LGMD-1D DNAJB6 Foundation

and

MYOSYND™

501 (C)(3)

7/29/2025

Summer edition

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LGMD Conference Summary

Orlando, Florida

7/18-7/20/2025

A Success By Any Measure!

The president of the LGMD1D Foundation and Registry, Natasha Lowery, represented us in Orlando and connected with so many people. I want to thank everyone who made her feel welcome too.

The picture below is the autosomal dominant break out group which was a first in the history of the conference and thanks to all that supported our effort!

Conference News and Other Updates:

There are new videos from the conference that you will find HERE ranging from trends in genetic testing, the latest therapeutic options, and a panel discussion with a FDA rare disease specialist.

Although news of a gene therapy being shelved by the FDA and Sarepta was a downer, there are still roughly 550 biopharm companies working on various gene therapies HERE. There is no reason to give up.

Our autosomal dominant registry is approaching a milestone of 200 members since inception in 2018. Our registry is worldwide and adding members. We need our members to remind their neuromuscular doctors to refer new AD individuals to us since we are research ready.

THERAPY NEWS FLASH: Our secret researcher weighed in on their view of ASOs as a treatment option for AD LGMD; we have discussed in our newsletters: "Regarding ASOs & siRNAs: Dyne & Avidity therapeutics look like the real deal for muscle delivery. Their myotonic dystrophy and FSHD trials are looking very promising. Ionis pharmaceuticals - a behemoth in the ASO space (nusinersen, tofersen, etc), had been avoiding any muscle related therapeutics, but has now developed their own chemistry for targeting muscle. It looks very promising. https://pubmed.ncbi.nlm.nih.gov/40207629/. Yes - ASOs & siRNA are absolutely a viable avenue. It's the main avenue I'm currently pursuing in my NIH funded research."

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: https://www.mda.org/care/connect

(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.

Thank you for your support and all the best from us!

William Lowery MD