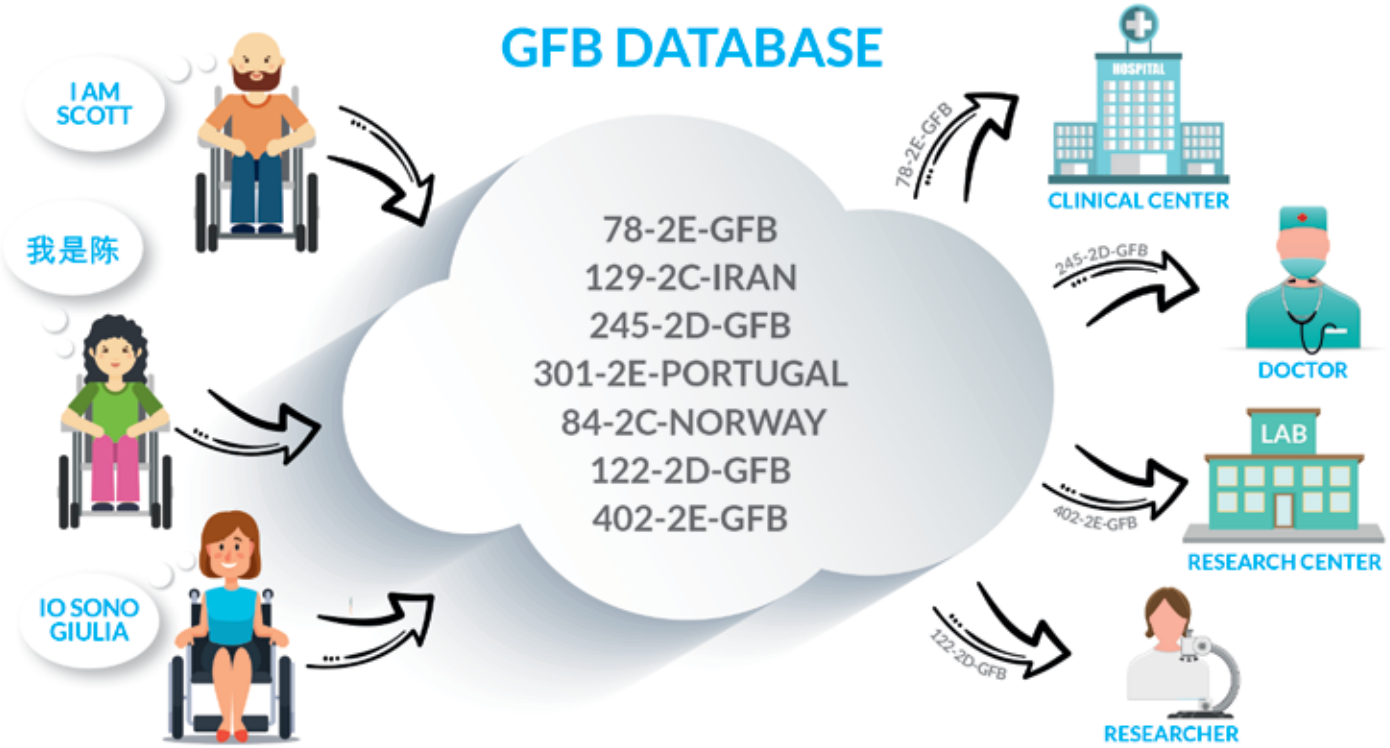
All the patients affected by LGMD2E/R4, LGMD2D/R3, and LGMD2C/R5 can take part in the Quality project.
Participating is simple: email info@beta-sarcoglicanopatie.it, and in return, you will receive a questionnaire to fill out. In the questionnaire, you'll find questions related to the following subject areas:

- Informed consent
- General information and other information on your disease
- Mobility
- Carrying out daily activities
- Fatigue in carrying out daily activities
- Psychological well-being.

The questions will be semi-structured with most in multiple-choice format.
If you have any problems filling out the questionnaire, you can also participate in a video call with GFB staff members who can help you. We are able to assist in the following languages: Arabic, Italian, Romanian, Russian, Spanish, Portuguese, French, German, English, and Persian.

Patients will be identified with alphanumeric codes.

The alphanumeric codes are needed to identify the patient throughout the data treatment at the clinical and research centers.



GFB will always keep any personal data of the patient confidential, which is the reason for these alphanumeric code. Patient data will be transferred to the clinical centers and researchers only through this alphanumeric code, in order to avoid identification of the patient. All these steps are well described in the Informed Consent that is provided at the beginning of the questionnaire.

The alphanumeric codes are composed of a number, the acronym which identifies the type of disease, and the origin of each patient (the name of the association or country to which the specific patient belongs). In this way, it will be possible to recognize patients coming from different associations and clinical centers, which is helpful in future publications of the study.

Possible Usefulness and Benefits of the Quality Project

This project presents several benefits. First, the Quality Project has the purpose of scientifically describing the quality of life of patients and will allow evaluation of the impact of potential new therapies. Moreover, it will allow patients access to new therapies and future clinical trials.

A Novel Therapeutic Solution for Sarcoglycanopathies

CFTR Correctors in Sarcoglycanopathies

Dr. Dorianna Sandonà, MD, PhD, Department of Biomedical Sciences, University of Padua, Italy

Sarcoglycanopathies or LGMDR3-6 are muscular dystrophies caused by mutations in the genes coding for sarcoglycans (α-, β, γ- and δ-SG). SGs form a complex that is crucial for protecting the sarcolemma from mechanical stresses elicited during muscle contraction. When the complex is absent or strongly reduced, because of the mutation of one subunit, the sarcolemma is fragile and muscle can be damaged.

Our findings show that most of the mutated sarcoglycans (particularly those carrying a missense mutation) are defective in the 3D structure (folding defective), or in trafficking toward cell membrane, and are therefore dismantled even though potentially functional.

Thanks to this knowledge, we envisage the possibility to help the folding process of these sarcoglycan mutants by using small molecules known as CFTR correctors, developed to treat cystic fibrosis. These compounds are able to correct a mutated CFTR that, like sarcoglycan, is defective in folding and or trafficking.

First, we successfully tested CFTR correctors in cellular models of sarcoglycanopathy. Subsequently, we treated primary pathological muscle cells from sarcoglycanopathy patients, evidencing that several compounds, and particularly corrector C17, rescued the sarcoglycan complex at the sarcolemma that resulted in being strengthened.

To validate the compound in vivo, we treated a novel mouse model of sarcoglycanopathy, ad hoc generated, observing the amelioration of the dystrophic phenotype at the histological, molecular, and, notably, functional level. Indeed, the muscle force generated by treated animals is nearly identical to that of the healthy mice and this occurred in the absence of evident signs of toxicity.

Our pharmacological approach aims at rescuing the endogenous protein and it is based on small molecules, easy to produce and deliver and, in this case, to optimize. It is evident the advantage over complex procedures such as gene or cell transfer, and could make patients more compliant to therapy.

Considering that missense mutations are the most frequent cause of LGMDR3-6, we are confident that the use of C17, or other correctors, will be of benefit for a large cohort of patients, representing a real advancement towards a cure for people suffering with these neglected diseases.

We Collaborated with Other Associations to Tour our Territory

Opportunities for disabled people in Valtellina and Valchiavenna

GFB and other associations in Valtellina and Valchiavenna collaborate in the organization of a lot of activities for disabled people.

Hiking in the Rhaetian and Orobie Alps

GFB and other associations promote accessible tourism in Valtellina and Valchiavenna, organizing both winter and summer outdoor activities with special mountain wheelchairs. Hikes are made accessible via these chairs.

Ski lessons for disabled people

Using necessary aids, the instructors of the Enjoyski School are involved to help make alpine skiing possible for the disabled.

Hiking on various trails, including the Valtellina and Rusca trails

Valtellina Trail: https://sentiero.valtellina.it
Rusca Trail: www.sondrioevalmalenco.it/it/itinerari/il-sentiero-rusca
Accessible trails map: www.dappertutto.org/percorsi

Other activities in Valtellina and Valchiavenna for the disabled are also possible

Trips on the red train of the Bernina, one of the most fascinating trains in the world: From Italy to Switzerland, from Valtellina to the Engadine, from the city to the pleasant landscape of the valley floor, to the peaks covered with glaciers. From Tirano to Saint Moritz, three hours of a breathtaking journey!

Helicopter trips, sailplanes and touring aircraft.
Rafting on the Adda river.
Boat cruises on Como Lake.

CONTACTS FOR EXCURSIONS:

Associazione dappertutto Odv: The Associazione dappertutto represents the will to restore fragility everywhere in society. It concretely helps to eliminate architectural barriers and sensitizes and promotes a culture that is attentive, available, accessible.
www.dappertutto.org
consulenza@dappertutto.org

- **Accessible accommodation map in Valtellina and Valchiavenna:** featuring 16 hotels and facilities for disabled people
  https://www.dappertutto.org/strutture-ricettive
- Accessible playgrounds:
  http://www.dappertutto. org/aree-gioco

Enjoyski Sport Onlus: Facilities, equipment, and resources are made available to give people with disabilities and their families the opportunity to practice different sports in the name of fun and inclusion.
https://www.enjoyskisport.it

Hope Through Rigorous Science

Thank you to all the participants in the ML Bio Solution-sponsored LGMD2I Lead-in (Natural History) Study and the Phase 2 Trial. You are making a significant contribution to furthering our understanding of LGMD2I and to developing a new potential treatment.

We are very grateful to all of you for your time and commitment!

Questions? Contact us at info@mlbiosolutions.com. Visit our web site at: www.mlbiosolutions.com.

ML Bio Solutions (ML Bio) is a biotechnology company founded by a family whose child was diagnosed with LGMD2I. ML Bio Solutions is a member of the BridgeBio family--a team of experienced drug discoverers, developers and innovators working to create life-altering medicines that target genetic diseases.

ML Bio Solutions

a bridgebio company

DYSFERLIN REGISTRY

by Jain Foundation

LGMD2B/R2/Miyoshi Myopathy 1

A registry focused on dysferlinopathy

www.jain-foundation.org/forms/dysferlin-registry-application/

Robust Responsive Ready

LGMD Awareness Day
September 30

- **Advocate**
- **Educate**
- **Celebrate**

By increasing awareness of and advocating for individuals living with limb-girdle muscular dystrophy (LGMD), we hope to assist in advancing the diagnosis, care, and treatment for individuals around the world.

Together We Are Stronger

LGMD-Info.org

@LGMDawareness

LGMD AWARENESS FOUNDATION

LGMD Awareness Day is a *project* of the LGMD Awareness Foundation, a 501(c)(3) advocacy organization.

LGMD-1D DNAJB6 Foundation

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

Beyond Labels & Limitations is dedicated to looking beyond the current status of Limb-Girdle Muscular Dystrophy 2A. Our objective is to raise funds for scientific research and educate people about the progression of the disease and how it affects those struggling with it. We believe there is a cure out there, and we will find it. We are hopeful for the future and the many possibilities it holds.

BEYOND Labels & Limitations
What Is Your Life Devoted To

BeyondLabelsLimitations.com

YouTube Search for John Graybill II

Listen on Apple Podcasts

We're Closer to a Cure

– with multiple potential treatments in clinical trials in 2021, your support means more than ever!

CURELGMD2i
Limb Girdle Muscular Dystrophy

Learn More, Donate, or Get Involved at

CURELGMD2i.org

Or follow us on

Edgewise Therapeutics

is proud to support the

2021 International Limb Girdle Muscular Dystrophy Conference

At Edgewise, patients are at the core of everything we do. We recognize that for patients with rare and debilitating diseases, every day without an effective treatment is a day too late. We are driven by this urgency to develop novel precision medicines for severe and debilitating rare muscle disorders.

For more information, please email info@edgewisetx.com or go to www.edgewisetx.com.

Edgewise THERAPEUTICS

Take part in the GFB Quality project

info@beta-sarcoglicanopatie.it

Limb girdle muscular dystrophy affects everyone
in the same way.
There are no differences among the different countries.
Among different cultures or languages.

Together we are stronger!

GRUPPO FAMILIARI GFB ONLUS BETA-SARCOGLICANOPATIE

GFB ONLUS

www.lgmd2e.org
info@beta-sarcoglicanopatie.it

The SPEAK Foundation
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ST JOHNS FL 32260-0062

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