# LGMD-1D DNAJB6 Foundation

## A Journey for Limb Girdle Muscular Dystrophy

**A 501 (c)(3)**

**The Newsletter from William S Lowery M.D.**

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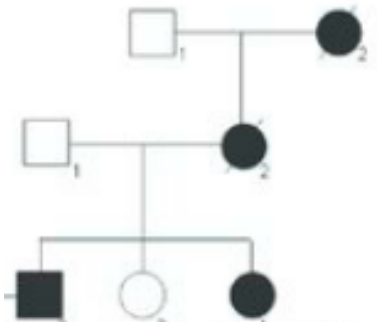
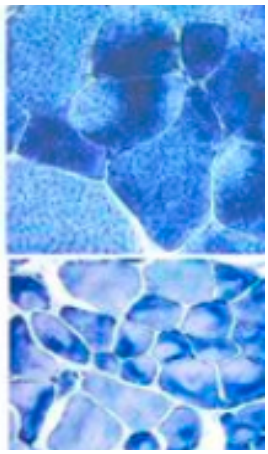
# Happy New Year to the Board!

**A big BIG thank you to all who donated to the foundation in 2018! And for those who are using Amazon Smile, your purchase contributions keep rolling in. We have reached our initial goal of 10,000 dollars and working on the next leg of our journey. Your generosity helps those affected with LGMD-1D to travel to Washington University at St Louis department of neurology to participate in a natural history study after which a genetic cure will be imminent. The Muscular Dystrophy Association and government grant organizations have contributed millions of dollars toward limb girdle muscular dystrophy (LGMD) for 2019 of which a fraction will be going to our sub type LGMD-1D. Our foundation will provide, in part, those expenses not covered by these grants.**

# What is LGMD-1D?

**Limb girdle muscular dystrophy is an adult onset muscular dystrophy with autosomal dominant genetic transmission to offspring (see the typical pedigree chart and muscle biopsy below). Offspring have a 50:50 chance of getting the abnormal dominant gene from one affected parent (dark circle or square). Since the gene is "dominant" it will dominate the normal gene donated by the normal parent and the disease will always manifest. This is analogous to an altered word in a sentence which confounds the reader and disrupts the intended meaning. A word editor is a common technology and the analogy to genetic editing will come into focus.**





# So LGMD-1D is a genetic disease?

**Yes, of the 10,000 estimated monogenetic diseases (one word altered in a sentence) LGMD-1D is rare compared to other common genetic diseases such as sickle cell anemia, cystic fibrosis and hemophilia. Estimates put the total LGMD population up to 500,000 affected worldwide with LGMD-1D being 6th place in the 30 subtypes of LGMD. LGMD-1D is the most common autosomal dominant LGMD as opposed to autosomal recessive or X linked genetic muscular dystrophies. The wheel diagram "A" below shows a rough incidence of the "DNAJB6"gene by the blue dot, which is the affected gene of the LGMD-1D subtype located on chromosome 7. Each altered gene in the wheel represents another type of muscular dystrophy in the LGMD spectrum. Chart "B" shows the red dot below the "DNAJB6" (the LGMD-1D subtype) bar indicating relatively few "altered words" that need to be "edited." This gives us hope that only a few "words" in our "story" will need editing.**

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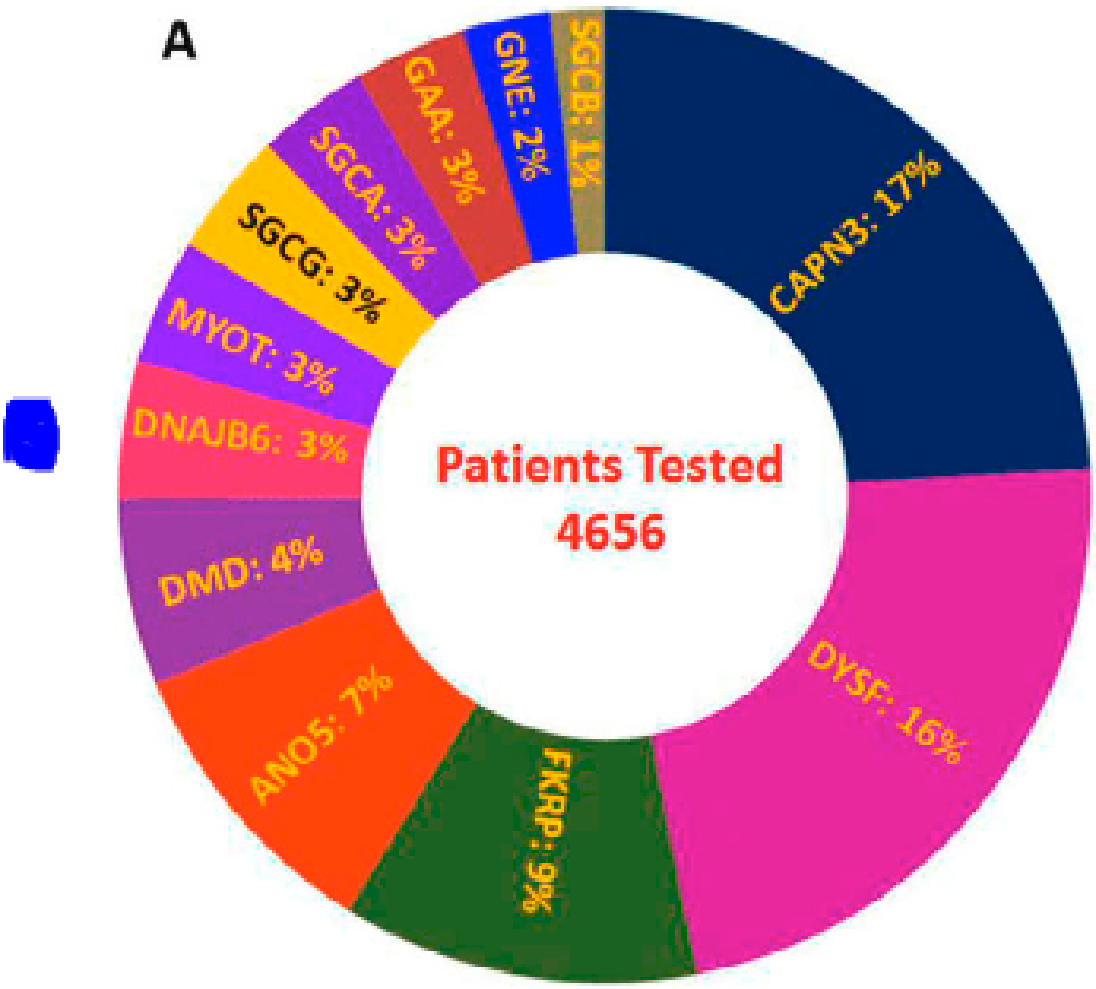
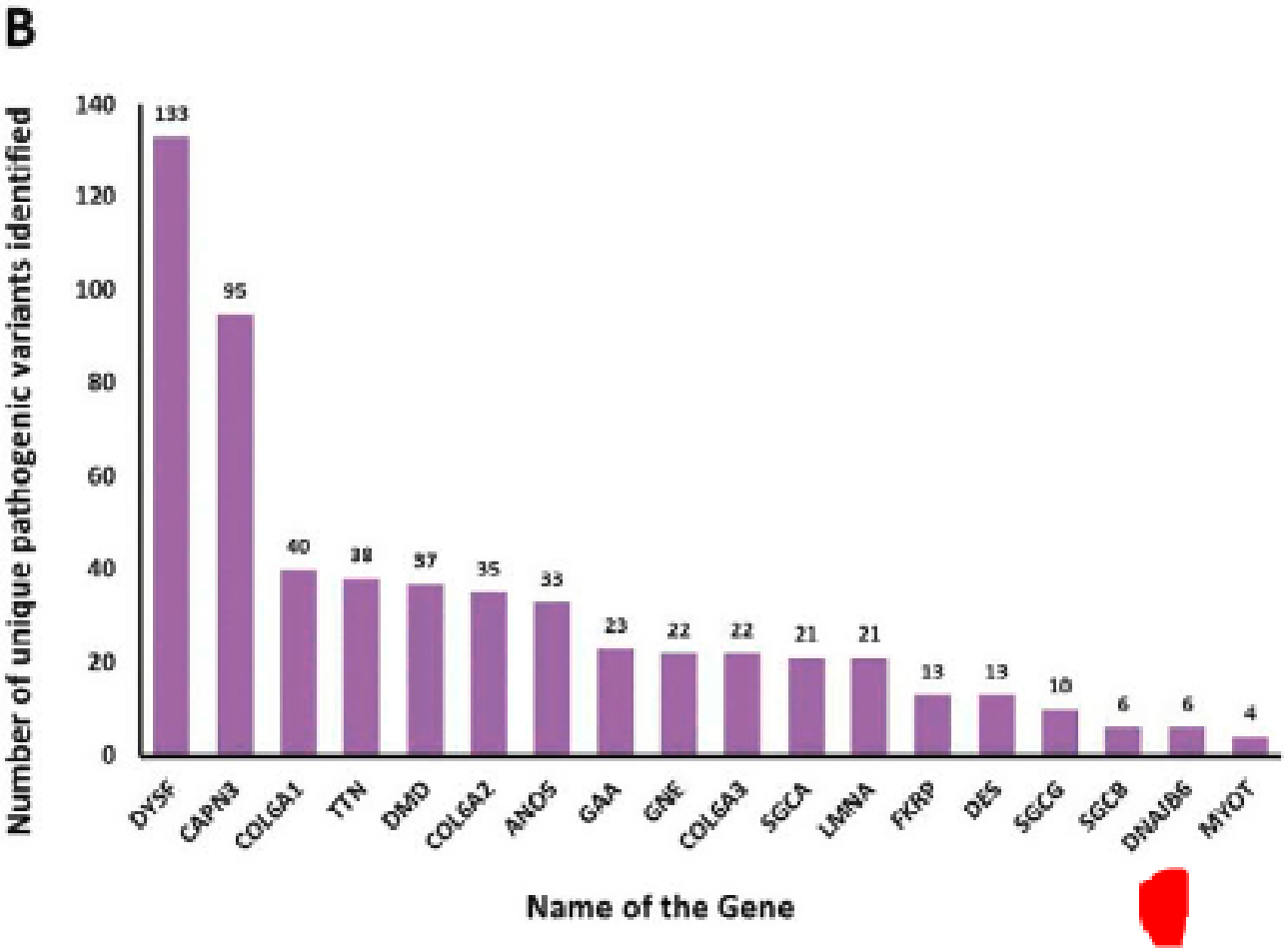

A
CAPN3: 17%
DYSF: 16%
FKRP: 9%
ANO5: 7%
DMD: 4%
DNAJB6: 3%
MYOT: 3%
SGCG: 3%
SGCA: 3%
GAA: 3%
GNE: 2%
SGCB: 1%
Patients Tested
4656


B
Number of unique pathogenic variants identified
140
120
100
80
60
40
20
0
133
95
40
38
37
35
33
23
22
22
21
21
13
13
10
6
6
4
DYSF
CAPN3
COL6A1
TTN
DMD
COL6A2
ANO5
GAA
GNE
COL6A3
SGCA
LMNA
FKRP
DES
SGCG
SGCB
DNAJB6
MYOT
Name of the Gene

# Are there genetic cures now?

**Yes, for example, autosomal dominant blindness and hemophilia have been cured by gene editing. That is, changing the one altered word in their sentence "cures" their whole story. Imagine a hemophiliac requiring 1 million dollars worth of blood products a year and countless hours in clinics not to mention their pain and suffering, all relieved and permanently cured by one 30 minute infusion of a genetic editor. Extrapolate that to the 10,000 monogenetic and orphan diseases languishing for this intervention and one can appreciate the power of this technology.**

# How do my donations help?

**Patients suffering from LGMD are unlikely to be fully employed or require special expenses to travel. Studies to find a genetic cure are located in distant regional centers of excellence therefore financial assistance is needed to transport patients to these centers. The number of patients we need for an adequate study requires us to locate everyone possible and facilitate their enrollment at a regional center. These logistical and travel concerns are usually not covered by research grants. Your donations help defray the costs to the most needy in the LGMD community to reach these centers of excellence. Finally, we represent the tip of the iceberg for many more people with other orphan genetic diseases who will benefit from the genetic tools and methods developed from this research.**

# How do I stay involved?

**There are many ways to help and donate:**

1. **Use Amazon Smile and list the LGMD-1D DNAJB6 Foundation for a donation with each purchase at no cost to you.**
2. **Consider us for an annual donation.**
3. **Visit our website www.lgmd1d.org for other donation avenues including IRA tax deductible contributions.**
4. **Contact me personally for your professional advice regarding any of our content including the website, newsletter, fundraising and medical to name a few.**
5. **Forward any of our information to people who would be interested in getting involved or have a muscular dystrophy that needs further help in diagnosis or referral.**

**Together we will make a difference!  Again thank you for for time and generosity.**

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