Additional Patient Comments from Surveys and Online Submission Portal

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September 23, 2022

MEETING HOSTED BY:
Coalition to Cure Calpain 3, the Kurt+Peter Foundation, the LGMD2D Foundation, the Speak Foundation, Cure LGMD2i, and the McColl-Lockwood Laboratory for Muscular Dystrophy Research

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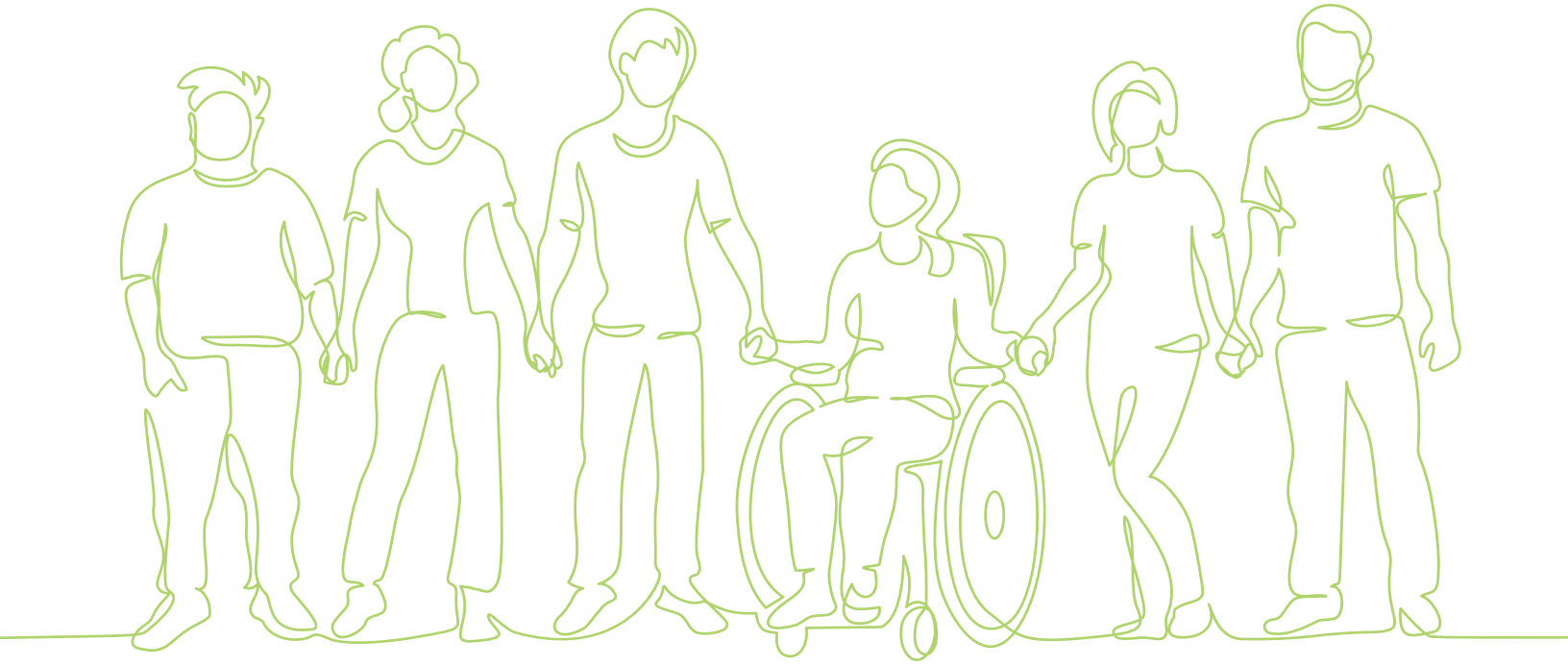
Contents



Additional Patient Comments from Surveys and Online Submission Portal

Limb-Girdle Muscular Dystrophy Externally-Led Patient-Focused Drug Development Meeting

LGMD Coalition hosted the LGMD Externally-Led Patient Focused Drug Development (EL-PFDD) meeting on September 23, 2022. This EL-PFDD meeting was modeled after the work of the FDA's Patient-Focused Drug Development (PFDD) initiative, a systematic way of gathering patient and caregiver perspectives on their condition and on available treatments. The information gathered at the meeting is presented in the *Voice of the Patient* report, which may be used to guide therapeutic development and inform the FDA's benefit-risk evaluations when assessing therapies to address LGMD. The report is publicly available for the many stakeholders in the LGMD community including the FDA, other federal agencies, academics, clinicians, advocacy and professional organizations, biopharmaceutical companies, and universities from across the world.

To ensure that as many voices as possible were heard, the LGMD Coalition as well as several international partner organizations conducted surveys prior to the meeting. In addition, an online comment submission portal was open for one week before and four weeks after the LGMD EL-PFDD meeting. Survey results and submitted comments are presented in this document, sorted by LGMD subtype and respondents are identified by their first name only (when known). Selected comments are included in the main body of the *Voice of the Patient* report. *The Voice of the Patient* report is available online at LGMDPFDD.com along with the meeting recording, the meeting transcript, as well as this document.

Comments were edited only slightly (if at all) for spelling and punctuation, and the following comments were not included in this document:

- Comments that did not address the Meeting Discussion Questions from **Appendix 4** of the *Voice of the Patient* report.
- Comments from individuals with LGMD subtypes other than 2C, 2D, 2E, 2F, 2A and 2i, as these subtypes will hopefully be included in a future EL-PFDD meeting.
- Comments perceived to be disrespectful to individuals, companies or organizations.
- Comments promoting commercial products or requests for resources.
- LGMD Coalition noted that many patients and caregivers used the comment portal to simply ask when a cure will be available; these comments were removed.

This EL-PFDD meeting had global participation. Many submitted their names, locations and the LGMD subtype represented, but didn't provide further information. These include: Dilek, Emre, Mukaddes (LGMD2C), Esra and Ishak (LGMD2D), Kemal (LGMD2E) and Bahar (LGMD2A) from Türkiye; Nina from Denmark (LGMD2i); Shukriah in Yemen; Ali and Mohamad (LGMD2i) from Iran; Carlos from Spain; Hamzh from Egypt; Gagan, Manish, and Syed (LGMD2A) from India; Tatiana and Sergey (LGMD2E) from Russia; Amal (LGMD2C) and Afaf (LGMD2A) from Tunisia; Asma (LGMD2A) from Libya; and Gulam (LGMD2A) from Pakistan.

LGMD2C / LGMD R5 γ-sarcoglycan-related

42-year-old woman living with LGMD2C (diagnosed at 9 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

1. *"Muscle weakness (limbs, trunk, breathing muscles)"*
2. *"Inability to walk, transfer on my own"*
3. *"Fatigue (upon waking up, exhausted feeling 24/7)"*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"BREATHING. I cannot lay down to sleep without non-invasive ventilation to help me breathe and the weakness is causing the need of ventilation during the day as well sometimes. BATHING. TOILETING. DRESSING. Basically, conducting daily living activities on my own is no longer possible due to the weakness. WALKING or MY MINUTE ABILITY TO TAKE A STEP OR TWO. It has recently become difficult to take a step to allow myself to be easily transferred to my w/c or elevated toilet chair. My future without bearing weight on my legs anymore is daunting."

What worries you most about LGMD?

"I've been fighting this disease since childhood, I am now 42. My fear is that a cure will never come. That I will continue to lose abilities people take for granted. LGMD2C is taking my breathing, my joy to sing and sometimes even to have enough air to speak. It's already changed the texture of my heart muscle, for now it's pumping okay but soon what I have to look forward to is it soon failing as well. I fear choking on my own fluids since the weakness doesn't allow me to cough productively anymore either. I fear not being able to swallow, eat the foods I enjoy. I fear the future needing care 24/7 and no one or no place humanly suitable being present for me. I'm living with a disease that isn't just satisfied taking my ability to move my limbs, to walk, sit and stand but it also wants my ability to breathe, swallow and heart beat."

Please describe your experience with treatments and other approaches to manage LGMD.

"I have never received treatments. I have always been told there was nothing to do. Only have had the routine examinations to observe progression. On my own I have taken supplements; I cannot say any have worked. I've had a day of PT off and on referred by muscle clinic maybe a total of 3 times in my life where I was shown and given exercises to do at home to continue doing myself but truthfully I never continued them. Maybe I would have continued the exercises with a physical therapist but I was never given that option."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"If the progression of the disease could just stop and maybe I could regain some percentage back to be able to breathe normally and have a productive cough on my own, I dare to say I could live with that. Even if it still meant not being able to toilet or bathe on my own or walk."

Parent/guardian/caregiver of a 4.5-year-old son living with LGMD2C (diagnosed at 1 year)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Slow to stand up"

"Slow movement"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Stand up"

"Movement"

What worries you most about LGMD?

"Fall down"

Please describe your experience with treatments and other approaches to manage LGMD.

"Vitamin"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Good movements"

Parent/guardian/caregiver of a 13-year-old son living with LGMD2C (diagnosed at 5 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Not walking"

What worries you most about LGMD?

"Immobility"

Please describe your experience with treatments and other approaches to manage LGMD.

"Job qualification"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Yes"

23-year-old woman living with LGMD2C (diagnosed at 22 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Not being able to run and get up from the ground and get tired quickly"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"I couldn't get a job, I wanted to be a doctor"

What worries you most about LGMD?

"I'm afraid that there will be no cure"

Please describe your experience with treatments and other approaches to manage LGMD.

"I would be worse if I exercised"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I want gene therapy"

40-year-old woman living with LGMD2C (diagnosed at 16 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Not being able to walk, raise my arms, or stand up from a seated position."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Yes, one of my biggest and hardest struggles is honestly accessibility to restrooms as well as always having to have help even at home. Every activity or task is a struggle of some kind and I'm constantly needing help. I would love to be able to do things on my own."

What worries you most about LGMD?

"I worry about it continuing to get worse and causing my health and quality of life to diminish. I also worry about dying at a young age as well as not having family to help support me and being all alone. Being alone really worries me."

Please describe your experience with treatments and other approaches to manage LGMD.

"Diet helps me feel overall better but there has not been anything to really help manage LGMD."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Stopping the progression"

27-year-old woman living with LGMD2C (diagnosed at 6 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Stiffness, Scoliosis pain, lumbar pain"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"I can't do all the simple daily human activities: walk, shower, get food, toilet brush, my hair clean my teeth, etc,"

What worries you most about LGMD?

"Get more sick. I wish I could stop my LGMD"

30-year-old woman living with LGMD2C (diagnosed at 15 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Inability to walk alone"

"Difficulty going up and down stairs"

"Inability to do some chores"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Yes, almost all activities"

What worries you most about LGMD?

"Disease progression over time"

Please describe your experience with treatments and other approaches to manage LGMD.

"I don't take any medicine, just vitamin D, I think exercise and swimming are good too"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Gene therapy is the solution"

Parent/guardian/caregiver of a young daughter LGMD2C patient

1. **How symptoms affect life:**

   "Symptoms have changed my daughter's life and the whole family has had to limit everything for her."

2. **The first 1-3 symptoms that have the most significant impact on your life:**

   "– Acute abdominal pain – Fatigue when climbing stairs – Difficulty running and playing like all children"

3. **Describe a good day and a bad day:**

   "– The best day was when my daughter came home from school all happy and proud because she talked to her classmates about her condition and that she would be bedridden and told them that none of this would make any difference to her because disability is not an impairment – The worst day for us was when I went with her to the kindergarten party in the theater, she has all the time because the director prevented her from participating with her colleagues in the show because she is sick and especially when just discovered the disease"

4. **Activities that you cannot do or that you do partially:**

   "climbing stairs"

5. **How your symptoms have evolved over time:**

   "– At the beginning it was a difficulty to climb stairs but now the lifting of the floor has become quite difficult with a loss of strength at the end of the day"

6. **How your ability to cope with the symptoms evolves over time:**

   "physical, psychological and emotional impacts: on the physical level the capacities slowly decrease with time but on the emotional level my daughter keeps a very good psychology because we have already made a lot of efforts to reach a stage where she is totally convinced and especially that she is different and is not inferior to any person"

7. **What you fear most is what worries you the most and what frustrates you the most:**

   "what really worries my daughter is that she tells me I don't want you to leave me and my daddy I can't do anything without you both."

8. **How you manage your symptoms:**

   "– Medical treatments: corticosteroids 10 days on / 10 days off with vitamin therapy – Physiotherapy: 3 sessions per week with a balneotherapy session (only one because the child goes to school),"

9. **The efficiency with which these treatments treat your most important symptoms:**

   "balneotherapy with an Arnica oil massage and a Magnesium salt bath are the best for the symptoms"

10. **The negative aspects of your current treatments:**

    "corticosteroids are not tolerated by the stomach"

11. **In the absence of a complete cure, what characteristics are you looking for in a treatment:**

    "we are looking for a curative treatment such as gene therapy"

Nikki, parent of a 13-year-old son living with LGMD2C

Comment 1:

> *Our son is almost 13, he has so many questions that unfortunately we, his parents, or his doctors can't answer. He reads online all the progress and steps being taken for other types of MD, but we need more focus on LGMD. It is heartbreaking to watch him struggle to do the many things we take for granted everyday, such as brushing his teeth, getting a drink of water, or getting in a car. Our son has the biggest heart and is very positive, but everyone has their bad days. I would love to be able to give him some great news for his 2C diagnosis.*

Comment 2:

> *Our 13 yr old son struggles emotionally. Not being able to play with friends, go to birthday parties, play sports or even have fun at recess. He has a great group of friends, but other kids are not as understanding, mean almost. He constantly feels like a burden and apologizes often. Walking and daily functions are a struggle. He just wants to be able to hang with his friends and not feel like crap for the next few days.*

Nahira, woman living with LGMD2C

> *I am a patient with LGMD2C from Puerto Rico, and I was misdiagnosed with Spinal Muscular Dystrophy for 25 years. I used to walk with difficulty, but I had overall body weakness. In my teenage years, I lost my ability to walk and stared using an electric wheelchair. In 2018, I was formally diagnosed with LGMD and been learning more about it every day. The most significant impact in my life is how I have seen that over time I lost my ability to be independent and I am not able to do basic activities of daily living such as getting in and out of the bed, dressing, bathing, using the toilet, grooming, doing my hair, doing house chores, meal preparation, among others.*
>
> *I am not able to walk or do any activities that involve body transfers or strength, including using my arms for any activities requiring lifting, carrying, pulling or pushing anything with my arms, which is very frustrating. I suffer from fatigue at the end of my days.*
>
> *My LGMD has affected too by feeling chronic pain, mostly in my hips and I have developed other conditions due to it such as osteoarthritis. The only functional ability I had was standing and in 2020, I lost my ability to stand too. It has been difficult to depend on family members to be able to continue living my life. My caregiver is my mother, and she lives day by day just caring for me and attending to needs 24 hours a day.*
>
> *My best day is when I don't feel muscle pain and I am able to work, spend time with my little niece and I am able to go out of my home.*
>
> *I have lost my ability to do so many things in my life. I stopped walking and being able to use my arms in my young 20s. I used to be able to be very athletic as I live in Puerto Rico, my life and hobbies involved swimming, surfing, and just being by the sea. Outdoor activities were the main focus in my life, including hiking, kayaking, camping, among other. Unfortunately, I am notable to do any of those activities anymore. Water was my home, and it was a therapeutic experience. Now, due to my LGMD, I can't even move with my wheelchair in the sand;it's almost impossible here. I used to be able to play the violin as I come from a music background family, and now I can't lift my arms to even hold my toothbrush in the mornings.*

I have tried not to let my Limb Girdle limit me. I love life and I have tried to keep fighting this disease to achieve all my goals. I have learned to cope with my condition by focusing on how to help other feel empowered and to assist other young individuals with MD.

Even if I can't do any of the activities of daily living, I successfully graduated with a master's degree in counseling and a law degree and I am in the passing the bar exam. I have dared to travel and see the world and have been able to visit other countries. I can drive with a modified vehicle, and it took me 10 years to be able to achieve that. My hope is that soon we will all be able to receive treatment to stop the impact and to redevelop my muscle and body, to be able to do everything I used to and to be able to walk and feel the sand in my feet, to swim as far as I used to, to be able to hold my violin and play, to be able to continue traveling around the world and to live life independently again.

Luca, 37-year-old man living with LGMD2C

Comment 1:

I'm Luca, I'm 37, I work as a mechanical engineer. I have LGMD2C. The only symptom affecting my life is a lack of muscle strength. Respiratory and cardiac symptoms are very limited. I can drive the car and I can move alone for my work and entertainment commitments. Most of the time spent at home is on the computer to work or play. The thing that worries me the most is that I no longer have enough strength to lead and lose my independence.

Sometimes, I am frustrated by the time it takes me to do activities that take seconds for an able-bodied person.

I do physiotherapy at home to maintain muscle elasticity and take some medications to compensate for heart symptoms. I don't do any particular treatment.

In the absence of a complete cure, I would want a drug that can improve muscle strength so that I can get back to standing and walking on my own.

Comment 2:

The only symptom that affects my life is the lack of muscle strength so I do physiotherapy at home to maintain the elasticity of the muscles. In addition to this, I pay particular attention to the positions during sleep to compensate for the contractions of the tendons.

In the absence of a complete cure, I would want a drug that can improve muscle strength so that I can get back to standing and walking on my own. the only factor in deciding whether to do a new treatment would be side effects versus efficacy

Emanuela, 47-year-old living with LGMD2C

"I am ill with LGMD2C, I am 47 years old, I write from Italy.

My worst symptoms now are the lack of respiratory autonomy, weakness especially at certain times of the month and the lack of cardiac resistance.

A good day is when I breathe well enough. Over time I have always gotten worse. I am only concerned about having the management of my life, now I live alone with caregivers. For my problems I use the ventilator, but I am unable to do physiotherapy for financial reasons. I am hoping for a cure that stops the worsening. Thank you"

Ana, living with LGMD2C

"I'm Ana from Serbia. After 3 muscular biopsies, I got the diagnosis of Limb girdle MD type 2C. First symptoms I felt on my dance classes, I had terrible pain in my gluteal area. Slowly I started to lose my strength and muscles weakness started to influence on my life. Now I'm 36 years old, I'm in the wheelchair and I need help for all things I do. I'm a mother of healthy girl and that's the best thing in my life. I'm doing the exercises and swimming every day in the pool. It's the most important thing. I hope that the medicine will be find at least to stop progress of the disease."

Raja, Family member/caregiver of individual with LGMD2C (Translated from French)

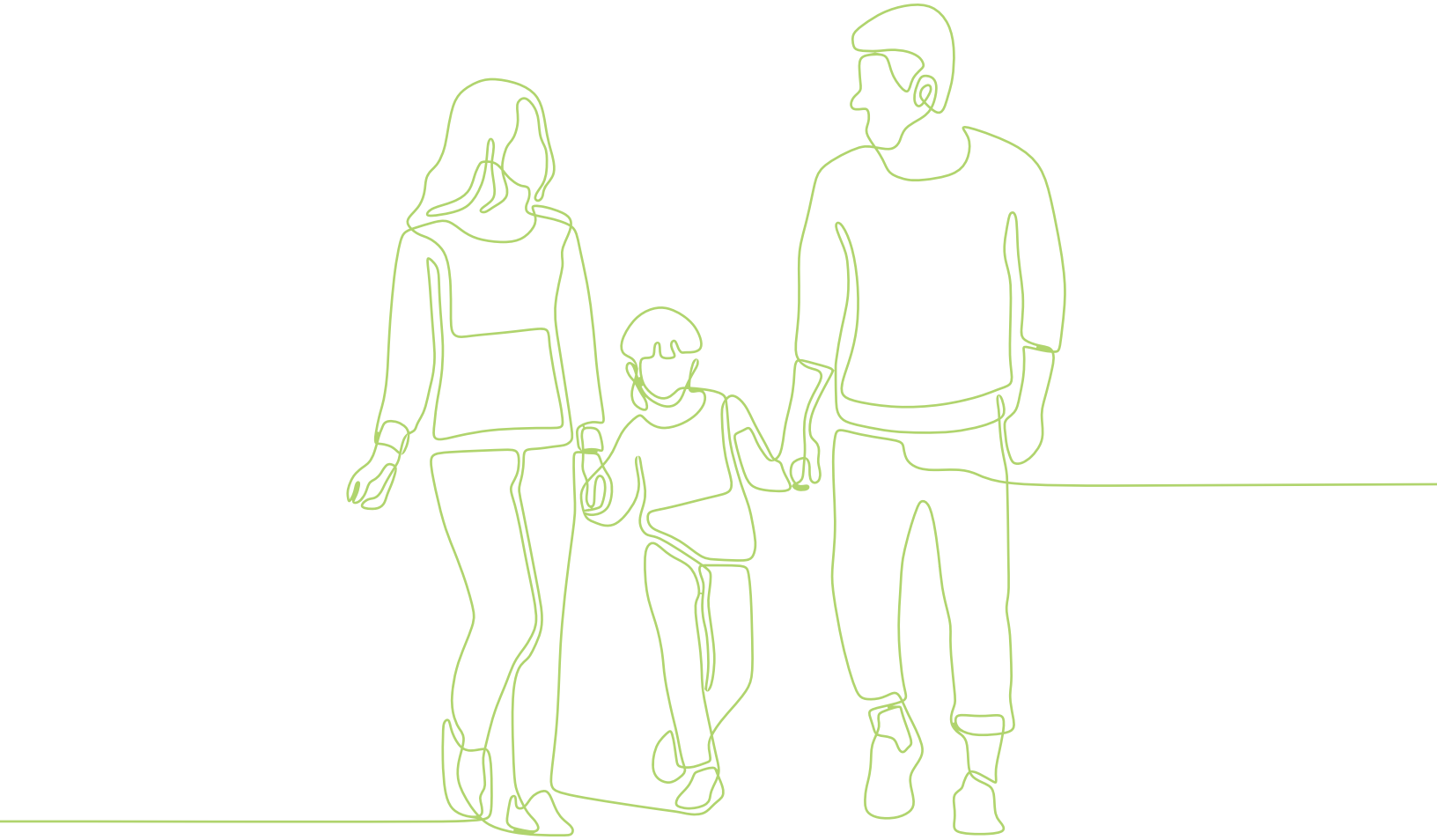
1. *"The three most common symptoms in my daughter are difficulty climbing stairs, lifting from the floor and running like children her age"*

2. *"The worst day in my life is seeing her unable to do her activities like all children when she was five and when we were not yet aware of the disease*

   But the best day in our life was when she found herself alone in school and she talked with her friends about her future and that she will lose the step and ask them not to laugh at her. We both me and her dad were happy because her illness is no longer a challenge for her"

3. *"Run or climb or jump like 8-year-olds my daughter is unable to do"*

4. *"Today the little one is used to the symptoms of muscular dystrophy and behaves according to the abilities she still retains"*



Charlotte, parent of a child living with LGMD2C

"I am a parent and worry about not being able to help my child as I get older."

Peter, man living with LGMD2C

"On my worst days I am unable to participate with my friends because of accessibility or fatigue."

Younssa, woman living with LGMD2C

"The symptoms that bother me the most beautiful daily life its difficulty to get up from a chair to move around and find my place in society you have the evolution of the disease. this research can help me feel more comfortable in my body and accept it"

Vanessa, woman living with LGMD2C

Comment 1:

"Symptoms that currently are impacting my life massively are all due to the ongoing muscle weakness especially now that it is taking away my air. That is my number 1. The inability to take a full deep breathe, the inability to hold a conversation without gasping for air, the need to be hooked up to non-invasive ventilation, the need to carry such device everywhere I go and the need to not only use at night but for a portion of my day. Number 2 would be how the weakness has taken away my ability to be self sufficient. I no longer can toilet myself, bathe myself, dress myself, prepare meals myself. I no longer have enough strength to put my hair up in my desired style since I can not elevate my arms. I slouch and prop my arms on counters to brush my teeth, to feed myself. Which leads me to my Number 3 symptom. The unrelenting fatigue. Everything is taxing. The little I do takes much effort even with help. Pushing through the day is a struggle when you feel so exhausted. It truly is a will of the mind to tell yourself to physically attend events, to hold down a job even part time, go out with friends, go shopping things that bring such enjoyment.

At my current state, I can no longer really distinguish between my best days to my worst. Maybe on a good day I suffer from a little less of the extreme fatigue where I can go longer in my day without using my non-invasive ventilation device and possibly leave the house to run an errand with my home care aide or loved one. My worst days would look like needing to be hooked up to my ventilation all day, being so tired I can't think to even have my assisted showers.

I really miss having the ability to bathe and toilet myself. It's so difficult to keep having strangers help with something so intimate. Needing the help to lotion your body and get dressed. It's awful to constantly depend on someone to place you on the toilet or help with a urinal. I cannot say how many UTI's I've had since you end up dehydrating yourself when your out to not use the facilities and hold it for long periods because no one is around. I truly hate now having to receive assistance in breathing. I love to blast music and sing along to a song but it all is defeating. I barely make it half way. Being attached to this vent and hoses gets in the way of transfers. I do not want it to be a permanent fixture in my life.

I don't believe the way I am coping with the disease has changed. Since I was diagnosed as a child based on symptoms I have felt I have always felt a grieving of the loss of ability. You have episodes of full days of tears but then you adjust and continue forward because there are no treatments and I and others with LGMD are given no other option but to grieve and cope."

Comment 2:

"The drawbacks I chose was limited availability or accessibility. For example, I've often heard shining reviews about the benefits of aquatic therapy but for me it's impossible. The pool used for therapy in my area doesn't have the right accessibility equipment to lower me into the pool. I can't transfer myself to the chair for pool lift. Also, some physical therapists don't wish to transfer me or can't. My last out appointment had my elderly mother transfer me onto a table for exercises. Just terrible."

LGMD2D / LGMD R3 α-sarcoglycan-related

50-year-old man living with LGMD2D (diagnosed at 38 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"difficulty walking, low muscle strength, low quality of life"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"life itself in all areas"

What worries you most about LGMD?

"become dependent on others"

Please describe your experience with treatments and other approaches to manage LGMD.

"I didn't have any experience [with treatment] other than keeping my spirits high and being active."

Parent/guardian caregiver of a 4-year-old son living with LGMD2D (diagnosed at 3 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Unable to match peers in sports"

"Declining ability"

"Constant worry about falling or other things"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"None as of now"

What worries you most about LGMD?

"Losing independence of my child"

Please describe your experience with treatments and other approaches to manage LGMD.

"Exercise"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Not lose ability to function independently in life"

Parent/guardian/caregiver of a 9-year-old son living with LGMD2D (diagnosed at 8 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Muscle weakness in his legs. He can't keep up with his friends and falls easily. He struggles to even play a simple game of tag.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Not being able to walk at a pace of a typical 9-year-old, Not being able to play sports, Inability to climb the stairs without difficulty or assistance*

What worries you most about LGMD?

> *I worry about him losing his ability to walk at all. Mostly I worry about his loss of independence.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *We see a physical therapist every week. She does a variety of stretches and some muscle strengthening moves with him. This has helped a lot. We also give him vitamin D supplements as he is quite deficient in it. We also mix creatine into smoothies. Otherwise, there is, unfortunately, no treatment available.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Anything to slow progression. These little kids need time to allow the wonderful scientists in the world to discover a cure. We have to slow down progression somehow to preserve the muscle they have!*

15-year-old female living with LGMD2D (diagnosed at 12 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Walk speed is slow. Bad stamina. Walking weird*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Cannot run at all*

What worries you most about LGMD?

> *My future and ending up in a wheelchair forever*

Please describe your experience with treatments and other approaches to manage LGMD.

> *The most I've done is physical therapy*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I'm not sure. Just something making me able to live a normal life."

Kelsey, caregiver for two children living with LGMD2D

"I have 2 children 9 and 16 with LGMD2D. I would like them to have hope for a better future."

Faran, parent of a nine-year-old boy living with LGDM2D

Comment 1:

"As a mother of a young child with LGMD2D, I am eager for some options to slow progression. There have been promising developments for other forms of LGMD. Some even approved by the FDA to be fast tracked. We would love to see more exploration of the cross functionality between subtypes for these treatments and more fast tracking of approvals. We are truly desperate to slow the progression of this disease to prolong our children's mobility as long as possible."

Comment 2:

"My son is 9 years old which is a very active age for a young boy. His LGMD2D prevents him from keeping up with his friends. He is unable to even play a game of tag without falling. His ability to stabilize his body with the smallest nudge or stumble leads to frequent falls. Some of which has lead to a strong impact to his head and limbs. We fear that he will break a bone and be unable to use that limb leading to more muscle loss that he will be unable to recover. Faran, parent of a child living with LGDM2D He is young now so preserving the muscles he has is critical to prolonging his mobility and independence. We are desperate for a treatment and ultimately a cure."

Comment 3:

"My son (9) starting showing symptoms of muscle weakness around 7 years old. We eventually pursued physical therapy. It was there that the PT noticed signs of myopathy. A blood test then showed ck levels around 10,000 which then directed us to a neurologist. They first thought it was Duchenne but then genetic testing confirmed 2D. He now has muscle weakness in his legs and arms, frequent muscle cramps, he struggles climbing stairs, getting into the car and playing with friends."

Prakasa, 32-year-old man living with LGMD2D

1. *"LGMD affects me everyday of my life and even more on my worst days. On my best days it would cause me an inconvenience that I could get on with the support of my family and friends. For example, I'd have to skip a restaurant or an event where everyone of my friends would be going just because the venue is upstairs and there's no elevator present."*

   On my worst days, it'd just throw me into a spiral of depression. I'd usually face these when I am all alone without anyone else's help. Most of the time my worst days would not just be a physical obstacle but also a lot of mental pressure that accompanies it. It causes me a lot of worry when I think about my future and the problems I'd encounter with the rapid progress of my disease.

2. *"There are a lot of activities I'd need to skip because of LGMD. Specifically speaking, I've always loved to be independent in my life, to do whatever I want to and to go wherever I want to, without anyone else's help. But LGMD has made it impossible for me to even accomplish the most basic activities of my life without someone else's help."*

3. *"I have had LGMD2D since I was 16 years old and now I'm 33. During the early 20s of my life it was relatively easy for me because there were so many friends and family members around me that helped me to forget that I even had this problem. But the late 20s and my early 30s have been becoming a lot of struggle for me because everyone around me are settling down in their lives and having their own life to worry about and it pretty much made me feel alone for the most part of my life. My physical strength is declining rapidly, so many physical problems like constant muscle cramps surrounded me and combined with that is the mental pressure. But I've been learning how to live with this problem by modifying my home according to my needs, traveling in cars with elevated seat height, using devices that would help me to get up from a seat etc.*

   Presently I am using 'Sit & Stand' device (Purchased from UK Amazon site by my friend living in London) which was gifted to me by my Close friends on this year march, which I am using that Sit & Stand device to get up from sofa seat, dining table chair, to get out or get in to car seat, to get up from movie theatre seats, etc etc."

Kelly, individual living with LGMD2D

Of all the symptoms of limb-girdle muscular dystrophy, which 1-3 symptoms have the most significant impact on you or your loved one's life?

"One would expect that losing the ability to walk, transfer, or lift my arms would have the most significant impact on my life as an individual living with LGMD2D. Yes, these symptoms have an impact on my life, but I have been able to adapt seamless to those changes. The symptom I have struggled the most with is the diaphragm weakness and it's role in lung function. I use a bi-pap at night and I have a non-invasive ventilator (sip and puff) attached to my wheelchair during the day time. I also use a cough assist and function device to assist with coughing. The loss of lung function has been devastating. You never realize how much you appreciate the ability to breathe until it is a struggle. I am fortunate enough to be able to use a non-invasive sip and puff ventilator instead of a tracheostomy, however, I still struggle emotionally and physically. I am a lawyer and I find myself in court giving lengthy oral arguments and it is difficult to talk for long periods of time due to my breathing difficulties. It is also embarrassing to use the sip and puff straw. I deeply miss the ability to breathe independently."

Rachel, parent of a six-year-old son living with LGMD2D

My son Jacob is 6 years old living with LGMD, type 2D. Last week in school he wrote he wanted to 'run faster' as something to work on this week. When I asked him about his response, he said he is always behind his peers when running in gym.

Michelle, living with LGMD2D

I live with LGMD2D and started using a power wheelchair since 4th grade. I currently need help with everything from getting out of bed, toileting, showering, meal prep, eating. I have personal care assistants here helping me with everything on the daily. I use a power wheelchair, ventilator at night, and other assistive devices like shower chair (I'm in the process of getting an electric lift to help with transfers).

This disease is life-altering, but you roll with the punches... you have to. The progressive nature of this disease is what makes this disease so hard physically and emotionally. What will be the next ability that I lose?

Losing these physical mobile abilities is hard, but during the past few years I have now realized the next ability that I'm losing is the ability to breathe. I've cried so many tears about this and have come to terms with it, but little me as a 4th grader would have never known the respiratory difficulties ahead. It's very, very hard.

Please know any progress towards finding a cure or treatment would be LIFE changing.

Anwar, 27-year-old living with LGMD2D

Comment 1:

I am LGMD2D 27 years old (still walking) am very upset about the very slow process and research and very little effort for this particular type of LGMD2D.

The experience of Dr. Mendell since 2015 we have not heard anything about it for 7 years. It is a very long time. The disease robs us of our muscles day after day. If we are in wheelchairs, what should we do with the treatment?

And another question, why don't researchers focus on ways to regenerate muscle that has been replaced by fat? It cuts off hope and puts a red streak when the muscle is scarred

Comment 2:

> *The worst symptoms are falling and not being able to get up, causing me great embarrassment and I feel like crying and the other symptoms are the inability to walk normally like others*
>
> *And the third is pain in the lower back, extreme tiredness and fatigue*
>
> *It affects me a lot. I feel sad every day every moment. I can't hang out with friends nor enjoy the life you enjoy. My worst days are the days when friends go on vacations and picnics and mountain climbing and I can't go. And the best days never exist.*
>
> *Yes, there are a lot of activities that I can no longer do because of LGMD2D. First of all, running and walking independently, climbing stairs, riding a horse, skating, getting up, sitting, visiting friends and patients, watching matches, shopping, hiking, and every activity that requires healthy muscles*

Ksenia, living with LGMD2D

1. *Malfunction of the movement of the arms and legs*
2. *good: go out for a walk, go to the pool*
   Bad: due to bad weather I don't go out for a walk, my parents are busy
3. *change clothes, wash, wash*
4. *all symptoms worsened and progressed over time*
5. *gets worse over time*
6. *have a detrimental effect on all spectrums of emotions*
7. *deterioration in health*
8. *progression of my diagnosis*

1. *I go to the pool, regularly go to the hospital for examination, go to sanatoriums*
2. *have not tried*
3. *do not heal*
4. *permanent moves*
5. *health maintenance*

Donavon, living with LGMD2D

> *Since 1999, when I started gene therapy in muscular dystrophy with Dr. Jerry Mendell, the outcome measurements haven't changed. All muscular dystrophy gene therapy outcomes are based on the North Star Assessment. The North Star Assessment includes the six-minute walk, getting up from the floor, and climbing a few stairs. These outcome measurements were used in my sister June's trial, with one patient improving over time who scores a 52 on the North Star. These outcome measurements might be good for some type of trials but not for all types of trials.*

If there were different outcome measurements approved, this would increase the number of patients available to be used in research. Many patient groups are limited for trials such as limb girdle muscular dystrophy since many of the patients are in wheelchairs and would rarely meet the three criteria of the North Star Assessment.

I've searched several gene therapy trials on ClinicalTrials.gov, and none of them had respiratory improvement as an outcome measurement, while some had respiratory impairment as an exclusion criterion. However, a company in France, Atamyo, has started to dose patients in a gene therapy trial for LGMD2i. One of the main inclusion criteria is to have moderate respiratory impairment (forced vital capacity below 80%).

Other than safety trials, all the trials for LGMD used patients that are below the age of 18. When I was 18, my respiratory system was not affected. I didn't start having issues until I was about 50 years old. So testing children below the age of 18 in LGMD trials most likely won't show any respiratory impairment. However, if an LGMD trial for gene therapy included patients whose lung capacity is affected, respiratory function improvement could possibly be shown.

I've been told that 200 muscles are involved in walking, and 100 of them are critical for walking. In contrast, breathing involves 18 muscles, and the major one is the diaphragm. It seems that putting emphasis on improving a function that involves fewer muscles could be worthwhile. For LGMD types such as 2D where respiratory failure is a main cause of death, maybe research should work towards strengthening the respiratory muscles in adults.

By the way, I've read about technology available today to assess diaphragm strength as presented in the 2016 article titled, 'Validation of ultrasonography for non-invasive assessment of diaphragm function in muscular dystrophy' by Drs. Nicholas Whitehead, Stanley Froehner and others in the Journal of Physiology.

If it can be shown that gene therapy can result in a stronger diaphragm and other muscles around the lungs can be strengthened, this would have a major impact on my quality of life. Not only would I be able to breathe easier, but it would allow me to then communicate better with a stronger voice, and I'd be able to talk longer without getting so tired. My weakened lung and diagram muscles severely reduce my lung capacity which means I cannot lay flat in a bed anymore or fly on an airplane without my ventilator. If I drink too much water or eat a little too much, it puts pressure on my diaphragm and makes it harder for me to breathe, so then I need to use my ventilator for a while to give my lungs a rest. With stronger breathing muscles, potentially I could decrease or even discontinue using a ventilator during the night and/or throughout the day which would also decrease the insurance burden of covering the cost of the ventilator, cough assist machine and associated masks/filters.

Because of changing technology, I believe the FDA should be working to help LGMD patients beyond just walking. I do believe that being able to walk is extremely important and should be measured; but being able to help somebody breathe better should be a target. To me, breathing is actually more important than walking for older patients. Research in that direction may be easier to do, in my opinion, as you're not trying to strengthen so many different muscles. Thank you, Donavon"

Caroline, caregiver to patient living with LGMD2D

> *"In Argentina there are a lot of patients with LGMD. We need to include in the treatment. Regards. Ignacio's Mother"*

LGMD2E / LGMD R4 β-sarcoglycan-related

Parent/guardian/caregiver of a 14-year-old son living with LGMD2E (diagnosed at 10 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Muscle weakness throughout the body, restriction in movement, low weight"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Movement, movement, breathing are very important. LGMD takes it away every day."

What worries you most about LGMD?

"Rapid progression, and there is nothing we can do to help our son. Other than warning for treatment."

Please describe your experience with treatments and other approaches to manage LGMD.

"We do stretches every day, exercise, proper nutrition, rest. The drugs used do not help in any way."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"We are waiting for the treatment of srp-9003. This is our main hope. To help this treatment, nutrition, physical exercise are mandatory"

Parent/guardian/caregiver of a 14-year-old son living with LGMD2E (diagnosed at 10 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Weakness of the muscles of the whole body, slowness of movement"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"It is very important to move independently, breathe. The disease every day takes the muscles of the whole body and leads to weakness of the whole body. I really want to walk, wash, dress, go to school, shop, hospital, communicate with friends, etc. on my own."

What worries you most about LGMD?

"rapid progression of the disease. Inability to move"

Please describe your experience with treatments and other approaches to manage LGMD.

> *We are doing our best to fight the disease. Swimming pool, exercise, stretching, velomoto simulator, breathing exercises, splints at night. We take care of food and rest. But no matter what we do, the disease progresses and very quickly.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *We are looking forward to treatment (SRP-9003). Every day, every minute we wait! The sooner we get treatment, the better. With treatment, restoration is necessary – standing support, good wheelchairs, seat supports, physical therapy, warm pool, splints, food. We kindly ask you to register as soon as possible (SRP-9003)!*

23-year-old woman living with LGMD2E (diagnosed at 6 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *sleep problems, hands and feet don't work, muscle pain*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *to go*

What worries you most about LGMD?

> *progressing*

Please describe your experience with treatments and other approaches to manage LGMD.

> *no*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Rehabilitation*

Parent/guardian/caregiver of a 11-year-old daughter living with LGMD2E (diagnosed at 8 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

Three main symptoms

> *Difficulty to squat*
>
> *Difficulty to get off the floor*
>
> *Progressively difficult to manage stairs*

> *A significant impact on every aspect of life both physically and emotionally, huge impact on her childhood is 'catching up' with her friends. We live in Kent United Kingdom, we had to gather lot of information ourselves to support us in making reasonable adjustments to continue at same school. We are successful in making our daughter complete her primary school in a normal school.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Unable to play with other children.*

What worries you most about LGMD?

> *Lack of treatment*
>
> *Lack of disease modifying drugs*
>
> *facing up to everyday pessimism especially professionals who look up on internet to explain LGMD lack of awareness about LGMD – my daughter was initially thought slow*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Vitamin D*
>
> *Multivitamins*
>
> *Good diet*
>
> *Stretching*
>
> *Activity based exercise like swimming*
>
> *Positive focus on abilities*

Parent/guardian/caregiver of a 11-year-old daughter living with LGMD2E (diagnosed at 8 years)

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Stop progress or at least slowdown progress. Delay in access to treatment is equivalent to children losing their childhood.*

57-year-old man living with LGMD2E (diagnosed at 19 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *resistance to exertion, too hot or cold*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *No*

What worries you most about LGMD?

"Heart"

Please describe your experience with treatments and other approaches to manage LGMD.

"Physiotherapy"

31-year-old man living with LGMD2E (diagnosed at 8 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"be dependent on others, be able to walk, muscle weakness"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"be able to walk, be able to wander freely"

What worries you most about LGMD?

"the drug is not released"

Please describe your experience with treatments and other approaches to manage LGMD.

"physiotherapy, nutrition, psychology"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"living my life freely"

Individual living with LGMD2E

Symptoms, how symptoms affect life

"Sometimes they limit me, but I try not to think about them and move on"

First 1-3 symptoms that have the + significant impact on your life

"– autonomy – lack of strength – breathing"

Describe a good day and a bad day

"A good day is when I feel good and have the energy to get things done, fortunately, I get that quite often. A bad day is when I lack the strength and will to tackle my"

Activities you can't do or do partially

> *I used to like swimming, but there are too many problems from moving to dressing and undressing, to the cold water not being suitable for my muscles.*

How have your symptoms changed over time

> *I have gotten worse in both my muscle strength and my lungs. Fortunately, morale is always high, at least I try to*

How your ability to cope with symptoms changes over time

> *Let's say you adapt to the changes, at least you try. It is by no means easy, but never give up on physical psychological, and emotional impacts*

> *The physical gets worse from year to year. Logically, there is also a psychological impact on the progression of the disease both for me and for those around me, but each person has his or her problems, so one has to be a little optimistic*

What do you fear most as you get older

> *Of getting worse*

What worries you the most and what frustrates you the most

> *When my parents are no longer able to care for me and I have to deal with caregiver topics. Maybe, in the meantime, I'll find myself a nice hottie.*

How do you manage your symptoms?

> *I don't manage them, I live with them. Day to day, I live by the day.*

Medical treatments, physical therapy, occupational therapy, equine therapy,

> *I use a night ventilator and a cough machine often, especially when I have phlegm. I gave up physical therapy for covid but still didn't get many benefits from it*

Non-traditional treatments: acupuncture...

> *I treat myself with color therapy.*

How well do these treatments treat the most significant symptoms

> *It makes me feel good, it doesn't hurt*

Negative aspects of your current treatments

> *No negative aspects*

In the absence of comprehensive care, what features do you look for in a treatment

> *I would like treatment that would not make the disease worse, especially breathing*

Individual living with LGMD2E

First 1-3 symptoms that have the most significant impact on your life

"respiratory insufficiency, intestinal difficulties, cold"

Describe a good and a bad day

"a good day when I breathe well and have no intestinal problems and a bad one when it's not like that"

Activities you cannot do or do only partially

"I do almost nothing myself"

How your symptoms have changed over time

"slow and continuous worsening"

How your ability to cope with symptoms changes over time

"accepting and being vigilant"

What you fear most as you get older

"losing your lucidity as well"

What worries you most and what frustrates you most

"I worry about society and I am frustrated by the lack of empathy"

Non-traditional treatments:

"insufficient physiotherapy, but as soon as I can"

How well these treatments treat your most significant symptoms

"they relieve joint problems and pain"

Negative aspects of your current treatments:

"cost"

In the absence of a complete cure, what characteristics do you look for in a treatment

"Constancy"

Sibling A, living with LGMD2E

Symptoms, how symptoms impact your life

"Very little mobility in limbs and trunk so I need continuous assistance"

First 1-3 symptoms that have the + significant impact on your life

"no strength in limbs, heart problems, severe scoliosis"

Describe a good and a bad day:

"a good day when I can do what I like without the help of others. A bad day when my limbs are cold and hurt as a result I have no strength"

Activities you can't do or do partially

"no I can walk, personal care management, eating, etc. But good control of wheelchair driving"

how have your symptoms changed over time:

"less and less muscle strength, worsening of heart problems, and back pain"

how your ability to cope with symptoms changes over time:

"I have less and less patience for I am losing my autonomy"

physical psychological and emotional impacts

"It is increasingly difficult because I need more and more care from my family members, which brings me anger and discouragement"

what you fear most as you get older:

"is that you will no longer have the muscle strength and therefore not be independent in the simplest activities"

what worries you most and what frustrates you most:

"not being able to do the things I enjoy anymore."

How do you manage your symptoms:

"I have checkups and doctor visits every other year. I take medication for heart failure. I do physical therapy twice a week and respiratory gymnastics four times a week. I try to feed myself properly. Physiotherapy relieves my pain and I hope it avoids joint tension long enough. – Sibling A, living with LGMD2E"

how well do these treatments treat the most significant symptoms:

"relieve worsening heart problems, maintain good limb mobility"

negative aspects of your current treatments:

"unfortunately, there is no improvement"

in the absence of a complete cure, what features do you look for in a treatment:

"In the absence of a complete cure I would at least like it to fortify me enough to be able to sit comfortably in a wheelchair and be able to continue to use my hands to drive it and to use device and technology. And not make me especially worse from a cardiac and respiratory point of view"

Sibling B, living with LGMD2E

Symptoms, how symptoms impact your life:

> *Very little mobility in limbs and trunk so I need continuous assistance*

First 1-3 symptoms that have the + significant impact on your life:

> *no strength in limbs, heart, and breathing problems*

Describe a good and a bad day:

> *a good day when I can do what I like without the help of others. A bad day when my limbs are cold and hurt as a result I have no strength.*

Activities you can't do or do partially:

> *no I can: walking, personal care management, eating. But I have good control of wheelchair driving*

How have your symptoms changed over time:

> *less and less muscle strength, worsening of heart, and breathing problems*

how your ability to cope with symptoms changes over time:

> *I have less and less patience because I am losing my autonomy*

physical psychological and emotional impacts:

> *It is increasingly difficult because I need more and more care from my family members, which brings me anger and discouragement*

what you fear most as you get older:

> *is that you will no longer have the muscle strength and therefore not be independent in the simplest activities*

what worries you most and what frustrates you most:

> *not being able to do the things I enjoy anymore.*

How do you manage your symptoms:

> *I have checkups and doctor visits every other year. I take medication for heart failure. I do physical therapy twice a week and respiratory gymnastics four times a week. I try to feed myself properly. Physiotherapy relieves my pain and I hope it avoids joint tension long enough.*

How well do these treatments treat the most significant symptoms:

> *relieve worsening heart problems, maintain good limb mobility*

Negative aspects of your current treatments:

> *unfortunately, there is no improvement*

In the absence of a complete cure, what features do you look for in a treatment:

"In the absence of a complete cure I would at least like it to fortify me enough to be able to sit comfortably in a wheelchair and be able to continue to use my hands to drive it and to use device and technology. And not make me especially worse from a cardiac and respiratory point of view"

Nicole, mother of children with LGMD2E

"My children are dying from limb girdle muscular dystrophy 2E. Exercising accelerates the muscle breakdown. Why won't the FDA allow the right to try act or any other way for us to dose with sarepta's gene therapy that successfully worked in the first 2 trials? There have been no catastrophic side effects. In two years they will have no muscle left to preserve."

"Please for the Love of God allow us access to this medicine"

Carles, individual living with LGMD2E (diagnosed at age 11)

"I have a LGMD2E and I was diagnosed at age of 11. I have most of the symptoms that have been related by my colleagues. Now, I am using the wheelchair almost for everything but not in home or in my job (small spaces that I know very well). The point is that as I am borderline walking and not walking I note that to me it is difficult to start moving. When I stand up. I need few minutes to start moving. I give the order to my legs but they do not answer. that is increasing time to time. For the rest I need assistance for personal issues as have been mentioned before."

"In a bad day I have difficulties to go from kitchen to bed (20 m). In a good day I can walk for 1 hour (slowly but 1 hour). Headache is normal during bad days, so I guess there is some circulatory issue not working very well."

Erik, 45-year-old living with LGMD2E

"I have LGMD2E, 45 years old. I have done yoga breathing and meditation for the last 25 years. Daily commitment to the breathing has really helped to keep my muscles going. Meditation to manage anxiety."

Serghey, living with LGMD2E

Answers to the questionnaire

1.1. *"weakness, difficulty in moving, difficulty in lifting things."*

1.2. *"a good day is when everything works out, even slowly and when people are understanding. When events bring joy. A bad day is when you can't do what you have planned, due to the illness."*

1.3. *"The activity that is impossible or difficult to do is everything that is connected with movement, and this is almost everything. It is impossible to go to a public toilet, most shops and institutions, transport, medical facilities, wherever there are steps. There are big restrictions everywhere."*

1.4. *"over time, the symptoms become more pronounced and stronger every day. The disease progresses very quickly, we do not have time to adapt to the condition, as the condition worsens again."*

1.5. *"Dealing with symptoms becomes difficult over time. No matter how hard we try."*

1.6. *"It's very emotionally difficult. There are many physical limitations."*

1.7. *"As we get older, we are afraid that our strength becomes less and less. But I really want to study, just live, fall in love, make friends, work."*

1.8. *"Most of all, it worries and frustrates me that moving is more difficult every day. Skills go away and we try our best, but the disease progresses and does not recede."*

2.1. *"We fulfill all the recommendations of both doctors and older patients. Swimming pool, stretching, bike exercises, bodyweight exercises, breathing exercises, to the maximum of our movement. Exercises that you can do around the house, wash the dishes, peel the potatoes."*

2.2. *"we do not use unconventional methods, the most important thing is not to harm ourselves"*

2.5. *"I really want the disease to stop, to begin the recovery as much as possible."*

Beatrice, mother of a son living with LGMD2E

Comment 1: Symptoms: how symptoms affect your life

"My son does not move his legs and arms, he can only move his hands and fingers with difficulty. This limits him in all activities that require movement. He has to be fed, washed, dressed, moved in the car, he is only independent in using the wheelchair with a joystick, mobile phone and PC."

First 1-3 symptoms that have the most significant impact on your life

1. *"lack of strength and motor difficulties, which limit him in daily activities"*
2. *"muscle retractions, which limit him for posture, sitting, and lying down."*
3. *"heart problem, which obliges him to take a lot of medication, wear a subcutaneous defibrillator, and frequent check-ups and examinations."*

Describing a good and a bad day

- *The days are all similar, with study activities and technical drawing of architecture on the PC, free time with various hobbies, following formula 1, outings, and parties.*
- *Bad days are those with colds, and fever, which happen 3-5 times a year.*

activities you cannot do or do only partially

- *Cannot walk, stand, eat alone, wash and dress himself, or drive a car.*
- *He can partially do the shopping.*
- *He is independent in studying, speaking in a group, telephoning, and video-chatting.*

how your symptoms have changed over time

- *Until the age of 6, he was normal, then he had difficulty climbing stairs, at 10 he stopped walking and at 14 he started having difficulty with his hands. At 16 his heart got worse and he was implanted with a defibrillator. Although my son doesn't perceive the heart problem, e.g. he has no difficulty breathing, and no headaches.*

How does your ability to cope with symptoms changes over time

- *My son is now more responsible for his illness, he trusts his caregivers, he has confidence and he takes very good care of himself.*
- *Physical, psychological and emotional impacts*
- *In addition to the motor difficulties, my son has become very thin and has a lot of retractions, which are very obvious to anyone looking at him.*
- *On a psychological and emotional level, he is always confident and positive.*

what you fear most as you get older

- *As parents, we fear his heart problems, but my son doesn't seem very worried.*

what worries you the most and what frustrates you the most

- *As parents, we fear losing our son, it frustrates me most to think that we did something wrong and that we could have done better.*

Comment 2: How do you manage your symptoms

- *Medical treatment takes medication for heart, bones, stomach, blood circulation, vitamin and food*
- *supplements.*
- *Physical therapy: he does motor physiotherapy twice a week at home*
- *He does respiratory physiotherapy twice a day with a cough machine.*
- *occupational therapy, I organize myself*
- *equine therapy, I tried oniotherapy 2-3 times*
- *non-traditional treatments, acupuncture NO*

how well do these treatments treat the most significant symptoms

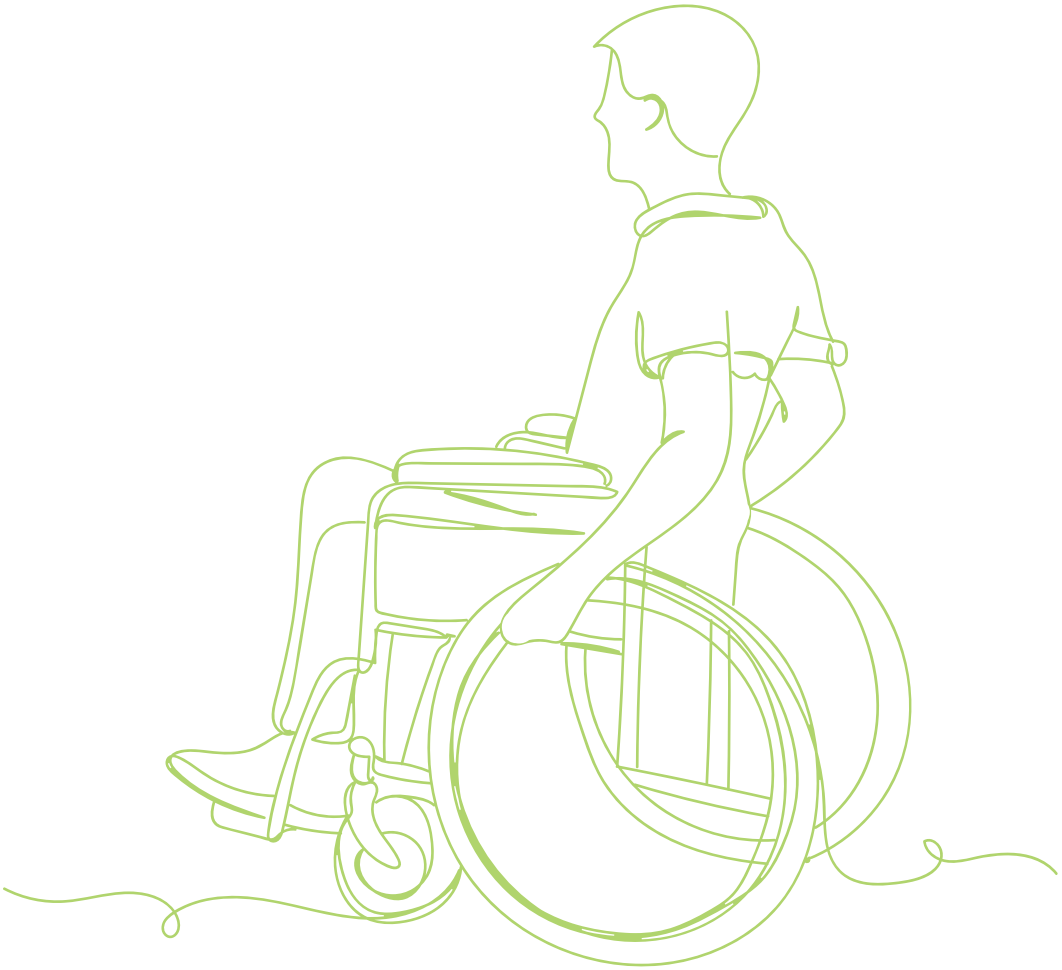
- *"These treatments only help to slow down worsening, which happens anyway"*

negative aspects of your current treatments in the absence of a complete cure.

- *"Taking steroids has caused eating difficulties and bone demineralization."*
- *"Taking many medications forces my son to frequent check-ups."*

what features you look for in a treatment

- *"Try to slow down the progression of the disease"*
- *"To stabilize the pathology"*
- *"Some improvement"*
- *"Complete recovery"*



LGMD2F / LGMD R6 Δ-sarcoglycan-related

64-year-old woman living with LGMD2F (diagnosed at 55 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

1. *"Managing stairs"*
2. *"Getting up from the floor or chair"*
3. *"lifting up hands and opening bottles"*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Walking, holding my grand children"

What worries you most about LGMD?

"To be wheelchair-bound"

Please describe your experience with treatments and other approaches to manage LGMD.

"Physiotherapy and steroids"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Medical Equipment"

LGMD2A / LGMD R1 calpain 3-related

58-year-old woman living with LGMD2A (diagnosed at 31 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Loss of mobility (lower extremities), upper-body weakness, fatigue"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Toileting independently in public restrooms, traveling/taking family trips with ease, driving"

What worries you most about LGMD?

"That I will progress to such a point that I will be unable to manage the tasks of daily living independently (beyond my already limited abilities)."

Please describe your experience with treatments and other approaches to manage LGMD.

"Power wheelchair, accessible van, remodeling home (no steps, wheel-in shower), weekly physical therapy (to try to maintain flexibility, strength, alignment)."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Being able to go from a sit-to-stand position to allow for safe and easier transfers to/from bed/chair/toilet, particularly toileting independently in public restrooms."

61-year-old woman living with LGMD2A (diagnosed at 9 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Loss of upper extremity strength, severe lordosis, pulmonary involvement"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Independent self-care (bathing, dressing, toileting), driving, food prep/eating, and hugging my family members and friends"

What worries you most about LGMD?

"Aging with LGMD and being placed in a facility for long term care if/when my husband can no longer care for my physical needs."

Please describe your experience with treatments and other approaches to manage LGMD.

> *Keeping active has been the most beneficial. Aquatics therapy has been very helpful for rehab after injury but is not readily available on a long term basis. Use of a ventilator (AVAPs) at night improves my sleep and fatigue levels. Weight loss and lifestyle changes to maintain the weight loss has improved my overall health and mobility.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *If only there was a way stop the unrelenting progression of this disease! I can deal with not walking but loss of upper extremity strength is the worst. I would great if I could improve my upper extremity strength...even just a little.*

36-year-old woman living with LGMD2A (diagnosed at 21 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *I can't get up from my chair. Can't walk on uneven surfaces. I can't bend over.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *I can't go outside without help. I can't get into the bath to wash myself. In winter it is difficult to walk in clothes. Do not hold objects weighing more than 1 kg.*

What worries you most about LGMD?

> *Weakness. Inability to take full care of oneself*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Charging and light exercise. Reception of a complex of vitamins gives cheerfulness, energy and endurance. But they do not stop the progression of the disease.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *A healthy lifestyle, proper nutrition, more proteins and fats, few carbohydrates, no sugar. Regular correct exercise. Little stress. Help and support from loved ones. Everything is important in our life. But this will not restore the ability to self-care and will not stop the disease.*

51-year-old woman living with LGMD2A (diagnosed at 8 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Arm/shoulder function, neuropathic pain, controlling weight, affect of cold*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Independent toilet transfers, cooking, socialising"

What worries you most about LGMD?

"The future – pain – when I am ill for any reason I have a taste of how little I will be able to do."

Please describe your experience with treatments and other approaches to manage LGMD.

"Diet and exercise – a strict keto diet helped me to keep excess fat off, and regular physiotherapy and hydrotherapy have helped me to keep flexible and feeling good. However I stopped the keto diet as I worried it was unhealthy and also prefer a vegetarian diet for ethical reasons – I have since put on a lot of excess fat which I find uncomfortable."

"At age 12 I had Achilles tendon release which made me much less prone to falls – 40 years later I have developed a similar contracture in my left ankle which affects pain and posture and would love to repeat it but have been told it is likely to be unsuccessful."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"weight control, treatment for contractures – maintenance of flexibility, support for arm function, pain control/treatment for neuropathy"

36-year-old man living with LGMD2A

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"I can't walk normally. I can't climb stairs. My legs are weak and sometimes I fall"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"I can't climb stairs."

What worries you most about LGMD?

"My biggest worry is that one day I will end up in a wheelchair."

Please describe your experience with treatments and other approaches to manage LGMD.

"I live in Africa and was diagnosed in Turkey and have never taken medication for this disease."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I hope one day to be cured of this disease."

21-year-old woman living with LGMD2A (diagnosed at 13 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Weakening of the hip, thigh, arm, shoulder muscles and not being able to run, and having difficulty jumping up and down the stairs and on the floor, and scapular dislocation, weakening of the lower back, which happens when it is in a bent position, it is not able to straighten the back.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Going out alone and not having a suitable job, limited transportation and giving up most of your interests.*

What worries you most about LGMD?

> *Not being able to do personal work, and severe depression, more limited, not being understood, and being isolated from society.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Proper nutrition and proper weight, being active, exercising, making people happy, thinking and having a good and useful perspective.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *More efficiency to do personal work, to be lively and fresh, and to prepare the body for definitive treatment and to get results sooner, to strengthen muscles and not to get help from others.*

26-year-old woman living with LGMD2A (diagnosed at 19 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Arm/leg pain & cramping, muscle wasting resulting in inability to walk, fatigue*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Traveling, cooking, gardening, cleaning my home, washing my hair, going up stairs, swimming, yoga, going for walks in my neighborhood, hugging my loved ones.*

What worries you most about LGMD?

> *The medical expenses of having to care for myself i.e. hire a caretaker, physical therapy, assistive devices.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *I get out of my house everyday and try to remain active. I have found it to be very beneficial; less fatigue and improved strength. I try to move for at least 6 hours. This was versus a sedimentary lifestyle that caused dramatic muscle fatigue and cramping.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Free, expansive, and all-encompassing healthcare. An accessible city, or greater world, that I can be free to move in.*

> *I also hope to be able to remain at this current muscle functioning capacity*

39-year-old woman living with LGMD2A (diagnosed at 12 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Lost the ability of walking, lost the ability to self transfer, Lost independence to eat and drink on my own.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *I used to do roller skating at a professional level participating in competitions. I would love to be able to move my arms to play the cello. I would love to walk by the seaside and feel the warm sand under my feet. I would love to find a job that I feel passionate about without thinking if I'm suitable because of my physical restrictions.*

What worries you most about LGMD?

> *My lungs and heart may fail at some point in the future, cutting short my life before I see my kids being born, grow up and their kids.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *It has been very helpful receiving physical therapy and stretching. Motomed physical therapy machine has been indispensable in contributing to feel well and healthy. A tier 3 and 4 weight loss management service is primordial for people with reduced mobility but not always eligible depending on where we live.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *More changing places available throughout the country and events. Hoists installed in hospitals for women to access smear tests as any other women. Support in finding places to live/buy that are at a ground level and accessible for people with reduced mobility/wheelchair users, the same way there are only houses available to buy for 60+. Access to physical therapy including machine to help us stand up.*

46-year-old woman living with LGMD2A (diagnosed at 38 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Pain, weakness, tiredness.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Movement in sufficient quantity, especially on an inclined plane."

What worries you most about LGMD?

"Aggravation of symptoms, inability to walk independently."

Please describe your experience with treatments and other approaches to manage LGMD.

"I feel the real effect only from pain pills."

Parent/guardian/caregiver of a 19-year-old woman living with LGMD2A (diagnosed at 13 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Climbing stairs, raising from a chair or the floor, very tight Achilles tendons"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Being really independent, always thinking if the place I am going to has stairs or rocks etc, If I fall down I cannot stand up without any help, I cannot use the bus because the stairs are too high. So in many cases I prefer to go nowhere alone."

What worries you most about LGMD?

"Not being independent. Not having all the wonderful experiences that young people have. Traveling, dancing, doing sports, meeting new people, walking long distance."

Please describe your experience with treatments and other approaches to manage LGMD.

"Staying in a good weight, doing physiotherapy twice a week going to the university having good friends and two helpful parents."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Getting stronger, being safe."

Parent/guardian/caregiver of a 16-year-old daughter living with LGMD2A (diagnosed at 13 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Walking on stairs, imbalance, not enough stamina."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Taking public transport, spending time with friends."

What worries you most about LGMD?

"No cure, doctors doesn't have knowledge."

Please describe your experience with treatments and other approaches to manage LGMD.

"Currently not doing anything."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Able to lift my legs."

19-year-old woman living with LGMD2A (diagnosed at 6 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Getting up from a chair and when I fall and go up the stairs, I want help with that."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"I used to go swimming but I didn't come back because I got sick."

What worries you most about LGMD?

"Everything worries me and I find it difficult."

70-year-old woman living with LGMD2A (diagnosed at 20 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Wheelchair 100% can't walk anymore. Loss of strength in my arms and getting pressure sores because skin is so thin on my butt it hurts everyday. I am always cold and very stiff my legs and arms just don't work for me anymore."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"I can no longer drive. It is really hard to get into a pool because no ceiling lifts and my portable one hurts me so much and my bones break so easily."

"I get tired so much and can't even using my CPAP."

What worries you most about LGMD?

"I want a cure and I can't do anything alone I always need someone to help me."

Please describe your experience with treatments and other approaches to manage LGMD.

"This is what I am trying to find help with. My MDA Dr's never have anything new to help me. I have tried different meds but I hate feeling dizzy and almost feels drunk when taking some that are not working."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I want to be able to sit in my electric wheelchair and get on an airplane and I can't transfer into their other seats because my bones would break doing that. I want a swimming pool to float in and get off my butt needs ceiling lift to transfer and need more people to help me!"

50-year-old woman living with LGMD2A (diagnosed at 26 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Muscle weakness, balance and, recently I have added pain as a symptom. I'm this symptom is exacerbated during cold seasons."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Work outside of the home, pick up my grandchildren, and transfer independently."

What worries you most about LGMD?

"Passing this along to my children."

Please describe your experience with treatments and other approaches to manage LGMD.

"Aqua therapy."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I don't know. I do appreciate these opportunities."

24-year-old woman living with LGMD2A (diagnosed at 13 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Getting up from a chair, going up the stairs, and lifting my arms."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Driving, getting up from a chair, walking long distances."

What worries you most about LGMD?

"That I will be immobile soon, or that it will affect my breathing."

Please describe your experience with treatments and other approaches to manage LGMD.

"Swimming helped in keeping me active and mobile."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Slow down the deterioration of the muscles."

38-year-old woman living with LGMD2A (diagnosed at 19 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

1. *"Extreme fatigue."*
2. *"Inability to perform simple daily activities and needing help in everything."*
3. *"Chronic acute pain throughout the body."*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"All the simple day-to-day activities like going to the bathroom without needing help or a lever. To take a shower without making a million transitions from chair to chair."

"Being more active for my little girls."

"Basically get out of jail. which is actually my body."

What worries you most about LGMD?

"When will they find a cure for this disease and will I get to be healthy soon for my little girls."

Please describe your experience with treatments and other approaches to manage LGMD.

"Proper nutrition and physical activity including hydrotherapy and physiotherapy. But the health insurance funds only approve 12 treatments per year and this cannot really maintain a useful sequence of treatments."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I pray every day that they find a cure for this disease."

"I hope that in this conversation on 9/23 we will hear good news that the research is progressing and that there is a cure with God's help."

63-year-old man living with LGMD2A (diagnosed at 40 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Weakness in legs"

"Severe back pain because of that can't bent much more."

"Little bit of weakness in both the hands."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Sitting on floor"

What worries you most about LGMD?

"As of now I am ambulatory patient my most worry is how long I will be ambulatory."

Please describe your experience with treatments and other approaches to manage LGMD.

"I am taking homeopathy medicine which I think slower down my progression. As such there is no medicine in my country which really treat symptoms expect steroid."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"I can live my normal life. Can do what all I wanted to do."

63-year-old man living with LGMD2A (diagnosed at 16 years)

"Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?"

"Inability to walk, very limited arm strength, inability to transfer."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Inability to walk or transfer self in and out of wheelchair."

What worries you most about LGMD?

"Becoming completely dependent on others for physical needs."

Please describe your experience with treatments and other approaches to manage LGMD.

N/A

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Any improvement in leg and arm strength."

48-year-old woman living with LGMD2A (diagnosed at 14 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

1. *"Difficulty to stand up from a seated position"*
2. *"Impossibility to climb stairs"*
3. *"Difficulty to walk without help"*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Yes. Run, dance, travel alone, everything that requires movement."

What worries you most about LGMD?

"Losing my quality of life"

Please describe your experience with treatments and other approaches to manage LGMD.

"I swim and do stretching almost everyday; I take vitamins and oxandrolone; I try to live as much 'normal' as I can, driving, going out with friends, taking care of my husband and kids. I still walk with a cane and I take care of my diet to keep slim. Unfortunately, I had to stop working because it was too tiring."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"To recover movements that bring back my physical independence."

42-year-old woman living with LGMD2A (diagnosed at 15 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Not being able to walk, unable to raise arms and hip/lower back pain from all-day sitting."

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"The most frustrating and affects my daily life and being able to enjoy life better is not being able to toilet myself. Affects my social life and my health and is so hard to deal with because it is an activity that is necessary multiple times a day."

What worries you most about LGMD?

"It will severely cripple me and make my quality of life terrible and I will probably need to be in a nursing home."

Please describe your experience with treatments and other approaches to manage LGMD.

> *I have always just been as physically active as possible. I only ask for help when necessary. I do chores as PT. My partner does range of motion on me five nights a week before I go to bed. That helps me and helps me sleep better. I am careful with my diet, so as not to gain too much weight or encourage other health problems. I have home modifications like a ramp, toilet with moveable arms, shower seat. I wear loose clothing. I use satin sheets and satin-type night clothes to help in rolling over through the night. Everything I need to access, cupboards, fridge, freezer, etc is in my reach/level.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *A treatment that stops or deters progression. Aids that are affordable, like standing wheelchairs or roll-in showers, etc. Better yet, insurances like Medicaid should cover them instead of considering them luxury, unnecessary items.*

31-year-old man living with LGMD2A (diagnosed at 24 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Weakness of Shoulder, Hip/lower back and thigh muscle*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Sitting up from chair is extremely difficult, unable to climb stairs, very difficult roll in the bed or get out of the bed. Easy fatigue, with only 5 minutes of work.*

What worries you most about LGMD?

> *There is very good research done by multiple organizations, however I'm afraid that it will never get tested on time by FDA which will cause me to go on wheelchair knowing that this could have been prevented.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Physical therapy has helped slow down progression.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Being able to be independent and do what I like for my career. Also being able to care for my family and not be burden to them.*

70-year-old woman living with LGMD2A (diagnosed at 32 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Standing & walking"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Getting dressed, combing my hair, waving my hand, walking to toilet & getting in & out of bed."

What worries you most about LGMD?

"I'm never going to do things on my own again."

Please describe your experience with treatments and other approaches to manage LGMD.

"Fighting Back Program for exercise. During pandemic lost a lot of strength. Back on program now."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"less costly caregiving"

34-year-old woman living with LGMD2A (diagnosed at 13 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Weakness in legs and shoulders"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Dressing standing up"

What worries you most about LGMD?

"Expensive cost of treatment / cure when there's one. It's equivalent to no hope if the treatment is very expensive."

Please describe your experience with treatments and other approaches to manage LGMD.

"Massage to promote blood circulation, stretching to maintain flexibility and vitamin Bs"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Affordable treatment / cure."

24-year-old woman living with LGMD2A (diagnosed at 15 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Lack of balance, struggles getting off the floor and lifting heavy objects and general weakness in muscles*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Running, doing stairs basically everything*

What worries you most about LGMD?

> *Losing all my muscle strength*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Physical therapy*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Medication or surgery to help stop it or make it better*

45-year-old man living with LGMD2A (diagnosed at 29 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Falling down, Unable to do any action against gravity, deteriorating basic functions like breathing, sitting*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *A whole bunch: Bending, gripping things, sitting during a meeting, typing for long, walking stability without falling down. Not to mention the lack of ability to play any sport with my child.*

What worries you most about LGMD?

> *Ultimate destruction of all muscles*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Prayer to cope mentally*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Any improvement that does not lead to newer or other health or mind complications*

21-year-old woman living with LGMD2A (diagnosed at 13 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Not bending the back and straightening it, not seeing"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Easy to go around and go out"

What worries you most about LGMD?

"Being exhausted"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Spiritual healing"

63-year-old man living with LGMD2A (diagnosed at 48 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Climbing Stairs"

"Not able to sit on floor"

"Not able to pick things from floor"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Little bit of running"

What worries you most about LGMD?

"To be non ambulatory patient"

Please describe your experience with treatments and other approaches to manage LGMD.

"Homeopathy medicine which I think has slowed disease progression"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Like today I may be able to live my remaining life as ambulatory patient."

50-year-old woman living with LGMD2A (diagnosed at 26 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *muscle weakness, (hip, shoulder, and back) pain, grasping*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Transferring, independent dressing, cooking*

What worries you most about LGMD?

> *Genetic concern for offspring*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Aqua therapy, wheelchair upgrades/technology, household & vehicle accommodations*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Pain management and mandated support with equipment from insurance*

49-year-old woman living with LGMD2A (diagnosed at 12 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Massive overall muscle loss and weakness, intense pain, exhaustion*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *The list of things I can no longer do far outweighs what I am still able to do. Some specifics are — the inability to: use the bathroom independently, dress myself, bathe myself, reposition myself/apply medicine/use devices in order to reduce pain, hold my head up, get in/out of bed, cook/make meals, drive for long distances, independently go where I would like and be able to do what I would like when I got there...the list could go on.*

What worries you most about LGMD?

> *The continued progression of this disease is like a ghoul always hovering over me. What will be stripped away next? What new pain will crop up and become a constant reminder of the devastation that is happening to my physical self? I am worried that if my caregiver becomes hurt or unable to care for me, I will be in an immediate state of horror. I have moved through this before and it is a nightmare. Caregivers do not fall out of trees, they are expensive and they are usually nowhere near as skilled as someone you have worked with for years.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *"There are such limited options for treatments for LGMD – are there technically any other than pain dampeners and durable medical equipment? Thankfully, I am a strong advocate for myself and I have an excellent PT. What has been helpful in managing my condition is having an excellent caregiver who provides exceptional insight and delivery of what I need for activities of daily living. I take baclofen for severe muscle spasms in my back. I often use a heating pad or Thermacare patches for my back. I have a TENS unit for my back. I use OTC drugs for bursitis in my shoulders and hips. I have had cortisone shots as well. I do some stretching. I have a brace to help slow the contractures I am getting in my ankles. I have a new brace that attaches to my chair to help me sit upright/support my trunk. I have and use the tilt feature on my power chair for helping to alleviate horrible pain under my ribs (from not being able to support my trunk any longer), my back, hips and neck. I have a Tempurpedic mattress to help me sleep for a few hours at a time before having to be repositioned – I also use a huge memory foam leg pillow for positioning at night. I use a heater on my legs in the winter to help not be freezing all the time. I also use toe and mitten warmers to go outside [even for short trips] because I am very susceptible to the cold. I generally no longer eat sugar/sweeteners or flours of any kind in order to reduce inflammation and cut down on my pain. I follow this diet also to keep my weight from getting too high. I used it to lose 50 pounds and maintain that weightloss when I stick to the plan. Lifestyle modifications are things like – not going places I would like when I would like because it might be too exhausting to do so. Just getting ready for an event takes so long and tires me out so much, I might not be able to enjoy myself when I get to the event. I have greatly limited my outings and doings because of MD – especially as I age with this disease."*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *"I have lived with this disease for most of my life and I have come to terms with there not being any sort of cure in my lifetime – especially for my type of MD. One of my friends asked me why there wasn't more happening for my type [vs. DMD for example] and I simply said that people with LGMD2A/RI tend to live longer. Perhaps this is my pessimistic/realistic view of how things have played out thus far and I agree to extend someone's life as much as possible is what is important. So I am happy that the people I know that have more severe forms of MD have gotten treatments sooner. However, I would still like meaningful treatment options for myself before I die. If there was something that would halt the progression – or even slow it [since it seems to ramp up the older I get] – that would be wonderful."*

26-year-old woman living with LGMD2A (diagnosed at 17 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *"Not being able to walk without holding on to someone due to losing balance, not being able to sit down and/or get up from a chair without help. Not being able to pick up anything from the ground due to not being able to bend over at all. Plus many many more."*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *"Yes, there are many activities, most of them."*

What worries you most about LGMD?

> *That one day I won't be able to move at all, that I won't be able to have children, I won't be able to live life to the fullest. That in the worst case scenario, god forbid, I would lose my family who help me with everything and who I depend on, and would simply not be able to live because I can't do almost anything by myself.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *As there's no treatment, drug, or cure for LGMD, there hasn't been anything that helped coexist with this condition. I recently bought a scooter and that's the only thing that helped at slightest.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *A meaningful benefit would be to be able to walk without holding on to someone or objects in order to not lose balance and fall, of trip and fall and not be able to get up on my own.*

63-year-old woman living with LGMD2A (diagnosed at 18 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Cannot raise arms, Walk slowly, Cannot lift*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *I cannot lift my grandchildren. Grocery shopping for items on tall shelves.*

What worries you most about LGMD?

> *Being so dependent on others for daily things.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *I try to keep my weight on the very slim side. I used to swim but now walk and do adapted water aerobics to try and keep as much muscle as I have. I have many adaptations in my home such as lowered counters, smart lights, no steps.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *If I could stop the progression so I could at least maintain what strength I have.*

60-year-old woman living with LGMD2A (diagnosed at 4 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

1. *"Loss of hip and leg strength which allows for walking and standing"*
2. *"Not being able to lift my arms"*
3. *"The fatigue from ordinary activities is limiting."*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

- *"Walking, getting out of bed, dressing and showering"*
- *"Opening doors, eating, drinking"*
- *"Driving"*

What worries you most about LGMD?

- *"My worry for what will happen after my husband is no longer able to care for me is constantly on my mind. At times it is all consuming and affects my quality of life."*

Please describe your experience with treatments and other approaches to manage LGMD.

- *"Using a power wheelchair to manage fatigue with a ramp van for mobility. Aleve as needed for pain. A roll-in shower with shower chair."*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

- *"Stop the progression to preserve any muscle I may have left! Or, figure out how to build muscle in a limb or all over. Hopefully to build it faster than the disease destroys it. I could use just one strong muscle!"*

21-year-old woman living with LGMD2A (diagnosed at 16 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

- *"I constantly lose balance both in private and in public which have hurt me some times. I can not walk for long periods and feel my legs getting tired very fast. I can't get up from a seated position easily and I can't get up off the floor at all on my own. So, when I fall, I need someone to help me up."*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

- *"Dancing. I would love to dance, but I can't because it requires too much movement which I can't do easily. Exercising as well. Also traveling because a lot of places has stairs, especially in different countries."*

What worries you most about LGMD?

> *That there will never be a cure. That one day, I will fall really badly and hurt myself to where I break/fracture something badly and it will make things worse. Also, if one day I decide to have children, I'm afraid that the gene for LGMD will be transferred to them.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *So far, there is nothing that has helped me yet and I'm trying to find something. I tried some form of IV for 6 months, but it didn't help with my strength at all.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *If I can become stronger in both my arms and legs. If I can go up stairs slightly easier and get up from a seated position easier as well.*

Ayşe, living with LGMD2A (translated from Turkish)

> *I am Ayşe, I am 24 years old I live in Manisa Demirci in Turkey I have LGMD2A genetic muscle disease I can't stand I can't stand I can't stand I have trouble walking I've been walking with support I've been struggling with this disease for 12 years please help me too I want to get well and regain my health please help me too for the sake of Allah.*

Jill, 43-year-old woman living with LGMD2A

> *I'm grateful for this opportunity to communicate with the FDA. I was diagnosed almost 20 years ago and am now 43 years old. The progression of this disease has affected my quality of life and mobility immensely, and our LGMD2A community is in dire need of effective treatment options. I think most of us feel that time is ticking while we age and our bodies worsen. The only treatment we currently have would be physical therapy, but it is unfortunately not enough in most cases. I can no longer walk upstairs, carry a pet or a child, ambulate without a rollator, lift my arms above my head, do a squat, dance, walk on the beach, roll over in bed unassisted, get up independently from a seated position, or give a proper hug. I have fallen more times than I can remember and can no longer get up from the floor without help. If it weren't for my husband, I would likely need to live with other family members or within a form of assisted living. I have fallen more times than I can remember and can no longer get up from the floor without help. If it weren't for my husband, I would likely need to live with other family members or within a form of assisted living. The inability to exercise enough to burn calories has led to weight gain over the last several years. I have so much for which I am grateful in life, but this disease has taken away many freedoms that I once took for granted. I would do just about anything to regain even half of the physical abilities I had prior to my diagnosis. I sincerely hope that we can impress upon you the importance of our physical, social, and mental well-being and the urgent need for additional treatment options within our lifetime. Thank you for meeting with us all today.*

Mara, parent of 12-year-old son with LGMD2A

"My son (12) is diagnosed with 2A and has his first symptoms just now. He is walking, running, biking, he can do still almost anything, but he starts to tire, the legs hurt more often, and there was the episode he lost ability to walk for some minutes because of sudden weakness in his muscles, last week. He was very scared. And it is so hard to watch the process being absolutely helpless. There are just three persons of the same subtype 2A here in Latvia. There is no hope for special treatment or participating in some research project. But we do not lose our hope for some chance for cure. Until is too late for my son. He is still on the beginning of this hard way... And I pray for people who are working to find help for all people like my son."

Valley, 42-years-old, living with LGMD2A

Comment 1:

"I have LGMD2A, I'm 42 yrs old. My daughter is 9 years old. In my childhood I was such a sporty person, basketball, driving, dancing etc. When I was 20 I came to USA and working so hard but later near 27 approx., when I was walking everyone in my family and friends start saying that why are you walking like that. but I never took it seriously because I don't felt any problems. I married in 30 then after my pregnancy (it was normal delivery) I feel that day by day I'm getting weaker, when to doctor and then they diagnosed me with CAPN3 (LGMD) I have my blood reports. I can send it to you. Now I'm on walker my situation is becoming worst day by day, getting up from toilet seat, taking shower, using of hands while eating, shortness of breath. I'm in too much depression Sir! Please tell me when will the cure come for this disease. I went to the neurologist but they said they are helpless, they don't give any medication nor suggest anything like which proteins you have to take, or what should you eat to keep you healthy. LGMD..Hoping for the cure to come fast. Always praying to GOD. please help me"

Comment 2:

"I'm 42 years old. I have LGMD2A. I have me genetic blood report. It's CAPN3. I was walking by taking a support but now from one week not able to walk, I am totally dependent on walker now, day by day it's getting worst, look like soon I will be on wheelchair, don't know what to do! Went to the neurologist, he said I'm helpless sorry! No medicine No cure .. therapy is also not helping, I'm a housewife very difficult for me to survive. My daughter is 9 yrs old. Before I was a sporty person, playing badminton, dancing, driving doing a job in a restaurant. In 2017 they diagnosed me with LGMD. Every time I'm praying for the cure to come. Please help me please! Hoping for the cure to come fast before I die. It's intolerable!!"

Natalie, caregiver of husband with LGMD2A

Comment 1:

"My husband was diagnosed with LGMD2A over 15 years ago he could walk when diagnosed and slowly got weaker and weaker. Now his life is carers toileting washing and dressing him and debilitating loss of strength and severe fatigue."

Comment 2:

"My husband is in his 3rd year of a electric wheelchair. He has little strength to stand now. He has one last movement which is a stand and fall into bed. His progressive downhill slide has left him now needing fulltime care and carers coming into the house regimenting our lives which is exhausting, robbing my husband of a fullfilled life and robbing me of the full life I should be able to live. I feel I'm not living the life I was meant to have. Having to become a carer for my husband leaving fulltime employment to do the basic care my husband requires which is toileting."

Comment 3:

"My husband can no longer walk at all. My husband always struggled getting up from a seated position. Now this disease has robbed him of mobility. Needs care to toilet wash and dress independently."

Comment 4:

"Can't walk at all now and needs carers to dress toilet and shower. Affected him mentally as depressed."

Comment 5:

"Can't dress toilet or shower anymore cant walk at all too weak. I used to shower my husband when he could walk into a shower if it was level. Now my husband needs to be hoisted onto a shower chair and helped to be washed and showered."

Comment 6:

"My husband has gone from living a healthy normal life to gradually over time losing his independence in areas. At first he needed help washing in the shower. So I washed him standing up. To no longer having the strength to stand walk and at first for safety reasons we had to wash sitting down in commode chair. Now we can't walk even in the house aided anymore. He totally in dependant on a wheelchair hoists to shower and carers to wash, dress and toilet."

Velma, woman living with LGMD2A

"I'm enrolled in the Fighting Back Scholarship Program since 2008. During the pandemic I couldn't attend the program for 2 years. I did virtual but I really needed hands on. I did experience weakness during that time. Now I'm back and doing better. I was able to use a walker before the pandemic so I'm working hard to use it again."

Brenda, woman living with LGMD2A

"Diagnosed in 2010 with LGMD2A R1 at Brigham & Woman's Neurology with Dr. Anthony Amato. He wrote the textbook on Limb-Girdle. At 50 years old I changed my whole world and living situation. The state assisted in ideas to modify my home and work situation. I am presently mobile and working as Activities director at Vermont's only 5 star resort, 28th year. I fly-fish, lead bike rides off property, moderate hikes, cross country ski instruction, sea kayak with resort guests and do Art tours in house. I have a good long life ahead with this slow progressing disease. Missing 2 genes, Calpain and 1 unknown."

Rosaria, woman living with LGMD2A (translated from Italian)

Comment 1:

> *Over time I have weakened. And weighted down. I have had 2 missed pregnancies. We do everything together with my husband helps me wash myself to go to the bathroom. He gets me out of bed and chair. Helps me cook. We can't dance. Or take a bath alone in the sea. Over time you get tired a little more. But the grit and the love and the will to live is always the same. Because we have confidence, Faith in God, and we hope for a cure.*

Comment 2:

> *I can't raise my arms. I cannot get out of bed by myself from the chair or to go to the bathroom without the help of my husband. Or take a shower. I lack autonomy. I hope in your cure calpain 3 2A. My husband quit his job and became my assistant.*

Comment 3:

> *You cannot dance or go swimming in the sea alone. As you grow, fatigue makes itself felt. We are in your hands. Give us a cure. Thank you*

Comment 4:

> *I can't get out of bed by myself from the bathroom chair without help. There are days where you are full of life and others where you feel tired and think you can't make it. Yeah. We can't dance. Running or bathing in the sea. Growing up fatigue begins to be felt. Also because the cure has not yet arrived.*

Camilla, woman living with LGMD2A

> *I live with LGMD2A for 35 years, since I was 14. I struggle everyday to maintain my mobility and keep walking, but it's getting harder and I don't know for how long will it still be possible. So the timing for me and for most of us who live with this disease about a future treatment is crucial. If it takes 10 years for a treatment to be available probably it won't find many of us here to help. A late treatment means no treatment. So my question is: should we keep our hope for the next few years to have something concrete as a treatment? If so, does it have the potential to regress the symptoms? Thank you all very much for the attention. Regards, Camila (from São Paulo, Brazil)*

Maureen, caregiver of a 21-year-old son living with LGMD2A

> *Very interested and hopeful to know the possible treatments for LGMD2A, calpainopathy. My son Alberto Zamora suffers from it. He is almost 21 years old, he still walks with difficulty but falls frequently.*

Narenda, caregiver for daughter living with LGMD2A

My daughter is almost at the end of her life struggling since 2007 with Calpainopathy. There is no retarding factors for the disease to stop the progression of the disease.

Angela, woman living with LGMD2A

Comment 1:

Symptoms that most affect me 1 – arm weakness limiting my ability to perform many tasks 2 – walking limiting my independence and ability to go particular places, do things that I once enjoyed and go up/down stairs 3–pain and fatigue. Due to compensating with other muscles, I've had chronic back pain and fatigue easily throughout the day causing me to spend more time resting in bed during the day. On best days I have energy to spend time with my family doing something fun or the energy to do everyday tasks around the home as independently as possible. On my worst days, I fall or do not have the strength or energy to get out of bed, and have back or leg pain. Due to LGMD, I can no longer work as a nurse especially due to arm weakness. This is saddening and difficult emotionally and financially. I also feel like I cannot travel freely due to limitations on airlines, limited mobility once I get to a travel destination. Finally, I cannot just go to public places without concern of accessibility and planning ahead of time. Over time, I have been able to cope better even though my disease has progressed due to having a good support system, counseling, and gradual progression. I have just found it easier to accept what I am dealing with the more that I understand it and live it and the more hope there is for some treatment.

Comment 2:

I have used PT primarily for pain management and see a pain specialist PRN. I have had injections in my hips and nerve ablation to my lumbar area which has been life changing. I take medications at night that help me relax and sleep. I have used counseling to cope at times with life changes and online support groups. My biggest challenges have been finding providers that are knowledgeable about LGMD, access to patient studies as the travel is not always re-imbursed and difficult to arrange or doesn't allow for a companion to go with me without expenses. For many years a big challenge was finding an accurate diagnosis (this remains an issue) It took almost 10 years to identify one copy of a CAPN3 defect it is believed that I still have another CAPN3 defect that has yet to be identified which is also the case for 2 of my siblings. I would like to see disabled individuals be able to travel by air remaining in their chairs or to be able to have a foldable travel electric chair covered by insurance to travel more easily.

Peter 1, 23-year-old man living with LGMD2A

LGMD2A impacts every portion of my life. As an ambulatory 23 y/o, the gradual deterioration of LGMD has taken a toll on my physical being as well as my mental health. However, I also feel that LGMD has strengthened me as an overall human being– we are resilient people, truly.

Of all of the symptoms of LGMD, leg weakness is definitely the one that impacts me the most. In public, not only do I walk slower and with a gait, but I feel a deep shame and oftentimes feel like everyone is watching me (although, this is becoming easier to cope with).

LGMD has affected my friendships tremendously. Many friends that I hadn't seen due to Covid in 2020 became more difficult for me to rekindle with– a lot of these people didn't know of my disability, but since the last time I saw them, it has become harder to hide. Spending time with people I have not seen in a few years is hard for me, so I push them off. On the other hand, my good friends, partner and family are easier than ever to spend time with; they are accepting, helpful and good people for me to have within my life. I strive to eventually re-connect with all of my friends from the past.

I always say I have bad days and worse days. It's harsh, but true. I did not experience symptoms until I was 18 years old, so it is easy to remember what being "normal" was like. My bad days I feel tired, but able. My worst days, I am still able but plagued by sadness and depression. In public, not only do I walk slower and with a gait, but I feel a deep shame and oftentimes feel like everyone is watching me (although, this is becoming easier to cope with). Luckily, I do not experience days like that often.

I went to and graduated culinary school and worked in restaurants until I couldn't anymore. I miss the calamity of a professional kitchen so much, but I am unable to do that work – what I love – anymore. However, I did move into food journalism and reporting, which is also a line of work I love. It combines my love for food and cooking with my talent of storytelling. I am thankful in a way that my disability led me here.

My ability to cope with symptoms has gotten better. Having a partner and friends who understand is then best therapy. I still hide a lot, but just two years ago I was hiding everything. Being open is a huge weight off my chest.

I thank you for your time today with this panel and I implore the FDA to approve genetic treatments so people like me can settle into a physical state rather than constantly dealing with change and more muscle loss. I am tired and I want stability in my condition more than anything else.

Karen, 62-year-old woman living with LGMD2A

Comment 1:

I have 2A. I am 62, diagnosed in 1988. Impacts often overlooked include increased difficulty getting to doctor appointments, dental care, toileting, loss of hands. Women can't find the doctors that are equipped for well woman visits. Not everyone has a caregiver or the financial means to get assistance. It's the loss of little muscles as well as large muscles. I am limited to traveling because of toileting. I had to leave work because I couldn't use the toilet at the office. At home, I am dependent on my husband for showering, meals, dressing, any household chores. I am left with my voice which I'm using to advocate for a treatment. This disease affects not just the patient but everyone. It's expensive as we usually require lots of equipment. We need not just hope but treatment and soon.

Comment 2:

2A, 62 years old. Chiropractor treatment helped with my scoliosis and balance, hip pain. Husband uses a massage roller on my hips, I use kinesio tape everywhere. I have soft orthotics for my feet and legs. I swam and worked out at the YMCA for years but it's too draining now but I greatly encourage it.

Janice, 70-year-old woman living with LGMD2A

"I am 70 yrs old. I have LGMD2A/R1, diagnosed at age 16 and I have thinning of skin on my bottom. Lots of pain in my bottom bones. I'm sitting on a special cushion but it hurts still. Is there any thing more I can do to help this?"

Baha, individual living with LGMD2A

"I'm originally from Sudan, I have 2 other siblings living with LGMD one of them passed away 4 years ago. I was diagnosed with LGMD2A at the NIH."

"I can't wait to start receiving the gene therapy, thank you for the hard work you do for us and we hope that the FDA will support this new treatment to cure LGMD!"

"There's always a hope and light at the end of the tunnel!"

Teresa, woman living with LGMD2A

Comment:

"Manual wheelchair, power wheelchair, grab bars around home, lowering cabinets, rising toilet, chair and bed yoga. Minerals and vitamins."

3rd poll question….

"very little, but just started supplements of minerals and vitamins."

Akriti, 29-years-old individual living with LGMD2A

"I am 29 years old."

"It was in 2004 when I was in 7th standard, that I started experiencing frequent falls and incidences of knee buckling. But at that time I didn't pay much attention to these symptoms thinking of it as some sort of temporary weakness."

"The symptoms became more noticeable an year later when I started using support for climbing up the stairs and my teachers and family started pointing it out."

"During early teenage when everyone around me were growing up, I could see myself becoming weak day by day and did not even know the reason behind it. I was getting slow, my gait was different, couldn't get up from floor and climbing stairs had become more difficult."

"I was diagnosed with LGMD2A calpainopathy in 2008 at age 16."

"However, I still pursued my studies, cleared pre-medical test and did MBBS, and MD."

"Presently I am a doctor living with LGMD2A"

"I stood first in my batch during my residency, but I did not opt for super-specialization because of my growing disability"

"Although I am a qualified doctor, I am restricted to work from home"

"Three symptoms that have had the most significant impact on my life are:

- *inability to get up from chairs and couch*
- *inability to walk independently*
- *inability to climb stairs"*

"Muscular dystrophy has affected my life in every possible away."

"It affects the life of my caregiver (my husband) enormously, as because of me he has to be by my side always. I am completely dependent upon him for bathing, toilet and other basic needs."

"It is heart breaking."

"I really hope that some treatment option comes soon"

Matt, man living with LGMD2A

"One of the hardest parts is slowly losing your independence and mobility. It is really hard to continually adjust my expectations for my life. It is hard to not feel defeated when I have to keep letting go of activities I used to be able to do. And I'm especially scared of the future because my muscles will only grow weaker without treatment."

Lisa, parent of individual living with LGMD2A

"I am a parent of a 30-year-old with 2A and I just want to reiterate that this is a physical disease but it also so strongly impacts a persons socialization and their mental health. Treatments and cures are very desperately needed."

Roman, man living with LGMD2A

"My name is Roman, I'm 34 years old, LGMD2A

I came 5 years ago from Russia to participate to research. My doctor asked me to stay here if I want to be as close as possible for future research. I sold everything I had, still sacrificing a lot to be here and wait for the treatment.

No support, no family. Surviving and wait for news from you guys. I hope you can develop the treatment asap. I don't know how long I can stay here."

Christopher, caregiver/spouse of individual living with LGMD2A

Comment 1:

"Having seen the rapid pace COVID vaccinations were developed once the FDA understood the necessity, it is frustrating to witness how slow the development of treatment options have taken. Especially considering the all-encompassing low quality of life at various stages across all types. I understand the need for funding and incentives for companies to actually work toward treatment options, but surely there are ways for the FDA to incentivize companies to push for treatments and to accelerate the approval process based on the body of research already in existence. I mean there's no treatments, not even to pause or reduce symptoms. Can you speak to the reasons why this is so? I'm sure a large percentage of individuals with the various types of this disease would be absolutely willing to participate in any promising study regardless of the risks."

Comment 2:

"As a spouse/caregiver, I'd say the most important aspect for 2A would be reducing or pausing the effects on mobility, especially on the hip flexors/legs. Watching my wife fall and having to lift her off the ground, often in front of crowds, is dreadful for her to endure. I also have to help her up from many chairs and toilets, as well as into and out of zero-entry swimming pools when a lift is not available. Swimming pools are the only time she gets to feel the freedom of normal movement and serves as her only real way to work out."

Heather, 60-year-old woman living with LGMD2A

"I am 60 years old with LGMD2A diagnosed when I was 30. what I most struggle with on a daily basis is when going out and the worry about if I suddenly have to use the restroom. I am wheelchair bound on my own with no caregiver and so I have to rush home to use the bathroom. I wish there was an easier way."

Aleksandrov, man living with LGMD2A

I have LGMD2A…whatever I write you have heard from thousands of other people like me. Every single minute is hard and I expect this year already to release the treatment for limb-girdle.

Mostafa, parent of a son living with LGMD2A (translated from Arabic)

My son has LGMD2A muscular dystrophy. It appeared when he was 5 years old and now he is 11 years old. The situation has come that he cannot raise his hand and cannot roll over on the bed at night and he is now sitting at home only walking but very carefully and I want treatment for him.

Emily, mother of a 16-year-old daughter living with LGMD2A

I am hopeful that the FDA understands how important it is for those with LGMD2A (and others) to have a treatment or cure. My daughter was diagnosed at age 7 and is now 16. We no longer can watch her run, walk, or stand up on her own. She needs our help transferring from her wheelchair to her bed. I am most concerned about loss of strength in her arms as it will impact her independence even further. She is a smart, thoughtful, talented girl who plans to pursue a degree in Chemistry; she has so much to offer to others. I want to know that she will always be able to share her gifts with her community. More importantly, I want to know that she can live a full, independent life. Our family thanks the organizers of the EL-PFDD meeting as well as those who are actively working on a treatment or cure.

Jessica, mother of a nine-year-old daughter living with LGMD2A

A cure would preserve the walking ability my nine-year-old daughter Sophie has left. She is fully aware of her muscle strength decline due to her LGMD/2A diagnosis and it scares her and causes immense anxiety. On bad days Sophie can't get out of bed for two days, she has severe pain, cramping, and chest pain. A bad day can happen after day of normal activity. She has spurts of decline and we never know how much will happen or if she will continue to be able to walk. Knowing a gene therapy was in development would give us all hope and she can continue to play with her friends and dance.

The ASCPA runs commercials for supporting and bringing awareness to animals constantly, St. Jude receives billions yet MD patients are often forgotten and there is a big population who can increase their quality of life significantly and remain more independent if these drugs are brought to fruition and available to patients like my 9 year old daughter. Sophie is fully aware that she is losing her abilities and muscle strength, she is in pain and needs help. The FDA must consider pushing along clearance and research for gene therapies for muscular dystrophy and all types not just the most well known types.

Bogdan, father of a five-year-old son living with LGMD2A

"Hello, I'm sorry it is hard for me to express my self in English. My only son, he is 5 years old, was diagnosed with LGMD2A this year and our lives have turned to a nightmare. Only my wife and I know this because if our parents find out the news would kill them. Again I am sorry I can express my self and say more. I hope we all will have a cure sure. Thank you and best regards."

Jillena, woman living with LGMD2A

"I would love to be able to drive but vehicles that are accessible are way too expensive to afford. I work so I don't qualify for help with that unless I haven't been informed of all the programs available which is definitely possible."

James, 52-year-old man living with LGMD2A

"I would love to have some of my mobility back, range of motion and able to sleep in my bed and take hot shower again just to relax my muscles."

Noni, woman living with LGMD2A

Comment 1:

"I know I won't be able to walk again, but if a treatment could help the next generation of kids not progress to the debilitating state that I'm in, that would be success for me."

Comment 2:

"Initially when I was a kid, my struggle was going up a flight of stairs, running, jumping. Then the next struggle was getting up from a chair or from the toilet, and getting up from the floor after one of the many falls. Then the struggle was walking even with assistance. As I have already lost the ability to stand up and walk, my current greatest struggle is losing the use of my arms, which cannot really be replaced and limit my life in the greatest ways.

If a treatment would at least preserve the limited use of my arms, I would consider that a success!"

Fatoş, individual living with LGMD2A, (translated from Turkish)

"There is no situation as difficult as needing a person who has always limited me in my life. My worst memory was the day I couldn't go out, and being needy of my old mother, please find my treatment."

Martha, woman living with LGMD2A

"It occurs to me that most of the 'treatments' being talked about are not really treatments but symptom control or 'dealing with' the weakness. A real treatment would at the very least, stop the progression. Current treatments at best, slow progression."

Melissa, woman living with LGMD2A

Comment 1:

"The symptom with the most impact on me is the muscle weakness. Due to muscle weakness I have a high risk of falling and need supports to get up from a seated position, which limits my ability to participate in activities professionally and socially. I cannot sit on the floor with my students because I cannot get up, I cannot visit colleagues' or friends' homes easily, I have to ensure that it is accessible for me which requires planning. This all leads to intense anxiety and fear on a daily basis because I have to be so aware of my physical environment; 'if I fall, what will I use to attempt to get back up? Who is available to lift me if needed?' 'If I go to –name–'s house, will they have stairs?'

Similarly, when I get sick, such as with a cold or the flu, my muscle weakness is intensified, and often leaves me bed ridden. I can't physically move because my muscles hurt, but not moving is also bad for the muscles, so I am in a no-win situation. Even a simple cold requires me to get help with tasks such as going to the doctor or getting medicine."

Comment 2:

"My independence has been severely affected because of limb-girdle muscular dystrophy. Doing any activity alone is risky because of the risk of falls, the worry that I may not be able to reach something, or anxiety that bathroom facilities may not be accessible.

I do not attend activities at other people's homes because of the risk of falling or possibility of stairs, which I cannot physically do. I avoid traveling with friends; I need assistance to navigate an airport due to the size, airport security is especially time consuming because I have to explain that I cannot get up to walk through. Airplane seating is very tight, which is uncomfortable and I have a lot of difficulty getting up from an airplane seat.

I avoid a lot of social activities, including dating, due to having muscular dystrophy because there are too many unknowns in new situations. I always volunteer to pick a restaurant because I know which ones are accessible, which frustrates my friends who want to try new things. I always volunteer to drive places because I know my vehicle is accessible to me, so I am using more gas and putting more wear on my vehicle without compensation from others."

Hisham, individual living with LGMD2A

"I'm from Iraq and I'm 29 years old. there are many activities such as hiking, camping that I used to do when I was younger but unfortunately now I can't do them anymore. Now I have a difficulty to climb the stairs, difficulty in working out. and my muscles are being weaker and weaker. in Iraq, we don't have a good awareness or knowledge about this disease in the society and health care workers too and that make it harder for me because of lack of support."

LGMD2i / LGMD R9 FKRP-related

35-year-old woman living with LGMD2i (diagnosed at 4 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Loss of the ability to breathe independently, loss of mobility due to the loss of strength, loss of upper arm function.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Caring for myself (showering, toileting, eating, etc.)*

What worries you most about LGMD?

> *That I might die soon because of my respiratory and cardiac involvement!*

Please describe your experience with treatments and other approaches to manage LGMD.

> *That's too broad for a question.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *To stop the disease progression and to gain lungs vital capacity!*

63-year-old woman living with LGMD2i (diagnosed at 35 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Weak legs: unable to stand up unassisted, weak core: lack of balance, fall risk.*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Going up stairs, walking up an incline, standing up from the toilet, chair or car seat.*

What worries you most about LGMD?

> *Heart and/or lung involvement and falling and breaking a bone. Also, needing a wheelchair.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *I take D-ribose and I think that it helps a little bit.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Anything that slows the progression.*

37-year-old woman living with LGMD2i (diagnosed at 2 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"leg weakness, loss of expanded movements, loss of individual abilities (required way more assistance)"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"walking unassisted, getting up and down from seated position or stairs"

What worries you most about LGMD?

"future degenerative losses or losses of what little I can still do independently"

Please describe your experience with treatments and other approaches to manage LGMD.

"I have not been able to participate in any trials due to a multiple sub type diagnosis. The only daily management I use is assistive devices and OTC pain killers as needed."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"Halting any new muscle loss/degenerative changes"

64-year-old man living with LGMD2i (diagnosed at 56 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Not walking and not havening the strength I used to"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Walk"

What worries you most about LGMD?

"That I will be too old when the treatment is available"

Please describe your experience with treatments and other approaches to manage LGMD.

"therapy but it's limited on its effects"

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"A treatment that helps improve strength"

32-year-old man living with LGMD2i (diagnosed at 4-5 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Weakness, imbalance, the visuals of atrophy*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Many. I'd like to do sports, but that's very far away. Even mundane things such as walking stairs, rising from chairs, or running to be in time, are hard.*

What worries you most about LGMD?

> *Not being self reliant.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *As far as I know, there are no real treatments. I've tried physical therapy, but the knowledge is limited and the results lacking.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *A slight improvement in walking stairs, and rising from the ground, to be able to participate in society. But even a halting of the progress is fine, then I will still be self reliant.*

Parent/guardian/caregiver of a 27-year-old man living with LGMD2i (diagnosed at 19 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

1. *can't get out of chair*
2. *can't walk long distances*
3. *must use walker full time*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *I can't walk on the beach, I can't get out of the chair at the movie theater, can't access lake or ocean water because I lose my balance / footing on uneven material. I can't enjoy the snow because I'm afraid of falling on the ice.*

What worries you most about LGMD?

> *Not being able to get out of bed, not being able to get in and out of the shower, not being able to get myself dressed, not being able to make my breakfast. Afraid of losing my independence.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Walking when I was younger has helped me, I think? No other treatments. Tried aqua therapy but was unable to safely get out of the chair lift.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Independently taking care of my basic needs. Preparing my food, dressing and bathing myself, walking without a walker or not needing a wheelchair.*

46-year-old man living with LGMD2i (diagnosed at 18 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *Walking ability loss, muscle pain, balance problems*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *Most of my work*

What worries you most about LGMD?

> *Cures are being tested right now, but the process is super slow. It would be nice to facilitate a speedier process, specially for non genetic cures.*

Please describe your experience with treatments and other approaches to manage LGMD.

> *Physical therapy is an underrated treatment. It helps me tremendously.*

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

> *Being understood as a patient with a complicated disease: we need custom help. Psychological support and physical therapy would be a must.*

40-year-old man living with LGMD2i (diagnosed at 39 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

> *weakness, movement restrictions, inability to lead an active lifestyle*

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

> *I can't go on a sailing yacht trip*

What worries you most about LGMD?

"inability to recover"

Please describe your experience with treatments and other approaches to manage LGMD.

"nothing"

54-year-old woman living with LGMD2i (diagnosed at 18 years)

Of all the symptoms you have experienced because of LGMD, what are the top three symptoms that you consider to have the most significant impact on your daily life?

"Broken bones, heart failure, restrictive lung disease"

Are there specific activities that are important to you but that you cannot do at all or as fully as you would like because of LGMD?

"Stand from a sitting position, walk without falling, reach above my chin, bathe, toilet independently"

What worries you most about LGMD?

"Being on ventilation 24/7, another femur break, heart failure, early death, loss of independence, need to go into a facility for care at a young age."

Please describe your experience with treatments and other approaches to manage LGMD.

"There is no medicine or treatment, but I take meds for heart issues. I try and keep my stress low, stay as active as possible, without further damaging my muscle, eat healthy, and attempt to keep my weight down."

Short of a cure, what would represent a meaningful benefit to you in a future treatment?

"If I could stop declining and stay where I am at, that would be huge. I am slowly getting worse every day. If I could just stay where I am, and maintain this quality of life, I would be happy."

Dianna, woman living with LGMD2i

Comment 1:

"One of my most important activities I'd love to do is to be able to run and play with my daughter."

Comment 2:

"I was diagnosed in 2019 with LGMD2 I. I was actually about four months pregnant with my daughter at the time. One of the first things I noticed was that I actually started having symptoms whenever I was about 23 years old and I am now 31. My first symptom was waking up one morning, and realizing my legs were weak, and throughout the years, it just kept progressing. I am able to walk with a cane.

I am not able to get up out of the floor without my lift, and if we go to restaurants, I have to have my husband to help me up out of any chair besides my lift chair, and when I was pregnant with my daughter, and even after I had her for about the first six months, I slept in my lift chair the whole time because it was just hard for me to get out of bed because we just have a standard bed. My fear is being a burden to my husband because before I got my lift to get off the floor when I would fall here at home, I would have to call him and he would have to leave his work to come pick me up and then go back to his job. I am a stay at home mom which I am glad for, but I know that my little girl would love for me to be able to get on the floor and play with her, go outside, run, jump and play with her but I can't. She's only two years old so she's a little energetic little girl.

Nick, caregiver of a mother living with LGMD2i

My name is Nick, I'm 29 years old living in Los Angeles. My mother, Anita was diagnosed with 2i in her 30's. I'm her only son and always helped with things as I grew up. We are a tight knit family unit, my mom, dad and I. The assistance I provided became and still is second nature. My mom has created a family culture of joy, happiness, and gratefulness. We have been fortunate and have spent time taking vacations over the years across the US and Europe. Growing up, my mom attended years of soccer, baseball, basketball and a short phase of paintball games. She never missed a chance to take my friends and I go out shopping or to the movies. She would host friend sleepovers when I was younger, making her legendary chocolate-chip pancakes in the morning. She was the first one to volunteer to be involved with my school activities. We have and continue to live overall happy lives. We laugh together, cry together, and support each other. We don't let little things get to us or hurt our morale. With that being said, it all boils down to how my mom is a relentless warrior, who will stop at nothing to live her life to the fullest. My father and I envy her amazing attitude and endless smiles. She keeps us going, she keeps us strong, she keeps us happy and not the other way around. We have had our fair share of struggles over the years but we never focus on those. We can only move forward and take the days as they come. I hope this short comment allows other families to see how we deal and cope with challenges presented by MD. We follow all news sources in hope of some type of breakthrough treatment. It seems like the science is getting close, if anyone reading this is a decision maker in getting things approved and through the system quicker – it is families like us it directly affects.

Kelly, mother of a daughter with LGMD2i

I think that it is so important to know that a 'successful' treatment would be to just stop the progression of this heartbreaking disease. Hitting a 'pause' button would be considered a win for our teenage daughter with LGMD2i. Anything that improves her strength and function would just be icing on the cake.

James, man living with LGMD2i

Limb Girdle Muscular Dystrophy 2i has impacted me the most with climbing stairs, running and my handwriting isn't the best. I get sore calves if I do too much exercise or walking.

Karen, 60-year-old woman living with LGMD2i

Comment 1:

> *The 3 symptoms of LGMD that affect me the most are inability to walk/climb stairs, rising from a seated position, and getting in/out of and turning in bed*

Comment 2:

> *Another thing my caregiver/husband does for me every day is provide emotional support and encouragement.*

Comment 3:

> *I use aqua therapy whenever I can and have home health PT to help with stretching and range of motion. I use a bipap every night. I use TENS, ice, heat, tylenol, and CBD to ward off debilitating chronic thoracic back pain.*

Comment 4:

> *LGMD2i, female, age 60. Living with LGMD is all consuming. One of the many concerns is the high costs of home adaptations, DME, accessible vehicles, copays are huge, ongoing obstacles that cause stress. It also is very stressful thinking about and planning for future needs. Having a treatment that would stop progression of this disease, would alleviate some of that anxiety.*

Margaret, caregiver for a son living with LGMD2i

> *Our sons declining lack of mobility*
>
> - *Can't get out of bed*
> - *Can't get out of a chair*
> - *Can't bend over to pick up things*

Kristen, woman living with LGMD2i

Comment 1:

> *I am a woman living with LGMD2i. The symptom that most drastically affects my life, all day everyday, is the struggle to get up from a seated position. I am unable to get up from many chairs, including many toilets. This severely limits my ability to go out and about as I never know if I will be able to go to the bathroom. I can't sit down at many of my friend's homes because I won't be able to get up from their furniture if it is too low. It is also extremely difficult to travel by plane because I really struggle to get up from the seat.*
>
> *A related symptom that makes life really hard is the frequent falls and inability to get up from the ground. I live in constant fear of a fall when I am outside my home. The instability and poor balance that results from the weakened muscles, means that if I stumble just a little I cannot catch myself*

and I will fall. Then once on the ground, I am stuck as I cannot get up unless there is a very strong person willing to lift me up. They have to be strong, because I am 100% dead weight until fully standing. I have been stuck sitting on the pavement of a parking lot freezing in the pouring rain waiting for a good Samaritan to come to my rescue.

The next symptom that most severely affects my life is the difficulty climbing stairs. I can't live in a home with stairs, so it is extremely difficult to find housing. I found that most apartments listed as first floor still have stairs to enter the building. I did find a 5th floor apartment with no stairs with two elevators. However, when a storm knocked the power out, I was stranded in my 5th floor apartment for 5 days because the elevators don't work without electricity. Because of the extreme difficulty with stairs I cannot access many buildings and facilities with my friends and family. You'd think ADA (Americans with Disabilities Act) would cover this, but many older buildings are somehow exempt from ADA requirements.

Comment 2:

I would love to regain some of my lost muscle strength and function. However, I would also be thrilled to simply stop the progression of the disease. Stopping the progression of this disease would be a huge success. The fear of things getting worse, and increased disability from this disease is intense. Having the ability to stop the disease from causing more muscle weakness would allow patients living with the disease to maintain whatever level of independence we have now and spare us from suffering further disability and further limitations. Please know that hitting the pause button is a major victory.

Michele, individual living with LGDM2i

Comment 1:

My top 3 symptoms that impact my life:

- *Balance*
- *lack of covered physical therapy*
- *Fatigue*

On my worst days my fatigue is my biggest complaint. I struggle with 'the more I do, the more tired I feel.' The overwhelming fatigue I feel makes it hard to commit to something.

My best days are few and I feel I can get a ton accomplished.

Something that I really would love to do is swim, I don't swim any longer as I don't feel safe anymore. I miss it.

Because of all the support groups I feel I am more equipped to cope with my symptoms. I would love to see more in person gatherings so that we can better support one another.

Comment 2:

I have LGMD2i. For me personally I would love to see some strengthening, but for younger people I would love to see a super slowing of progression. To keep kids and young adults walking as long as they can.

Edith, woman living with LGDM2i

"The future for us is always changing with our muscles progressively getting weaker. It's hard to plan for future events or life because we don't know what will our physical ability look like. I am in the process of designing a home to build. The process is frustrating because I could design for my abilities today but what about in a year or in 5 years? It is hard to know what I will need in the near future. I don't need many accommodations now but feel like I have to be selective and smart with my build. I also find that designing for special accommodations can be costly. Where able-bodied people tend to avoid those extra costs, we are always in need of something to help us live and continue to perform our activities of daily living, which can change quickly. The costs add up and can be a burden for many who can't afford it. Those that have to go without equipment, because of cost, are also having to go without living freely in this world, and are missing out.

If there was even a way to stop the progression of this disease, then I feel like more of us with LGMD could better plan for our future (even week to week) and participate in the world better with our loved ones without missing out.

A cure is the best outcome but anyway to stop the progression would mean the world."

John, 45-year-old man living with LGMD2i

Comment 1:

Q: Of all the symptoms of limb-girdle muscular dystrophy, which 1-3 symptoms have the most significant impact on your life?

"I have 2i and am 45 years old. In the past four to five years, my ability to walk and traverse normal, everyday situations has significantly diminished—walking from car into my office, walking through an airport, walking into a venue to attend performance such as a child's graduation ceremony or performance, walking around the yard to do light work with my kids, or picking something up from the ground. I've begun relying more on walking sticks this year. These are things that I felt I'd always be able to do, and if I could retain them and simply push the pause button I wouldn't have to begin the transition into assistive devices."

Comment 2:

Q: How does limb-girdle muscular dystrophy affect you or your loved one on best and on worst days? Describe your best days and your worst days.

"I used to be involved (at least present) in most family activities: helping coach a little-league team, attending outdoor activities, playing catch, exploring a new place while travelling, throwing the frisbee or other backyard games. Now I can't walk in grass, sand nor on any inclined slope. My necessity to prevent falls now overrides my ability to be with them in many activities, or requires we change plans or not participate in some activities. Now I must plan every step and action before we get to a new place. Google maps has become my friend to help me pre-navigate new scenarios. On good days we successfully navigate a new experience in a new place together. My bad days include me often being alone by choice or necessity while those I want to be with are doing something fun together, I'm no longer able to join in."

Steven, 64-year-old man living with LGMD2i

"I am 64-year-old male with 2i living on my own. I started having symptoms in my 40s, I was misdiagnosed with Beckers, then about 10 years ago with the saliva test I had a new diagnosis. 7 years ago, I took a bad fall down the stairs and broke c-1 I was very lucky to have no paralysis from the neck, but my muscles never recovered again to walk. I was accepted in a clinical trial for biomarkers with Dr Mathews. My concern at my age with new therapies or treatments for 2i on the horizon will I live long enough to see any of these I am not ready to give up my independence. I am slowly getting weaker. I do not want to end up in a nursing home. I am hoping that I will be able to try some of these clinical therapies soon. time is not on my side. Thank you for listening"

Luther, parent of a daughter with LGMD2i

"My daughter Jane was one of the presenters. Fortunately, she is in a clinical trial that seems to be working with a drug that is safe. She did not receive the placebo, but we have concerns for patients who continue to deteriorate who have to take a placebo for an extended period of time. If a drug has proven to be safe, it's inhumane to require patients to continue on a placebo and deteriorate when they could benefit by taking the drug."

Lacy, 49-year-old woman living with LGMD2i

"Thank you for taking the time to hear our stories and what daily life is like for all of us with LGMD. I am 49 with LGMD2i/R9. I started showing symptoms at a young age and was diagnosed at age 16 by way of a biopsy. I am married with six kids. My story is similar to the ones that have already been shared. My husband is my caregiver. He is 15 years older than I am and it is a constant fear that something will happen to him. We have an 11-year-old and 15-year-old still living in our home. I have been using a power chair full time for the last 7 years. I have pulmonary and heart failure. I use a Bipap at night and a sip and puff during the day. I take medications trying to stabilize my heart. My husband and I were foster parents for years, but we surrendered our license a year ago after I took a fall and sustained a concussion. This was my 4th concussion. I was transferring to the toilet and when I landed on the toilet seat my upper body didn't have the strength to maintain my body upright-my head went back and hit the wall. I haven't been the same since. Cognitively, emotionally, physically. The disease robs our abilities and we never know when we are going to lose something else. I live in a hyperawareness that contributes to my fatigue. I can no longer care for myself, lift my arms, hug my children when they need comforting, it's affected my marriage both emotionally and physically. My children live in fear not knowing if I'm going to be here to attend their graduations, their weddings, meet my grandchildren, they don't know when they won't have a mom anymore."

Stephen, father of two daughters living with LGMD2i

"We are very concerned with clinical trial designs for ultra-rare diseases such as LGMD2i/LGMDR9 that include long placebo periods. For example, the 3-year placebo period designed into the upcoming BBP-418 Phase 3 clinical trial for LGMD2i. A placebo period longer than a few months, given a natural history study and knowing the characteristics of BBP-418, seems unreasonable and is a concern.

There appears to be no precedent for how a Phase 3 should be designed for an investigational drug for LGMD2i. (Within the neuromuscular disease community, there is precedent of DMD trials without placebo.) The closest prior example would be Pfizer's terminated Phase 2/3 of a myostatin inhibitor which did not require a placebo; and instead, all participants saw their strength decline as expected over a short period of time. While that outcome is disheartening, all participants remained in the trial, and even continued during the dose escalation and extension phase. We stuck with it out of sheer hope.

Based on observations and the Phase 2 data released earlier this year, participants will know if they are on the placebo after a few months.

An anticipated trial design of a 3-year placebo destroys any sense of hope a patient with this ultra-rare disease holds onto as they physically struggle to get out of bed in the morning.

Participation in a clinical trial is always voluntary and a patient can withdraw at any time. Are you going to stay in a trial for 3 years once you realize you are on the placebo? Over the course of 3 years, the body is atrophying with each sip. Participants may drop out so they can be eligible for another trial that is enrolling during that time frame. Those other trials will be a source of hope for patients.

We ask that the FDA reconsider and direct the placebo period in clinical trials such as the upcoming BBP-418 Phase 3 clinical trial for this ultra-rare disease, LGMD2i, be eliminated or reduced to a minimum, months, not years. The long placebo period would appear to be unnecessary for an investigational drug for an ultra-rare disease. Our concern is that within months a patient can become non-ambulatory with no way to reverse the loss.

Sue, 49-year-old woman living with LGMD2i

I am a 49-year-old mother of five children, ages 8-18. I have been diagnosed with LGMD2i. My hope is that some treatment will become available soon, that if not to cure this disease, then to at least stop the progression so that I can maintain the function that I currently have.

Stephanie, woman living with LGMD2i and mother of a son living with LGMD2i

So many ways it affects everyone. I've noticed my lack of abilities has created anxiety in my children. They also have a lot of guilt whenever something they want to do, I can't go, when I have them do it without me. My husband feels greater stress to provide more all the time because of what the future looks like for me and also our son who has the same LGMD2i. I feel at a loss and sad a lot because of all the things I can't help my kids do, or won't be able to do with them like even be the volunteer mom at a class field trip because the school bus stairs are too big and I can't get on them, I never know what to expect and if in the middle of something I will end up letting people down because I'm not able to do things. I have to give a lot of trust to people I do not know in so many ways. The sadness and stress, worry and toll it takes is high and knowing we just have to deal makes it seem like we can never get ahead.

Cyanne, 31-year-old twin living with LGMD2i

My story started back in 1991 when my twin sister Zia and I (Cyanne) were born. We live in South Australia. Just before we were 2 years old, we had a muscle biopsy done to determine why we were behind the average milestones. We weren't walking or crawling as others our age were. We couldn't even hold our heads up while sitting in a baby walker. We struggled a lot; physically. Just after our 2nd birthday, we were diagnosed with Muscular Dystrophy. The doctors weren't sure what particular muscular dystrophy we had, they went for the worst case scenario and told our parents we would never be able to walk or talk, that we wouldn't be able to live a normal life basically.

But hey, I'm stubborn. I did all those things. It might've taken me longer to reach the normal milestones, but I managed to walk, and I can certainly talk. As we were growing up, walking was difficult. We fell over a lot, but my sister and I had this weird twin vibe, where I'd fall over and hurt myself, but my sister would cry, and vice versa. Although I can no longer walk (I am now 30), I live a fairly normal life. My condition has never stopped me from achieving what I want in life. It is a part of me, but it doesn't define me. My goal in life is to be happy and to make a difference, and every day I achieve that goal.

We tried to look further into the diagnosis, we had to drive several hours outside of our home town to see a specialist. But after many horrendous tests, missing out on so much school, we decided it wasn't worth being a pin cushion. The doctors were baffled. They had never seen anything like it.

In mid primary school we started using a walking frame to get around school, it gave us more stability when walking, but as walking became too difficult, we moved onto manual wheelchairs. By the end of primary school, early high-school we relied on our electric wheelchairs all the time. As we become too tired, too quickly. As we used our electric wheelchairs all day, we began to develop scoliosis. My sister, Zia, has it more severely than I. Over the last few years we could no longer reach to the sky. We can feed ourselves and brush our own hair by leaning on the kitchen table. But eventually, even this became too difficult.

We looked into having surgery to fix our curvature of the spine, but decided against it due to the many complications this would bring. The medical world back then wasn't as advanced as it is now.

Through primary school, my sister and I used a hoist to be able to use the toilet. As we had support workers, they have a no lift policy, meaning they had to use a hoist. This meant instead of taking 5 minutes to go to the toilet, it took half an hour. That is a lot for a kid in primary school. Who had to go twice a day. It meant I missed out on a lot of school and friends social time. Even through high school. This also included taking time out to receive physio, go swimming and seeing doctors who would pull us out of class to talk to us.

Around the age of 16, we decided to go back down the road of diagnosis; to find out what type of MD we had. By this point, my sister and I were fully dependent on our wheelchairs. We needed help to transfer to the toilet, to get in and out of bed, to prepare food. Everything that involved strength basically. We continued to decline. As no one with LGMD is the same, my sister and I are very similar in our progression. My sister is just a few milestones weaker than I am, or by a few years so to speak, having to go on a bi-pap machine first, and almost dying because we didn't know that a cough assist machine existed.

Just before our 22nd birthday, we finally found out we had Limb-Girdle Muscular Dystrophy type 2i or R9. So what did that mean for us? Absolutely nothing! It didn't change a thing, but it was nice to just know. You know? To put a name to it. Gave us something we could google and research. It gave us

something we could share with our friends and family. It gave us a better chance at understanding the specifics of what to expect in the future. It didn't change the challenges we would have to face. And it certainly didn't change my determination to live my unique best life.

It is hard living with this condition, having an intellectual mind, but having your body fail you. It makes you feel sad, annoyed, frustrated and even lonely at times. But you can't let that get you down. You have to keep on rolling forward! Set goals, and look at what you can achieve in life. And if there's something you can't achieve, then what can you change to MAKE it achievable?

People say life doesn't really begin until after school. We either have career paths we want to follow or we are unsure on the direction we want to take. I didn't have a direction or a goal in mind. I was lost. Back then I didn't have the funding for support workers like I do now and I was stuck at home. I couldn't follow a career choice as I couldn't relocate to another city. I wasn't seen as a person, wanting to live a normal life. I was seen as a disabled person. Someone who should be at home. I was seen as someone that had no value to add to the world outside!

When school finished, so did our physio and swimming. The only way we could continue our therapy was if we went back to school. Who would want to continue studying down a grade once you've graduated? How is that fair? So we told them to shove it. And moved on. We sought other options. We used Bowen therapy and often saw a chiropractor.

Our mum was working from 9-5pm everyday so we could have food on the table, and a house to live in. She had no choice but to come home from her lunch break to take us to the toilet and get us lunch. Sometimes she got to eat, sometimes she didn't. We asked for more hours. Someone to come in and help us. Saving mum from having to come home in her only hour lunch break. But we were living at home. In their eyes, they saw mum as our carer. So we didn't need the help. Regardless if she was working or not. They didn't care that she had her own life to lead. Mum often got told to quit her job to look after my sister and me. We weren't living. We had very little reason to wake up. We were surviving. Just. It took us years to fight for what we have today. Many frustrated tears and sleepless nights, feeling like we had no hope. No future. If it wasn't for our mum and our positive mindset, I don't know where I would be right now.

We did have some funding for personal care in the mornings. To shower. Everyday. To start our day. But that often didn't get used. Sometimes they didn't turn up, other times we would turn people away at our front door. Would you let someone come in to shower two young girls, when you can visibly see and smell that they couldn't even look after themselves? Sometimes we even had cleaners sent in for personal care. That's not right.

My sister and I were still seen as one person. Not two. We shared hours. We couldn't even get funding to leave the house for an hour to be part of our community. We were stuck at home for many hours during the day. Like a bird in a cage. What teenager wants to stay at home all day? Or hang with their mum when catching up with friends? There were only so many movies to watch, books to read, and pictures to draw on a daily basis. I needed something different. I needed something to do to keep me busy.

Art is something I have always enjoyed, (and coffee), whether it's drawing, painting or getting my hands dirty. It's one passion that I've had my whole life and I don't think that's ever going to change. Art to me is my outlet and my escape from reality, it's something that I crave and is something I can do without the help of others. Unless it's to sharpen a pencil or need more paint. But it's something that I can personally do. I think that's one reason why I love art so much.

But living in a small town, you don't have a wide range of study options. The kind of art I wanted to study wasn't available at our local Tafe or Uni campuses. I physically could not relocate to Adelaide, away from my family, my home. I didn't have the funding, even if I wanted to move. Again something I wanted but couldn't because of my LGMD.

Our care has increased a little bit over the years, but still not nearly enough. Things didn't really change until my mum was 6-8 months pregnant with my little brother. We could see this coming. We tried getting more funding before it got to this point. But it wasn't seen as something we needed. It was a dangerous game we were playing. By the time mum could no longer physically lift my sister and me, she was 7 months pregnant. The Dr. told her no more or you'll hurt yourself and the baby. It wasn't until then that we were given funding for more support hours. Leading up to that, my brother used to kick me from inside mum's belly when she was lifting me. But he wouldn't move much for my sister.

Mum was no longer lifting us. She was on strict instructions to rest until she popped. So our care had increased. Mum was no longer in danger from hurting herself and the baby. And I was no longer in danger of being dropped. It gave mum much needed rest. And it gave me the freedom to do more! To leave the house! It was a whole new life! A lot of people don't realise how much I depend on my support workers or how much they help me live a fulfilling life.

In December 2012, my mum gave birth to a healthy baby boy. Who we call Jayden. Or Bob. Depending on his mood. When mum got her 6 week check up, and got the all clear by the doctors, we got all our hours taken away from us again. Mum was in no shape or form to start lifting us, but having no other choice, that's what she did. In doing so, she injured herself badly, something she will never fully recover from. We ended up getting those hours back eventually, but not easily. We went through the appropriate steps to complain. Again. Then we went higher. Finally someone started to listen. But it wasn't soon enough. What would happen if anything ever happened to our mum? We don't know. We try not to think about it. All I hope for is that I'll die first.

I am now 31 years old and am declining in strength everyday. I have very bad contractures that can not be fixed. I use an electric wheelchair all day, slowly starting to develop pressure sores. I only weigh 25kgs as I struggle to gain weight. I use a cough assist machine when sick. I use a bi-pap machine every night while I sleep, I also use a toilet / shower chair, and a hoist, and I receive nearly 24/7 hour care. Having modifications done to my home to try and live life. My mum is always sacrificing herself to make sure my needs are met, which breaks my heart.

I hope for a cure, or something to stop the progression of muscle weakness but I don't think that will happen in my lifetime. I only hope that it can help others in the future, so they don't have to go through what I have.

You've got to follow your heart and your dreams, even if everyone disagrees with you, because it's your life. Not theirs. And though the journey may not always be easy, nothing beats the feeling of reaching your personal goals. Make goals of your own and start taking steps to achieve them. Goals can change and grow as you do. Advocate for your peers and the people around you. Be inclusive and celebrate differences in each other. Our voices are strong. They are loud. They are powerful. I have overcome so many obstacles in my life, and community. It took bravery, determination, patience and perseverance but in the end I feel like I've made a difference. I HAVE made a difference! And I hope by hearing my story you will be inspired to continue making a difference in your community and leading your best possible life."

Lindsay, 65-year-old man living with LGMD2i

"Hi, thanks for the opportunity to submit some comments. I was unable to listen live to the PFDD meeting as time differences here in Melbourne, Australia made it a bit too difficult. Congratulations to those involved. I am a 65-year-old male, widower, father of a 26-year-old son, and 23-year-old daughter. I was formally diagnosed with LGMD2I/R9 approximately only 11 years ago via muscle biopsy and genetic testing. I am fortunate that I have been mobile enough to continue to live without any 'formal' assistance or care from others, however my legs and core and lower back areas are now becoming much weaker and I am now much slower in all I do. After listening to the many stories shared in the EL-PFDD meeting I wanted to relate my experiences and hope that it may be of some use to all. I have never been a fast runner or able to lift heavy weights etc. But I have always been easily involved in sports – tennis, cricket, Australian Rules Football, swimming, snow skiing, gym work, running etc etc. I played sports from age 5 until my early 50's – still skiing, tennis, running until then. I ran a marathon at age 49. I have always had a strange running style and that's now obviously due to the LGMD. Until 3 years ago I was still able to climb stairs, ladders, get in and out of vehicles, without any need for assistance. Now I must use handrails or walk up ramps rather than stairs. Getting in and out of cars, in particular low to the ground vehicles, is much more difficult. I am still mobile and try to walk every day but anything with slopes or rises is becoming more and more tiring each month. I have had a few falls, luckily not too much damage, though I did have to go to hospital for a concussion test last year. I cannot roll over in bed and am very slow in getting out of bed. I am still able to use the bathroom and shower without assistance although I am very conscious of using other people's facilities as they may be more slippery or have less access. I still travel, both interstate in Australia, but also overseas, having been to North America, Europe, and UK in the last 5 years – obviously missing the Covid years. I seem to be lucky so far with my heart and respiratory being strong, although I have sleep apnea which means I snore a fair bit.

If you would like to have more information about my symptoms, lifestyle etc, rather than typing a thesis here on this platform, please feel free to email me directly. My final hope is that one day Australian patients can be included in formal trials. Surely it isn't that difficult in this this day and age. I am concerned that my children may also suffer from a 'late onset diagnosis' when they are older. I am very keen to be involved. Best regards, Lindsay"

Cindy, mother of a 34-year-old son living with LGMD2i

"Thank you for providing us the opportunity to give voices to you all on what it is like to have to live with LGMD2i/R9. Other LGMD2i/R9 families had given some very informative testimonials on how this disease affects every minute of their lives. I would like to add to this.

My son, Joshua is 34 years old. He was diagnosed at age 12. He has been non ambulatory for the last 5 years. He has been a heart failure patient, due to cardiomyopathy since age 20, due LGMD2i. His lung function is limited as well, due to LGMD2i.

While it is so very sad, devastating and heartbreaking to lose skeletal muscle, one can still exist. Heart failure and limited lung function is a whole set of very scary and dire complications, with little to no recourse from the medical standpoint other than some blood pressure drugs to allow the heart muscle to not have to work so hard, thereby hopefully slowing down progression of muscle cells being turned into non functioning scar tissue/fatty cells.

We need all pharmaceutical companies working on this vastly important aspect of LGMD2i/R9, along with the current trials for treatments for skeletal muscle groups. We need the FDA to review these

options as soon as they become available.

Time is not on our side.

Thank you

Julia and Marcus, parents of three daughters living with LGMD2i

We have 3 daughters aged 21, 18 and 16, who all have LGMD2i. They have all been diagnosed as children while pre-symptomatic due to blood tests during diagnosis of Coeliac Disease (which all 3 also now have). Symptoms have begun in the pre-teen or early teen years (about 11-13 years).

Our eldest:

- *Has deteriorated a lot in the past 3 years while at university. She now describes herself as disabled.*
- *Has had to give up driving as she can no longer lift her legs.*
- *It is a huge effort to climb stairs, hauling herself up using two handrails and a wide stance. We do not have a downstairs bathroom, toilet or bedroom.*
- *She has a pronounced waddling gait and her right knee turns in when she walks.*
- *She uses a manual wheelchair for longer walking outside.*
- *She cannot rise from a chair without using hands and arms. She struggles to get up from the floor or low chairs/sofas, and has to be helped. She can't squat but 'falls' into seated position – so has to sit down on public toilets, even if they are dirty (a fear during Covid).*
- *She regularly falls as her leg gives way under her – she calls it having a 'weak leg.' Her boyfriend says that she goes down like a deckchair!*
- *She struggles to lift her feet to step into showers, up steps or slopes etc.*
- *She has a weak grip and needs help to chop food, hold heavy pans and bags etc.*

Our younger two:

- *They struggle with energy levels and have to regulate their days, or they won't be able to complete everything they would like to.*
- *They can no longer run or participate in PE (PT) lessons.*
- *They struggle with longer walking (over 10/15 mins) and have a slow pace. They use a wheelchair for longer walking.*
- *They use rails to climb stairs. This can be a problem in school, where the stairs get very busy and they may not be able to reach the rails. Our youngest has been shouted at by the head teacher for crossing the stair traffic to reach the rail.*
- *Combing, washing and styling hair can cause arm and shoulder pain. Our middle daughter has very long hair.*
- *They are susceptible to myoglobinurea (rhabdomyolosis) eg. from taking part in physical games with their friends at church youth group.*
- *They have kept their condition hidden from all but a few close friends, but the struggle to hide it can take an emotional toll.*

As parents:

- *We have had to become experts in medical issues, from Coeliac, other food allergies and stammering, to LGMD2i. We have to be organised (we have a shelf of files of all the girls' medical letters) and proactive. This takes a huge amount of time and energy.*
- *Between them, the girls have had about one hospital appointment per month for many years, which has meant time off work for us, and now for them. We have two cars to enable us to take them to the hospital while one parent works.*
- *We feel well-supported by the National Health Service locally and nationally, though at times we have had to educate our own doctor. We also have to explain LGMD to family, friends and schools.*
- *Form-filling for extra support at home, school and University takes many hours. Without being pro-active, the girls would not get this support. We have therefore had to educate ourselves on "the system."*
- *Online patient and family groups on Facebook have been a great source of information and support. We attended the Iowa Dystroglycanopathies conference in August – we only knew about this through Facebook.*
- *We are excited to hear about clinical trials for treatments, and would like to help. While improvements in strength and muscle tone would be a dream outcome, to just halt deterioration would be a great result of any treatment.*

Julie, woman living with LGMD2i

Comment 1:

The grief cycle of LGMD is cruel. Each loss is heavy. We constantly make adjustments only to wonder how long this adjustment will last before the next one comes. I am trying to keep as active as possibly without injury. I have fractured my pelvis twice, nose, shoulder, ankle, and most recently my sacrum. After the sacrum fracture I was not able to regain the ability I have to walk safely inside my home, or shower independently. As hard as these things are I see a future of continued loss where this reality feels like the good old days. I am constantly worrying about making sure I am pushing myself to maintain function but not getting injured.

Comment 2:

I am hearing ambulatory patients talk about alternative treatments like physical therapy, chiropractic, supplements, and I think it shows how traumatizing this disease is. We do not want the cruel reality of what this disease takes from us. There is no treatment and I have personally spent so much money and energy trying anything and everything. I now use a wheelchair full time with a cruel reality that harder days are coming and only so much is within my control.

Pam, 52-year-old woman living with LGMD2i

"I was diagnosed at age 30-22 years ago. While it was relieving to get a diagnosis it also came with the knowledge that there was no cure, and no real treatment. I am now at the point where I can't go out of my house without help. I've had a few leg breaks in the last few years, and while I use a walker around my house, I can't go out of the house on my own. So I'm now in the process of figuring out new mobility options, and how much it will cost. And also how can I majorly adapt my house to fit future issues. But one of the biggest issues is the loss of independence and having to rely on others for simple everyday tasks – especially bathroom needs. Mentally, it is constantly a struggle as my body keeps going through new levels of decline, and I have to go through constant mental shifts as I decline to a new normal. Sometimes I really struggle because I don't see anything in the near future that will help, and I'm getting older and declining constantly. I'm constantly wondering what condition will I be at next year, and how much more isolating will it be. It's becoming harder to visit family and my kids because they live in places where all homes have stairs to enter. I've also learned to live with always being sore or some part of my body hurting."

Richard, man living with LGMD2i

"As possible treatments are studied, how is the data from various natural history studies being utilized to reduce the need for control groups or to reduce the burden on patients especially during phase 3 drug trials?"

Beth, woman living with LGMD2i

"Every aspect of my life is affected by this disease.

I have already lost so much strength and ability and I continue to lose what little mobility and independence I do have left. Since I don't have much left now, I notice the loss greatly.

My husband and children are having to do more and more for me and around the house as I lose the ability to do so.

I am at high risk for falling and a fall can put me down for weeks at a time in pain and I fear breaking a bone which would be devastating.

Fatigue seems to rule my life and is very unpredictable. I can not predict how I will feel from day to day or even hour to hour. Emotionally this brings feelings of complete failure and worthlessness. I cannot commit to anyone to do even small tasks because I just don't know how I will feel to do anything at any given moment.

I'm hoping for a gene therapy trial to begin while I could still qualify. Every day that goes by I fear that a fall would eliminate me from the opportunity."

Rachel, caregiver to individual living with LGMD2i

Comment 1:

> *These day to day 'treatments' that patients try might help to manage symptoms have not proven to do anything significant to stop the progression of this disease. The MD will win in the end. As a caregiver I have felt false hope in some of these 'treatments.' Stretching or physical therapy shouldn't be referred to as treatments because they are just ways to manage the disease. Nothing that I know of has proven to actually treat the disease yet. Despite trying different 'treatments' the disease is moving too fast.*

Comment 2:

> *Stopping or even slowing the progression of the disease is a HUGE hope. Anything beyond that would be AMAZING.*

Jane, caregiver for an individual living with LGMD2i

> *As far as future trials are concerned we urge you to expedite the process for rare disease. Double blind trials where the efficacy and safety if already proven only hurts those you are trying to help. It will also be harder to recruit patients who know they might be taking a placebo. This patient population doesn't have time on their side.*

Cynthia, mother of a 41-year-old woman living with LGMD2i

> *Our daughter was diagnosed in 1997 at age 16. She is now 41 and in a motorized chair. She is a college graduate and is now almost finished with her Masters in psychology. She has a good husband and three wonderful children. We are so proud of her and her accomplishments! Life is getting harder for her and she would Love it if she could just stop her progression! She's not looking for a miracle, just doesn't want to get any worse.*