Vol 5 / Issue 3

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

YOUR VOICE MATTERS!

Patient-Driven Advocacy is Always at the Heart of what The Speak Foundation Does

ORLANDO BOUND!

We Welcome You to the 2025 International Limb Girdle Muscular Dystrophy Conference

THINK GLOBAL, ACT LOCAL

How GRASP-LGMD Works Together Towards a Common Mission

CRACKING THE CODE OF GENETIC TESTING REPORTS

Explaining the Key Components of a Genetic Testing Report

2025 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE

ORLANDO, FL

JW MARRIOTT ORLANDO
JULY 18–20, 2025

CONFERENCE PROGRAM
INSIDE

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

SPECIAL CONFERENCE EDITION

Every person deserves excellent healthcare.

This is why The Speak Foundation's Healthcare Access Grant program exists.

Many individuals with physical health conditions struggle with healthcare insecurity, marked by lack of access to quality care and the financial inability to travel. At the Speak Foundation, we believe that every person deserves excellent healthcare and that financial struggles should not prevent anyone from world-class care. The Healthcare Access Grant program provides travel grants for qualified LGMD patients living in the United States to visit approved, leading centers of LGMD excellence. This program is open as of July 1, 2024. To find out if you are eligible for a travel grant, please contact ***Jessica@thespeakfoundation.com***.

GRASP-LGMD Principal Investigators and Sites in the US

- University of Iowa
- University of Colorado
- University of Florida
- Atrium Health
- Nationwide Children's Hospital
- Washington University in St. Louis
- Kennedy Krieger Institute
- University of Kansas Medical Center
- University of Minnesota
- Indiana Community Health Clinic
- University of California, Irvine
- Virginia Commonwealth University

** If you have another center in the US that you would like to be seen at for LGMD, please connect with us.*

Eligibility Criteria:

- US residency
- Suspected or confirmed diagnosis of LGMD
- Lacking access to an excellent facility for medical care/in need of multidisciplinary care
- In need of financial assistance
- Must be able to travel

** Please also note that the grant program involves reimbursement of travel expenses, and recipients must cash the check within 90 days of receipt.*

The SPEAK Foundation

TheSpeakFoundation.com/grant-programs

LGMD News

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How To Donate

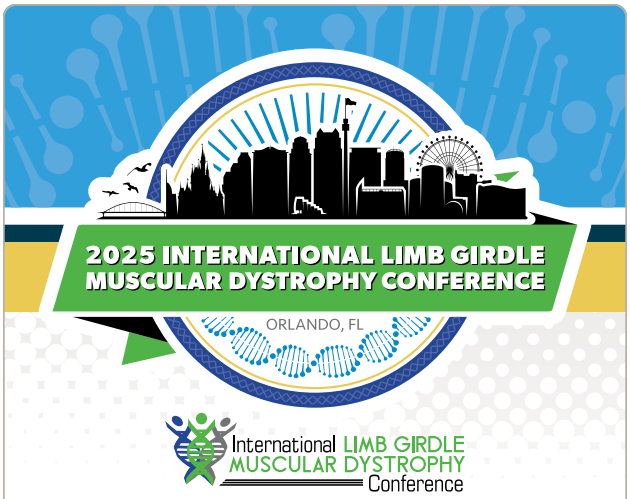
Visit **TheSpeakFoundation.com** and select "Donate" to support our mission and programs. The Speak Foundation is a 501(c)(3) nonprofit charity. All donations are secure and tax deductible.

Thank you for your support!

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

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

Contents



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AT THE SPEAK FOUNDATION, WE BELIEVE YOUR VOICE MATTERS.

INDIVIDUALS WHO LIVE WITH LIMB GIRDLE MUSCULAR DYSTROPHY DESERVE TO BE HEARD. WE KNOW THE ONLY PERSON WHO CAN SPEAK FOR YOU IS YOU. SO WHEN WE BEGAN THE INTERNATIONAL LGMD CONFERENCE IN 2019, WE WANTED TO MAKE SURE THE VOICE OF THE PATIENT WAS THE LOUDEST IN THE ROOM. PATIENT-DRIVEN ADVOCACY IS ALWAYS AT THE HEART OF WHAT WE DO. BE A PART OF THE SPEAK FOUNDATION'S ADVOCACY EFFORTS – JOIN OUR LGMD PATIENT NETWORK, STAY CONNECTED WITH *LGMD NEWS* MAGAZINE, AND PARTICIPATE IN OUR MANY ADVOCACY EVENTS. SPEAK UP AND SHARE YOUR VOICE TODAY!

TheSpeakFoundation.com

International Consortium of LGMD Organizations

United States

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TheSpeakFoundation.com

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CURELGMD2I
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LGMD AWARENESS FOUNDATION
LGMD-Info.org

LGMD-1D DNAJB6 FOUNDATION
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LGMD2L-Foundation.org

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TitinMyopathy.com

THE JAIN FOUNDATION
Jain-Foundation.org

Italy

CONQUISTANDO ESCALONES ASSOCIATION
ConquistandoEscalones.org

"GFB ONLUS"/ FAMILY GROUP OF BETA-SARCOGLYCANOPATHY
Beta-Sarcoglicanopathy.org

GRUPPO CINGOLI OF UILDM – UNIONE ITALIANA LOTTA ALLA DISTROFIA MUSCOLARE
UILDM.org

ITALIAN ASSOCIATION CALPAIN 3
AICA3.org

Japan

PATIENTS' ASSOCIATION FOR DYSFERLINOPATHY JAPAN
PADJ.jp/index.html

Korea

KOREAN DYSFERLINOPATHY PATIENTS ASSOCIATION
Cafe.Naver.com/UniteDsyferlinopathy

Argentina

ADM ARGENTINA MUSCULAR DYSTROPHY LGMD GROUP
ADM.org.ar

Spain

CONQUISTANDO ESCALONES ASSOCIATION
ConquistandoEscalones.org

France

"GI LGMD"/LGMD PATIENT GROUP OF AFM-TELETHON
LGMD.AFM-Telethon.fr

Netherlands

STICHTING SPIERKRACHT
StichtingSpierkracht.com

Australia

DANIEL FERGUSON LGMD2A FOUNDATION
DFFoundation.com.au

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

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

LGMD Day on the Hill is an advocacy event and program of the Speak Foundation. This event is for delegates living with limb girdle in the USA to meet with their legislators on **September 9, 2025**.

Let Your Voice Be Heard. Selected delegates will need to travel to Washington, D.C., the day before to participate in mandatory advocacy training on September 8th in preparation for LGMD Day on the Hill. Limited travel stipends are available, provided by **The Speak Foundation** and the **LGMD Awareness Foundation**.

Delegate Applications Accepted through July 22, 2025.

APPLY TO BECOME A DELEGATE TODAY!

The Speak Foundation's Inaugural

LGMD Congressional Champion Award

FOR THE FIRST TIME, The Speak Foundation is presenting awards to Members of Congress who have gone the extra mile for patients with LGMD. This new award, the LGMD Congressional Champion Award, will be presented to bipartisan leaders in Congress who are advancing legislation that is important to our patients and community. The difficult issues facing limb girdle muscular dystrophy patients are not going to be solved by one political party or the other, but instead, by individuals who seek common ground solutions that keep the patient voice and perspective at the center. With these inaugural awards, The Speak Foundation is proud to recognize Sen. Roger Wicker (R-MS) and Sen. Amy Klobuchar (D-MN) for their tireless commitment and leadership with the 2025 LGMD Congressional Champion Award.

Senator **Roger Wicker**

Senator Roger Wicker (R-MS) is the Republican co-chair of the Senate Rare Disease Caucus. He has been committed to policy goals that advance innovation for rare disease treatments, and in this role, Senator Wicker has been a leading voice on the legislative needs of the rare disease community, including his support of the FDA Rare Disease Innovation Hub. For these reasons, we are proud to present Senator Wicker with this award.

Senator **Amy Klobuchar**

Senator Amy Klobuchar (D-MN) is the Democratic co-chair of the Senate Rare Disease Caucus. She is also the lead Democratic sponsor of the Scientific EXPERT Act, along with Senator Wicker, which would provide further direction to the Rare Disease Innovation Hub to convene key meetings to streamline the development process for rare disease therapies. Senator Klobuchar is also a leader on the long-term reauthorization of the Pediatric Priority Review Voucher program. For these reasons, we are proud to present Senator Klobuchar with this award.

Congratulations!

Welcome!

We all know the world is changing — and for the LGMD community that change brings hope. Treatments are now in development for many forms of LGMD, and we are facing a future where real progress — and even effective treatments — may be within reach. This is why our conference theme is **Believe there is Hope for a Cure**.

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. 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

2025 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE

ORLANDO, FL

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

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

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Schedule

FRIDAY JULY 18, 2025

2025 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE
ORLANDO, FL

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

3:00 – 6:00 PM	**General Registration — Check In**	Grande Registration Desk
4:00 – 6:15 PM	**Exhibitor Setup**	Palazzo Foyer
6:30 – 8:00 PM	**Welcome Reception for Individuals Living with LGMD**	Valencia/ Terrace/ Del Lago

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SATURDAY JULY 19, 2025

8:00 – 11:00 AM	**General Registration — Late Check In**		Grande Registration Desk
8:15 – 9:00 AM	**Breakfast Snacks**		Palazzo Foyer
9:30 – 10:30 AM	**Session 1**	**Welcome, Overview, and Clinical Care** **Conference Welcome** • Kathryn Bryant Knudson, President, The Speak Foundation **LGMD Advocacy and Organization Recognition** • Kathryn Bryant Knudson, President, The Speak Foundation **LGMDs: What Should Patients Know to Get Productive Clinical Care?** • Matthew P. Wicklund, MD, University of Texas **Developing Formal Standards of Care for LGMDs** • Volker Straub, MD, PhD, University of Newcastle, UK • Meredith James, PhD, University of Newcastle, UK	Palazzo Ballroom
10:30 – 10:45 AM	**Break**		

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Schedule

SATURDAY JULY 19, 2025

Continued

10:45 – 11:45 AM	**Session 2: Breakout**	**Understanding Diagnosis and Genetic Test Reports** • Lee Kugelmann, NMSc, CGC, University of Florida • Conrad (Chris) Weihl, MD PhD, Washington University School of Medicine • Peter Kang, MD, University of Minnesota • Brad Williams and Sara Yemm	Palazzo Ballroom
		Life Hacks for Successful Living with LGMD • Tips & Tricks: Carol Abraham & Victoria Nedza • Driving Independence: Cerys Davage & John Faver	Palazzo A
		Raising a Child who has LGMD — Meetup for Parents and Family Members • Rachel DeConti, Faran Day, and Aleksandra Leijenhorst Le Belle	Palazzo C
11:45 AM – 12:00 PM	**Break**		
12:00 – 12:50 PM	**Lunch**	**Sarepta Therapeutics Symposium** *Sponsored by Sarepta Therapeutics*	Palazzo Ballroom
12:50 – 1:00 PM	**Break**		
1:00 – 1:45 PM	**Session 3: Breakout**	**How Does an Idea Become a Drug? Moving from the Lab to Clinical Trials** • Kevin Campbell, PhD, University of Iowa • Sharon Hesterlee, PhD, Muscular Dystrophy Association • Nicholas E. Johnson, MD, Virginia Commonwealth University	Palazzo Ballroom
		Genetic Counselors: Q&A • Lee Kugelmann, NMSc, CGC, University of Florida • Brianna N. Gross, MS, LCGC, Children's Hospital of Philadelphia	Palazzo A
		Living with Dominant LGMDs and Treatment Approaches • Jeffrey Statland, MD, Kansas University Medical Center • Victoria Nedza, Patient • Natasha Lowery, LGMD-1D DNAJB6 Foundation	Palazzo B
		Building Community Connections: Adolescents — Meetup for Ages 12–17 (No Parents) • Peter Frewing, Brooklyn Garza, and Sammy Brazzo	Palazzo C

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International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

1:45 – 2:00 PM	**Break**		
2:00 – 2:45 PM	**Session 4: Breakout**	**Current Drug Development Pathways for LGMDs** • Barry Byrne, MD, PhD, University of Florida • Matthew P. Wicklund, MD, University of Texas	Palazzo Ballroom
		Physical Therapy and Rehab: Areas to Watch with LGMD • Lindsay Alfano, PT, DPT, PCS, Nationwide Children's Hospital • Vovanti Jones, MD, University of Missouri	Palazzo A
		Building Community Connections: Young Adulthood with LGMD—Ages 18–25 (No Parents) • Cerys Davage & Serena Desiderio	Palazzo C
2:45 – 3:00 PM	**Break**		
3:00 – 3:45 PM	**Session 5: Breakout**	**Ask the Expert: Q&A** • Lindsay Alfano, PT, DPT, PCS, Nationwide Children's Hospital • Barry Byrne, MD, PhD, University of Florida • Vovanti Jones, MD, University of Missouri • Peter Kang, MD, University of Minnesota • Volker Straub, MD, PhD, University of Newcastle, UK • Bjarne Udd, MD, PhD, Tampere Neuromuscular Center • Matthew P. Wicklund, MD, University of Texas	Palazzo Ballroom
		Exercise, Weight Management, and Nutrition in LGMD • Tina Duong MPT, PhD, Stanford Medicine	Palazzo A
		Navigating Mental Health with Changing Disability • Melissa Grove MS, LPC	Palazzo B
3:45 – 4:00 PM	**Group Photo**		Palazzo Ballroom
4:00 – 5:30 PM	**Break**		
5:30 – 6:30 PM	**Cocktail Reception, Awards Ceremony, and Scientific Poster Session**		Palazzo Ballroom
8:00 PM	**"In Search of Strength" Documentary**		Palazzo Ballroom

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Schedule

SUNDAY JULY 20, 2025

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

8:15 – 9:00 AM	**Breakfast Snacks**		Palazzo Foyer
9:30 – 10:45 AM	**Session 6**	**Updates from Clinical Trials** **ML Bio Solutions** • Douglas Sproule, MD, MSc, Chief Medical Officer **Sarepta Therapeutics** • Louise Rodino-Klapac, PhD, Head of R&D, Chief Scientific Officer **AskBio** • Samuel Hopkins, PhD, Senior Vice President of Therapeutics **Edgewise Therapeutics** • Joanne M. Donovan, MD, PhD, Chief Medical Officer	Palazzo Ballroom
10:45 – 11:00 AM	**Break**		
11:00 – 11:50 AM	**Session 7: Breakout**	**Cardiac and Respiratory Management** • Frank Raucci, MD, Virginia Commonwealth University (Cardiac) • Beth McNally, MD, PhD, Northwestern University (Cardiac) • Megan Lowery, MD, University of Florida (Pulmonary) • Amy Fulford, RT, Millennium Respiratory Services	Palazzo Ballroom
		Rare Recessive LGMD Subtypes • Christina Pacak, PhD, University of Minnesota • Monkol Lek, PhD, Yale School of Medicine • Bjarne Udd, MD, PhD, Tampere Neuromuscular Center	Palazzo A
		Natural History Studies: What to Expect, and Why Should I Participate? • **Moderator:** Carla Zingariello, DO Montefiore/Albert Einstein College of Medicine • Ruby Langeslay, MPH, Virginia Commonwealth University • Dan Pope, Patient • Sujethra (Suji) Vasu, MD, Caregiver • Kevin Warf, BS, Nationwide Children's Hospital	Palazzo B
11:50 AM – 12:00 PM	**Break**		

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2025 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE
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International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

12:00 – 12:50 PM	**Lunch**	**ML Bio Updates BBP-418** *Sponsored by ML Bio Solutions*	Palazzo Ballroom
12:50 – 1:00 PM	**Break**		
1:00 – 1:35 PM	**Session 8**	**LGMD Advocacy Opportunities** **Rare Disease and Patient Roundtable Discussion with Amy Comstock Rick from the FDA** • Amy Comstock Rick, J.D., CDER's Associate Director for Rare Disease Strategy and the Director of Strategic Coalitions for FDA's Rare Disease Innovation Hub • Kathryn Bryant Knudson, Brad Williams, and Carol Abraham **Laying Out the Upcoming LGMD Advocacy: Lessons from the Rare Disease Community** • Annie Kennedy, Chief of Policy, Advocacy, and Patient Engagement, EveryLife Foundation **Call to Action 2025 LGMD Day on the Hill** • Kathryn Bryant Knudson, President, The Speak Foundation	Palazzo Ballroom
1:35 – 1:45 PM	**Break**		
1:45 – 2:45 PM	**Session 9**	**Emerging Treatments** **Stem Cells + Gene Editing** • Simone Spuler, MD, Charité — University Medicine **Repurposing Treatments** • Mohan Viswanathan, PhD, Massachusetts Institute of Technology **Development of Novel Gene Therapy for FKRP Disorders** • Jeffrey Chamberlain, PhD, University of Washington **Prime Editing** • Scot A. Wolfe, PhD, UMass Chan Medical School	Palazzo Ballroom
2:45 – 2:50 PM	**Closing Remarks**	**"We Cannot Do This Without Everyone"** • Kathryn Bryant Knudson, Brad Williams, and Carol Abraham	Palazzo Ballroom
2:50 PM	**Adjourn**		

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Featured Session

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LGMD POSTER SESSION

Saturday • July 19, 2025
5:30 P.M. • Palazzo Ballroom

Don't Miss Our First-Ever
LGMD Poster Session

At the 2025 International Limb Girdle Muscular Dystrophy Conference

The 2025 International Conference will feature a special scientific poster session showcasing 25 research studies focused on limb girdle muscular dystrophies (LGMDs). We invite you to join us at the session taking place at 5:30 P.M. Saturday in the Palazzo Ballroom, where attendees will have the opportunity to view and discuss the work of researchers who are making important strides toward advancing the development of therapies for these conditions. Posters will be presented in clear, non-technical language to encourage meaningful engagement and foster constructive relationships between attendees and researchers.

For those unable to attend in person, we plan to share a post-conference addendum with more in-depth summaries of the research presented, to be published in a future issue of *LGMD News*.

Stay Tuned!

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ORLANDO, FL

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

Clinical Research

1 ***Motor Function in LGMD R1/2A (Calpainopathy): Validation of Clinical Outcome Assessments for Clinical Care and Trial Readiness***

Authors: Meredith K. James (University of Newcastle, UK), Lindsay Alfano, Megan Iammarino, Natalie Reasch, Chris Steiner, Audrey Beale, Melissa Smith, Dionne Moat, Jassi Sodhi, Karen Wong, Emma Grover, Emma Robinson, Anna G. Mayhew, Michelle Eagle, Volker Straub, Michela Guglieri, Chiara Marini Bettolo, Robert Muni Lofra, Jordi Diaz-Manera, Linda Lowes

2 ***Progression of Walking Impairment in Patients with Dysferlinopathy Over 12 Months: Clinical Outcome Study for Dysferlinopathy (LGMD R2/2B)***

Authors: Meredith K. James (University of Newcastle, UK), Karen Wong, Mark Richardson, Emma Grover, Emma Richardson, Heather Hilsden, Anna G. Mayhew, Volker Straub, Lisa Alcock

3 ***Development of Pediatric and Adult LGMD R9/2I Disease Specific Physical Function Questionnaires Using PROMIS Item Banks***

Authors: Douglas Sproule (ML Bio Solutions), Beth Leiro, Katherine Mathews, John Vissing

4 ***Integrating Patient Insights into AB-1003 Gene Therapy Clinical Trial Design in LGMD R9/2I***

Authors: Samuel Hopkins (AskBio Inc.), Pooja Merchant, Stacey Abby, Kara Witcoff

5 ***Provisional Standard of Care Guidelines for the Diagnosis and Management of LGMD R9/2I***

Authors: Kelly Brazzo (CureLGMD2i), Katherine Mathews, Gorka Fernández-Eulate, Karim Wahbi, Robert Muni Lofra, Nicholas Johnson, Conrad Weihl, Marianne de Visser, Teresinha Evangelista, Lindsay Alfano, Linda Lowes, John Vissing, Kristin Ørstavik, Meredith James, Kate Adcock, Sam Geuens, Tracey Willis, Lone Knudsen, Erik Niks, Andreas Dybesland Rosenberger, Menno van der Holst, Elise Dupitier, Aleksandra Leijenhorst, Jean Pierre Laurent, Kathryn Bryant Knudson, Melanie Bordes, Tanya Stojkovic, Volker Straub

6 ***Using Quantitative MRI to Understand Muscle Changes in LGMD R1/2A and Becker Muscular Dystrophy***

Authors: Kelly Rock (University of Florida), Prathyusha Bellam, Donovan Lott, Rebecca Willcocks, Alison Barnard, Sean Forbes, Alexa Harris, Oneema Kamal, Julia Hinkle, Claudia Senesac, Krista Vandenborne, Glenn Walter

Demographics and Quality of Life

7 ***Quality Project for Sarcoglycanopathies: LGMD R3, R4, and R5***

Authors: B. Vola (GFB), M. Bianchi, M. Cerletti, O. Sermeryak, C. Sanchez Riera, C.C. Triki, Yvan Torrente

8 ***The Patient Experience of Living with LGMD R1/2A: Findings from a Focus Group***

Authors: Nicole LaMarca (Sarepta Therapeutics), Tamara Wyzanski, Chloe Carmichael, Catherine Bottomley, Jennifer Levy, Monique Dabbous

9 ***Understanding the Burden of Living with LGMD R9/2I and the Impact of a Potential Therapy***

Authors: Cybele Gouverneur (ML Bio Solutions), Mallory Harden, Doug Sproule, Ariel Rosen, Lucy Nelson, Kathryn Bryant Knudson, Kelly Brazzo

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Diagnostic Tools

10 ***Improving Genetic Diagnosis in LGMD Using Nanopore Long Read Sequencing***

Authors: Christine C. Bruels (University of Minnesota), Hannah R. Littel, Carrie Walls, Seth A. Stafki, Elicia A. Estrella, Lauren Brady, Mark Tarnopolsky, James J. Dowling, Carla D. Zingariello, Basil T. Darras, Partha S. Ghosh, Peter I. Karachunski, Georgios E. Manousakis, Randal C. Richardson, Louis M. Kunkel, Christina A. Pacak, Christopher Faulk, Peter B. Kang

11 ***Improving LGMD Diagnosis through Systematic Variant Resolution***

Authors: Conrad C. Weihl (Washington University), Amanda Clause and the ClinGen LGMD Variant Curation Expert Panel

12 ***MRI-Based Criteria to Differentiate Dysferlinopathies from Other Genetic Muscle Diseases***

Authors: Holly Borland (University of Newcastle, UK), Carla Bolaño-Diaz, José Verdú-Díaz, Alejandro Gonzalez-Chamorro, Sam Fitzsimmons, Gopi Veeranki, Volker Straub, Jordi Diaz-Manera

13 ***Myoguide.org: A Web-Based Portal Supporting the Analysis of MRIs for the Diagnosis of Neuromuscular Patients***

Authors: Holly Borland (University of Newcastle, UK), Carla Bolaño-Diaz, Jose Verdu-Diaz, Alejandro Gonzalez-Chamorro, Goknur Kocak, Sam Fitzsimmons, MYO-Share working group, Jordi Diaz-Manera

Disease Knowledge

14 ***Sarcoglycans are Enriched at the Neuromuscular Junction***

Authors: M. Gloriani (University of Milan), B. Cheli, C. D'Ercole, V. Ruggieri, M. Cosentino, M. Serrat Pineda, B. Blozanoska-Ochser, F. Grassi, M. Bouché, L. Madaro, Carles Sánchez Riera

15 ***Comparing Phenotypes in Limb Girdle Muscular Dystrophy Disease Progression***

Authors: Sara A. Marshall (Nationwide Children's Hospital), Megan A. Iammarino, Melissa A. Smith, Christopher L. Steiner, Natalie F. Reash, Audrey B. Knight, Meredith K. James, Stephanie M. Hunn, Shelley RH. Mockler, Heather Hilsden, Lindsay N. Alfano, Linda P. Lowes

16 ***Miyoshi Myopathy and LGMD R2/2B are the Same Disease***

Authors: Holly Borland (University of Newcastle, UK), Ursula Moore, Heather Gordish, Jordi Diaz-Manera, Meredith James, Anna Mayhew, Michela Guglieri, Roberto Fernandez-Torron, Laura Rufibach, Jia Feng, Andrew Blamire, Pierre Carlier, Simone Spuler, John Day, Kristi Jones, Diana Bharucha-Goebel, Emmanuelle Salort-Campana, Alan Pestronk, Maggie Walter, Carmen Paradas, Tanya Stojkovic, Madoka Mori-Yoshimura, Elena Bravver, Elena Pegoraro, Linda Lowes, Jerry Mendell, Kate Bushby, Volker Straub

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Research Tools

17 ***Human iPS-Derived Model to Study Sarcoglycanopathies***

Authors: B. Vola (GFB), G. Pompilio, M. Cerletti, C. Sanchez Riera, F. Gros-Louis, Yvan Torrente

18 ***The LGMD 2A/Calpainopathy Registry: A Patient-Powered Natural History Study and Trial Recruitment Tool***

Authors: Jennifer Levy, (Coalition to Cure Calpain 3) Jordan Boslego, Michela Guglieri, Ann Martin, Katherine Mathews, Michele Wrubel

Therapy

19 ***Development of Gene Therapy for FKRP Disorders***

Authors: Hichem Tasfaout (University of Washington), Theodore Ric Louie Reyes, Jeffrey S. Chamberlain

20 ***Design of Synthetic Promoters for AAV-Driven Treatment of LGMD R1/2A***

Authors: Ruby Goldstein de Salazar (UCLA), Irina Kramerova, Jesus Perez, Stephen D Hauschka, Jeffrey S. Chamberlain, Melissa Spencer

21 ***Use of Lamotrigine to Treat Painful Muscle Contractions in LGMD 1C "Rippling Muscle Disease"***

Authors: Morgan Jordan (University of Florida), Carla D. Zingariello

22 ***Developing a Modified CRISPR-Cas9 Editing Approach to Correct a CAPN3 c.550delA Mutation Associated with Limb Girdle Muscular Dystrophy***

Authors: Logan Gauthier (Yale University), Shushu Huang, Kaiyue Ma, Kenneth Ng, Katherine Koczwara, Nicholas Johnson, Monkol Lek

23 ***FORTIFY: A Phase 3 Study to Evaluate Efficacy & Safety of BBP-418 in Individuals with LGMD R9/2I, FKRP-Related***

Authors: Ada Lee (ML Bio Solutions), Tricia Blankenbiller, Amy Rainey, Lindsay Reklis, Douglas Sproule

24 ***Correcting Mutations Responsible for LGMD R2/2B (dysferlinopathy) with Prime Editing***

Authors: Camille Bouchard (University of Laval), Jing Jiang, Joël Rousseau, Jacques P. Tremblay

25 ***Developing Prime Editing Therapeutics for LGMD R9/2I***

Authors: Anya T. Joynt, (University of Massachusetts) Dongsheng Guo, Katelyn Daman, Jing Yan, Charles P. Emerson Jr., Scot A. Wolfe

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

Thank You!

2025 INTERNATIONAL LIMB GIRDLE MUSCULAR DYSTROPHY CONFERENCE
ORLANDO, FL

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THE SPEAK FOUNDATION

International LIMB GIRDLE MUSCULAR DYSTROPHY Conference

Thank You to Our Incredible 2025 International LGMD Conference Leadership Team:

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Kelly Brazzo

Jennifer Levy, PhD

Jessica Evans, PsyD

Melina Garza

Vovanti Jones, MD

John Faver

Faran Day

Dan Pope

*Special recognition to: **JP Laurent, PhD**, **Jennifer Levy, PhD**, **Brad Williams, PhD**, **Rachel DeConti**, **Kelly Brazzo**, **Carol Abraham**, and **Laura Rufibach, PhD**. We thank all of the organizations who have sponsored, with a special thank you to **LGMD Awareness Foundation** for their highest organizational sponsorship.*

Congratulations!

2025 Award Winners

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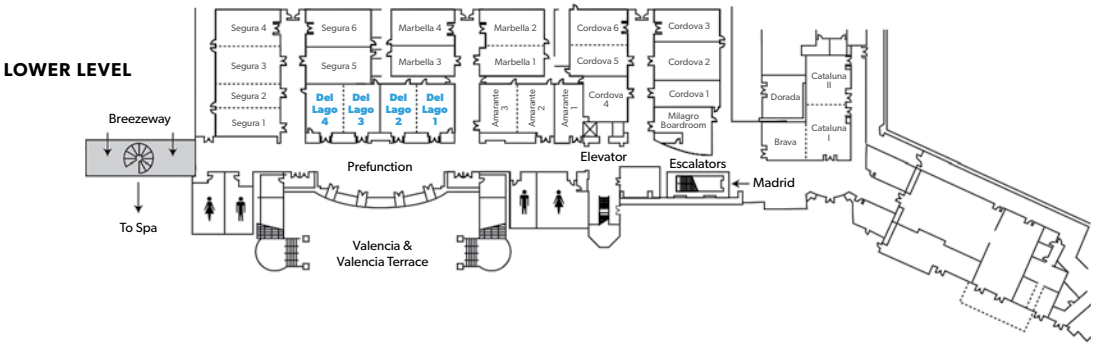

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fortify

Thank You

to the participants and clinical sites in FORTIFY, our Phase 3 study. We appreciate the entire LGMD2I/R9 community for all you continue to do on behalf of LGMD drug development.

bridgebio MLBioSolutions

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Episode 14

"Love isn't love till it's given away."

Determination has helped Lacey fulfill her dreams, but it hasn't stopped the progression of LGMD21/R9.

on rare

Check out the amazing stories of our On Rare podcast guests living with LGMD2I/R9. We thank Lacey and the other guests for sharing their perspectives.

Know Your Subtype!

bridgebio

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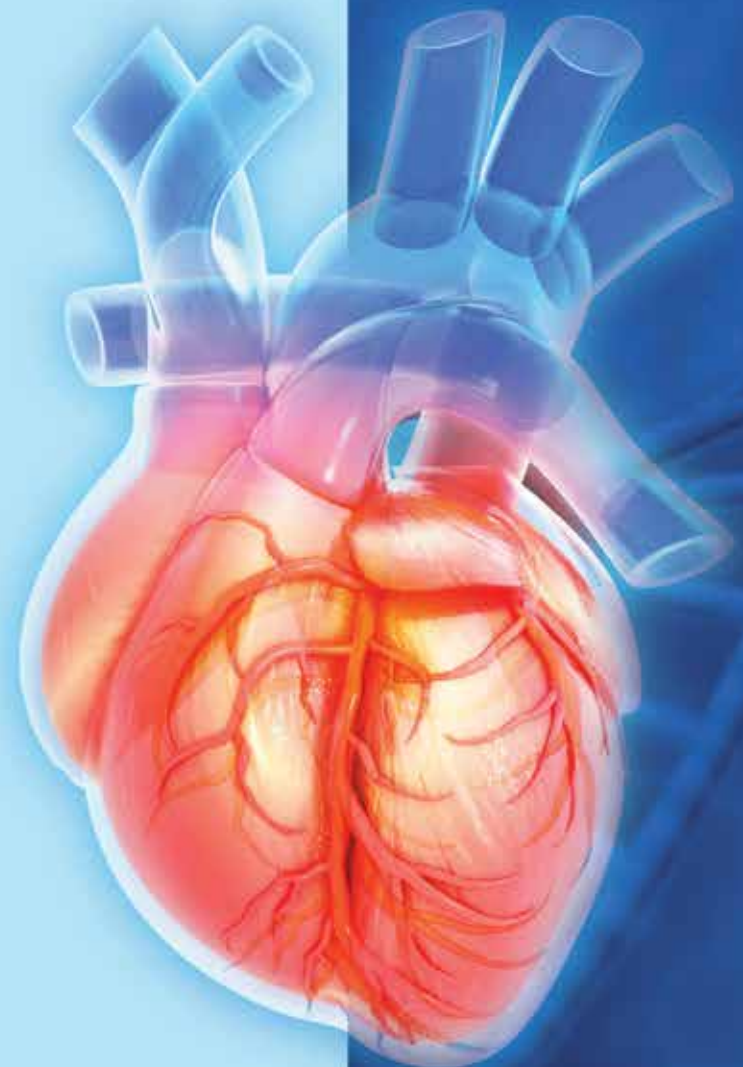
Limb-Girdle Muscular Dystrophy Type 2I/R9 (LGMD2I/R9) (FKRP-related) is a rare genetic disorder that causes progressive muscle weakness around the hips and shoulders. However, LGMD2I/R9 is one of the LGMD subtypes that can also impact the heart. This is one reason why getting genetic confirmation of your LGMD subtype is important.

Some individuals with LGMD2I/R9 may develop cardiomyopathy, weakening of the heart muscles, and conduction abnormalities (irregular heartbeats). Obtaining confirmation of your subtype will allow physicians to monitor your heart and provide optimal care.

Knowledge is power – speak with your physician about confirming your LGMD subtype and *stay proactive about your heart health!*



bridgebio MLBioSolutions
a bridgebio company

Come find us at the 2025

International Limb-Girdle Muscular Dystrophy Conference

Industry Symposium

Sunday, July 20, 2025

Palazzo Ballroom

12:00PM

3 Posters

Booth

Congratulations

Congratulations to the LGMD community on the **acceptance of a new ICD-10-CM code** for LGMD2I/R9 (FKRP-related)!

This new diagnostic code will go into effect in October, so look for more details coming later this year.

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Together we are Stronger!

Mark your calendar for the 11th Annual LGMD Awareness Day

Elevate Awareness with the LGMD Awareness Day Toolkit!

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

Join the LGMD Awareness Foundation in our mission to elevate global awareness for limb-girdle muscular dystrophy.

Together we advocate for better access to diagnosis, care, and treatment for those affected.

LGMD AWARENESS FOUNDATION

lgmd-info.org

Thank you to our sponsors:

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SAREPTA THERAPEUTICS

You're Invited to the LGMD2I/R9 Networking Event

FUNDING PROGRESS & FOSTERING COMMUNITY

CURE LGMD2i

Limb Girdle Muscular Dystrophy

curelgmd2i.org

Saturday July 19th

6:30-8:30pm

Palazzo Room B

If you are living with LGMD2I/R9 and attending the International LGMD Conference, please join us for a reception to meet and greet your LGMD2I/R9 patient community in a smaller setting.

RSVP :

Questions? Email: info@curelgmd2i.org

Sponsored by:

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LEARN MORE ABOUT PARTICIPATING IN NON-INTERVENTIONAL RESEARCH STUDIES FOCUSED ON LGMDS

JASMINE
Living with
LGMD2A/R1
(calpainopathy)

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

When you participate in non-interventional studies, like interviews and surveys, you can help increase 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:

Support clinical research decisions, such as informing clinical trial design based on lived experiences and preferences of individuals

Advance drug development for researchers and regulators, who often rely on patient-reported data to make decisions regarding drug development

Evaluate the patient's perspective of current disease management and assess the impact of potential interventions to help enable better patient-centric care

Visit LimbGirdle.com/non-interventional-studies or scan the QR code to learn more

Please note: *Participation in these types of studies will not impact your ability to enroll in future clinical trials for potential treatments. If you are eligible and participate in a non-interventional research study, you may be compensated for your time.*

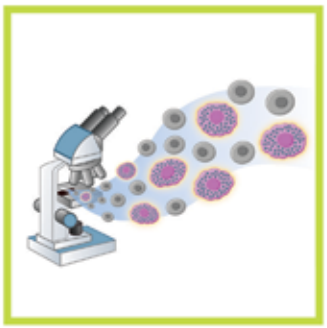
Intended for U.S. audiences only.

SAREPTA THERAPEUTICS

HOW DO SCIENTISTS MEASURE IF GENE THERAPY IS WORKING?

One important measurement is called protein expression.

PHILLIP
Living with
LGMD2B/R2
(dysferlinopathy)



What is protein expression?

Protein expression refers to the production of proteins by cells.

Gene therapy is designed to help the body express, or produce, a functioning version of the protein which is either missing or not properly working in the individual. After a person receives gene therapy, a scientist may measure protein expression to determine how much protein is being made. To measure protein expression, scientists collect a sample from the part of the body that is impacted by the disease. In the LGMDs, this may occur through a muscle biopsy. Researchers can then perform a variety of tests, like Western blot or immunofluorescence, to quantify how much of the new protein is inside the cells. This may help them evaluate whether the gene therapy is working as intended.

Why is protein expression important?

Gene therapy aims to slow or stabilize a disease, which may require tracking disease progression over time. In diseases like the LGMDs, it may take years to properly evaluate changes in muscle function.

Protein expression, which can be a biomarker or surrogate endpoint, can help predict intended clinical benefit. Regulators like the FDA may consider biomarker data when determining whether to allow a drug on market while additional studies are conducted to confirm clinical benefit.

Learn more about gene therapy research at

LimbGirdle.com

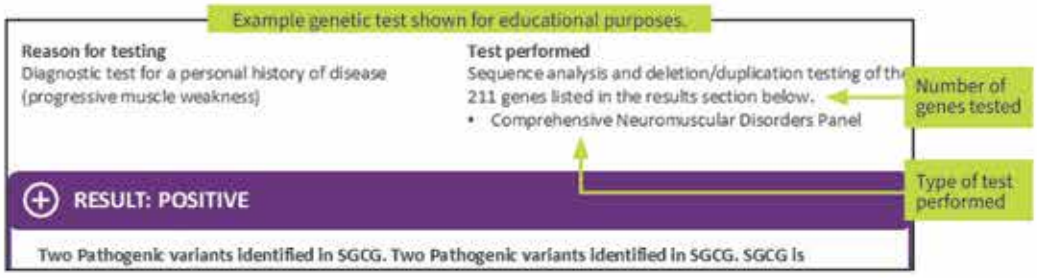
DO YOU HAVE YOUR GENETIC TEST REPORT?

You can self-advocate as a member of your care team.

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

Clinical trials for LGMDs are subtype-specific and require a genetic diagnosis. If you suspect you have an LGMD (or if your doctor made the diagnosis without genetic testing), or if your genetic test was done years ago, **it's critical to ask your doctor about genetic testing**.

What information can you learn from a genetic test report?



Test results for autosomal recessive subtypes of LGMD can be:

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 should I do next?

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.

START THE CONVERSATION

Talk to your doctor and find the LGMD support you need.

It's important to discuss your diagnosis with your doctor(s) but it can be challenging to remember everything to discuss at your appointment.

We've prepared a discussion guide that you can use, including questions like:

- Do I have a definitive (genetic) LGMD subtype diagnosis?
- Is the test result of my genetic test "Uncertain"?
- What other providers, such as cardiologists and pulmonologists, should be included as part of my care team?
- How often should I see my care team providers?

Stop by Sarepta's table at the conference to speak with our Patient Affairs team and pick up a copy of the discussion guide!

The guide is available to download on

LimbGirdle.com

SAREPTA THERAPEUTICS

Written By Ruby Langeslay

Think Global, Act Local:

How GRASP-LGMD Works Together Towards a Common Mission

The Genetic Resolution and Assessments Solving Phenotypes in LGMD (GRASP-LGMD) consortium was created in 2018 and currently includes 16 sites in the United States, Europe, and New Zealand. Its overarching mission is to hasten therapeutic development in the LGMDs.

This is done by working together on a variety of natural history studies for multiple LGMD subtypes with the goal of developing standardized outcome measures to track disease progression amongst the populations. The consortium sites collaborate with one another on data analyses, central specimen storage, academic presentations, and manuscripts as a single entity. Past studies have included participants with LGMD R1/2A through LGMD R6/2F, LGMD R12/2L, and LGMD D1/1D. There has also been interest in studies for rarer types of LGMD and other neuromuscular disorders such as Becker Muscular Dystrophy and Late-Onset Pompe Disease. The consortium hopes to expand on long-term natural history studies for all subtypes of LGMD and more sites in other states and countries to help participants access these opportunities. ■

GRASP-LGMD Principal Investigators

- Dr. Lindsay Alfano **Nationwide Children's Hospital**
- Dr. Zineb Ammous **Indiana Community Health Clinic**
- Dr. Urvi Desai **Atrium Health**
- Dr. Jordi Diaz-Manera **University of Newcastle, UK**
- Dr. Stacy Dixon **University of Colorado**
- Dr. Nicholas Johnson **Virginia Commonwealth University**
- Dr. Peter Kang **University of Minnesota**
- Dr. Doris Leung **Kennedy Krieger Institute**
- Dr. Kathy Mathews **University of Iowa**
- Dr. Tahseen Mozaffar **University of California, Irvine**
- Dr. Richard Roxburgh **University of Auckland**
- Dr. Valeria Sansone **Centro Clinico NeMO Milano**
- Dr. Jeff Statland **University of Kansas Medical Center**
- Dr. Conrad Weihl **Washington University in St. Louis**
- Dr. Carla Zingariello **University of Florida**

Active GRASP-LGMD Natural History Study

Updated: July 1, 2025

Recruiting:

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

Inclusion Criteria

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

Exclusion Criteria

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

Interested? If you'd like more information on existing and future studies, you can reach out to **Ruby Langeslay** (**Ruby.Langeslay@vcuhealth.org**) and **Jennifer Raymond** (**Jennifer.Raymond@vcuhealth.org**), who are the main points of contact for the GRASP-LGMD central coordinating center.

JAIN FOUNDATION
LGMD2B/R2 | DYSFERLINOPATHY | MIYOSHI

DYSFERLIN REGISTRY
by Jain Foundation

The dysferlinopathy community is invited to a private reception!

Who: For all attendees and caregivers of the 2025 International LGMD Conference affected by LGMDR2/2B/dysferlinopathy

When: Saturday, July 19th at 6:45 pm

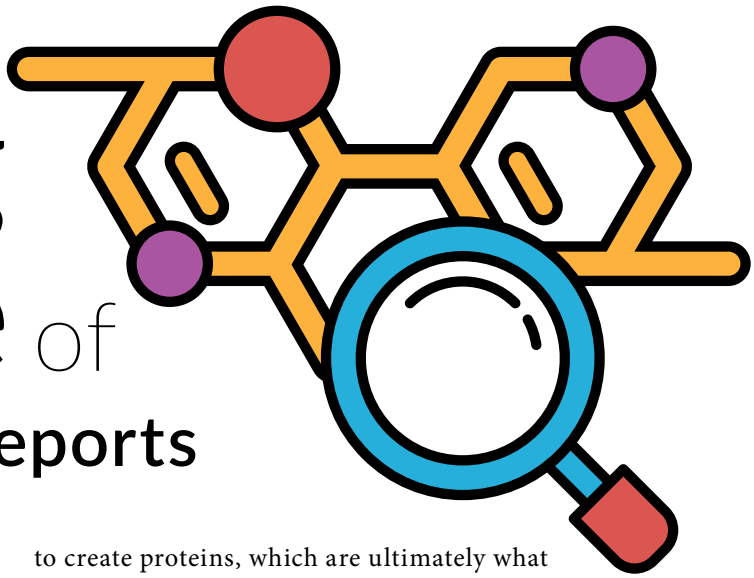
Where: Del Lago rooms 1&2 Lower level of the JW Marriott Orlando, Grande Lakes Hotel

Join the Jain Foundation, patients, family members and friends affected by dysferlinopathy for food and drinks. The LGMD Conference includes hundreds of individuals with dozens of subtypes of LGMD. This is an opportunity to get to know others with the same subtype of LGMD in a smaller setting.

RSVP at patients@jain-foundation.org or online

Written By Brianna Gross, MS, LCGC

Cracking the Code of Genetic Testing Reports



Genetic testing is an increasingly common tool for diagnosing muscle diseases, as it often allows determination of the specific cause of a person's symptoms. However, for many, the results can be difficult to interpret. Typical genetic testing reports contain a mix of technical language, classifications, and lab-specific terminology that may not be intuitive.

Knowing how to read and interpret this type of report is essential — whether you are a patient trying to make sense of your own results or a healthcare provider helping someone navigate their genetic testing report. This article breaks down the key components of a genetic testing report and explains each one in clear, accessible terms.

Before discussing a genetic testing report, it is helpful to understand genes and their roles in the human body. Humans have 46 chromosomes, which come in 23 pairs. Each pair has one chromosome inherited from the person's mother and one from their father. Our genes are located within our chromosomes, and each chromosome has on average, about 1,500 genes. Genes are like instruction manuals that provide the body with directions to create proteins, which are ultimately what makes up everything about the human body. These genetic instructions are written in an "alphabet" of four different nucleotides, which are designated by the letters A, T, C, and G. Each "word" in our genes' instructions is comprised of three letters (nucleotides). We call these three letter "words" codons; each codon specifies a particular amino acid. Amino acids are the building blocks of proteins (21 different amino acids occur in proteins) and fit together like a string of pearls. Each protein is made up of a specific sequence of different amino acids strung together (see Figure 1).

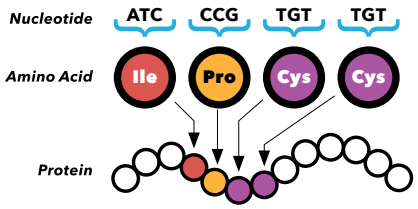


Figure 1: Genetic Instructions

Our genes are all spelled a bit differently which is why humans are different from one another. Many times, variation in the genes' instructions is harmless and well-tolerated by the body. For example, there are many variations influencing skin color, hair color, and bone structure.

However, some changes can be harmful if they alter the genetic instructions too much. When this happens, the resulting protein may not function properly — or may not be made at all — leading to disease symptoms.

These symptoms are what may ultimately lead a healthcare provider to order genetic tests to try to determine the cause of the person's disease. They can then create a healthcare plan that will ensure the person is being treated appropriately and being offered proper medications, treatments, and monitoring.

To better understand how to interpret a genetic testing report, this section will take a closer look at a real-world example and break down the report's components (see Figure 2). Recent genetic reports produced in the US will usually contain most sections of this example; however, reports from elsewhere may look quite different, although they will have some of the common elements.

1 Demographics

Always ensure that your name and date of birth (DOB) are accurate before getting to the "meat" of the report. In the demographics area of the report, you will also be able to see the name of the test, your ordering provider, the institution that your provider is associated with, the date your sample was collected, the date the report was printed, and laboratory identifiers or accession numbers.

2 Result Summary

Often, the laboratory will provide their own brief interpretation of the results and put a small summary together before inputting the actual genetic changes found. They may label the test results as positive or diagnostic, which means that the lab found a genetic change that is known to cause symptoms in a person. They may label the test results as uncertain or unknown, meaning they found a genetic change but are not certain that the genetic change is contributory to symptoms. Lastly, they may label it as negative or non-diagnostic, which means they did not find a genetic change. In the example report, the result summary identifies the report as positive.

3 Results

The results section is where you will find the exact genetic change that was identified. The laboratory will list the gene that was identified to have the variation within it. In the example report, the *gene* listed is *FKRP*, which is associated with LGMD R9/2I. *Variant* will list the exact spelling change that was found in the gene. The variant is listed here in two different ways. There is a c. and a p., which both have a combination of numbers and letters. The c. indicates the nucleotide change in the DNA sequence. In this example, the nucleotide change is c.826C>A. This means that at the 826th letter or nucleotide, this individual has an "A" where most people have a "C." The p. is the amino acid change in the protein. In the example report, the amino acid change is p.Leu276Ile. This means that at the 276th amino acid in the FKRP protein, where most people have a leucine (Leu), this individual has an isoleucine (Ile). Amino acids can be written in one of two abbreviations — a three-letter abbreviation, like Ile, or a single letter abbreviation, like I. *Zygosity* refers to the number of copies of a specific spelling change a person possesses. If a gene is on an autosome (chromosomes 1-22, which everyone carries two copies of, one from each parent), the zygosity will be either *homozygous* or *heterozygous*. Homozygous means that both copies of the gene (one from each parent) have the same identical spelling variation. Heterozygous means that only one of the two copies of the gene has that spelling variant. However, in a recessive disease, the other copy often has a different variant; individuals with two different variants in the same gene are referred to as *compound heterozygous*.

The report will also include a variant *classification*. All variants in a gene are categorized based on what impact they have on the body (see Figure 3). Laboratories use a 5-point scale to identify a genetic variation based on the impact to the body. Variants can be classified as benign, likely benign, a variant of uncertain significance (VUS), likely pathogenic, and pathogenic, meaning disease-causing. There are specific criteria that laboratories use to classify variations. Variants that are classified as pathogenic contribute to symptoms in a person with certainty, while variants that are benign are known not to cause disease. Reports are generally summarized by the laboratory as

1

Name: Julles Ritten
DOB: 11/07/2018
Hospital Identifier: 268890
Accession Number: PTC1995
Sample Type: Buccal

Sample Collection Date: 01/31/2025
Sample Accession Date: 02/02/2025
Ordering Institution: IBC
Report Date: 02/20/2025
Test Performed: Neuromuscular Disorders Panel

The Genetic Testing Company

2

⊕ RESULT: POSITIVE

A homozygous pathogenic variant was identified in FKRP. This variant in FKRP is associated With autosomal recessive Limb Girdle Muscular Dystrophy Type 2I (LGMD2I).

3

Gene	Variant	Zygosity	Classification
FKRP	c.826C>A p.Leu276Ile	Homozygous	Pathogenic

4

Clinical Summary:

The *FKRP* gene is associated with autosomal recessive dystroglycanopathy spectrum disorders. Limb Girdle Muscular Dystrophy type 2I [LGMD2I/type R9 (LGMDR9)] (OMIM; Brockington et al. Hum. Molec. Genet. 10:2851-2001), Merosin-Deficient Congenital Muscular Dystrophy type 1C (MDC1C), Muscle-Eye Brain disease (MEB) and Walker-Warburg Syndrome (WWS). LGMD2I/R9 is characterized by progressive muscle weakness and wasting, particularly affecting the proximal muscles with a large phenotypic range and age of onset (Strensland et.al. 2010). Individuals with MDC1C most often have an early age of onset, an inability to walk, enlarged calf muscles, contractures and intellectual disability, and can often have brain abnormalities. They may also have respiratory failure, calf and quadriceps hypertrophy, and macroglassia (OMIM 606612; Brockington et.al.AM. J. Hum. Genet. 69:1198-1209, 2001). Individuals with MEB and WWS are born with muscle weakness and structural abnormalities of the brain and eyes. Patients with WWS often die at birth or shortly thereafter due to complications from severe CNS structural abnormalities.

5

Variant Summary:

***FKRP* c.826C>A, p.Leu276Ile: homozygous; pathogenic**

The c.826C.A, p.Leu276Ile variant in FKRP is located in exon 4/4. This variant is considered a known founder mutation in the Hutterite population (Frosk 2005 PMID:15580560). This variant has been reported in population databases and is present in 0.2% if European chromosomes (2820/1135906 chromosomes). This variant has also been reported in ClinVar (Variation ID: 4221) and is reported in both compound heterozygous and homozygous states in many individuals (PMID: 11741B28; Walter 2004 PMID: 15060126, Computational prediction tools and conservation analyses suggest this variant may impact the protein. Mice animal models have shown Limb Girdle Muscular Dystrophy (Krag 2015 PMID:26574668). In summary, this variant meets criteria to be classified as pathogenic for autosomal recessive FKRP-related muscular dystrophy. Criteria applied: PM3_Very Strong, PS3, PP3.

Figure 2: Genetic Testing Report

positive if the variant is pathogenic or likely pathogenic, and negative if the variant is benign or likely benign. VUS means that the lab recognizes a spelling variation, but there is not enough data to support a positive or negative classification. This might be due to factors such as frequency in the general population, if the variation is rare, or lack of information due to few functional studies on this particular variation. If a VUS is a classification on a genetic testing report, it is vital to go to an expert to see if there is some data that is suggestive of either a pathogenic or benign classification, as well as having an expert perform a clinical exam to see if the condition that the gene is associated with matches with the individual's clinical picture, also referred to as phenotype. Expert healthcare professionals have the ability to help shed further light on a VUS.

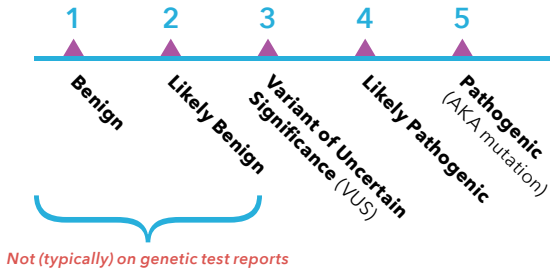


Figure 3: Variant Classification

Sometimes, the results will include the inheritance pattern of the condition that the gene is associated with. All genetic conditions can be inherited in different ways, the most common being autosomal dominant inheritance, autosomal recessive inheritance, and X-linked inheritance patterns. Autosomal dominant inheritance is where only one of the two copies of a gene having a pathogenic variant is enough for a person to be symptomatic with the condition. The most recent system of naming LGMDs designates these subtypes starting with the letter D. Autosomal recessive inheritance is where both copies of a gene must possess a pathogenic variant for a person to be symptomatic with the condition. For LGMDs, these subtypes are labeled starting with the letter R. In some cases, a particular gene can be associated with both autosomal dominant and recessive inheritance, depending on the specific genetic variation. For instance, mutations in the Calpain-3 (CAPN3) gene cause both LGMD R1 with recessive inheritance, and LGMD D4 with dominant inheritance. X-linked inheritance means the genetic variation is located on the X chromosome, causing different inheritance patterns for genetic females and males — for example, in Duchenne Muscular Dystrophy.

4 Clinical Summary

A clinical summary of the genetic condition that a gene is associated with will usually be listed. This will detail some of the clinical picture of the condition, including the name of the genetic condition and common symptoms. Because this part of the report can carry the most emotional weight for patients, it may be helpful to have support when reviewing it. It's also important to remember that while multiple conditions linked to the gene variant may be mentioned, the patient will only be diagnosed with the one that matches their specific symptoms and test results.

5 Variant Summary

A variant summary is always listed on a genetic testing report and details specific facts about the variant. This includes location, frequency, whether it has been reported to cause disease, functional prediction models, whether it has been reported in medical literature, and the classification of the variant.

Genes Analyzed

The genes that were specifically looked at for errors will be included in the "Genes Analyzed" section. This is specifically for single gene or genetic panel tests. Whole exome sequencing and whole genome sequencing will not include this information on their genetic testing reports. If this is the case, it is always possible to contact the laboratory to confirm whether a specific gene was looked at fully and would have been reported on.

Methods

The methods section will detail the type of technology that the genetic testing used. This is mostly helpful for clinicians, especially when genetic testing has been performed, but no explanation for an individual's symptoms has been found.

Testing Limitations

The testing limitations section explains the technology used for testing and what each laboratory is specifically validated for. Information is especially useful for clinicians when a person remains undiagnosed despite having clear symptoms of a genetic condition.

References

The references section includes all publications that have been utilized for the report.

Genetic testing reports contain a lot of information, much of it in very technical language, and can be quite overwhelming to receive. Familiarity with the structure and terminology used in a report can empower individuals to better understand their health and make informed decisions. Whether you are navigating these reports as a patient, family member, or healthcare provider, a clear understanding of what the results mean and how they fit into the larger clinical picture is essential for moving forward with proper healthcare management. ▪

Brianna N. Gross will be participating in a Q&A Session with Genetic Counselors in the LGMD conference.

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

Lisa, living with LGMD2I/R9

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

Find out if you qualify today

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

We invite you to learn more

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

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

AskBio
A Bayer Cell & Gene Therapy Platform Company

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

Our mission
is to accelerate and influence
the path to a cure.

Make a Difference

LGMD-1D DNAJB6 Foundation
lgmd1d.org

Proud *Friend of LGMD* Sponsor

We are proud to stand with the global LGMD community as a *Friend of LGMD* Sponsor for the 2025 International Limb-Girdle Muscular Dystrophy Conference.

By supporting this important event, we help amplify patient voices, fuel meaningful connections, and drive forward the mission of improving lives for LGMD2D/R3 patients.

Together, we move toward a future of continued hope and progress for all impacted by LGMD.

LGMD2D Foundation

Limb-Girdle Muscular Dystrophy

The LGMD2D Foundation is a registered 501(c)3 non-profit foundation providing research through funding to help expedite effective treatments and a cure for those living with Limb-Girdle Muscular Dystrophy, type 2D/R3.

We provide education, advocacy and support to individuals and families living with LGMD2D.

Our International Patient Registry is free to join and is the only database for patients afflicted with LGMD2D.

For more information and to join our community, visit us at LGMD2D.org

Follow us:

FUNDING PROGRESS & FOSTERING COMMUNITY

CURE LGMD2i

Limb Girdle Muscular Dystrophy

curelgmd2i.org

Celebrating 15 years!

We need your help to support the next generation of gene editing treatments. Donate $15 (or more) today for the 15th year of CureLGMD2I (R9) Foundation:

Donate here »

What have we accomplished?

- Funded 6 Advocacy/Support Programs
- Sponsored 18 Scientific Conferences
- Supported 9 Awareness Programs
- Co-funded 20 Scientific Research Grants
- Supporting the International FKRP Registry
- Invested in 4 Biotech Industry Startups
- THREE Ongoing Clinical Trials

Want to get involved in making a difference?

We need advocates to help support drug development for rare diseases like the LGMDs.

Get involved »

CONNECTING FOR A CURE

CURELGMD2i

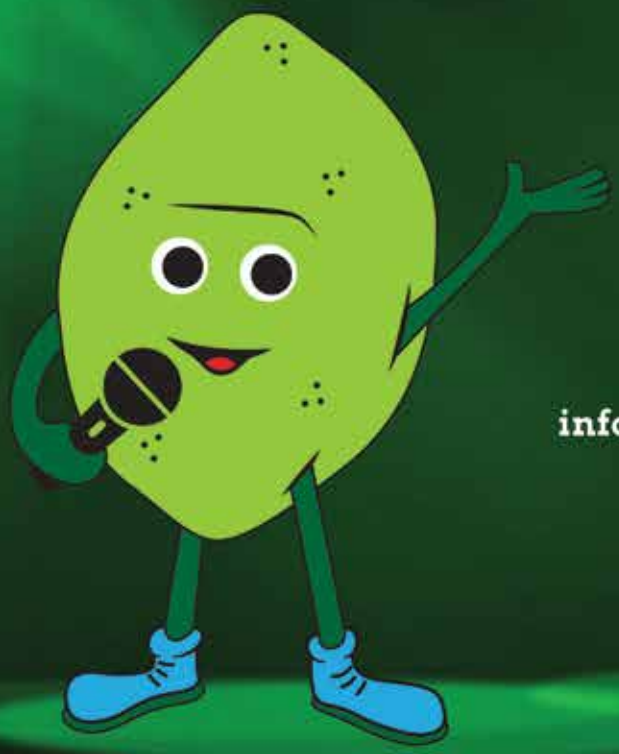
Limb Girdle Muscular Dystrophy

Save the date:

Get our newsletter for info on upcoming 2025 Connecting for a Cure events.

Sign up »

Share Your LGMD Spotlight Story



Your story has the power to inspire, inform, and connect others in the LGMD community—and we want to hear it.

Whether you're living with LGMD, caring for someone who is, or contributing to care, research, or awareness — your experience matters.

Submit your story and join us in celebrating the journeys, challenges, and triumphs that make the LGMD community so extraordinary.

Together We Are Stronger

SEPTEMBER
30

Save the Date for LGMD Awareness Day!

Join us for the 11th year of amplifying voices, sharing stories, and driving change.

Visit lgmd-info.org/awareness-day to learn more today!

LGMD
AWARENESS FOUNDATION

Together, we can advocate, educate, and celebrate — one story at a time.

Visit our website to share your spotlight today.

lgmd-info.org/spotlights

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

Global Advocacy Meeting

We Are Interested In Hearing From You!

All LGMD organizations outside the USA are invited to attend this meeting.

October 17th 12:00 pm ET

Sign Up Today to Attend!

TheSpeakFoundation.com/advocacy

The SPEAK Foundation