LGMD-1D DNAJB6 Foundation

and

MYOSYND™

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7/29/2025

Thanksgiving Edition

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Thanksgiving Message: Building Family Connections in the LGMD Community

Thanksgiving reminds us of what truly matters—family, connection, and the support that carries us through life's challenges. For many families in the limb-girdle muscular dystrophy (LGMD) community, those connections are not just emotional—they're also scientific and deeply personal.

At this time, many families still do not have a confirmed genetic diagnosis. Sometimes testing is not yet available or affordable. Other times, results show what's called a Variant of Uncertain Significance (VUS)—a genetic change whose impact isn't yet fully understood. This can be frustrating, especially when symptoms are clear, but the science isn't there yet to confirm the diagnosis.

That's where family activism comes in. When families work together—sharing information, testing relatives with and without symptoms, and building a clear family pedigree (see family treeon page 4)—they help turn uncertainty into understanding. These efforts can reveal patterns of inheritance, especially in autosomal dominant LGMD, where the condition appears in multiple generations. By connecting the dots, families help scientists and clinicians upgrade uncertain variants to "diagnostic"—transforming "maybe" into meaningful answers.

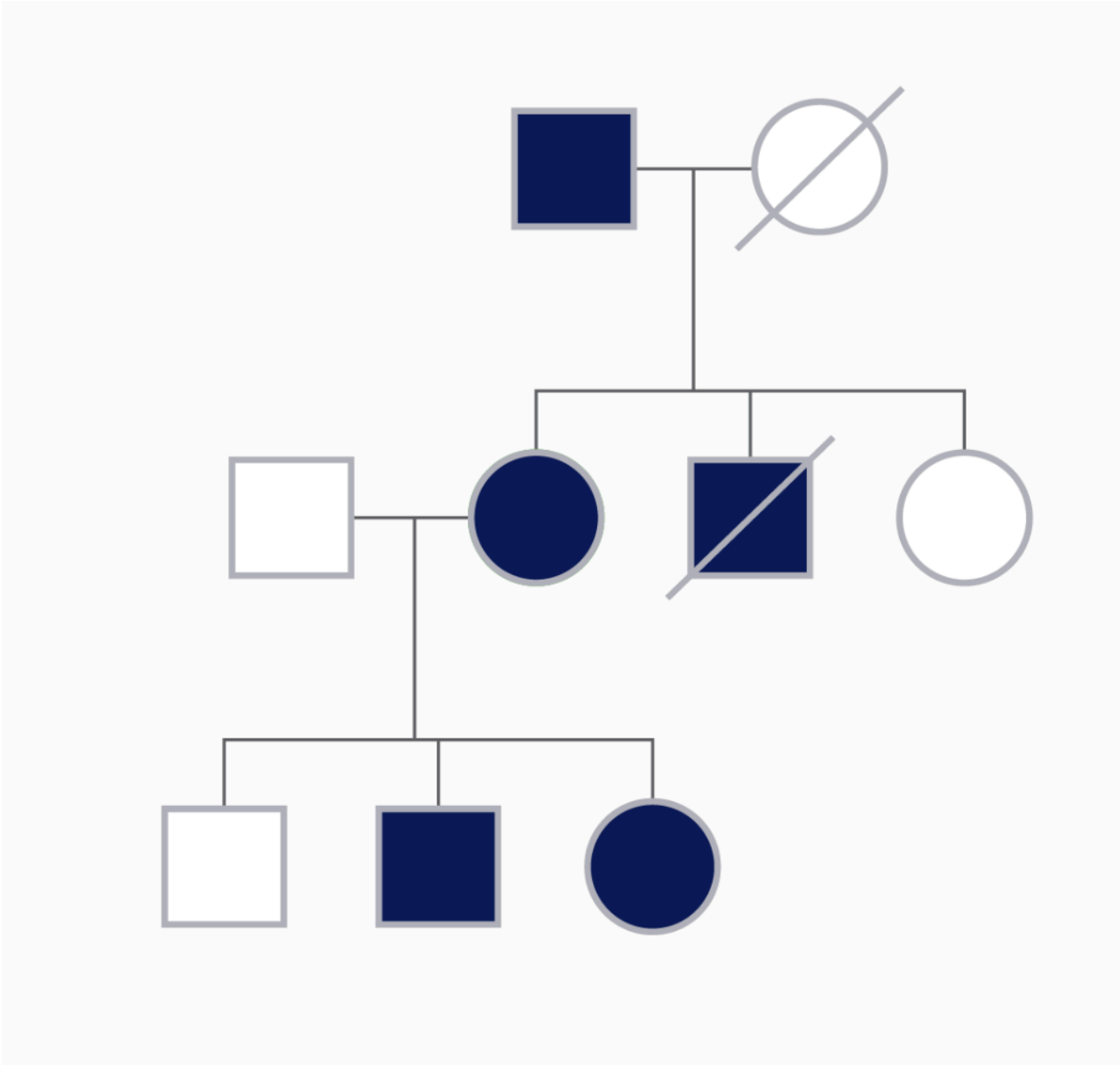
Since 2018, our foundation has partnered with sponsored genetic testing programs to make this process easier and more accessible. But the most powerful progress often begins at home—with families willing to explore their shared story.

Think of it like the Oklahoma Land Rush—every family staking its claim, marking the ground where their genetic story belongs. It takes effort, but the reward is lasting: knowledge, clarity, and the power to help future generations.

This Thanksgiving, we honor every family working together toward that goal. Your courage and collaboration are helping build the map that guides all of us forward.

Please see our evergreen links below and reach out to us for help.

Simple Family Pedigree Chart for a Family with Autosomal Dominant LGMD



Important AI Links

The foundation is heavily into AI through Chat GPT and others. Apparently our QR access codes to our custom built GPTs for the LGMD community were a big hit at the conference so we will offer an array of them in this newsletter. You will need a free Chat GPT account to use them. As always, we will make these for you.

LGMD ASSISTANT (find a clinic): Like most of us with LGMD or suspected LGMD, finding a neuromuscular doctor or clinic in your state or country can be problematic, with LGMD Assistant 3.0 you can type in your suspected variant, if known, and your state or country (be sure to ask for all addresses and phone numbers to be included) and the custom GPT will provide you with local MDA clinics and university hospital centers near you:

THERAPY: Autosomal dominant therapy questions for specific genetic variants answered here:

PLANNING A TRIP: LGMD person planning a trip to Atlanta, Paris, Rome or Riyadh? Check out Access Ally. Just type in the city and country, sites you want to visit and equipment you will need (ask for addresses and phone numbers of vendors to be included). The response will direct you to accessible facilities, rentals and all accommodations you will need. Do not forget to keep chatting with it to get all your questions answered:

LGMD INSURANCE NAVIGATOR: Having trouble getting authorizations for medications, DME or dreading the next insurance interaction, just type in your insurance and what you need and you will get answers. (again it may take several follow up chats but this will be very helpful, include available ICD 10 codes for LGMD subtypes and CPT codes below for what you need:

ICD-10-CM Diagnostic Codes for Limb Girdle Muscular Dystrophy (Effective for use beginning October 1, 2022–per CMS update)		
ICD-10-CM Code	**LGMD Subtype**	**Description**
G71.032	LGMD2A / R1	Calpain-3 dysfunction (calpainopathy)
G71.033	LGMD2B / R2	Dysferlin dysfunction
G71.0341	LGMD2D / R3	Alpha-sarcoglycanopathy
G71.0342	LGMD2C / R5 or LGMD2F / R6	Gamma or delta-sarcoglycanopathy
G71.035	LGMD2L / R12	Anoctamin-5 dysfunction
G71.038	Other genetically confirmed LGMD	Includes LGMD21 (FKRP), LGMD2 (TTN), etc.
G71.039	LGMD, unspescified	For cases wcithut confirmation
G71.031	LGMD1 (dominant)	Includes LMNA, MYOT, CAV3, etc.
G71.039	LGMB, unspesestified	Inlcudes LMNA, MYOT, C/

Common CPT codes for the LGMD community:

Service Type	CPT / HCPCS Codes
Walker / cane / platform	E0100–E0159
Seating / cushioning	E0181–E0199
Wheelchair accessories	E0992–E1002 etc.
Oxygen / respiratory support	E0424/E0431, E0468, E0470 etc.
Home ventilation	E0466
PT Evaluation	97161–97164
PT/OT Treatments	97110, 97530, 97542
Electrical stimulation	97032
Caregiver training	G0539, G0542
Psychosocial support	81404, 81405, 81406, 81408

OUR WEBSITE:

Important MDA Links

MDA Connect: HERE and HERE

(Helps find a neuromuscular doctor near you!)

MDA camp for attendees and volunteers HERE

MDA college scholarship program HERE

An emergency preparedness plan is crucial for those with LGMD

Important Patient Care Links

1. **I have weakness but my genetic panel just shows "variants of uncertain significance" (VUS).**

   Step 1: Test other family members with and without symptoms and take results to a genetic counselor.

   Step 2 Consider sponsored whole exome sequencing (WES) and whole genome sequencing (WGS)

   Harvard, Stanford Center for Undiagnosed Diseases

2. **Do I really have a genetic muscle disorder? See HERE**

3. **Do I have a mitochondrial muscle condition? Video1, Video2**

   United Mitochondrial Disease Foundation (UMDF) Genetic Testing For Suspected Mitochondrial Disease Program HERE . Sponsored testing that clinicians can order if a mitochondrial disease is strongly suspected.

4. **Are you having a muscle biopsy?**

Tissue referral to University of Iowa

University of Iowa Muscle Biopsy Requisition

5. **Gene matching sites, you must register all your VUS variants to connect with others to help establish a genetic diagnosis: MyGene2, GenomeConnect, Rare-X**

6. **Sponsored flights for rare conditions HERE and HERE.**

7. **Joe's House (discount lodging for patients with cancer and rare conditions)**

Important Foundation Links

1. LGMD1D DNAJB6 Foundation
2. Foundation YouTube Archive
3. Foundation assisted sponsored genetic testing
4. Foundation Autosomal Dominant Registry
5. Foundation: Solving Your Variant of Uncertain Significance (VUS)
6. LGMD1D AI Assistance and LGMD Apps

Ways to Donate

1. **OUR WEBSITE** **(a secure site with all the listings below)**
2. **PAYPAL** **(Our foundation's secure site)**
3. **CREDIT CARD** **(Network For Good credit card portal)**
4. **VENMO** **(@lgmd1d) Foundation Account**
5. **EVERY.ORG** **(ALL CRYPTO CURRENCIES)**
6. **SQUARE**
7. **If you are over 72 consider a Qualified Charitable Distribution (QCD) from a traditional IRA and lower that dreadful RMD and avoid that higher tax bracket. Also available to Roth IRA participants.**

   RMD calculator HERE, medicare income bracket HERE.
   tax bracket for SS HERE.

Thanks for your support and Happy Thanksgiving from us!

William Lowery MD