Vol 1 / Issue 3

LGMD News

Uniting the Limb-Girdle Muscular Dystrophy Community

UNDERSTANDING DYSTROGLYCANOPATHIES

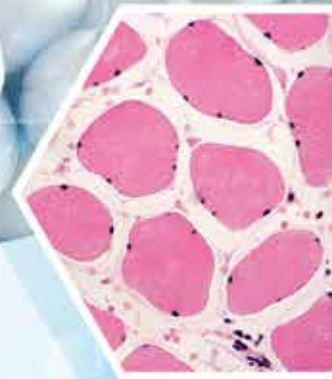
An Interview with Dr. Katherine D. Mathews and Dr. Nicholas Johnson, Two Key Players in the Field of LGMD2I/R9 Research

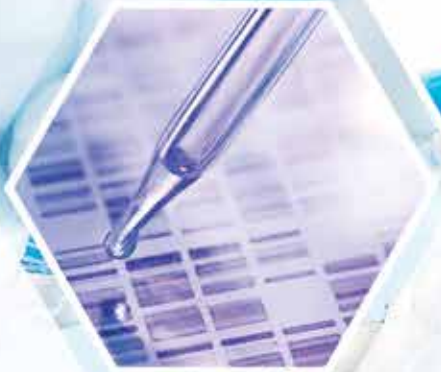
A Diagnosis Turned Into a Mission

Carles Sánchez Riera, PhD, with LGMD2E/R4, Changed His Career Course to Better Understand the Disease and to Help Others

GRASP CONSORTIUM

A Call to Participate in Natural History Studies





TEAM TITIN

A Titanic Job of Making Sense of Titin Variants

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.

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Twitter.com/LGMDFoundation

An Accurate Genetic Diagnosis is Vital

This proactive step to find your accurate genetic diagnosis could make a difference in the quality of your life.

Featured Resource

Invitae.com

In this issue, we highlight the importance of an accurate genetic diagnosis. The story of Patrick Moeschen demonstrates how misdiagnosis can take you on a frustrating journey of confusion. Often misdiagnosis leads to a great loss of time and money, too. "Trust but verify" should be a patient's motto when it comes to diagnosis because many forms of muscular dystrophy mimic one another. Early in the disease process, you might appear to have a particular form phenotypically, however, as time progresses, certain features may begin to emerge that do not fit that pattern. That is why one must insist on genetic testing.

Be vigilant and do not pursue testing just once. A verbal diagnosis is not enough when it comes to LGMD. We are at a crossroads where many forms will have treatments in the near future. Time wasted can potentially interfere with your ability to get into a clinical trial. If you have never been given confirmation of a genetic diagnosis, then I would urge you to pursue it.

There are many different forms and routes of genetic testing available to patients. Invitae's (**Invitae.com**) Detect Muscular Dystrophy program offers sponsored, no-charge genetic testing for patients suspected of having a form of muscular dystrophy. No insurance or out-of-pocket costs are associated with testing through this sponsored program. This program is currently available for eligible patients in the US, Canada, Argentina, Australia, Brazil, Chile, Colombia, and Mexico. For more information on whether or not you qualify for this program, visit **Invitae.com/detect-muscular-dystrophy** or contact Invitae client services directly at 877-688-0992.

There are many aspects of living with MD that may feel, or in fact are, out of our hands. With genetic testing having become more widely available and even at no charge to the patient in some cases, pursuing knowledge about our diagnosis is an empowering opportunity that many of us can choose to take advantage of. This proactive step to find your accurate genetic diagnosis could make a difference in the quality of your life. ▪

Kathryn Bryant
Editor In Chief

Our Mission

The Speak Foundation was based on the principle of "Speak up for those who have no voice."

Speak up for those who cannot speak for themselves. — Proverbs 31:8


Connecting and Reaching Everyone
C.A.R.E.
A Program of The SPEAK Foundation
Connecting and Reaching Everyone
Are you a newly diagnosed patient with limb-girdle muscular dystrophy?
We understand LGMD and we are here to help. The C.A.R.E. program gifts newly diagnosed patients with a box of helpful tools that includes smart technology and items donated from the International Consortium of LGMD Organizations. This program is available to patients newly diagnosed with limb-girdle muscular dystrophy on or after May 1st, 2021. Open to U.S. residents only.
The SPEAK Foundation.com


Do You Need Financial Assistance for Mobility or Durable Medical Equipment (DME)?
Through **The HOPE Project** by The Speak Foundation, individuals living with limb-girdle muscular dystrophy can receive up to a $500 stipend per qualified applicant to help cover the costs of mobility and DME equipment. Applications are available at **TheSpeakFoundation.com** and will be accepted through September 30th. Open to U.S. residents only.
The HOPE Project
A Program of The SPEAK Foundation

The Importance of Supporting Organizations Led by Individuals or Families Living with the Disease

About fifteen years ago, I came upon an organization that was created to cure a disease, but the organization had very few meaningful relationships with individuals actually *living* with the disease. As far as I knew, they had not one person on staff with the condition. I soon learned that this is a trend among some nonprofit organizational systems. The problem is that patients need to be a part of the decision-making process so that the results reflect what we feel is important.

In recent years, patient-led organizations have emerged in order to advocate for disease-specific programs and research. Patient-led organizations all have the following traits in common: they were started either by an individual or a family member of the individual with the disease; they are focused on finding a cure for one specific or a very limited group of diseases; they maintain low overhead to free up monetary resources to direct it towards a cure; and they make an intentional effort to recruit disabled employees and volunteers as well as able-bodied employees.

Research shows that organizations led by individuals or family members affected by the disease are often the ones attached to innovations in the field. Patient-led cooperatives often invest in new research and even start companies to find cures. Many of these types of organizations exist in the LGMD space. In fact, every single organization on our Connect page (page 7) was started by a patient or a family member of someone with the disease.

The Speak Foundation makes it a priority to support nonprofit organizations and companies that encourage patient-led associations. The Speak Foundation itself is a patient-led organization. The philosophy is that individuals directly affected by the disease are leading the organization and driving the mission. This in no way means that only disabled individuals work for the organization. In fact, many patient-led organizations hire just as many able-bodied individuals as they do affected individuals. There is a mutual respect in patient-led organizations where individuals with disabilities are considered equally of value as those without disabilities.

We encourage pharmaceutical companies to become leaders in this issue. Disabled individuals are a minority group and companies must intentionally seek to support their efforts. Get behind patient-led organizations. Subscribe to and monetarily support patient-led initiatives. Hire disabled individuals and add to your company's website that you support nonprofit organizations that are led by individuals with the disease. Work through these organizations and empower them. Strengthen the voice of these patient-run cooperatives with *your* voice. Together, we can make a change. ▪

Kathryn Bryant
Founder, The Speak Foundation

Kathryn Bryant

In recent years, patient-led organizations have emerged in order to advocate for disease-specific programs and research.

Do You Have Something to Voice?

Send Your Thoughts To:
ContactUs@TheSpeakFoundation.com

"I Am Not Alone"

Reaching Out for Meaningful Support

There is something almost indescribable about the "me too" experience of seeing someone else walk, drink from a glass, or get up from a chair just like you. The sense of belonging can foster such hope!

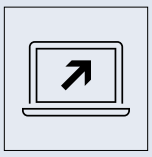
Do you remember what it felt like that first time you connected with someone else living with limb-girdle muscular dystrophy? I do. There is something so powerful about the experience of realizing, "I am not alone."

The Speak Foundation grew out of a desire to connect lives and unite a community. At that time, the landscape for the LGMD community looked very different than it does today. There were no patient-led organizations for muscular dystrophy. There were no treatments for LGMD on the horizon. It was not at all uncommon for people to have never met someone else with muscular dystrophy, let alone LGMD, or their particular subtype. This is why The Speak Foundation started holding our annual patient support conferences in Atlanta, Georgia, in 2008. These weekends became a sacred place where a "family" was formed. There is something almost indescribable about the "me too" experience of seeing someone else walk, drink from a glass, or get up from a chair just like you. The sense of belonging can foster such hope!

Today, the means to connect with others who share similar circumstances and challenges is at our very fingertips. We do not want you to miss out on the many communities of support that exist specifically for you and your subtype — communities where you can share and learn from others how to navigate life with LGMD. In this issue, on the page to your right, you will find a helpful list of many patient and family-led organizations that are subtype-specific who can offer you meaningful support.

Here are just some of the many ways that you can connect with those who "get it." There are Facebook groups associated with many of the organizations we have highlighted for you. Most of them also have a patient registry that we encourage you to join. Patients are getting creative as a result of the pandemic and are even initiating and facilitating their own Zoom meetings. The International Limb Girdle Muscular Dystrophy Conference coming up in September is yet another opportunity to connect virtually with thousands of people with LGMD from all over the world. You can also visit **TheSpeakFoundation.com** to register for the conference, subscribe to *LGMD News*, and join our patient network so that we can keep you informed of the latest research, clinical trials, and opportunities to be active in the LGMD community. ▪

Contributed by Jessica Evans, PsyD
Assistant Director, The Speak Foundation



LGMD Patient Network

Register at **TheSpeakFoundation.com** to receive updates on the limb-girdle muscular dystrophy patient community. By registering, you will receive our magazine and many important resources to help you. We offer multiple programs to enhance your quality of life, such as the International LGMD Conference and the Personal Care Attendant Stipend Program. The Speak Foundation is a patient-led team of individuals who are living successfully with LGMD — we know firsthand the importance of ***community***.

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

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

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

COALITION TO CURE CALPAIN 3 (C3) is committed to treating and ultimately curing limb-girdle muscular dystrophy type 2A (LGMD2A, also called LGMDR1 Calpain 3-related or calpainopathy). Our mission is to fund high potential research and clinical trials as we educate the global community about this rare disease.

DO YOU LIVE WITH LGMD2A/R1? WE WANT TO CONNECT WITH YOU!

JOIN OUR REGISTRY LGMD2A.org
to be alerted when research studies are seeking participants

VISIT OUR WEBSITE CureCalpain3.org
to learn more about how C3 is making a difference and driving progress towards a cure

FOLLOW US ON FACEBOOK Facebook.com/CureCalpain3
for up-to-date research news

JOIN THE "C3 COMMUNITY" Facebook.com/CureCalpain3/groups/LGMD2A/
a private Facebook group that is a vital hub connecting patients from around the world to help navigate the challenges inherent in living with a rare disease

COALITION TO CURE CALPAIN 3
OVERCOMING WEAKNESS WITH STRENGTH
10 Years of Progress

C3 is a 501(c)(3) US-based tax-exempt charity

JAIN FOUNDATION
ORCHESTRATING A CURE
LGMD2B/R2 DYSFERLINOPATHY MIYOSHI

The Jain Foundation is a nonprofit, scientifically led foundation whose mission is to cure muscular dystrophies caused by dysferlin protein deficiency, which includes limb-girdle muscular dystrophy type 2B/2R (LGMD2B/2R) and Miyoshi Muscular Dystrophy 1 (MMD1), collectively called Dysferlinopathies.

DYSFERLIN REGISTRY
by Jain Foundation

The Jain Foundation curates the Dysferlin Registry, an international registry for people with dysferlinopathy. The registry is critical to successful future trial recruitment. It also serves to educate and connect individuals who are navigating life with dysferlinopathy. The registry platform provides a quality, private media for open discussions. For more information about the registry, email ***Patients@Jain-Foundation.org***.

Jain-Foundation.org • DysferlinRegistry.Jain-Foundation.org

Tahseen Mozaffar, MD

Professor of Neurology and Pathology and Laboratory Medicine
Vice Chair for Research, Department of Neurology
University of California, Irvine

Q

Where can I get whole genome sequencing? I live in the US and am wondering if there is a free resource?

A

The whole genome studies are not currently a free resource unless you participate in a research study. They are not offered for clinical diagnostic purposes as free resources, however, there are laboratories that provide whole genome sequencing for a fee.

Q

Is it possible to get a genetic test for international patients? I live in India.

A

There are programs, often research-based, within countries like India, where free genetic testing can be obtained. There was a recent program that was implemented in India, through Perkin Elmer. Additionally, Invitae was involved in testing in South America through a Sanofi-Genzyme-funded research study.

Q

Can you explain the difference between whole genome sequencing and other types of genetic tests?

A

Most of the free genetic tests that are currently available are panel-based, next generation, sequencing-based studies. These are the most common genetic tests, but are only targeted to look at genes of interest at 10-20x coverage. Most panels cover around 35-70 genes, but some panels are larger. These panels have limitations and cannot pick up deletions, duplications or repeat expansions. They also do not examine the non-coding regions of the gene very well. Whole genome sequencing, however, does not have these limitations. Whole genome is also unbiased, which means that it looks at all genes and not just the genes of interest. Therefore, it may pick up abnormalities in genes with which we were not anticipating a problem, or a novel gene altogether that was previously not known to be associated with muscular dystrophy. ■

Meet the Expert

Tahseen Mozaffar, MD is Professor of Neurology and Pathology and Laboratory Medicine and the Vice Chair for Research in the Department of Neurology at University of California, Irvine. He is the Director of the UC Irvine-MDA ALS and Neuromuscular Center and the Director of the Division of Neuromuscular Disorders.

Have a Question for Our Experts?

Send Questions To:
ContactUs@TheSpeakFoundation.com

Udd group at the Folkhälsan Research Center, Helsinki, Finland. **(L to R):** Per Harald Jonson, Mridul Johari, Sabita Kawan, Helena Luque, Jaakko Sarparanta, Peter Hackman, Anna Vihola, Marco Savarese, Bjarne Udd

A Titanic Job: Making Sense of Titin Variants

We are on the move from the diagnosis to the understanding of the molecular mechanisms, with an ambitious final goal: therapy.

"Je cherche après Titine" ("I am looking for Titine") is a famous song that was composed by Léo Daniderff in 1917 and became famous thanks to Charlie Chaplin's interpretation in *Modern Times* (1936).

Titin (*titine* in French) hunting has been an enduring activity in medicine, too. In the early 90s, 23 muscular dystrophy patients were identified within one large Finnish family. Interestingly, the disease course amongst the patients within this family appeared to follow two very different patterns. The symptoms in one form had an onset in childhood, with weakness presenting in the hip and shoulder muscles and progressing to wheelchair confinement within twenty years. In the other form, however, onset occurred in adulthood (at around 30-40 years), presenting with weakness in the lower limbs and without advancing disability. Here were two different diseases with the same causative gene. Ten years later, this gene was identified: titin!

Titin is one of the most abundant proteins expressed in heart and skeletal muscles. Titin plays a crucial role in muscle formation and is essential for a proper contraction. Titin is also the biggest human protein. However, a mutation (a small change in the last part of the gene and, thereby, a small change in the terminal portion of the protein) was the cause of the observed diseases in the Finnish family. When a single copy of the mutated gene is inherited, the mild disease with an adulthood manifestation, named tibial muscular dystrophy or Udd myopathy, is observed. If a child inherits two mutated copies of the gene, the more severe disease, the limb-girdle muscular dystrophy LGMD2R titin-related, occurs.

Since then, a large number of additional mutations (errors in the gene) have been

found. Moreover, in the last ten years, the list of titin-defect muscle diseases has grown substantially. Some titin mutations increase the risk of developing a dilated cardiomyopathy, a disease characterized by heart dilatation and failure, while others will never have any cardiac defects in their lifetime. Other mutations cause skeletal muscle diseases, some of them with a clinical manifestation already at birth or in childhood, others with the first signs and symptoms only apparent later in life.

We all have small changes in the titin gene and protein, and most of these small changes do not have any impact on the protein function. How to identify the few disease-causing mutations among the vast number of harmless alterations is a scientific challenge that we are currently facing. IDOLS-G is an EU-funded project that we are coordinating, which aims at improving the diagnostic process and our understanding of the titinopathies. And we are on the move from the diagnosis to the understanding of the molecular mechanisms, with an ambitious final goal: therapy. Finding a cure for a rare genetic disease is a multi-step process that starts from the identification of the disease-causing genetic defect.

Such titanic efforts require experts with different backgrounds, including clinicians, geneticists, molecular biologists, biophysicists, and more, all working together for a better tomorrow for this disease community. ▪

Contributed by Marco Savarese, PhD
Folkhälsan Research Center
University of Helsinki

Connect with Us

Inquiries or Questions?
CureMyopathy@gmail.com

TitinMyopathy.com
CMDIR.com

Facebook.com/Groups/TeamTitin

Team Titin

Variants in the massive gene titin (TTN) can cause a spectrum of muscle and heart disorders including LGMD 2J, also known as LGMD R10 Titin-related.

Team Titin is a consortium of scientists and affected community members aimed at making a worldwide difference in Titin-related muscle and heart disorders by: collaborating with other organizations, raising awareness, connecting with families, providing education, and supporting research. Our goal is to serve as a catalyst for stakeholders to develop a better understanding of Titin-related disorders, leading ultimately to a cure.

To learn more visit titinmyopathy.com
EMAIL:
CureMyopathy@gmail.com
FACEBOOK:
facebook.com/groups/teamtitin
REGISTRY:
Congenital Muscle Disease International Registry (cmdir.com)

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

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.

SAREPTA THERAPEUTICS

Natural History Studies: A Call to Participate

We have an opportunity to work together to make sure we can design the best trials to ensure therapies meaningfully impact quality of life and slow the progression of disease.

Our understanding of how the limb-girdle muscular dystrophies progress over time, and how to measure that progression, relies on natural history studies. These studies are often designed to measure strength, physical function, and ambulation over a certain period of time. They often also use questionnaires to understand how limb-girdle muscular dystrophy affects an individual's life. The information gained from these studies lets researchers know how to design treatment trials.

Without information gained from a natural history study, researchers may not know what tools to use to measure improvement in a therapeutic study, nor how long they need to wait to see whether the drug will change the course of the disease. More importantly, natural history studies guide both clinicians and patients in what to expect, and how best to manage the condition.

While these natural history studies are a critical step in drug development, I, like all of you, cannot wait to put these studies behind us. When that happens, it is because we will have moved onto treatment trials. Until then, I am writing to ask you to participate in these studies because they will accelerate drug development in limb-girdle muscular dystrophy. We have an opportunity to work together to make sure we can design the best trials to ensure therapies meaningfully impact quality of life and slow the progression of disease. We, within the GRASP-LGMD consortium, are working on multiple natural history studies to achieve our shared mission of advancing therapies in limb-girdle muscular dystrophy. Please consider joining one of these studies. ■

Contributed by Dr. Nicholas Johnson
Associate Professor and Vice Chair of Research
Virginia Commonwealth University

UPDATED: JUNE 1, 2021

GRASP-LGMD Clinical Trials

STUDY 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)

STUDY 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: **Brittney Holmberg** | Project Manager, Grasp-LGMD Consortium | (804) 997-9384 | *Brittney.Holmberg@vcuhealth.org*

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John Vissing, MD
University of Copenhagen


Katherine Mathews, MD
University of Iowa


Linda Lowes, PT, PhD
The Research Institute at Nationwide Children's Hospital


Matthew Wicklund, MD
University of Colorado Denver


Nicholas Johnson, MD, MSci, FAAN
Virginia Commonwealth University


Peter Kang, MD
University of Minnesota Medical School


Tahseen Mozaffar, MD, FAAN
University of California Irvine


Urvi Desai, MD
Atrium Health


Volker Straub, MD, PhD
Newcastle University


Monkol Lek, PhD
Yale University, Non-Clinical Site

GRASP-LGMD Researcher Spotlight

Dr. Nicholas Johnson | Virginia Commonwealth University

Dr. Nicholas Johnson is Associate Professor and Vice Chair of Research at Virginia Commonwealth University. After medical school at the University of Arizona, he completed a residency and fellowship in neuromuscular experimental therapeutics at the University of Rochester. His research program includes a translational laboratory and coordinating center for the consortium. He is one of the co-founders of the GRASP-LGMD consortium, formed to improve diagnosis in LGMD and accelerate therapeutic development. He has led multi-center natural history studies to develop and refine clinical endpoints. These include the GRASP-LGMD study and ML Bio studies listed here, as well as studies in myotonic dystrophy. His clinical practice in Richmond, VA, focuses on inherited nerve and muscle diseases.

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Bed Mobility

Getting enough sleep helps to keep your mind and body healthy. As mobility becomes more difficult, some individuals with LGMD may face challenges getting a good night's sleep, ranging from difficulty in getting comfortable, repositioning, and/or getting in and out of bed. Thankfully, there are a growing number of products designed for individuals who have limited bed mobility. Some of these sleep products and tips include:

Draw Sheet

A draw sheet can help someone to roll and reposition you with ease. The bed sheet is placed underneath you, perpendicular to the bed and extending from shoulder level to buttocks, with at least 6-8 inches of sheet remaining on each side. Someone can then lift and pull on one edge of the fabric to assist in rolling or repositioning you.

Satin Sheets and/or Pajamas

This is an option that may increase the ease of positioning and turning. Bed sheets with a higher thread count can also reduce friction and therefore improve bed mobility.

Adjustable Frame Beds

An adjustable frame bed is recommended for those who have limited bed mobility. Using a remote control or voice control, you can elevate the foot of the bed and raise the head of the bed.

Above Left: Bed Rail Advantage Traveler (MSRP: $89.99). Above Right: 30" Safety Bed Rail (MSRP: $199.99). Learn more at **Stander.com.**

Bed Assist Rails or a Spindle Headboard

Bed assist rails come in a variety of sizes and configurations. A person who still has some upper extremity abilities may find that grasping a bed rail or the spindle of a headboard is beneficial to help provide leverage to assist in rolling. Many bed rails are portable and slide under the mattress for added stability.

Mattress Overlays

Specifically designed to prevent discomfort from immobility and encourage good blood flow to the skin, mattress overlays are fabricated from foam, rubber, gels, or in an innovative mechanical design. These greatly increase comfort and can help prevent painful bedsores.

Pillow Support

Similar materials and technology used in mattress overlays are also used in special pillows that provide support for the head, neck, hips, spine, and surrounding muscles. Pillows can play a vital role in supporting your body for added comfort. ■

Thankfully, there are a growing number of products designed for individuals who have limited bed mobility.

Featured Resources

AssuredComfortBed.com
Stander.com

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

Left: Assured Comfort® offers Hi-Lo adjustable beds that allows the height of the bed to be adjusted, which can make getting in and out of bed easier. Learn more at **AssuredComfortBed.com** (MSRP: starts at $4,400).

Patients Who Have Gone Into Science Series

PART 1 Carles Sánchez Riera, PhD

LGMD2E/R4

By Rebecca Lucas Gregg
By Brad Williams, PhD
Director of Research, Jain Foundation

A Diagnosis Turned Into a Mission

Being diagnosed with a rare disease can be life-changing in so many ways. This experience has even led some to completely redirect their career paths in order to learn more about the disease itself and help others who are affected by it. This was the case for Carles Sánchez Riera, PhD, who has LGMD2E. He turned his life's work into learning why some of our muscles resist the degeneration process if all of them have the same genetic mutation.

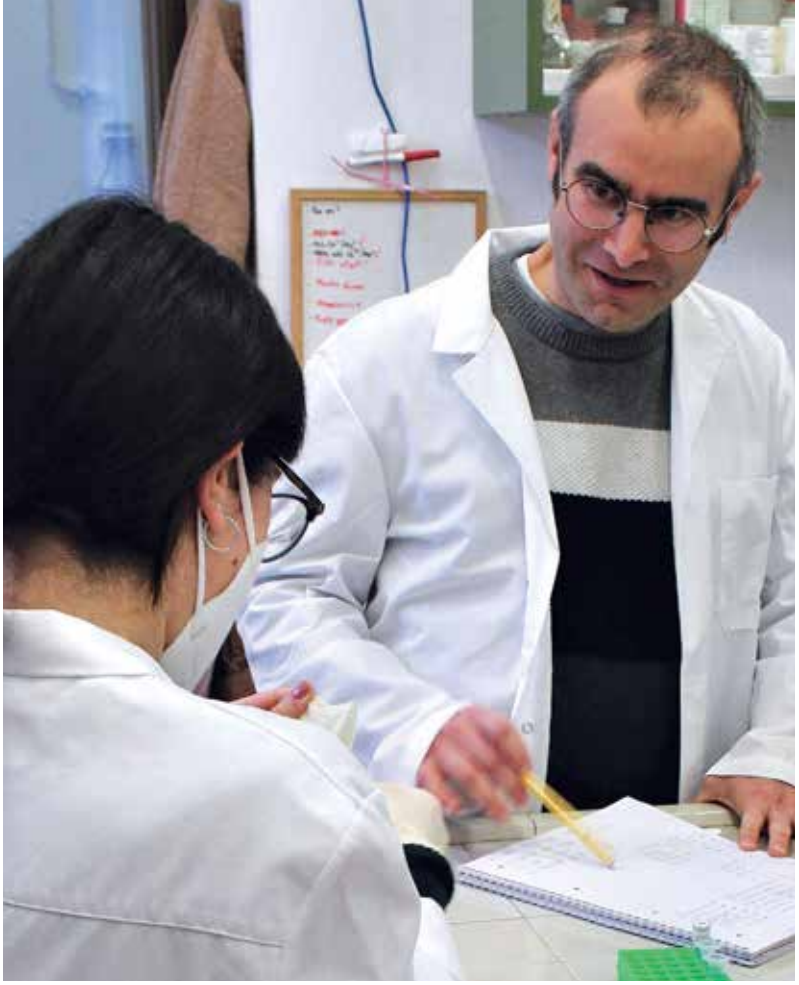
Carles has always been interested in science. As a child, he was fascinated with the idea of becoming an explorer, an animal documentarian, and an advocate for the planet. However, as time went by, his aspirations started evolving into a sense of social responsibility. He felt empathetic to injustices in many areas and motivated to make a meaningful difference in the world. By then, LGMD symptoms began affecting his daily life. He was not satisfied with his doctor's explanations, as there was a huge lack of knowledge in the muscle field, and so Carles decided to focus on his own disease and to make that focus his "social war."

"Knowing myself better allows me to accept my situation," says Carles. "At the age of 23, I came to the point that I wanted to decide about my health and not let my health decide for me. Therefore, I started to study biology."

> **"I came to the point that I wanted to decide about my health and not let my health decide for me. Therefore, I started to study biology."**

When asked if having LGMD has played a role in pursuing science as a career, Carles says, "The impact of LGMD in my life is totally determinant and positive, but it is not due to LGMD itself. It is because it pushes me to understand who I am. It challenged me to seek a better version of myself."

Carles is currently working at the Laboratory of Muscle Histogenesis and Remodeling at Sapienza University of Rome. His current focus is learning why some muscles



Left: Dr. Carles Sánchez Riera in his laboratory, collaborating with a student assistant.

To Learn More About Carles' Work:

DiversitaMuscolare.it

are protected in muscular dystrophies, as all forms of muscle dystrophy show specific patterns of some muscles being more affected, while others are relatively spared. Muscles degenerating at different rates, even though all share the same genetic mutation, is evidence of natural protection mechanisms against the degeneration process. Understanding how this protection occurs may help make treatments more effective. In collaboration with other scientists, Carles has been working on a project studying muscle reconstruction with biomaterials. Loss of muscle mass and fibers in our diseases continues as time goes by, making restoration of muscle more difficult. Therefore, understanding the mechanisms which protect some muscles will be necessary, as well as using new technologies such as 3D printing and biomaterials to create specific matrix environments to allow new fibers to be incorporated into the damaged muscle.

> **"Our duty is to be honest with everyone (including ourselves), to find a balance between our necessities and our responsibilities."**

Living with LGMD as a scientist does present challenges for Carles due to physical limitations from the disease. Carles explains, "In my case, I can still stand by myself. However, it is difficult to remain standing for a long time at the bench, to stand up from a chair, to pass through narrow doors, to get to objects stored up high, or to manage heavy ones. But we always work to find solutions. For example, I concentrate my efforts into analyzing data: microscopics, images, graphics, bioinformatics. I am also working in coordinating students and, for instance, we are looking to make the lab wheelchair accessible. Our duty is to be honest with everyone (including ourselves), to find a balance between our necessities and our responsibilities."

Regarding our patient community, Carles says, "It is okay for us to be angry, and to feel isolated or confused given our circumstances. However, we need to transform this energy into a source for collaboration. Our muscle disease community is small yet mighty, and there are more things [that] unite us than divide us. Hence, before starting to fight alone against windmills, turn around and look if there are others doing similar things. Collaboration is the key to be[ing] stronger." ■

Have you or someone you know been diagnosed with Limb-Girdle Muscular Dystrophy Type 2I?

ML Bio Solutions is developing an oral therapy for limb-girdle muscular dystrophy Type 2I (LGMD2I/LGMDR9).

A Lead-in Study (Natural History Study) is currently enrolling participants with LGMD2I. While there is no intervention in this study, participants will be eligible to participate in the late phase clinical trial of BBP-418 (Ribitol).

You may be eligible to participate in the Lead-in Study if you are 10-65 years old and have genetically confirmed LGMD2I.

Remote enrollment is available, all travel/study expenses are covered, and strict COVID safety precautions are observed.

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

ML Bio Solutions
a bridgebio company

Contact: Brittney.Holmberg@vcuhealth.org | 804-552-0014
info@mlbiosolutions.com | mlbiosolutions.com | clinicaltrials.gov

Mark your Calendar!

LGMD Awareness Day September 30th

Awareness is power, and power brings cures, treatments, and changes policies. LGMD Awareness Day is an annual effort to globally raise awareness of individuals living with Limb Girdle Muscular Dystrophy. The seventh annual Limb Girdle Muscular Dystrophy Awareness Day will be celebrated worldwide on September 30th, 2021. As an Ambassador for LGMD Awareness Day, we encourage you to adapt activities in accordance with your customs. Our goal is to reach the widest audience possible. Make an impact — get involved today!

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LGMD-Info.org

LGMD AWARENESS FOUNDATION

LGMD Awareness Day is a project of the LGMD Awareness Foundation, Inc.

By Kelly Brazzo
Co-Founder and CEO, CureLGMD2i Foundation

Understanding Dystroglycanopathies:

An Interview with the Experts

Dr. Katherine D. Mathews and Dr. Nicholas Johnson are two of the key players in the field of LGMD2i research and development.

Kelly Brazzo, Co-Founder and CEO of the CureLGMD2i Foundation, has a 13-year-old daughter, Sammy, who is diagnosed with LGMD2i/R9. The Brazzo family was fortunate to meet Dr. Mathews nine years ago when they first attended the Dystroglycanopathy Patient & Family Conference at the University of Iowa. They have been actively attending the conference each year, where Sammy participates in the natural history study. Sammy was also enrolled in the ML Bio Solutions lead-in study at VCU, which is being led by Dr. Johnson and the GRASP Consortium. Sammy has gone on from the natural history study to the ML Bio Phase 2 study, designed to test a disease-modifying therapy in LGMD2i.

Kelly posed a few questions to Dr. Mathews and Dr. Johnson, and then wrote this article to share their knowledge and expertise with our LGMD community.

Below: The Brazzo Family


Dr. Nicholas Johnson


Dr. Katherine D. Mathews

How did you become interested in studying the LGMDs?

Dr. Johnson: I have had a longstanding interest in muscle diseases due to gene mutations. The LGMDs caught my attention as a group of disorders with a significant need to develop the tools to conduct clinical trials and take advantage of the novel gene replacement technology that offers such promise for these conditions.

Dr. Mathews: I was trained in both neurology and genetics, and like Nick, have always been interested in genetic diseases of muscle. When we started the Iowa natural history study, FKRP mutations had just recently been discovered as a cause of muscular dystrophy and I was eager to help learn more about FKRP-related muscular dystrophy to improve patient care and lay the groundwork for future treatments. This has expanded to include all the dystroglycanopathies and other causes of LGMD.

What are your most challenging experiences in working with this population?

Dr. Johnson: Unfortunately, we do not currently have any effective therapies to slow progression. I am hopeful that will change soon, but it can make it challenging to see someone lose their strength and not have the current tools to stop it.

Dr. Mathews: I have loved the opportunity to get to know many people with LGMD, those who are my patients and those I have met through their research participation. It is hard when patients and families have had questions about their disease that we did not yet have answers for, but that is changing. I hope that we will be able to offer more specific treatments in the future.

Can you explain the prevalence of 2i in the world?

Dr. Johnson: The estimated prevalence is around 4.3 per million people, which makes it a rare condition. In other LGMDs, the prevalence has likely been underestimated, as it was previously difficult to obtain genetic testing. The impact of improved diagnostic testing is unclear for LGMD2i.

Dr. Mathews: The prevalence varies somewhat by geography and heritage of the population. There are some mutations that arose many, many generations ago and have persisted currently. One of the most common mutations in FKRP arose at the time of the Vikings and is relatively common among those with Scandinavian heritage. In Iowa, we found that ~1/330 chromosomes carry this mutation. There are other ancient mutations in other populations.

Can you explain issues that can affect heart and lungs for 2i?

Dr. Johnson: Weakness of chest muscles may impair breathing. This happens in about 30% of individuals with this disease. The heart muscle may also become weak with LGMD2i. In both cases, regular monitoring and follow-up by your multidisciplinary team in the clinic is critical to ensuring early detection and management.

Dr. Mathews: We recently published what we have learned about heart problems in people with FKRP mutations. Half of all participants had their first abnormal echocardiogram (ultrasound looking at heart function) by age 33 years. Those who have two copies of the "Viking founder mutation" have later onset of heart function (half had abnormal echocardiogram by age 54 years), while those with other genetic test results had earlier abnormality of heart function, on average. It is important to note that there was a wide range of ages in both groups. Our results are very similar to results from a group in Italy. The weaker heart function can be treated with medicine, so everyone with LGMD2i should have their heart monitored regularly with echocardiogram and/or cardiac MRI. Breathing problems (particularly at night) and weak cough tend to occur late in patients with LGMD2i, but disorders like obstructive sleep apnea can affect anyone. Breathing and cough strength should also be monitored regularly, as noted by Nick.

What are the warning signs that LGMD2i patients should be aware of as the disease progresses?

Dr. Johnson: I am not sure about warning signs, but you definitely want to make sure you are seeing your physician once a year to get your breathing checked. If you feel like you are having more difficulty breathing or are waking up in the morning not feeling rested, you should discuss this with your physician.

Dr. Mathews: I agree with Nick about the symptoms that might suggest not breathing well during sleep. A few other symptoms that might suggest weakness of breathing muscles are consistently waking with a headache in the morning, or feeling like your cough is getting weak or is not effective. We have also found that people with LGMD2i have a higher rate of difficulty with bladder control than family members. This is usually treatable with medicine. Swallowing is also sometimes a problem, and typically is managed by working with a speech pathologist. Fatigue can become a problem, similar to other chronic diseases. LGMD2i is a slowly progressive disorder, so any sudden change in function should prompt attention.

Can you share about the problem of exercise and rhabdomyolysis, as well as how to find a safe balance for someone with an early diagnosis of this disease?

Dr. Johnson: I'm leaving this one for Kathy as she knows more about this than anyone else!

Dr. Mathews: When we exercise or move, everyone gets little tears in the muscle cell membranes that are rapidly repaired by the body. At the same time, the body sends messages to make those muscles bigger and stronger. Rhabdomyolysis refers to muscle cell breakdown and is typically used to mean extreme muscle cell breakdown. People can become aware of muscle cell breakdown if a very high level of muscle cell breakdown products in the urine make urine brown-colored (like tea or cola). People with LGMD2i have ongoing muscle cell breakdown with normal activity, and we detect that by measuring the CK level in the blood. If people with muscular dystrophies are very active, it causes an increase in the muscle cell breakdown and sometimes that makes urine turn brown. We recommend pushing fluids, and normally it clears very quickly, by the next time urine is passed. This can be viewed as a sign that the activity was too vigorous. If brown urine persists, or if it is accompanied by weakness or severe pain, patients should notify their doctor or go to the ER as it can cause kidney injury. Because movement is required to keep muscles healthy, and activity is the signal to build muscle, it is important that patients keep as active as possible, and learn to recognize the signs that they have done too much.

Will you explain the difference between heterozygous and homozygous mutations?

Dr. Johnson and Dr. Mathews: As you may know, you need two abnormal copies of the FKRP gene to have LGMD2i (one copy from the mother and one from the father). This is known as an autosomal recessive disorder (compared to autosomal dominant disorders where you only need one abnormal copy of a gene to have a disease). If both mutations in an autosomal recessive disease are the same, it is called homozygous. If the mutations from the mother and father are different, it is known as heterozygous. The most common mutation in LGMD2i came from ancestors in northern Europe (the "Viking mutation"). About 90% of individuals with LGMD2i have at least one copy of this common variant (genetic change: c.826C>A, protein change: L276I). If they have 2 copies of the c.826C>A variant, the genetic change is homozygous. The rate of disease progression is somewhat related to the specific mutations.

What is the typical age of onset and the typical progression of the LGMD2i?

Dr. Johnson: Onset typically occurs in late childhood or the teenage years, though there can be a wide range in age of onset. The progression itself is slow, measured over the course of years.

Dr. Mathews: In the Iowa natural history study, half of participants with FKRP mutations had their first symptoms by age 8 years, but there was a range from infancy to well into adulthood. It sometimes takes years after the earliest symptom before a diagnosis of muscular

dystrophy is made. About 25% of the people in the natural history study have great difficulty walking or have lost the ability to walk independently. The earliest we have seen this is in late childhood.

Why is it so important to participate in natural history studies?

Dr. Johnson: Some of the answers described above are based only on clinical experience and individual patients. The natural history studies allow clinician investigators to measure change more accurately and understand the factors that may lead to slower or faster disease progression. This will lead to more standardized care, but more importantly, there is an opportunity to design better drug trials.

Dr. Mathews: Thanks to the commitment of people who participate in natural history studies, we have learned a lot about LGMD2i, some of which is mentioned above. We hope that what we have learned will help people with muscular dystrophy stay healthier. By doing the same tests every year, we are able to measure how rapidly certain functions change. This tells us how many people we need to study on a new potential treatment to know if the treatment is effective or not.

Can you list and describe the different clinical trials that are or will be available for 2i?

Dr. Johnson: Outside of the natural history studies, there is a Phase 2 clinical trial sponsored by ML Bio to determine if their compound can slow disease progression. There are several other companies that have stated their intention to begin gene replacement trials in the near future, but these are not open yet.

Dr. Mathews: A trial looking at a steroid in LGMD2i was closed after not enough people enrolled.

What is the importance of participating in the FKRP registry?

Dr. Johnson: This provides a good tool for investigators to contact you about these studies.

Dr. Mathews: The registry also collects information about people with the same disease all over the world. Some of the information about the disease can be compared to the information from natural history studies to develop a more complete picture of who has LGMD2i and how the disease affects people.

What are you most excited about for our LGMD2i community?

Dr. Johnson: I am very excited for what looks like multiple, potentially promising new therapies!

Dr. Mathews: I have been working on this disease for about 15 years and so I am also very eager to be able to test new treatment options!

We offer special thanks to Dr. Johnson and Dr. Mathews for their contributions to this article. On behalf of our entire LGMD2i community, we would like to extend our appreciation for all of their efforts to provide knowledge and hope to LGMD2i patients and their families. We are closer than ever to having a successful treatment for this condition and we look forward to hearing positive outcomes from the clinical trials that are starting for LGMD2i! ▪

A "Voice" for LGMD

The Voice Season 20 contestant Savanna Woods

> *Watching my mom live with this illness has taught me to take each moment and live it to the fullest.*

For those of us with limb-girdle muscular dystrophy, we understand how little light our rare disease shines on the grand stage of the neuromuscular disease community. Savanna Woods, however, who is competing on Season 20 of *The Voice*, is using her platform to shine light on LGMD in honor of her mom, Lacey Woods, a woman living with LGMD2i. Savanna considers Lacey her inspiration and calls her "[my] favorite person in the world."

Growing up, Savanna closely watched Lacey struggle daily. She also watched her never let those struggles define her. She says, "My mom has always taken care of our family, just like any other mom does. She is such a strong woman and has so much inner strength that it has influenced me in so many ways."

The Woods family is from Stanwood, Washington, and music, family, culture, and spirituality have always been woven into their lives. Singing has always been a big deal in the Woods' household. Savanna's father, Stewart, has been a singer-songwriter and musician for decades, and instilled that love for music in his children, too. In fact, one of Savanna's two bands includes her sisters, Ireland and Paige, and is called "The Woods Sisters."

In 2018, Savanna combined her love of singing and songwriting with traveling, and created videos on Facebook every Wednesday which featured her performing songs solo, or with family and friends. She coined these "Wandering Wednesdays," often putting a hashtag in front of the moniker. These videos would be recorded in places like Scotland, Spain, and Italy. When the pandemic hit in 2020, and travel restrictions occurred, she started live streaming these recordings from her home in Washington. Since the pandemic also caused live music venues to shut down, Savanna found herself with more free time to share her music online. It was then that she decided to try out for the virtual auditions on *The Voice*.

Regarding her singing style, Savanna says, "I'm not curating things to be a certain style or to be a certain way. I sing through my feelings. I just kind of follow my heart wherever it goes."

When it was her turn to compete in the blind auditions, she sang "Zombie" by The Cranberries, which wowed hosts Nick Jonas and Kelly Clarkson. Kelly said, "She is cool as hell. I love a creepy kind of vibe. I don't know if anyone like her has ever been on my six seasons of *The Voice* before. She's like if Patty Griffin and Nirvana had a baby."

After a tense battle between Kelly and Nick both vying for Savanna to choose them as

Below: Savanna and her mom, Lacey. Lacey has also been working to raise awareness and to help educate and support others with LGMD2i.

her coach, Savanna chose Kelly in the hopes that she would improve herself by learning more about the music industry.

Winning *The Voice* is not the ultimate goal for Savanna. Her goal is to reach and heal people through her music. That is what drives Savanna to keep writing and sharing her music with the world. She also hopes that her music will help raise awareness for LGMD.

Lacey has also been working to raise awareness for this disease. She created the LGMD2i website (**LGMD2i.com**) with the goal of educating and assisting others with the condition, and she started the LGMD2i Facebook group, which she moderates. She says, "These channels have given others who are on a similar journey as me a place to get answers, build friendships, and find hope and support."

Recently, *The Voice* featured Savanna and her family, an opportunity Savanna took to talk about her mom. "Watching my mom live with this illness has taught me to take each moment and live it to the fullest," said Savanna. "I just want to connect with more people and get my music out there."

Lacey said of her daughter, "People ask me all the time, 'Aren't you so proud of her getting on *The Voice*?' and I say, 'No! I am proud of her every day for waking up, working hard, and being a beautiful human being.'"

Savanna continues to advance on *The Voice* at the time of this writing. By the time this article is published, we will know more about how the show played out. One thing is for sure: whether or not she competes to the end, we have not heard the last of Savanna Woods!

You can follow Savanna on Facebook and Instagram at **Savanna Woods Music**. You can also join Lacey's LGMD2i Facebook group at **Facebook.com/groups/LGMD2i**. ▪

Contributed by Rebecca Lucas Gregg

Connect with Savanna

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Savanna Woods Music

Savanna Woods Music

The Importance of Genetic Confirmation

No one needs to tell Patrick ("Pat") Moeschen how life-changing genetic testing can be. From childhood to his mid-thirties, he thought he had Becker Muscular Dystrophy. Becker Muscular Dystrophy (BMD) symptoms include generalized weakness that first affects muscles of the hips, pelvic area, thighs, shoulders, and heart.

Pat Moeschen with wife Vanessa and sons Ted and Tim

When treatments start arriving, I want my name to be called, and knowing my subtype certainly helps. If you don't know your subtype, it is important to get a genetic test to find out. It is a simple blood test, and for me, it literally was life-changing!

In 1985, at the age of twelve, Pat was diagnosed with BMD by way of a muscle biopsy. Pat and his family learned how to live with the diagnosis and they treated every birthday as an achievement. He grew up and became a middle school music teacher, married Vanessa, and they built their life together in New Hampshire. They wanted to have children but were unsure about this decision since BMD can be so unpredictable.

Pat's disease did not stop him from fulfilling his love of travel. He traveled the globe and often spoke about muscular dystrophy at conferences, telling his story to many. It wasn't until 2011, when he was speaking at a muscle disease-related conference in Rome, that a doctor started up a conversation with him about his condition. The doctor listened to Pat's medical history and told him he felt Pat had been misdiagnosed and recommended a genetic test to find out for sure. Pat soon tested and learned that he actually has LGMD2E, which has a completely different disease course. Pat said, "My family and I felt like we had discovered a new relative with this diagnosis. My future now looked completely different and [it] changed my outlook. My wife and I felt better about having children since this disease course shouldn't affect my heart or end my life early. It gave me a new lease on life."

Since that new lease on life, Pat and Vanessa had two boys, Tim, age 6, and Ted, age 4. They still live in New Hampshire where Pat continues to work as a middle school teacher. He is incredibly grateful to know which subtype he has, especially given the excitement in the LGMD community about upcoming clinical trials for gene therapy. "When treatments start arriving, I want my name to be called, and knowing my subtype certainly helps. If you don't know your subtype, it is important to get a genetic test to find out. It is a simple blood test, and for me, it literally was life-changing!"

Pat's story is all too common for those diagnosed with muscular dystrophy and other rare diseases. The only way to know for sure which form of muscular dystrophy a person has is through genetic testing. Genetic testing consists of analyzing the cells in a blood, tissue, or saliva sample, and looking for specific mutations in the DNA that are known to be associated with a given disease or condition. If you are interested in obtaining a genetic test, we have included resources in this issue that may help you. ■

Contributed by Rebecca Lucas Gregg

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