LGMD-1D DNAJB6 Foundation

and

MYOSYND™

A 501 (C)(3)

11/14/2022

Thanksgiving Edition

Volume: 1, issue: 10

Table of Contents:

Thanksgiving Reflections	**Pages 2**
Home Medical Care for LGMD	**Page 3-4**
Important Links	**Page 5**
Ways to Donate	**Page 6**

Thanksgiving Reflections

The Foundation was started in 2018 as a registry for autosomal dominant limb girdle muscular dystrophy, a group that was poorly organized and deserving of research and therapeutic options. To date we have 124 persons with genetically proven autosomal dominant LGMD including LGMD 1A to 1H (older classification) and all varieties of Myofibrillar Myopathies. Our group includes people from Russia, Middle East, Europe, UK, US, Mexico and Australia to name a few. We are thankful for the explosion of genetic therapies including blocking oligonucleotides which are now in routine clinical practice requiring only a subcutaneous injection monthly or quarterly to eliminate the damaging effects of a deleterious autosomal dominant gene.

In addition, since 2019, the Foundation has engaged in sponsored genetic testing to help others in their arduous journey to find a genetic diagnosis for their muscle weakness. To date we have helped 350 people including their family members in an attempt to resolve their "variants of unknown significance", (VUS). In 30 % of cases a definitive diagnosis was established on initial testing. There were cases of Duchenne's Muscular Dystrophy (DMD), and Spinal Muscular Atrophy (SMA) where therapy was offered immediately. Regarding the 70% of persons who have VUS and weakness, we have been able to resolve a few VUSs to a definitive genetic diagnosis which is very important if genetic therapy is an option. We are thankful that we have met so many people with rare muscle conditions, in underserved areas and populations; and given them hope that someone is listening.

Home Care for the LGMD Community

As a practicing physician for 35 years in internal medicine, pulmonary and critical care, I can attest that I have seen a few things and learned how to efficiently navigate patients through the complexities of our monolithic medical morass. However, changing hats, I am writing as a physician with late onset LGMD-1D. I have had to give up the critical care component due to weakness and now do pulmonary and outpatient internal medicine at a more comfortable pace.

With that in mind and understanding how difficult it is to visit the doctor with LGMD, I have put together a series of short videos regarding the things you need at home to supply the doctor with all the data he/she needs for a complete virtual visit. There is a list of devices and over the counter medications that can make it less likely that you need to go to the office or to a pharmacy to take care of routine problems. You will need a smartphone and a computer with internet access and a doctor who accepts email.

Full disclosure: I have no financial arrangements with any of the products displayed.

Legal: Final approval and appropriateness of medications and medical applications presented here are between you and your physician.

Home Care for the LGMD Community

Videos

These videos are not professionally done, the message is more important than the medium. The first three are narrated pictures followed by live demonstrations. These are helpful for anyone trying to save time, money and the inconvenience of medical offices.

1. Introduction
2. Over the counter medications
3. Medical application pictures
4. Taking your temperature and pulse oximetry
5. Taking your blood pressure
6. Home otoscope (ear) and oral scope
7. Home EKG
8. Home colonoscopy: Cologuard

   Detailed instructional video

9. Do you have a urinary tract infection: the dipstick
   Detailed instructional video

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

Are You Having a Muscle Biopsy?

Tissue referral to University of Iowa

University of Iowa Muscle Biopsy Requisition

Do I really have a genetic muscle disorder? See HERE

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

Ways to Donate

1. **OUR WEBSITE** **(a secure site with all the listings below)**
2. **AMAZON SMILE** **(list the LGMD-1D DNAJB6 Foundation for donation with each purchase at no cost to you.)**
3. **PAYPAL** **(Our foundation's secure site)**
4. **CREDIT CARD** **(Network For Good credit card portal)**
5. **VENMO** **(@lgmd1d) Foundation Account**
6. **EVERY.ORG** **(ALL CRYPTO CURRENCIES)**
7. **SQUARE**
8. **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