LGMD-1D DNAJB6 Foundation

and

MYOSYND™

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11/22/2023

Thanksgiving Edition

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3rd International LGMD Conference

Happy Thanksgiving to all of our members and especially for your enduring support through 2023. We look forward to 2024 and the expansive progress that will continue in the diagnosis and therapy for LGMD.

The 3rd International LGMD Conference in Washington , DC, October 27-29 offered in person and virtual attendance. It was a success by any measure and the video highlights can be found at the organizer's website, The Speak Foundation HERE. The Foundation also provides selective videos HERE. The highlights include genetic diagnostics for variants of unknown significance and steps for diagnosing LGMD. On the therapeutic front, Duchenes and autosomal recessive LGMD disorders received the most attention, however, progress for autosomal dominant LGMD is catching up and equally viable. Finally, gene editing, cell therapy, blocking RNAs and their respective vectors are all in the final stages for clinical utility. In summary, the event was a culmination of industry support and academic excellence with more hope to follow. Again, Happy Thanksgiving!!

The MDA Conference

The next MDA conference is March 3-6, 2024 in Orlando, Florida with virtual attendance available. I will be attending virtually and list specific highlights and videos. Please see the registration link HERE.

This conference incorporates more neuromuscular conditions than LGMD, such as ALS, SMA, Charcot Marie Tooth disorder, mitochondrial disorders to name a few. There tends to be therapeutic reports including autosomal dominant conditions of all disorders noted above. Finally posters and abstracts are presented in person and virtually which finds a wider audience. Please see some videos from prior years HERE.

AI, Chat GPT, LGMD and You!

You cannot escape the hype but this is actually a thing. Although I am a physician, I have been programming computers off and on since age 15. Now, a company named Open AI developed a Large Language Model (LLM) called Chat GPT 3.0, 3.5 and 4.0 which provides a way to write instructions to Chat GPT without knowing any computer programming language. This automates and organizes just about any life chore. Imagine writing simple instructions to a search process that returns an organized list of tomato salads from around the world.

I have written several GPT apps including "LGMD Assistant 3.0" that shares genetic analysis, lists MDA and university neuromuscular clinics in your state and free genetic testing sites in one response. Also "Access Ally" which reports accessible wheelchair venues, disability equipment rentals and transportation needs for all wheel chair travel plans in one response for any city in the world. The bad news is that I cannot share the direct app link for you to access without an account. I can share the output by an email link.

Chat GPT 4.0 which is 20 dollars a month, allows me to write and link my programs to anyone with the same account. Alternatively, there is a website called YouAI which is free to develop and share these apps on a limited basis. I am happy to pursue any of these options with those who are interested.

See LGMD Assistant 3.0 HERE and HERE, Access Ally HERE and 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.

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

MDA Connect: Helps find a neuromuscular doctor near you!

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.

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