

LGMD-1D DNAJB6 Foundation

and

MYOSYND™

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2023 MDA Conference

The Muscular Dystrophy Association's annual conference was in person and virtual this year. There was a spectrum of rare neuromuscular conditions and more common conditions including ALS, Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA) presented. Many exciting genetic therapeutic approaches were discussed. In summary, genetic therapy is helping to stabilize deteriorating muscular and neuropathic conditions with a few observations of my own listed below.

1. Biopharm interventional trial (BIT) presentations were liberal in patient inclusion, especially patients in wheelchairs with emphasis on arm and upper body strength improvement or preservation. In addition, the studies were international in some cases to enhance numbers and ethnicities.
2. BIT presentations whether cell, genetic or small molecule therapy showed mild to modest functional effect.
3. Two methods of gene delivery to muscle cells are evolving: using a neutered virus as a delivery system, or in some cases, a non viral package encasing the therapeutic gene to enhance entry into the muscle cell. Both work and are actively being refined. The issue now is the high doses of gene with either vector needed to deliver enough good gene inside the affected muscle cell. Finally an individual's own cells can be reprogrammed to function correctly.
4. The high doses of gene product and the use of a neutered virus as a delivery system are the main causes of side effects in my own medical opinion and there seems to be a consensus. Side effects are usually not severe and include flu symptoms, nausea, mild kidney and liver injury which subside.

2023 MDA Conference

5. Although few LGMD studies were presented, the lessons being learned from the ALS, DMD and SMA experience is valuable to the LGMD universe.
6. New techniques for VUS (variants of unknown significance) resolution are emerging.
7. Patients with symptoms of LGMD can be connected with their local MDA clinics and in some centers this can be virtual. See link or QR code.

MDA Connect: <https://www.mda.org/care/connect>



8. Video recordings of some of the lectures can be watched on the Foundation YouTube channel [HERE](#).

Important Links

Since our inception in 2018 we have collected links to help everyone's journey. Due to the onboarding of new members and new links I am adding this as an evergreen list to our newsletter.

[LGMD1D DNAJB6 Foundation](#)

[LGMD1D Foundation YouTube Archive](#)

[LGMD1D Foundation assisted sponsored genetic testing](#)

[LGMD1D Foundation Autosomal Dominant Registry](#)

[LGMD1D Foundation: Solving Your Variant of Uncertain Significance \(VUS\)](#)

Sponsored Whole Genome Sequencing: (next step for VUS resolution)

[NIH](#)

[Harvard](#)

[Stanford Center for Undiagnosed Diseases](#)

Do I really have a genetic muscle disorder? See [HERE](#)

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.

Important Links

Are You Having a Muscle Biopsy?

[Tissue referral to University of Iowa](#)

[University of Iowa Muscle Biopsy Requisition](#)

Gene matching sites, you must register all your VUS variants to connect with others to help establish a genetic diagnosis: [MyGene2](#), [GenomeConnect](#), [Rare-X](#)

Sponsored flights for rare conditions [HERE](#) and [HERE](#).

[Joe's House](#) (discount lodging for patients with cancer and rare conditions)

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

