Kourtney

LGMD "SPOTLIGHT INTERVIEW"

Name: Kourtney **Age:** 23 yrs. old

Country: USA

LGMD Sub-Type: LGMD2B / Miyoshi Myopathy

At what age were you diagnosed:

I was diagnosed at the age of 14.

What were your first symptoms:

The very first symptom I noticed was that my calf muscles began to swell up uncontrollably after a huge dance performance. I couldn't see my ankles after a while and we started to worry. The doctor told us to measure my calves' daily and to keep a log. They kept getting bigger and bigger. The doctors thought it could be because of a blood clot, so they did lots of ultrasounds on my legs, heart, and stomach to see if there was anything wrong. They couldn't find anything wrong. I then got my blood tested and they saw my CK levels were high, so they decided to do a muscle biopsy, and that's how they diagnosed me.

Do you have other family members who have LGMD:

Yes. My older sister, Stephanie Garrett, has the same type as me. She was diagnosed just a few months before I was. She

seems to have more problems in her arms (2B) and I seem to have more problems with my legs (MM).

What do you find to be the greatest challenges in living with LGMD:

I think the greatest challenge living with MD is coping with the idea that you can't do something that you used to be able to do. There are a lot of things that are hard to do physically, such as dance, climb, walk, etc. that I used to be able to do just fine. The hard part is being okay with it. I think MD is more emotionally challenging than physically challenging. But, you just have to stay positive and think of all the amazing things that you still can do.

What is your greatest accomplishment:

This is a hard question because I feel like I have accomplished a lot in my life. I graduated high school, then college, I have two jobs that I absolutely love, I got married to my high school sweetheart, but most importantly, I became a mom. I have always looked forward to becoming a mom ever since I was little. Once I was diagnosed with LGMD, I didn't know if it was possible or not. As I got older and my body started getting weaker, I questioned things. I have had so many fears about what harm I could possibly put on my family because of my physical situation. But, my friends and family have been nothing but supportive, loving, and helpful. My greatest accomplishment is my baby girl. I have found so many ways to make things work so that she is happy, healthy, and safe. I know that the future is going to be hard with kids, but it will be more than worth it.

How has LGMD influenced you into becoming the person you are today:

Even though LGMD has definitely made me weaker physically, it has more importantly made me stronger emotionally and spiritually. I would never wish anybody to have this disease,

however, I do think that it has been a blessing in my life. I am grateful that I have it, that I can teach others about it, and that I can be an influence and example to others.

What do you want the world to know about LGMD:

I want the world to know that we are normal people, that we have feelings, and that there are things you should be careful about doing or saying. Please don't stare, please don't tell me I walk funny, please don't laugh if I fall, and please don't judge me based on the things I can and can't do. Please don't make handicap jokes. Please don't assume I can do something. Instead, please help me, lift me up, and encourage me. Please make me feel loved and important. And please love me the same as if I was physically fine.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

If I could be cured tomorrow, the first thing that I would do is dance. Dancing was my heart, my soul, and my passion, and I miss it more than anything.

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Becky

LGMD "Spotlight Interview"

Name: Becky **Age**: 35 yrs. old

Country: USA

LGMD Sub-Type: LGMD2B

AT WHAT AGE WERE YOU DIAGNOSED:

I was diagnosed in 2009 at the age of 27.

WHAT WERE YOUR FIRST SYMPTOMS:

My first symptoms were difficulty walking up steps, getting up from the ground or getting up from a seat/toilet.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

No other family members have LGMD.

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:

The biggest challenge for me is the mental part of understanding my limitations. Knowing that my life is not the way I had ever imagined it would be. Knowing the things my husband and I had to change in order to cope and deal with this continuously progressing disease.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

I think my greatest accomplishment after being diagnosed with this disease is my marriage. We got married 2 months before I started having all of the tests to determine what was wrong.

In sickness and in health came a lot sooner than we could have ever imagined, but we are stronger today because of it.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:

This disease is life-changing. Despite how dependent I am on my family and friends I am still fairly independent. I drive with hand controls, I am resourceful in the ways that I find to keep doing things for myself, and in a way, even though my muscles are weaker, I am stronger. This didn't happen all at once. It took time to realize that I am a strong person even though my muscles are weak.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

I want the world to know and understand that this disease is so challenging. Sometimes my brain doesn't remember that I can't do something and I will just start to do it only to realize I can't. I want them to know that from the moment someone with LGMD wakes up they have to immediately think about how they are going to tackle the day. The world should know that this disease, while somewhat invisible, is real and is extremely difficult to deal with. That just because someone is out and about with this disease does not mean that they are OK or cured. We still have to live our lives, even if behind our smile we are exhausted or hurting. The world should also know that THERE WILL BE A CURE. There are so many people working on this and the strides made in just the few short years I've been diagnosed have been huge.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:

This list is LONG….but first I would run. My legs haven't moved that way in such a long time.

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Ralph

Name: Ralph **Age**: 24 yrs. old

Country: South Africa

LGMD Sub-Type: LGMD2B

At what age were you diagnosed:

I was diagnosed at the age of 23 – just after finishing my Rescue Diving Course.

What were your first symptoms:

I live a very active, adrenaline pumping lifestyle and after I noticed I was running slower and slower every day, I started training harder and harder as well. But in the end my body just kept getting weaker and weaker. After completing my diving I decided to get some help.

Do you have other family members who have LGMD:

No, I am the only one and my family took quite a hit when they

heard my diagnosis. Didn't affect me really. Just saw it as another challenge. But after a while, reality really sank in…if you know what I mean.

What do you find to be the greatest challenges in living with LGMD:

Acceptance. The fact that I can't do all the activities that I used to do is quite a challenge. Hard to give up skydiving, white water rafting, and off-roading because it took too much strain on my arms and legs.

What is your greatest accomplishment:

Living the life most people dream of and being an inspiration to everyone around me. When you have the right mindset and a strong will there is nothing you can't accomplish.

There's a story that inspires me to live a full life. Publius Horatius Cocles was an officer in the army of the ancient Roman Republic who famously defended the Pons Sublicius during the war between Rome and Clusium. The poem goes: Then out spoken brave Horatius, the Captain of the gate: "To every man upon this earth. Death cometh soon or late. And how can man die better than facing fearful odds."

How has LGMD influenced you into becoming the person you are today:

I always believed that I was unbreakable. Since my diagnosis, I've realized that I am unstoppable. There's only life and death and by choosing life, my dreams are only a short breath away. And I intend to close that gap.

What do you want the world to know about LGMD:

Never look down on people who suffer from any form of a disease. The mindset you need to live with and overcome such an obstacle might just be too much for you to handle.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

I would start running and never stop until my legs break because for the moment…I'll be free!

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Adriana

LGMD "Spotlight Interview"

Name: Adriana **Age:** 48 years old

Country: Argentina

LGMD Sub-Type: LGMD2B

WHAT AGE WERE YOU DIAGNOSED:

I was diagnosed at the age 20.

WHAT WERE YOUR FIRST SYMPTOMS:

My first symptoms included: difficulty climbing stairs,

tiredness, and some falls.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

Yes, my brother also has LGMD.

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:

In the beginning the greatest challenge was the process of accepting the disease. Given that I am a very independent person with a very active life, today my biggest challenge is making my desires compatible with what is possible, and finding strategies that permit me to reach my goals, understand my limits, and find solutions.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

Being able to overcome the obstacles I was facing, being a mother, being surrounded by my loved ones, and working every day at something I like. Working in an NGO for neuromuscular diseases, ADM Argentina, of which I was its president in 2011 and 2016.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:

I had to redefine my projections for my future and in what fields I wanted to work, that was a big challenge. But it was possible, I was able to grow as a person, to live my daily life with more interest, to value what life has offered me, and to try to help other who are going through similar situations.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

I would like the whole world to know about the existence of LGMD, its possible treatments, above all fight for early detection to improve our quality of life. Support diagnosis and research.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:

I would go skating again.

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Melanie

LGMD "Spotlight Interview"

NAME: Melanie **AGE**: 33

COUNTRY: USA
LGMD Sub-Type: LGMD2b (Dysferlinopathy/miyoshi myopathy)

AT WHAT AGE WERE YOU DIAGNOSED:
I was diagnosed one week before my 29th birthday.

WHAT WERE YOU FIRST SYMPTOMS:
I could not rise up on my toes, I had hammer toes (foot deformity) and was in pain and fatigued all the time.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:
Nope, not one!

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGE TO LIVING WITH LGMD:
Losing the ability to do everyday tasks and activates has made life quite challenging. You have to plan ahead for every little thing. It also makes it difficult when people do not understanding this fact. When you do not look sick, people do not take your needs seriously.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
Getting my Master's Degree and securing a great job in the Federal Government (while being sick every day).

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
After I was first diagnosed I felt helpless and alone. I knew of not one other person with this form of MD and I was scared. Instead of sulking or letting this get me down, I thought "why not go out and do something about it?!" So I started looking into the MDA and how to fundraise. I started an MDA Muscle Walk Team and have has one every year since. Then I decided that I will do everything I can while I still can (since I am losing the ability to walk). And so I did. I have traveled all over and to some places alone. I have even gone sky diving. I believe that this diagnosis has made me an even more positive person. It has certainly made me an even stronger person. Many of the experience that I have had in the last 4 years, are a directly result of me sticking it to the LGMD and proving the odds wrong.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:
First, it does not define us. We are who we are, and we only happen to have this awful disease. Second, just because we look okay, does not mean we feel okay. Third, muscular dystrophy is not MS.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD DO.
Dance like I used to! Then hike in all the National Parks and take lots of amazing photographs. Then run a marathon or do an obstacle race.

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Emily

03/28/2016

LGMD “SPOTLIGHT INTERVIEW”

Name: Emily **Age**: 29 yrs. old

Country: United Kingdom

LGMD Sub-Type: LGMD2B

At what age were you diagnosed:

I received confirmation of my LGMD2B diagnosis at the age 28 but at the age of 24 it was already suspected.

What were your first symptoms:

My first symptom was struggling to climb stairs and I also noticed that I couldn’t stand on tip toes to reach books on my bookshelf

Do you have other family members who have LGMD:

No, it’s just me – all by myself!

What do you find to be the greatest challenges in living with LGMD:

There are many challenges. Like trying to be accepted as “disabled” when I don’t use a wheelchair. I find people don’t want to talk to me about it in general, which can be very isolating! At times I get self-conscious and anxious. Falling over in front of people is the worse and struggling to do things I once could is quite sad. I miss being able to run! Also the need to try and explain yourself. Having the odd wobble and feeling pathetic or weak.

What is your greatest accomplishment:

At the moment, leaving home and living independently in the countryside. I also volunteer for Muscular Dystrophy UK as a peer supporter, which to me is an amazing accomplishment.

How has LGMD influenced you into becoming the person you are today:

Having LGMD has definitely given me a different outlook! I’ve learned to appreciate smaller things in life and become more determined to achieve things on any scale. I feel very positive and want to turn my diagnosis around to focus on the good things not the bad. It’s made me more aware and empathetic, I want to help others like myself. It’s aided my ‘wicked’ sense of humor too!

What do you want the world to know about LGMD:

Everything! I just want people to understand the complexities of these conditions and the condition itself.

If your LGMD could be “cured” tomorrow, what would be the first thing that you would want to do:

This is a tough question, there are so many!

I'd probably want to run first but also get on a sporty motorbike and learn to ride!

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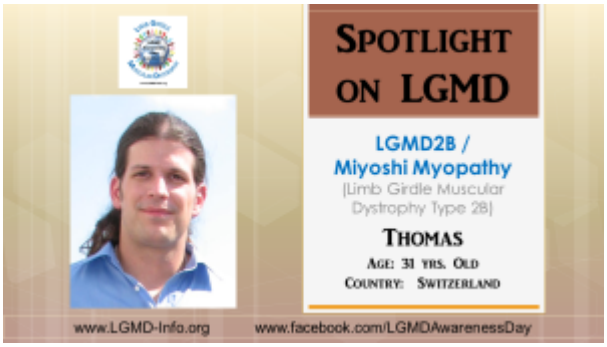
Thomas

01/30/2016

LGMD "Spotlight interview"

Name: Thomas **AGE**: 31



COUNTRY: Switzerland

LGMD Sub-Type: LGMD 2B / Miyoshi Myopathy

At what age were you diagnosed:

My first diagnosis (biopsy) was at 28, followed by the proof with genetic test one year later.

What were your first symptoms:

That is not so easy to say. Now I know where I have to look at, so many examples out of my memories came up; in my childhood we had a steeply rising stair around our home, where I always had difficulties to climb because the steps are very short. In teen age I started snowboarding, there I was not able to give much pressure to the front edge of the board. As young adult I could not jump to the music in discos. At age 24 / 25 I realized that I was not able to stand on my toes and my calve-muscles are actually inexistent. In the last few years I saw that my leg and arm muscles are shortening.

At 18 my doctor told me that I have too high creatine kinase values in my blood. It was around 250 – 300 somethings, too low for well-known diseases and too high to be normal. Now I know that this was the first medical indicator to Miyoshi.

Do you have other family members who have LGMD:

Yes, my sister has LGMD too. Her symptoms (stand on toes, creatine kinase) are the same, but she seems to be less affected.

What do you find to be the greatest challenges in living with LGMD:

In my opinion, the greatest challenge is to accept this challenge! At the first time after diagnosis, it was hard to accept. You need psychological strength to deal with that. Also when you look at your course of disease, not to overrate or underrate the symptoms and effects. By looking into the future, you always have to plan things with LGMD in mind (maybe you need a wheel chair in 5, 7, or 10 years).

What is your greatest accomplishment:

To accept LGMD and enjoy doing things I won’t be able to do in future.

How has LGMD influenced you into becoming the person you are today:

My diagnosis came just at the beginning of my Master study. Beside my Bachelor and Master study (fulltime study) I was working at weekends and school holidays. Now I work part time and have a good reason to enjoy my leisure time with my wife, our dogs and our garden. I value more what I have now.

What do you want the world to know about LGMD:

That it exists. I think there are many unrecorded cases of LGMD.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

I have just lost my driver license for trucks and coaches because of Miyoshi. It was not only a job to finance my studies, it was also my hobby. So I would get it back and drive around.

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Janet

LGMD "SPOTLIGHT INTERVIEW" – 01/07/2016

NAME: Janet **AGE**: 33 yrs. old

COUNTRY: United States

LGMD Sub-Type: LGMD2B

AT WHAT AGE WERE YOU DIAGNOSED:

I was originally misdiagnosed with Polymyositis at the age of 17. Years later at the age of 28, I was diagnosed with LGMD2B.

WHAT WERE YOUR FIRST SYMPTOMS:

I started having difficulty climbing stairs, running, dancing and unable to play soccer as well as any other sports.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

There is no family history of LGMD until me.

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:

Losing my independence has been very difficult. I am no longer able to walk long distances. I now need a power chair to move around. I am unable to independently care for my two children, 2 years old and 3 months old. I need someone to assist me in taking care of them.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

My greatest accomplishment has been accepting my disease. I

mentally prepared myself for my transition to a power chair, where now things can be a little easier when I'm outside my home.

HOW HAS LGMD INFTUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:

I have become a much stronger person. Despite the obstacles that come along with my condition, life goes on and I have and will make the best of it for myself and for my family.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

It's a rare disease, but it is out there and it's important to raise awareness that can help find treatments and a cure.

IF YOUR LGMD COULD BE "CURED',TOMORROW WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:

I would run towards my kids and lift them up into the air!

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Molly

11/30/2015

NAME: Molly **AGE:** 29 years old

COUNTRY: United States

LGMD Sub-Type: LGMD2B: dysferlinopathy/Miyoshi Myopathy

At what age were you diagnosed:

I started presenting symptoms when I was 17 and at the time I was told I was suffering from Guillain-Barre. When I was 19, my initial symptoms returned and worsened, however, this time I was diagnosed with polymyositis. After 3 years in and out of the hospital I was finally properly diagnosed via DNA testing with Miyoshi Myopathy and the age of 22.

What were your first symptoms:

My first symptoms were difficultly walking up stairs and getting out of chairs. I noticed a significant decrease in athletic activity at around age 15 but chalked it up to teenage laziness. It wasn't until I was a junior in High School and could not complete my P.E. requirement and started falling frequently that I knew there was a problem.

Do you have other family members who have LGMD:

I have no other family members who are affected by LGMD.

What do you find to be the greatest challenges in living with LGMD:

Some of the greatest challenges are watching friends and

family participate in physical activities such as running, hiking, daily exercise, and even dancing. It makes me sad that I cannot participate with them and sometimes I feel like I am missing out on quality time. Another perhaps harder struggle is the daily struggle, it takes much more effort to get ready each morning and also to accomplish daily tasks that many take for granted such as picking up your room, cleaning your house, laundry etc. I have had to learn to set my pride aside and rely on friends and family more, which in and of itself is a challenge.

What is your greatest accomplishment:

I think that my greatest accomplishment to date is that in-spite of being in and out of the hospital during my college years, I kept working and finally received a degree. Now, I put that degree to use everyday as a teacher. Teaching was always my dream and I achieved it. My students and colleagues are more that supportive and it makes it worth it every single day.

How has LGMD influenced you into becoming the person you are today:

LGMD has made me a more purposeful person. Purposeful with my own life, but also purposeful with who I surround myself with. When you live with a condition that affects your everyday life it is important to surround yourself with people and also activities that have meaning and fulfill your life, and not to waste time on anything petty and insubstantial. I am not sure that I would have realized this as early as I did if not for what I experience every day.

What do you want the world to know about LGMD:

Well, first of all I want everyone to stop thinking I have some strange form of MS. I also want everyone to know that we as a community are valuable and have a lot to offer. Our bodies may not cooperate with us, but we are a very bright and

passionate community. Also, I would just like the world to know that LGMD exists, but I think we are making great strides in that area.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

There is so much that I would like to do and obviously most of them have to do with physical abilities that I don't possess. I would love to hike the Appalachian Trail, or anything else that involves being outside for extended periods of time. O, and I would also love to jump again, there is something freeing about that.

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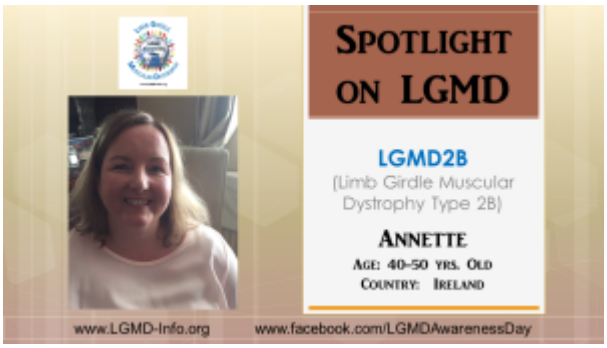
Annette

10/16/2016

NAME: Annette **AGE:** 40-50 yrs old



COUNTRY: Ireland

LGMD Sub-Type: LGMD2B

At what age were you diagnosed:

I was diagnosed at the age of 38 yrs. old

What were your first symptoms:

I was stumbling and falling. I also had problems rising from a seated position and climbing stairs.

Do you have other family members who have LGMD:

No, I am the only person in my family with this diagnosis.

What do you find to be the greatest challenges in living with LGMD:

Lack of awareness is prevalent. In addition, many facilities in Dublin (transport, restaurants, some theaters) are not anywhere near wheelchair accessible.

LGMD has impacted my professional life as I am no longer able to work full time and there have been financial consequences of this.

What is your greatest accomplishment:

I try to remain positive and optimistic. I am fortunate that I am able to maintain an almost independent life. I am delighted to be taking part in a clinical outcome study in Newcastle, UK (the people there are wonderful).

How has LGMD influenced you into becoming the person you are today:

I appreciate what I can do and try not to get downhearted regarding the things that are no longer possible.

What do you want the world to know about LGMD:

I am blessed with a wonderful family and core group of friends who never let me down.

LGMD is a challenge but with a good support system in place, life can be good. Greater funding allocations are imperative in order for LGMD to be treated and not be a life altering condition.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

I would play with my beloved niece Emily, bring her to the park and do all the things a good auntie should do.

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Brad

09/30/2015:

Name: Brad **Age**: 51 yrs. old

Country: United States

LGMD Sub-Type: LGMD2B/Miyoshi Myopathy

At what age were you diagnosed:

I first started having symptoms at 18. But it wasn't until about 10 years later that a neurologist told me I had Miyoshi. Then I had to wait another 10 years for the gene to be discovered before there was a way to confirm that that was what I have.

What were your first symptoms:

I couldn't stand on my toes, initially just on one side.

Do you have other family members who have LGMD:

No

What do you find to be the greatest challenges in living with LGMD:

That there is a lot more planning involved in scheduling any activity. A lot of everyday activities (travelling, shopping, going to a restaurant) involve a lot more things that have to be paid attention to.

What is your greatest accomplishment:

In 2000 I put together a website about dysferlin (the gene for my LGMD subtype). I didn't know anything about web design, but I wanted other patients to have a source of information about the disease and what research was going on. Through that, I was contacted by the Jain family after one of their children was diagnosed—they later started the Jain Foundation, which supports research on my subtype. In 2007 the Jain Foundation organized first research conference on dysferlin. I kept pinching myself the entire time thinking that this couldn't be real—there was actually a conference dedicated to this disease!

How has LGMD influenced you into becoming the person you are today:

I think it has made me more considerate and aware of others. I was always very independent when I was young, but needing help with some things, and also wanting to work with others to find cures for LGMDs, has taught me the importance of asking for help when you need it, offering help when you can, and working together.

What do you want the world to know about LGMD:

First, that it exists. I think a lot of people have the idea that MD only affects young boys, or at least children. I did before I started having symptoms. I want them to know that there are many different types of MD, and that symptoms can start at any age.

Secondly, I think society tends to view disability as a black and white thing. That is, one is either disabled and may need help, or they are completely fine and don't. But for a condition like LGMD where symptoms often progress slowly, when exactly does one become disabled? I think the way things are set up often discourages people who are able and who want to work from doing so, and from doing all they can with their lives

I want other patients to know that they aren't alone. When I was in grad school, I was extremely worried about my MD (which wasn't really diagnosed), but I didn't feel like I could share that with anyone. Many years later, I met another patient who also has LGMD, who was in grad school at the same university at the same time as me! Even though LGMD is a rare disease, there are other people out there and it's good to connect with them.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

Either go ice skating or skiing. Before I started having symptoms I was a competitive figure skater, and it would be really nice to be able to do that again. I also did a lot of

skiing when I was younger, and loved it.

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Katsuya

09/28/2015:

NAME: Katsuya AGE: 45 yrs. old

COUNTRY: Japan

LGMD Sub-Type: LGMD2B / MMD (Dysferlinopathy)

At what age were you diagnosed:

I was diagnosed at the age of 24.

What were your first symptoms:

I was not able to jump.

Do you have other family members who have LGMD:

Yes

What do you find to be the greatest challenges in living with LGMD:

It is to support a researcher.

What is your greatest accomplishment:

I have established PADJ (Patients Association for Dysferlinopathy Japan) which was established for the communication between Japanese and international patients with dysferlinopathy (Miyoshi myopathy, Miyoshi muscular dystrophy 1 (MMD1) and limb-girdle muscular dystrophy type 2B (LGMB2B), development of early treatment for dysferlinopathy aimed at a complete cure.

How has LGMD influenced you into becoming the person you are today:

I had to change a dream. It was a very difficult problem. However, I was able to meet the friend who was in the world.

What do you want the world to know about LGMD:

The dysferlinopathy (MMD/LSMD2B/ DACM)) is a rare disease. I do not know the exact number of dysferlinopathy patients in Japan.

It is a form of LGMD. Let's do our best together.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

I want to celebrate with family and friends.

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Gitika & Gopika

09/17/2015

NAME: Gitika & Gopika AGE: 38 yrs old

COUNTRY: India

LGMD Sub-Type (if known): LGMD2B

At what age were you diagnosed:

We were diagnosed at the age of 21 yrs.

What were your first symptoms:

Our first symptoms included difficulty standing on our toes and needing to hold onto a handrails after ascending 2 floors.

Do you have other family members who have LGMD:

We are a set of identical twins suffering from Limb Girdle Muscular Dystrophy. There is no family history within the past 5 generations.

What do you find to be the greatest challenges in living with LGMD:

Our greatest challenge is to overcome our gears, growing with this progressive disease and remaining positive. Gitika had

to quit he successful career and become a homemaker. Gopika still continues running her tutorials at and education center. She use to teach senior classes in a reputed school earlier.

What is your greatest accomplishment:

Our greatest accomplishment has been to come to terms with living this disease and working around our daily chores with continued enthusiasm – in spite of several of several let downs.

How has LGMD influenced you into becoming the person you are today:

We both have become more tolerant of people and situations, as we have to let go of our inhibitions and ask for support several times.

What do you want the world to know about LGMD:

Often, in the absence of any external visual feedback, it becomes difficult for people to comprehend the level of disability (unless using a power wheelchair) with this disease. We want the world to be equally passionate about LGMD patients and provide some benefits as given to other disease sufferers.

If your LGMD could be "cured" tomorrow, what would be the first thing that you would want to do:

Gitika would like to resume her career and go on a beach destination.

Gopika wishes to run on a treadmill and resume exercising at the gym.

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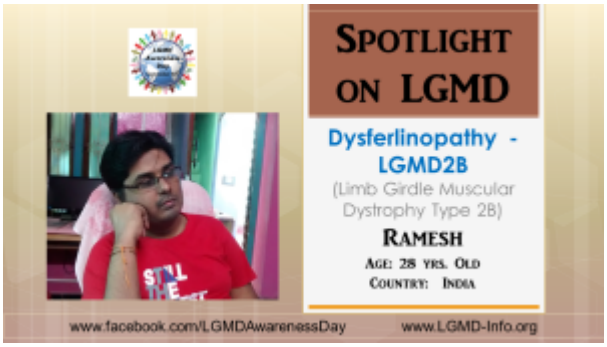
Ramesh

07/06/2015:

NAME: Ramesh **AGE:** 28 yrs. old
COUNTRY: India
LGMD Sub-Type: LGMD2B / Dysferlinopathy



AT WHAT AGE WERE YOU DIAGNOSED:
I started having symptoms at the age of 19 but was not diagnosed until I was about 22 years old.

WHAT WERE YOUR FIRST SYMPTOMS:
My first symptom was, when I was going to office suddenly I fell down. I consulted my doctor, he suggested physical therapy but that did not help much. After that, we found it might be best to consult the doctor in Bangalore (India). I had muscle & nerve biopsy and was in a cast for two months on each leg, one at a time. She sent me to a neurologist for further testing.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD: No

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:
I find that the greatest challenge in living with LGMD is that people have a hard time understanding it. It is not easy to describe LGMD to someone. Many people think that I am exaggerating or even lying about having this disease. In addition to the condition, I find the lack of accessibility and social attitudes toward disabled people as very limiting.

I wish people would be more open to learning about these diseases and understanding how get rid of this disease.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
My greatest accomplishment is, managing to work and keep working in my own way.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
I've learned to appreciate people helping me, to have patience, to keep being hopeful and avoid feeling depressed, to be thankful for everything, to understand people who aren't in the same situation as me and who are in a difficult situation, to value what I have and to be a saver, to give more love to my family each day as if it were the last.. I have learned to be patient and accept the fact that everyone needs a helping hand!

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:
Even most medical professionals are unaware of this condition they have to aware of this and teach the patients how to be safe and challenging towards life.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:
First of all I would like to congratulate each and every doctor who helped me, in person. The main thing is, I would like to open one blog about this disease and I will guide and support the patients how to overcome through the disease. I know how I am suffering with this disease. I will not give a chance to others suffer again.

Julianna

03/01/2015:

NAME: Julianna AGE: 24
COUNTRY: United States
LGMD Sub-Type: LGMD2B

AT WHAT AGE WERE YOU DIAGNOSED:
I started having symptoms at the age of 16 but was not diagnosed until I was about 22 years old.

WHAT WERE YOUR FIRST SYMPTOMS:
My first symptom was tight tendons. My doctor sent me to physical therapy but that did not help much. After that, we found it might be best to have my Achilles tendons lengthened. I had surgery and was in a cast for two months on each leg, one at a time. While recovering and beginning in physical therapy again, my physical therapist noticed that I was not gaining any of my strength back. He sent me to a neurologist for further testing.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD: no

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:
I find that the greatest challenge in living with LGMD is that people have a hard time understanding it. It is not easy to describe LGMD to someone. Many people think that I am exaggerating or even lying about having this disease. I am still mobile therefore they do not think I have any challenges. I wish people would be more open to learning about these diseases and understanding how difficult it is.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
My greatest accomplishment was graduating from college. I was juggling school work with medical testing's that I was having done. It was difficult to keep up with at times but I managed to pull through and become involved in school organizations. I joined Gamma Sigma Sigma(a service sorority) and I was events council chair of the United Students Government. I had the opportunity to plan events such as a prestigious speaker and a concert. Limb Girdle did not stop me from going after what I truly wanted. Walking across that stage with that diploma in hand was a feeling that I will never forget.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
LGMD has taught me to be more understanding of others and their stories. Everyone has a story we do not know. It has also taught me a lot about myself. I have learned what I can accomplish despite minor setbacks. A sense of humor is definitely a must in getting through the hard times. I have learned to be patient and accept the fact that everyone needs a helping hand!

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:
I want people to know that just because you cannot see it, doesn't mean it is not there! Try to be patient and understanding of people who explain this disease to you. It is just as hard for us to explain as it is for you to understand.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:
The first thing I would want to do is travel without all the difficulties involved. I would want to go to the beach and swim without needing help. It would be nice to be able to go where I want without having to plan every little detail ahead of time; just not worry and have a good time!

Suzanne

02/18/2015:

NAME: Suzanne

AGE: 45

COUNTRY: England

LGMD Sub-Type: LGMD2B – Miyoshi Variant

AT WHAT AGE WERE YOU DIAGNOSED:
I was correctly diagnosed at around 29 yrs. old.

WHAT WERE YOUR FIRST SYMPTOMS:
I couldn’t stand on tip-toes.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD: No

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:
The greatest challenge is finding the energy to do everything I want to do. I find that something like even getting out of a chair is such hard work.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
My greatest accomplishment is getting married and having two wonderful children. I have also learned to sail a boat with Sailability and have been ice skating in my wheelchair!

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
It has helped me to realize what is really important in life such as family and friends and appreciate what I have as there are always people worse off than you.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD: There are currently no treatments or cure for LGMD 2B Miyoshi. The Jain Foundation is currently helping with clinical studies and research to find out more about the disease and try to find treatments or a cure!

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:
I would give my kids a "proper full-on" hug, run down the road and take dance lessons.

Lydia

02/12/2014:

NAME: Lydia

Age: 45

COUNTRY: United States

LGMD SUB-TYPE: LGMD2B / Miyoshi

AT WHAT AGE WERE YOU DIAGNOSED: 41

WHAT WERE YOUR FIRST SYMPTOMS:
My first symptoms included: knees popping out of place, leg, foot, chest, neck, and arm pain, falling, lethargy, strange duck-waddle gait.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:
No

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:
The progressive nature of the condition is frightening. The lethargy LGMD causes is debilitating as well. In addition to the condition, I find the lack of accessibility and social attitudes toward disabled people as very limiting.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
This is a hard one. Managing to work, care for small children and a mother with dementia and my elderly father (along with the assistance of my siblings), and earning a Bachelor's of Social Work. Looking back, I am amazed that I was able to do it all.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
It has increased my empathy toward others and has been the impetus for my development of a disability consciousness.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:
Firstly, that they know about it at all. Even most medical professionals are unaware of this condition.

Secondly, it is progressive and debilitating.

Thirdly, I feel fairly safe in saying that most of us who have it would prefer otherwise, which is why research and public awareness are so vital.

Fourthly, and this is an example, recently in one of our

online groups, people with my condition were discussing the five things that they would like to do if cured. Things such as holding their children and giving loved-ones hugs. This is the pernicious reality of LGMD.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:
I would love to be able to go on a hike in a wooded area with my family.

José Antonio

02/06/2015:

NAME: José Antonio

AGE: 48

COUNTRY: México

LGMD Sub-Type: LGMD 2B/Dysferlinopathy

At what age were you diagnosed: at age 46

WHAT WERE YOUR FIRST SYMPTOMS:
I first noticed muscle weakness at age 30. I was found to have a heart problem in my aorta, and the cardiologist thought my muscle weakness was due to that. I had an EMG and the results came back fine, but I kept falling, bumping into things, not

having strength in my legs to climb stairs, or in my arms to lift heavy objects. I think the most alarming symptom was when I discovered that I couldn’t jump, then I knew something was wrong.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD: No
WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:
Everything! Once people help me to get out of bed I have to get into a special chair to use the bathroom, then people have to lift me so I can get into a wheelchair. I have physiotherapy from Monday to Friday. The physiotherapist lifts me using a belt and I walk with him about 10 meters several times. He has me flex my arms with 1 kg weights, then they lift me into my vehicle and they take me to my workplace where I stay in my vehicle. The saddest part is that I watch my children grow up, and I can´t go with them to enjoy simple things: beach, movies, parks, etc. My wife is the one who does this; she is very devoted to me and helps me a lot.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
Being able to stay in contact with my customers and to keep working in my own way.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
I’ve learned to appreciate people helping me, to have patience, to keep being hopeful and avoid feeling depressed, to be thankful for everything, to understand people who aren’t in the same situation as me and who are in a difficult situation, to value what I have and to be a saver, to give more love to my family each day as if it were the last.
WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:
How difficult it is to accomplish small things that are insignificant for other people, for them to know about these rare diseases, and for there to be more interest in studying why they occur and how they can be avoided. Also early diagnosis so patients can take vitamins to avoid health

problems, because when we become less mobile our bodies become weaker and our immune system weakens. Finally, and most importantly, to find a treatment or cure for this disease!

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:

I would walk on the beach, run, climb stairs, travel places with my children without them having to worry about me. Take my wife out to a beautiful place to eat and thank her for all the love she has for me by being able to help me and care for me. I would hug my children to thank them for all the support they give me every day.