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LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

Make Your Voice Heard

Upcoming EL-PFDD Meeting Will Be a Historic Opportunity for the LGMD Community

THE SEARCH FOR RARE FORMS OF LGMD

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VOLUNTEER BIOCURATORS NEEDED

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The SPEAK Foundation

2022 LGMD GLOBAL ADVOCACY SUMMIT
LIMB GIRDLE MUSCULAR DYSTROPHY

MAY 20, 2022

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WWW.MALONE-MEDIA.COM/VIDEOS/LGMDSUMMIT

GET READY FOR THE

2022 LGMD GLOBAL ADVOCACY SUMMIT

LIMB GIRDLE MUSCULAR DYSTROPHY

Brought To You By The SPEAK Foundation

A FIRST FOR THE LIMB-GIRDLE MUSCULAR DYSTROPHY COMMUNITY

ARE YOU AN INDIVIDUAL LIVING WITH LGMD?

JOIN US for the virtual **2022 LGMD Global Advocacy Summit**, where you, as the patient, will have the exclusive opportunity to interact directly with the pharmaceutical companies that are designing clinical trials and developing treatment drugs for LGMD.

AT THE SUMMIT YOU WILL:

- Learn about numerous upcoming clinical trials scheduled for 2022 + 2023.
- Learn about the regulatory drug process in Europe and in the United States.
- Share your voice with the invited companies so they can hear what matters to you most.

YOU DON'T WANT TO MISS IT!

MAY 20, 2022 | 12:00 PM – 7:00 PM EASTERN DAYLIGHT TIME (EDT)

WATCH THE LIVESTREAM EVENT AT

WWW.MALONE-MEDIA.COM/VIDEOS/LGMDSUMMIT

LGMD News

Vol 2 / Issue 2

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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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We want to thank **Julia Pokrovskaya**, a fellow LGMD warrior, for her painting to the right.

We Can All Make a Difference!

Many of us in the LGMD community feel burdened by the lengthy process of drug development. This frustration has motivated many of us to fierce activity. However, we can be more effective if we collectively find ways to actively join the process to find treatments. The 2022 LGMD Global Advocacy Summit and the Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting with the FDA and other stakeholders are both great opportunities to get involved this year.

How do these opportunities make a difference? When we hold events for the LGMD community, stakeholders observe several things by our participation. For example, the amount of patients involved helps them to determine if this disease is worth investing in, and the degree to which we are organized as a community informs them about our preparedness for clinical trials. Consequently, our failure to collectively represent communicates an unfavorable message.

Sometimes it can feel like a treatment will never get here. But significant progress is being made! Just this past month, Phase Two results were released for LGMD2I/R9 demonstrating that the new ribitol treatment works! Additionally, in the last month, I have seen two new companies share that LGMD is in their pipeline. So, we must persevere.

We at the Speak Foundation want to do even more. This is why in the second half of 2022, we are starting a fundraising campaign to raise funds for research and advocacy for ALL forms of LGMD. We want to help accelerate treatments, so we are throwing a wide net to researchers in the US to find new innovations. We cannot just wait on one approach.

Join us now in representing an energized LGMD community. The 2022 LGMD Global Advocacy Summit is coming next month, on May 20th. We invite you to partner with us for this virtual event and all of the opportunities to follow, because together we are stronger! ▪

Kat Bryant Knudson

Kathryn Bryant Knudson
Editor In Chief

Join us now in representing an energized LGMD community.

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International Consortium of LGMD Organizations

United States

The Speak Foundation
Uniting the entire LGMD community
TheSpeakFoundation.com

Beyond Labels & Limitations
Funding research for LGMD 2A/R1 and educating on its disease course
BeyondLabelsLimitations.com

Breathe with MD
Educating and raising awareness about breathing muscle weakness in neuromuscular disease
BreatheWithMD.org

CamronsCure
Funding research for LGMD 2S/R18
CamronsCure.com

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

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

Kurt + Peter Foundation
Funding research for LGMD 2C/R5
KurtPeterFoundation.org

LGMD Awareness Foundation
Join us for LGMD Awareness Day
LGMD-Info.org

LGMD-1D DNAJB6 Foundation
Representing LGMD 1D/D1 and DNAJB6 subgroup
LGMD1D.org

LGMD2D Foundation
Funding research for LGMD 2D/R3 and educating patients and physicians
LGMD2D.org

LGMD2I Research Fund
Funding research for LGMD 2I/R9 and educating the patient community
LGMD2IFund.org

LGMD2L Foundation
Representing the LGMD 2L/R12 Anoctamin5-related community
LGMD2L-Foundation.org

Team Titin
A consortium of scientists and affected community members for LGMD2J/R10 Titin-related
TitinMyopathy.com

The Jain Foundation
Funding research for LGMD 2B/R2 and educating the patient community
Jain-Foundation.org

Argentina

ADM Argentina Muscular Dystrophy LGMD Group
Funding research for neuromuscular diseases
ADM.org.ar

Australia

Daniel Ferguson LGMD2A Foundation
Funding research for LGMD 2A/R1 and educating the patient community
DFFoundation.com.au

France

"GI LGMD"/LGMD Patient Group of AFM-Telethon
Focusing on all subtypes of LGMD, supporting research and educating the patient community
LGMD.AFM-Telethon.fr

Italy

Conquistando Escalones Association
Funding research for LGMD 1F/D2
ConquistandoEscalones.org

"GFB ONLUS"/ Family Group of Beta-Sarcoglycanopathy
Representing the LGMD 2C/R5 Gamma Sarcoglycan-related, LGMD 2D/R3 Alpha Sarcoglycan-related, LGMD 2E/R4 Bèta-Sarcoglycan-related, and LGMD 2F/R6 Delta-Sarcoglycanrelated communities
Beta-Sarcoglicanopathy.org

Gruppo Cingoli of UILDM – Unione Italiana Lotta alla Distrofia Muscolare
Focusing on all subtypes of LGMD, raising awareness and providing support for the entire Italian community.
UILDM.org

Italian Association Calpain 3
Funding research for the LGMD 2A/R1 Calpain3-related community
AICA3.org

Japan

Patients' Association for Dysferlinopathy Japan
Representing the Japanese and International LGMD 2B/R2 Dysferlin-related and Miyoshi Muscular Dystrophy 1 (MMD) communities
PADJ.jp/index.html

Netherlands

Stichting Spierkracht
Raising awareness and supporting the LGMD2D/R3 Alpha Sarcoglycan-related community
StichtingSpierkracht.com

Spain

Conquistando Escalones Association
Funding research for LGMD 1F/D2
ConquistandoEscalones.org

Proyecto Alpha
Funding research for LGMD 2C/R5 Gamma Sarcoglycan-related, LGMD 2D/R3 Alpha Sarcoglycan-related, LGMD 2E/R4 Bèta-Sarcoglycan-related, and LGMD 2F/R6 Delta-Sarcoglycanrelated
ProyectoAlpha.org

Jeffrey M. Statland, MD

Professor of Neurology
University of Kansas Medical Center Research Institute

Meet the Expert

Jeffrey M. Statland, MD is Professor of Neurology at University of Kansas Medical Center in Kansas City, Kansas. His research background has centered primarily on describing the natural history of and response to therapy for inherited neuromuscular diseases. His specific research interest over the last six years has been preparing for clinical trials in muscular dystrophy. He is Director of the KUMC Muscular Dystrophy Association clinics, Co-Director for the FSHD Clinical Trials Research Network, and PI and Clinical Liaison for the GRASP-LGMD research network.

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Q

I am 32 years old and have LGMD2B/R2. My condition is very weak, as I cannot walk, stand, or move my hands well. Do you foresee a treatment for LGMD2B/R2? In the meantime, is there a supplement I can take to help me with my muscle weakness?

A

I think they are working on strategies to replace the dysferlin gene using retro-virus-delivered gene replacement drugs. This approach might stop further progression and allow some muscles to heal but is unlikely to help muscles that are already very weak. There are other research groups, however, who are considering cell-based or other approaches for drugs which may help with muscle growth or regeneration. I think we likely need both. But it may be many years before we see them available to prescribe. Many of my patients try supplements. There is no supplement that I think currently has enough information for me to recommend using it specifically. Many people consider antioxidants or energy carriers. Generally, I will support trying a supplement if I do not think there is a safety concern. I would discuss this with your doctor.

Q

What are the expected long-term side effects of COVID on LGMD patients, particularly those infected by the Omicron variant?

A

We do not know if there are any long-term consequences specific for LGMD. I tell patients to make sure they are vaccinated. If their breathing is affected, then I would consider them at high risk for complications from COVID-19 infection, so they should be sure to wear a mask indoors around other people, maintain social distance, and get the vaccine or booster per CDC guidelines.

We did conduct a survey in 2020-21 of muscular dystrophy patients, including LGMD, regarding the COVID-19 pandemic. (It can be read at **ncbi.nlm.nih.gov/pmc/articles/PMC8242695/**.) There is no current evidence that someone with LGMD would be more likely to catch COVID or have long-term side effects compared to non-LGMD individuals. That being said, the survey was prior to the Omnicron variant and we are still learning more about COVID-19 (and LGMD) every day.

Q

I have been diagnosed with LGMD2I/R9. I am homozygous with two diseased genes. My daughter is 20 years old, and she is heterozygous, but since she was little, she has had difficulties and muscle pain like mine. Doctors tell me that it is impossible for her to be sick with just one gene. Do you think that healthy carrier patients can express the disease?

A

What you are describing would be a manifesting carrier for LGMD2I/R9. I have not seen this, but I never ignore symptoms in my patients. If you are followed by a neurologist for muscular dystrophy, he or she can examine your daughter to assess her strength, and if anything seems abnormal, can order additional testing (for example, a serum CK or electromyography).

Q

Will taking Urolithin A help those of us with LGMD?

A

It is too early to tell. When muscle cell cultures grown in a lab were exposed to it, or when it was fed to mice who have mutations to mimic Duchenne muscular dystrophy, it suggested Urolithin A might be beneficial. But further studies would be required, including clinical trials to make sure it is safe and helpful for people with LGMD.

Q

Will any of the treatments in development reverse muscle damage or will they only retard or arrest it?

A

The disease-targeted therapies really target the mechanism which causes muscle damage. To the degree someone still has muscle left, it may be able to regenerate. However, there are several classes of drugs meant to either reduce the workload of muscle, encourage muscle growth, or improve performance. Additionally, there are stem cell therapies which may be protective or help muscle growth. I am encouraged that companies are pursuing these therapies, as ultimately, we may need both — a drug to target the disease mechanism as well as a drug to help with regrowth.

Q

Does LGMD-D5 impact bone density?

A

There is preclinical evidence in mice that mutations in the collagen VI gene might affect bone, but I don't know if this is the case for humans. Any muscle disease can potentially impact bone density. One thing that maintains our bone density is physical activity and weight loading. The bone density of someone who is too weak to remain physically active can be impacted. Your doctor can order a bone mineral density study or check your vitamin D levels and provide supplementation, if needed.

"Additionally, there are stem cell therapies which may be protective or help muscle growth. I am encouraged that companies are pursuing these therapies, as ultimately, we may need both — a drug to target the disease mechanism as well as a drug to help with regrowth."

Question

I encourage everyone to get involved in the ongoing trial preparedness studies if they are near a recruiting center and meet the inclusion criteria. It takes everyone working together to help make these treatments become available.

Have a Question for Our Experts?

Send Questions To:
ContactUs@TheSpeakFoundation.com

Q

Does the severity of muscle weakness associated with LGMD1B decrease with each generation? My older sibling has worse mobility than me, and my daughter, 39, just had an ICD implant and shows no sign of weakness yet.

A

As the nomenclature is changing, I am going to assume you mean your family has an LMNA mutation. I think there is considerable variability from one generation to the next in laminopathies, but do not think there is actual attenuation. Everyone's progression is different, even those within the same family (and same mutation). This is something we are currently researching.

Q

Can we not speed up the mechanism for cellular repair to minimize muscle loss and preserve functionality for an individual with LGMD? For example, are there not any drugs/super drugs/ nano molecules that can prevent inflammation and/or delay cell death to prolong muscle life?

A

We cannot do it in a convincing fashion yet, but many companies are pursuing compounds which are muscle-protective, anti-inflammatory, or meant to promote muscle growth. It is encouraging to me to see all the creative ideas being pursued in this area, but we will still need clinical trials to help us know if these compounds are safe and effective. In the meantime, I encourage everyone to get involved in the ongoing trial preparedness studies if they are near a recruiting center and meet the inclusion criteria (e.g. the GRASP-LGMD study at **Clinicaltrials.gov/ct2/show/NCT03981289?cond=lgmd-&draw=2&rank=10**). It takes everyone working together to help make these treatments become available.

Q

How will Calpainopathy change my muscles and body over time?

A

LGMD2A/R1 is a chronic, progressive condition, meaning it changes over time. The pace of that change can vary considerably from one person to the next. As time passes, the absence of calpain protein makes the muscle more susceptible to damage, and eventually, enough damage builds up that you experience this as weakness. Like with any wound, the body sends in cells to help clean up the damage, and like a scar, you can see fibrosis form in the muscle. Over long periods of time, the muscle may become replaced with fat and connective tissue. Therefore, we are working hard to better understand LGMD2A/R1 and develop therapies. I do think it is a hopeful time and we will see clinical trials in the next five years. In the meantime, for chronic, progressive conditions like LGMD2A/R1, small adaptations to your behavior now can potentially provide benefits. I would work with your neuromuscular doctor and physical/ occupational therapist to develop a personal plan that is right for you.

Q

How often should patients with LGMD be checked for heart and/or respiratory function during the winter season and through the COVID pandemic? Also, is there a risk to LGMDs with the flu shot?

A

The risk of having cardiac or lung involvement is different based on the LGMD mutation you have. Cardiac involvement is not common (except for some LGMDs with mutations in sarcoglycan or FKRP). Breathing problems are more frequent (but also depend on the mutation) and can affect your diaphragm or may become worse as your body weakens or loses mobility. You should work with your muscular dystrophy doctor to determine what your personal risk is and how often screening would be appropriate. If you are having specific symptoms, let your doctor know. I do not check breathing or cardiac function more frequently because of the pandemic, but I do think getting vaccinated is important. I also recommend people get the flu shot, especially if they have difficulties with their heart or respiratory function. ■

Contributed by Michaela Walker
Project Manager, Department of Neurology
KU Medical Center, The University of Kansas

> *You should work with your muscular dystrophy doctor to determine what your personal risk is and how often screening would be appropriate.*

The Search for Rare Forms of LGMD

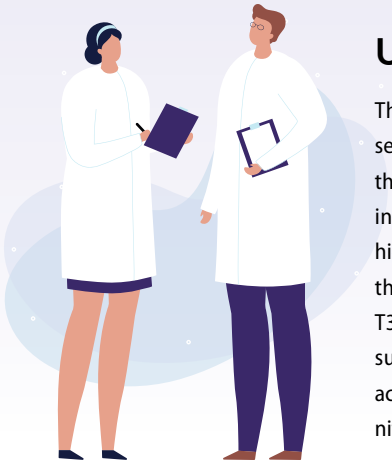
Having a full diagnosis is vital to participating in future trials and being eligible for new treatments that are approved by the US Food and Drug Administration.

"Be kind. Pursue excellence. Make a difference." These bold statements belong to the University of Minnesota Medical School's vision statement. Dr. Peter Kang, a pediatric neurologist and neuromuscular specialist at the university, is truly living out these words in his everyday work to help children and adults with muscular dystrophy (MD) get the answers they need.

It was during Dr. Kang's research training in the genetics laboratory of Louis M. Kunkel, PhD at Boston Children's Hospital in the 2000s that his interest in genetic dilemmas in MD was sparked. He first pursued gene expression studies in skeletal muscle, teasing out which genes are active in human skeletal muscles under different conditions. Before long, he became drawn to diagnostic dilemmas, both in the laboratory and in the clinic. The overarching problem in both settings centered around patients with MD whose cases were resistant to genetic explanations. He saw how frustrating it was for patients to have incomplete diagnoses, and with the help of Dr. Kunkel, Dr. Basil Darras, and Elicia Estrella, a genetic counselor, Kang began to gather these cases and analyze them systematically. He continued this work when he moved to the University of Florida in 2013, and more recently when he moved to the University of Minnesota in 2021.

Kang says, "Not having a complete genetic diagnosis can be very frustrating for a patient. With various natural history studies and clinical trials occurring for rare diseases, including muscular dystrophy, having a full diagnosis is vital to participating in future trials and being eligible for new treatments that are approved by the US Food and Drug Administration (FDA)."

In addition to treating patients with neuromuscular conditions, Dr. Kang is also Director of the Paul and Sheila Wellstone Muscular Dystrophy Center (MD Center) at the University of Minnesota. Founded in 1995, the MD Center coordinates research, training, and clinical care for a broad range of neuromuscular diseases including muscular dystrophy,



University of Minnesota Medical School

The MD Center has significant clinical, educational, outreach, and research footprints in the state of Minnesota and several neighboring states. For the past 10 years, the MD Center has co-sponsored the Annual Muscle Symposium that attracts more than 100 researchers and participants and features distinguished speakers that are recognized internationally. For the past 15 years, the MD Center has hosted the annual Greg Marzolf Jr. Symposium, which highlights the work of undergraduate students who work in the field of muscular dystrophy research. Additionally, the MD Center faculty members hold numerous National Institutes of Health (NIH) grants, including the largest T32 training program in the country for muscle research, supporting predoctoral and postdoctoral trainees pursuing laboratory and clinical research studies. The MD Center also hosts three annual community engagement activities: Laboratory Day, Family Camp, and a Community Picnic. These events provide opportunities for community members to connect with each other and with scientists, physicians, students, and trainees at the MD Center.

> **Once I became involved with the LGMD patient community, I realized this was a place I was supposed to be. This is a wonderful community to be part of, and there is a great need for these diseases to receive more attention from the medical and scientific fields.**
>
> **—Peter Kang, MD, FAAN, FAAP**

amyotrophic lateral sclerosis (also known as Lou Gehrig's disease), spinal muscular atrophy, inherited neuropathy, and congenital myopathy. The MD Center has more than 30 affiliated faculty members and dedicated staff, including an administrator, regulatory specialist, research study coordinators, a physical therapist, and a research genetic counselor.

"Once I became involved with the LGMD patient community," Kang says, "I realized this was a place I was supposed to be. This is a wonderful community to be part of, and there is a great need for these diseases to receive more attention from the medical and scientific fields."

Dr. Kang has used a range of genetic tools to study unresolved LGMD cases over the years. These tools have included Sanger sequencing, linkage analysis/homozygosity mapping, exome sequencing, genome sequencing, and most recently, a newer form of genome sequencing known as long read sequencing, or third generation sequencing. His numerous publications on these topics have included studies on the power of exome sequencing in the diagnostic approach for LGMD, the association of the muscle disease gene *PYROXD1* with LGMD, and the discovery in 2021 of a novel muscular dystrophy gene, *JAG2*. Along the way, he has engaged with many patients

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Neurogenetics@umn.edu

Med.umn.edu/mdcenter

Through this research, it is certain that Dr. Kang will continue to make a difference for the LGMD community, one diagnosis at a time.

and families affected by LGMD in his clinic and at the 2019 National LGMD Conference and 2021 International LGMD Conference, programs of The Speak Foundation (TSF). He also serves as the Medical Editor for *LGMD News* magazine and the Scientific Advisor to TSF. His work is taking on an even greater sense of urgency as novel therapies are being developed and tested for LGMD.

Dr. Kang's team is currently using cutting edge genomic techniques to study children and adults with LGMD who have incomplete or absent genetic diagnoses. Among numerous members of his laboratory over the years, Christine Bruels, Hannah Littel, Audrey Daugherty, and Seth Stafki are presently dedicating significant effort to this project. The testing, for which there is no charge for participation or travel required, is conducted on a research basis, and his team then discusses how any relevant findings can be confirmed and relayed to patients. Kang says, "I feel that it is important for as many LGMD patients as possible to be fully diagnosed, so that they can have options such as volunteering for clinical trials or being eligible for new therapies when they are approved by the Federal Drug Administration (FDA)."

If you have LGMD with an incomplete or absent genetic diagnosis, you can inquire about participation in Dr. Kang's research. Information may be found at **Med.umn.edu/mdcenter**, or by contacting the research team at **neurogenetics@umn.edu**. Through this research, it is certain that Dr. Kang will continue to make a difference for the LGMD community, one diagnosis at a time. ■

Written by Rebecca Lucas Gregg

Below: MD Center Colleagues (left to right): Andrew Thesing, Natalya Alassy, Peter Kang, Peter Karachunski, James Ervasti.

ADVERTISEMENT

JOURNEY

We welcome you to join our Journey...

Journey is a clinical outcomes assessment study, also referred to as a natural history study. Journey studies the natural progression of the disease and how it affects the muscles, lungs, and heart over a period of time (~3 years). The study does not involve the use of an investigational study drug. Individuals affected with Sarcoglycan Limb-girdle muscular dystrophy are invited to participate.

The Journey to uncover your potential treatment options

Currently there are no therapies for individuals with Limb-girdle muscular dystrophy. Participation will provide you access to highly experienced clinical trial physicians and clinicians with expertise in your condition and knowledge about future drug therapy research. Your participation in Journey will not hinder your ability to participate in future clinical studies, including those with investigational study drugs.

Who may be eligible

- Male or Female
- Age 4 years and older
- Genetic diagnosis of LGMD2E/R4, LGMD 2D/R3, or LGMD2C/R5

*Other inclusion/exclusion may apply and will be discussed with a study doctor at screening.

Journey Participation

Study participants will undergo medical and functional procedures during the screening visit, and at scheduled study visits throughout the lifetime of the study.

On-site visits: For screening and every 6-month visit:
- meet with the study doctor and research team
- complete motor assessments.

In between on-site visits: For every 3-month visit:
- study research team will contact you every 3 months by phone
- ask you questions on your health and wellbeing.

Journey Locations

The Journey study is currently enrolling at research centers in the United States and Europe, and is planned to be active in Canada, and parts of South America and Asia.

To learn more about the study and how you can join the Journey, go to journeyLGMD.com

You can also find more information about this study and participating locations, by visiting ClinicalTrials.gov and searching NCT04475926.

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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.

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

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.

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.

WHY

A genetically confirmed subtype diagnosis opens up new options to:

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

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

ADVERTISEMENT

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.

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SAREPTA THERAPEUTICS

Small Increases in Activity Level Could Help Improve Overall Fitness

Keeping up an exercise program can be difficult for anyone but can be especially challenging for individuals with muscle diseases such as limb-girdle muscular dystrophy.

As we move further into 2022, many people may be having trouble maintaining their New Year's Eve fitness resolutions. Keeping up an exercise program can be difficult for anyone but can be especially challenging for individuals with muscle diseases such as limb-girdle muscular dystrophy. I would like to suggest you shift your perspective from focusing on any exercise plan failures. Instead, consider setting the goal of increasing your activity level and then congratulate yourself on any small changes you make to become more active.

Any significant changes in your exercise routine or activity level, however, should be discussed with your physician or physical therapist prior to starting. You could ask to discuss your plans at your next visit to your neuromuscular physical therapist to identify any potential limitations and to get ideas on how to find an activity that fits into your lifestyle and interests. By identifying activities you can easily incorporate into your daily life, you are much more likely to maintain your new routine. This could be as simple as taking an evening walk, incorporating full arm movements when folding laundry, or doing breathing activities when you are watching television.

While activities such as swimming, riding a stationary bike, or using an arm ergometer are a better way to improve your cardio-pulmonary health, even small movements in the right direction are beneficial. I encourage you to speak with your healthcare team and set small goals. And remember to reward yourself for small victories!

For additional exercise tips for adults living with limb-girdle muscular dystrophy, visit **MuscularDystrophyUK.org/Exercise-Advice**. ■

Written by Linda Lowes, PT, PhD
Center for Gene Therapy
Research Institute at Nationwide Children's Hospital
Columbus, Ohio

UPDATED: MARCH 1, 2022

Active GRASP-LGMD Clinical Trials

RECRUITING: Defining Clinical Endpoints in Limb-Girdle Muscular Dystrophy (LGMD) (GRASP)

Inclusion Criteria:

- Age between 4–65 at enrollment
- Clinically affected (defined as weakness on bedside evaluation in either a limb-girdle pattern, or in a distal extremity)
- A genetically or functionally confirmed mutation in ANO5, CAPN3, DYSF, DNAJB6, or SGCA-G
- Ambulatory

Exclusion Criteria:

- Non-ambulatory at the time of enrollment
- 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

Subtypes:

- CAPN3 (LGMD 2A/R1)
- DYSF (LGMD 2B/R2)
- ANO5 (LGMD 2L/R12)
- DNAJB6 (LGMD 1D/D1)
- Sarcoglycan (LGMD 2D/R3, LGMD 2E/R4, LGMD 2C/R5, LGMD 2F/R6)

NOT RECRUITING: Biomarker Development in LGMD 2I/R9 (MLB-01-001)

Inclusion Criteria:

- Age between 10–65 at enrollment
- Clinically affected (defined as weakness on bedside evaluation in either a limb-girdle pattern, or in a distal extremity)
- A genetically confirmed mutation in FKRP (LGMD 2I/R9)
- Up to 60 participants will complete the10-meter walk test in greater than 4 seconds
- Up to 40 participants will complete the 10-meter walk test in over 12 seconds
- Up to 20 participants may be non-ambulatory

Exclusion Criteria:

- 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
- History of a bleeding disorder, platelet count <50,000, current use of an anticoagulant
- Positive pregnancy test at start or at any time during the trial

Subtype:

- FKRP (LGMD 2I/R9)

Contact: **Jessica St. Romain** | Project Manager, Grasp-LGMD Consortium | (804) 828-7887 | *Jessica.Stromain@vcuhealth.org*

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Newcastle University


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Yale University, Non-Clinical Site

GRASP-LGMD Researcher Spotlight

Dr. Linda P. Lowes | Nationwide Children's Hospital

Dr. Linda P. Lowes is Associate Professor of Pediatrics at The Ohio State University and a principal investigator in The Center for Gene Therapy in the Abigail Wexner Research Institute at Nationwide Children's Hospital in Columbus, Ohio. As part of the Center for Gene Therapy, she has been a pivotal co-investigator on numerous, first-in-human gene therapy clinical trials in rare diseases, including the first-in-human trial that led to FDA approval of the first gene therapy for spinal muscular atrophy. Her research interests focus on optimizing functional outcomes in adults and children with neuromuscular disorders. She is also interested in promoting clinical trial readiness through data drive outcome measure selection. To facilitate clinical trial readiness, she has conducted numerous natural history studies in LGMD and other disorders. She serves on the executive board for TreatNMD and was awarded the Excellence in Innovation award for her two inventions that use technology to quantify movement.

Scan to visit our website.

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.

mlbiosolutions.com
info@mlbiosolutions.com

ML BioSolutions
a bridgebio company

How Smart Is Your Wheelchair?

Over the past few years, SMART technology (**S**elf-**M**onitoring, **A**nalysis, and **R**eporting **T**echnology) has become an integral part of our lives. This advanced technology is now frequently found in household thermostats, lights, speakers, appliances, and some home assistant devices to make our homes more comfortable, safe, and secure. And when it comes to vehicles, newer driver assistance technologies help to keep you and your passengers safe, as well as other drivers and pedestrians, with integrated backup cameras, lane departure warnings, various sensors, and collision warning systems.

With all of the new technology being incorporated into our cars and homes, power wheelchairs have generally lacked similar technological advances. But that is finally beginning to change!

Recently, I was able to test-drive a power wheelchair that was equipped with LUCI smart wheelchair technology. I was intrigued by LUCI and wanted to learn how it could potentially make my life safer.

LUCI, an accessory for power wheelchairs that is an attachable hardware/software product, uses cloud and sensor-fusion technologies. This system combines data from cameras, ultrasonics, and radar to help the wheelchair "see" its environment and provides features like collision avoidance (such as with narrow doorways, walls, and furniture), drop-off protection for curbs, and anti-tipping alerts for steep ramps or uneven surfaces. The technology works with riders' steering inputs and reaction times to create a personalized, secure driving experience. LUCI consists of 6 sensors and a small dashboard, plus a phone app that goes with the system. LUCI is also compatible with Alexa® and Google Assistant.®

The hardware for LUCI's technology fits onto the base of powerchairs. Currently, LUCI is compatible with Permobil M3, M5, F3, and F5, and the Quickie Q300 M Mini, Q500 M, and Q700 M power wheelchairs. In the near future, LUCI should also be available for users of the ROVI and Amy Systems powerchairs.

If you are interested in exploring how LUCI smart wheelchair technology could improve your wheelchair mobility safety, contact your local power wheelchair provider to set up a LUCI demo and a test drive. To learn more about LUCI, go to **Luci.com** (MSRP: $8,445). ■

Written by **Carol Abraham**, *Retired OTR;*
Director of Community Outreach, Coalition to Cure Calpain 3
President, LGMD Awareness Foundation

Above: LUCI's patented system combines stereo vision, infrared projector, inertial measurement unit (IMU), ultrasonic, and radar data into a single view of the world, enabling never-before-seen possibilities for power wheelchair users. The sensor coverage is best illustrated from a bird's-eye view: **light blue** represents coverage by the stereo vision cameras, **purple** represents radar coverage, and **green** represents ultrasonic sensor coverage.

Featured Resources

Luci.com

What Is A Smart Power Wheelchair?

A smart power wheelchair is integrated or retrofitted with technology that provides enhanced, independent mobility to a wheelchair user, with user health and wellness data collection capabilities, and/or the aptitude to integrate with the connected world.

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Do you or someone you know have LGMD2I/R9?

AskBio will be conducting a clinical study (LION-CS101) of an investigational gene therapy for individuals with a confirmed genetic diagnosis of LGMD2I/R9.

- LION-CS101 is one-time intravenous infusion of gene therapy designed to produce fukutin-related protein (FKRP) in the body, primarily in muscle.
- This part of the study, to assess the safety of LION-CS101, will be conducted only in adults (ages 18 and 65 years). The study is a placebo-controlled, cross over study.
- The study is designed to investigate at least 2 different doses of LION-CS101 as compared to placebo.
- The initial phase of this first-in-human dose-finding study will be conducted in the U.S.
- Travel to LION-CS101 study sites may be reimbursed; local and home-based testing will be used when possible.
- Information on LION-CS101 can be found on clinicaltrials.gov.

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

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GET READY FOR THE

2022 LGMD GLOBAL ADVOCACY SUMMIT

LIMB GIRDLE MUSCULAR DYSTROPHY

At the **2022 LGMD Global Advocacy Summit**, a first for the limb-girdle community, you will learn directly from pharmaceutical companies about the clinical trials coming in 2022 and 2023. Additionally, you will hear from patient advocacy leaders from around the world about the regulatory drug process in both the U.S. and Europe. You will also have the opportunity to listen to LGMD patients and caregivers share with the invited companies about what matters most to them in trials and in drug development. **You do not want to miss it!**

SUMMIT SCHEDULE

12:00 PM

Welcome
European Advocacy

Learn about updates on drug development and the regulatory process in Europe, as well as the various structures that exist in Europe for patient advocacy and how you can get involved.

- **Rob Camp** | Director, International Patient Collaboration, EUPATI (Spain)
- **Aleksandra Leijenhorst** | Chair, Stichting Spierkracht (Netherlands) and Coordinator, LGMD CAB (Europe)


Rob Camp


Aleksandra Leijenhorst

1:00 PM

International Updates

Hear the latest LGMD updates from AFM-Téléthon.

- **Mandine Casado** | Expert Patient, GI LGMD, AFM-Téléthon (France)
- **Mélanie Bordes** | Expert Patient, GI LGMD, AFM-Téléthon (France)

Hear updates about The Quality Project by GFB Onlus, which seeks to improve the quality of life for patients living with sarcoglycanopathies.

- **Carles Sánchez Riera, PhD** | Researcher, Sapienza University of Rome (Italy)


Mandine Casado


Mélanie Bordes


Carles Sánchez Riera, PhD

1:10 PM

Roundtable Discussion 1

In this unique discussion, five individuals living with LGMD from around the globe will share what matters most to them in clinical trials and drug development.

- **Moderator: Kathryn Bryant Knudson** | Founder/CEO, The Speak Foundation (USA)
- **Brandi Benton** (USA)
- **Melissa Grove** (USA)
- **Kathryn Newnham** (England)
- **Carles Sánchez Riera, PhD** | Researcher, Sapienza University of Rome (Italy)
- **Joshua Thayer** (USA)

ALL TIMES ARE IN EASTERN DAYLIGHT TIME (EDT), UTC -4. | Summit schedule is subject to change.

WATCH THE LIVESTREAM EVENT AT

WWW.MALONE-MEDIA.COM/VIDEOS/LGMDSUMMIT

1:45 PM US Advocacy

In this session, you will gain insight on the various structures that exist in the United States for patient advocacy so you can be an effective voice for LGMD. You will also learn about the drug development and regulatory process from start to finish and find out how you can be involved.

- **Paul Melmeyer** | Vice President, Public Policy and Advocacy, Muscular Dystrophy Association (MDA) (USA)
- **Sharon Hesterlee** | Chief Research Officer, Muscular Dystrophy Association (MDA) (USA)


Paul Melmeyer


Sharon Hesterlee, PhD

Q&A

- **Brad Williams, PhD** | Director of Research & Diagnostic Innovation, Jain Foundation (USA)
- **Sarah Shira Emmons** | Director of Global Patient Outreach & Community Strategies, Jain Foundation (USA)

2:45 PM US Updates

Hear updates from the Principal Investigator of the GRASP-LGMD Consortium about active studies and clinical trials.

- **Nicholas Johnson, MD, MSci, FAAN** | Virginia Commonwealth University; Principal Investigator, GRASP-LGMD Consortium (USA)


Nicholas Johnson, MD, MSci, FAAN

Research Updates for LGMD2B/R2

Listen to research updates and information about preclinical progress for LGMD2B/R2.

- **Douglas E. Albrecht, PhD** | Co-President, Jain Foundation (USA)
- **Laura Rufibach, PhD** | Co-President, Jain Foundation (USA)

3:00 PM Roundtable Discussion 2

In our second roundtable discussion for the Summit, you will hear from four more individuals who are ready to share their voices with the companies designing clinical trials and developing treatment drugs for LGMD.

- **Moderator:** Kathryn Bryant Knudson | Founder/CEO, The Speak Foundation (USA)
- **Moderator:** Sarah Foye | Founder/Organizer, Team Titin (USA)
- **Mélanie Bordes** | (France)
- **Rachel DeConti** | (USA)
- **John Faver** | (USA)
- **Keisha Greaves** | (USA)

3:30–7:00 PM Pharmaceutical Updates

3:30 PM Sarepta Therapeutics, Inc.

Sarepta will present updates on their gene therapy development programs for multiple LGMD subtypes.

- **Louise Rodino-Klapac, PhD** | Executive Vice President, Head of R&D and Chief Scientific Officer (USA)


Louise Rodino-Klapac, PhD

4:00 PM Atamyo Therapeutics

Atamyo will share on their upcoming gene therapy trial for LGMD2I/R9, as well as their plans for their next programs for other LGMDs.

- **Sophie Olivier, MD** | Chief Medical Officer (France)


Sophie Olivier, MD

4:30 PM Vita Therapeutics

Vita Therapeutics will offer information about their platform and plans for LGMD2A/R1, as well as their goal to file a first-in-human IND study later this year.

- **Douglas Falk, MS** | Cofounder and Chief Executive Officer (USA)
- **Steve Brooks, MD, MBA** | Senior Vice President, Regulatory Affairs (USA)


Doug Falk, MS


Steve Brooks, MD, MBA

5:00 PM Asklepios BioPharmaceutical, Inc. (AskBio)

Hear from the AskBio team about their current clinical study (LION-CS101) of an investigational gene therapy for individuals with a confirmed genetic diagnosis of LGMD2I/R9.

5:30 PM ML Bio Solutions

The team at ML Bio Solutions will be sharing on their development of BBP-418, potentially the first oral treatment for patients with LGMD2I/R9.

- **Doug Sproule, MD, MSc** | Chief Medical Officer (USA)


Doug Sproule, MD, MSc

6:00 PM Edgewise Therapeutics

Edgewise Therapeutics will share about their current pipeline of precision therapeutics that regulate key muscle proteins to address LGMD.

- **Alan Russell, PhD** | Chief Scientific Officer (USA)


Alan Russell, PhD

6:30 PM Myogenica

Hear from a professor at the University of Minnesota about preclinical studies to enable a phase 1 clinical trial of iPSC-derived myogenic progenitors for the treatment of muscular dystrophies.

- **Rita Perlingeiro, PhD** | Cofounder (USA)


Rita Perlingeiro, PhD

7:00 PM Closing

ALL TIMES ARE IN EASTERN DAYLIGHT TIME (EDT), UTC -4. | Summit schedule is subject to change.

Written By Conrad Chris Weihl, MD, PhD
Professor of Neurology, Washington University School of Medicine, St. Louis, Missouri

and Kelly Macalalad, MSc
ClinGen Biocurator/Coordinator, Department of Neurology, Washington University School of Medicine, St. Louis, Missouri

Volunteer Biocurators Needed

Do you want to get involved with genetic research as a citizen scientist while also being able to help the LGMD community, or do you know someone who does? If so, then being a volunteer biocurator for ClinGen may be the right move for you!

As a volunteer biocurator, you will be tasked with Baseline Annotation, a curation process that involves annotating biomedical literature in a standardized form to aid ClinGen LGMD Expert Panels in evaluating the pathogenicity of Variants of Unknown Significance (VUS). ClinGen provides biocurators with all necessary training materials, so all you need is computer access and an interest in LGMD genetics. No experience is necessary.

Your role as a biocurator will help to reclassify LGMD genetic variants that have limited information or have conflicting classifications and do not have their pathogenicity established. Uncertainty in a variant's pathogenicity impacts LGMD clinicians who are unable to provide a clear diagnosis to LGMD patients, potentially leading to misdiagnosis, as well as LGMD patients who may harbor diagnostic uncertainty. As a result, patients may undergo inappropriate treatment and testing. However, with help from the work that is done by biocurators, the ultimate VUS pathogenicity that is determined by LGMD Expert Panels goes on to improve LGMD-specific genetic testing panels, resulting in highly improved treatment outcomes for LGMD patients. For more information on curating for ClinGen, please visit **ClinicalGenome.org/Working-Groups/Clingen-Community-Curation-C3**.

To take our LGMD volunteer survey, visit **CCDB.Clinicalgenome.org/apply/group/LGMD-Baseline**, or simply snap the QR code above.

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with multiple potential treatments in clinical trial in 2022, your support means more than ever!

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Limb Girdle Muscular Dystrophy

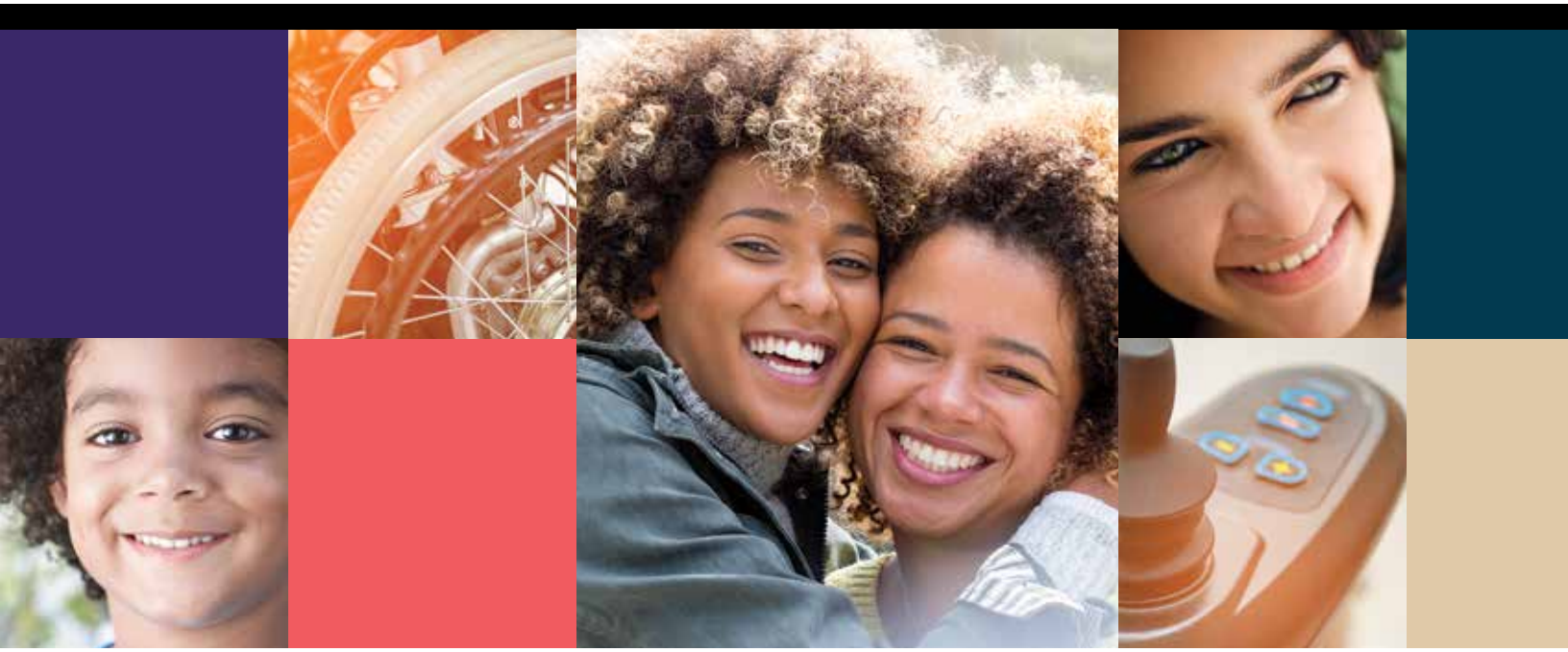
Learn about potential treatments and donate to support additional research at

CURELGMD2i.org

and follow us on

Written By Jane M. Lockwood

Cofounder, McColl-Lockwood Laboratory for Muscular Dystrophy Research
Atrium Health, Charlotte, NC



Make Your Voice Heard

You might have asked yourself, "What is an EL-PFDD meeting and why should I participate?" This article should help you understand more about this historical event for the limb-girdle community and why **your voice matters**.

Q

What is an EL-PFDD meeting?

A

The acronym EL-PFDD stands for **Externally-Led Patient-Focused Drug Development**. It is a meeting that is hosted, led, and presented by leaders in our LGMD community to the Food and Drug Administration (FDA) and other stakeholders such as product developers, clinicians, industry, and academic researchers. EL-PFDD meetings facilitate integration of patient insights into the drug development approval process. We will highlight patient and caregiver perspectives for six subtypes of LGMD, including 2A/R1, 2C/R5, 2D/R3, 2E/R4, 2F/R6, and 2I/R9. We will cover a range of topics from symptoms, health effects, struggles, and impact on daily living, to perceptions on existing and future treatments.

Q

What is the goal of the meeting?

A

The goal is to enable the FDA and the whole drug development community to understand the needs of our patient community and our expectations for future treatments.

Q

Why is it important to educate the FDA and other key stakeholders?

A

When the FDA is determining whether to approve new drugs, they weigh and assess benefits and risks. The FDA will consider scientific factors and evidence based on clinical trials, while also utilizing subjective judgment when considering the benefits versus risks assessment. Learning about and understanding the anticipated tolerance level of risks versus benefits from the LGMD community is critical in helping them to make informed decisions. Product developers, including researchers and pharmaceutical companies, will use the patient perspectives gathered during this meeting when developing drugs and designing clinical trials.

Q

What is the format of the meeting?

A

The meeting is virtual and will be held on Friday, September 23, 2022. The agenda will consist of speakers and panelists and will feature live polling and discussion forums from our virtual audience. It is a full-day meeting (from 10:00 a.m. to 3:00 p.m. EDT) dedicated to educating the FDA and other stakeholders about living with these six subtypes of LGMD. In the morning we will explain symptoms, health effects, and the toll the disease takes on daily living. In the afternoon we will discuss our perspectives on current and future treatments and communicate our benefit-versus-risk tolerance through more live polling and audience discussion.

Q

Why is it important for me to participate?

A

This is our opportunity to inform the FDA and other stakeholders about what matters most to individuals living with LGMD. Your voice will have an impact on future clinical trial designs, outcome measures that are more meaningful to patients, and greater awareness of the hardships and challenges of living with LGMD.

Q

Who should attend the EL-PFDD meeting?

A

YOU! Your voice is critical to a successful meeting. LGMD community members with 2A, 2C, 2D, 2E, 2F, and 2I should join us on September 23rd. We encourage affected individuals, caregivers, and family members to participate. We need your support and your partnership. With your participation, we will show how dedicated our community is to finding treatments and, ultimately, a cure.

Any member of the public can watch the meeting live or access the recorded footage later, with the ability to download the "Voice of the Patient" (VOP) report (a summary of the meeting). However, only patients with sarcoglycanopathy, LGMD2A, LGMD2I, or caregivers of these patients will be able to provide testimony and/or participate in polling.

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. We utilize induced pluripotent stem cell (iPSC) technology to engineer specific cell types designed to replace those that are defective in patients.

The first target is LGMD2A/R1, a muscular dystrophy caused by a defective gene for the protein Calpain-3. The lead asset, VTA-100, is an autologous cell therapy using gene editing technology to replace the defective gene which is drawn from the blood of a patient. The goal is to repair and replace defective muscle tissue in affected patients, regenerating healthy muscle.

VTA-100 is currently in the pre-clinical stages and working towards initiating IND-enabling studies with the goal to file a First-in-Human IND study in the second half of 2022.

VITA Therapeutics

Vitatx.com

clinicaltrials@vita-therapeutics.com

Q

How were the subtypes selected to be presented?

A

Sixteen representatives from ten LGMD organizations participated in a community discussion to determine how best to move forward with the EL-PFDD process. Participants agreed that sarcoglycanopathies (LGMD2C, 2D, 2E, and 2F), LGMD2A, and LGMD2I would request the first EL-PFDD due to their overlapping presentations and current programs in clinical and late-preclinical stages of development. This structure allows for other patient advocacy organizations representing different LGMD subtypes (e.g., dysferlinopathies) an opportunity, beyond this meeting, to generate patient experience data and meet with the FDA and others, when their community feels it is ripe for their engagement.

Q

How do I sign up to participate so that my voice can be heard?

A

Go to **TheSpeakFoundation.com/PFDD** or scan the QR Code below to register.

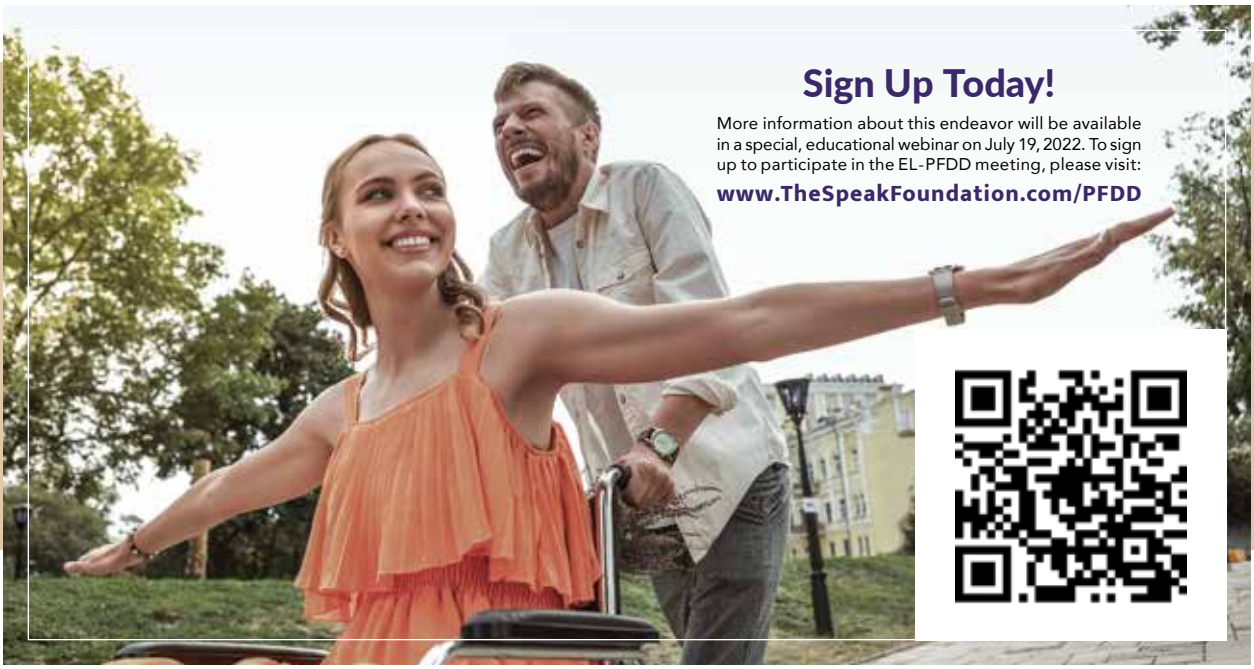
Q

What happens after the EL-PFDD meeting?

A

After our EL-PFDD, a "Voice of the Patient" (VOP) report will be compiled and made available to the public. The VOP report is a meeting summary and will be used as a reference document by the FDA, other stakeholders, and our patient community. This educational resource will be available to the FDA when reviewing potential, new drug therapies or treatments.

We need you to join our team and have your voice heard! Sign up to participate, mark your calendar for September 23rd, join our live discussion, and engage with the FDA and other key stakeholders about what matters most to you. ▪



Camronscure Foundation for LGMD2S/R18

Camronscure has raised funds for various organizations that are dedicated to researching gene therapy for limb-girdle muscular dystrophy.

Camronscure Foundation is a parent-founded organization named after a beloved son, Camron. It was created to fund research for limb-girdle muscular dystrophy type 2S/R18, Camron's diagnosis. LGMD2S/R18 is an extremely rare type of muscular dystrophy that causes muscle deterioration, as well as intellectual impairment, over the lifespan of the patient. There are only four confirmed cases of LGMD2S/R18 in the United States to date.

Camron's parents started the foundation when he was diagnosed at the age of 14. When Camron was a baby, his family noticed that he was not developing at the typical rate of a child his age. He was delayed in developmental milestones such as rolling over, walking, and talking. Although Camron eventually learned to do these things, he continued to experience delays through his adolescent years. He also began to show signs of intellectual impairment. Muscular dystrophy was not originally suspected until he started to lose muscle mass in his early teens. Finally, after months of appointments, a muscle biopsy, and genetic testing, Camron was officially diagnosed with LGMD2S/R18. Although it was a relief, in one sense, to finally have answers to explain Camron's many difficulties, the diagnosis still came as a shock to his family.

LGMD2S/R18 is so rare that the doctors had very little information they could provide about the disease. Unsure of what to do next, Camron's dad turned to Facebook in an effort to find others living with limb-girdle muscular dystrophy. That is when Camron's family learned about The Speak Foundation, which has been instrumental in providing support and guiding them to resources to help their son.

From there, Camron's parents decided it would be best to start their own foundation to raise awareness for their son's rare disease. Camronscure Foundation, which was founded in 2018, is the first foundation specifically dedicated to LGMD2S/R18. The mission of Camronscure Foundation is to raise awareness and funds for further research in the hopes of finding a cure or treatment for those affected by LGMD2S/R18. The foundation's kickoff fundraiser raised over 15,000 dollars, a tremendous success for the organization's inaugural event. Since then, Camronscure has raised funds for various organizations that are dedicated to researching gene therapy for limb-girdle muscular dystrophy.

Camronscure Foundation hopes to identify and connect with more families affected by a diagnosis of LGMD2S/R18 in the hopes to build a bigger support community for this disease. To learn more about Camronscure Foundation, please visit them on Facebook or at their website at **Camronscure.com**. ■

Written by Jessica Gerardo

Above: Camron with his family at the Camronscure fundraiser. **(From left)** Alberto, Jessica, Camron, Michael and Sarrie.

Connect with Us

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Facebook.com/LGMDCC

BELIEVE THERE IS HOPE

BELIEVE THERE IS HOPE FOR A CURE

Limb Girdle Muscular Dystrophy™

Be a Part of Our New LGMD Research and Advocacy Fundraising Campaign!

It's personal. If you are reading this, you or someone you know is living with a form of limb-girdle muscular dystrophy, and that affects all of us. We are family in the LGMD community. With over 30 different subtypes of LGMD, there is a need for ALL forms to see innovation in research and advocacy. The 2022-23 **BElieve THEre Is HOPE for a Cure** campaign endeavors to empower the entire LGMD community by raising funds for new research, advocacy efforts, and to directly assist individuals living with LGMD.

Join Us! When you participate, a Speak Foundation team member will reach out to you to help you personalize your fundraising experience and goals. You will "Raise Your Way"— that is, find and use the unique fundraising method that works best for you. To aid you on your journey, a fundraising kit will be available via digital download to everyone, everywhere (US and international), or mailed directly to your home (US residents only). **Help Us Make History for the LGMD Community!**

COMING SOON!

The **BElieve THEre Is HOPE for a Cure** fundraising campaign is a project of The Speak Foundation.

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Your Voice Will Make a Difference!

The LGMD community will be leading an Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting with the Food and Drug Administration (FDA) and other stakeholders on September 23, 2022.

What will happen at this EL-PFDD meeting? This important, monumental effort will give individuals diagnosed with limb-girdle muscular dystrophy types 2A, 2C, 2D, 2E, 2F, and 2I an opportunity to share with the FDA and other stakeholders about the experiences and challenges of living with LGMD.

What do we hope to gain? The EL-PFDD meeting is designed to engage patients and elicit their unique perspectives. With this information, our goal is to enable the FDA and other stakeholders to knowledgeably review and approve LGMD therapies that meet the needs and expectations of our patient community.

Coalition to Cure Calpain 3 — Overcoming Weakness With Strength
kurt+peter Foundation
LGMD2D Foundation
The SPEAK Foundation
CURELGMD2i
McColl-Lockwood Laboratory for Muscular Dystrophy Research

More information about this endeavor will be available in a special, educational webinar on July 19, 2022.
To sign up to participate in the EL-PFDD meeting, please visit:

www.TheSpeakFoundation.com/PFDD