

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.

* * * To read more "LGMD Spotlight Interviews" or to volunteer to be featured in an upcoming interview, please visit our website at: <http://lgmd-info.org/spotlight-interviews>

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.

* * * To read more “LGMD Spotlight Interviews” or to volunteer to be featured in an upcoming interview, please visit our website at: <http://lgmd-info.org/spotlight-interviews>

Lori

LGMD “SPOTLIGHT INTERVIEW”

Name: Lori **Age:** 47 yrs. old



Country: United States

LGMD Sub-Type: LGMD2i

At what age were you diagnosed:

I was diagnosed at the age of 41.

What were your first symptoms:

The first symptoms that I noticed were muscle weakness and falling although at the time I didn't realize that they were anything serious.

Do you have other family members who have LGMD:

Yes, my younger sister also has LGMD2i.

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

LGMD:

My greatest challenge is lacking the energy and physical strength to do everything I used to do and everything I want to do.

What is your greatest accomplishment:

My greatest accomplishments are raising my two sons (ages 17 and 24) and becoming an attorney.

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

Having LGMD causes me to be more grateful for what I can do and for the people in my life.

What do you want the world to know about LGMD:

I would like people to know that LGMD is a genetic disease for which there is no treatment or cure at this time.

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

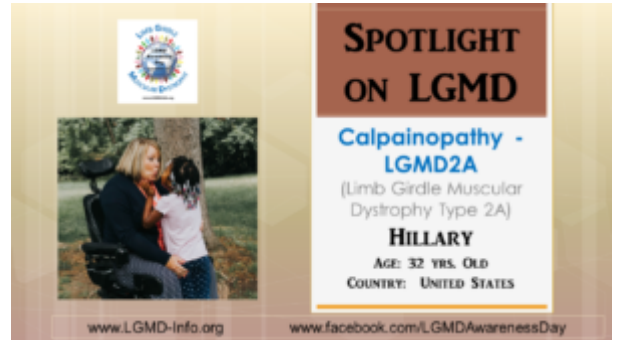
If I was cured tomorrow, I would like to go hiking or for a long walk.

* * * To read more “LGMD Spotlight Interviews” or to volunteer to be featured in an upcoming interview, please visit our website at: <http://lgmd-info.org/spotlight-interviews>

Hillary

LGMD "SPOTLIGHT INTERVIEW"

NAME: Hillary **AGE:** 32 yrs. old



COUNTRY: USA

LGMD Sub-Type: LGMD2A / Calpainopathy

At what age were you diagnosed:

I was diagnosed at the age of 13.

What were your first symptoms:

My first symptom were Tight Achilles tendons, high arches, and a slow runner. I started running just like my older brother, who was diagnosed with LGMD2A at the same time I was.

Do you have other family members who have LGMD:

Yes, my older brother (5 years older) also has LGMD2A.

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

The hardest thing about LGMD2A is that it is constantly changing. Over the past 20 years, I have figured out creative ways to accomplish daily tasks. Tasks as easy as going to the restroom independently have needed to be adjusted and

recreated many times. At one point I would use my forearms, forehead, and grab bars to literally climb up the wall from a seated position on the toilet. Because the disease is constantly changing, my workarounds and "solutions" only work for a few years and then I am back to the drawing board to figure out something new.

What is your greatest accomplishment:

My family is my greatest accomplishment. I got married at age 23 when I was still a very able-bodied individual. In the span of 9 short years, I am now a full-time wheelchair user. My husband and I have had the privilege to adopt two beautiful children. My husband and two kids are my everything. Life isn't always easy, but with these 3 by my side (or on my lap) I am reminded how beautiful life really is. When I was first diagnosed I always worried how I would chase after my kids or save them from running into the street. I never pictured myself in a wheelchair chasing them! Did you know that wheelchairs can cruise up to 7 miles per hour?! Turns out I had nothing to worry about. I am able to work part-time from home and balance my responsibilities as a wife and mother.

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

When I was 13 yrs. old I was playing basketball, volleyball, and softball. Sports were my everything. As one can imagine, the diagnosis was a total shock and I was in denial for a while. During tryouts for the JV volleyball team, I was completely shocked that my coaches didn't think I could keep up. They decided to let me play, but at the end of the season, I had accepted the fact that I wasn't as fast as my teammates. This came at quite the emotional blow. Luckily, with the influence of my parents and friends, I decided to try new things and I ran for student government and was an editor on the yearbook staff. Keeping busy helped and I found new things that I loved. The experiences and opportunities I had helped

me get a scholarship for college and lead to more leadership opportunities on my college campus. Additionally, I have had the opportunity to work in the hotel industry for the past 14 years and now have a position that allows me to work from home. Staying busy has always been one of the best remedies for my mental health, and to be honest I don't remember a time I wasn't busy!

What do you want the world to know about LGMD:

Raising awareness for rare diseases is never an easy task. When you see the firefighters out Filling the Boot, or see the Shamrocks out in local businesses, please keep donating. That money helps further research and it helps send kids to MDA camp. I have hope that the research being performed today will change lives.

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

The first thing I would do is chase after my kids, throw them up in the air, and play until I was dizzy!

* * * To read more "LGMD Spotlight Interviews" or to volunteer to be featured in an upcoming interview, please visit our website at <http://lgmd-info.org/spotlight-interviews>

Monkol

LGMD "SPOTLIGHT INTERVIEW"

Name: Monkol **AGE:** 38



Country: United States

LGMD Sub-Type: LGMD2G (also known as Telethoninopathy)

At what age were you diagnosed:

I received an official genetic diagnosis at 33, where they identified frameshift mutation in TCAP. I have since created the LGMD2G Foundation to find more LGMD2G patients and help them on their journey.

<http://www.lgmd2g.org/>

What were your first symptoms:

I started really taking notice when I was 19. I was having a difficult time standing up for long periods on public transport and going up stairs. I would be really sore and exhausted at the end of a long day of traveling to University, which took nearly 3 hours total each day.

Do you have other family members who have LGMD:

Yes, my second oldest sister. There are 7 siblings in our family, the two affected is roughly the 25% expected to be affected by a recessive disease. Guess my sister and I got a shitty roll of the dice!

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

At this stage of my life, it would be maintaining friendships. It's easy for me to make friends but I can't maintain them as I can't go to bars all night, I can't hike, ride a bike and do other physical things and lastly, it's difficult for me to go from A to B unassisted. All my social outings have to be very well planned to the detail and sometimes takes the fun out of things. People realize I'm too much of a bother and just don't invite me to things anymore and it hurts to be left out. It hurts my wife even more as she isn't handicap and she gets lumped in with me.

What is your greatest accomplishment:

Starting my own research lab at Yale School of Medicine as an Assistant Professor of Genetics. <http://www.leklab.org/>

[Lek Lab](#)

It has been a long and difficult 13-year journey but I got there! One of the aims of the lab is to improve the diagnosis rate of people with muscular dystrophy, particularly limb girdle muscular dystrophy. I'm really looking forward to working with LGMD patients directly that are still looking for answers as I also know how it's like to go on a decade-long diagnostic odyssey and the frustration of here we go again on each clinical visit!

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

It's given me purpose and a desire to live for others and not just for myself. I don't think I would have achieved as much in my life, if I didn't have the struggles of LGMD and being driven by the hope that one day, no one should need to struggle with this disease again. I want to be part of that effort and be there that day this becomes true!

What do you want the world to know about LGMD:

People with LGMD are beaten down every day both physically and mentally. We get beaten down each day by our family and friends that don't understand why we can't smile and get on with life when our muscles are aching and we are denied one more thing we can't do in life. We get beaten down by the workforce, who expects us to compete with our peers on an unfair playing field, where equal opportunity is lip service so privileged people can feel good about themselves. We get beaten down by the general public, who can't quite empathize exactly how it's like to struggle but think they do. Lastly, we get beaten down by ourselves as we don't have enough self-belief that we can give a lot back to society instead of the burden people make us to be!

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

The younger and immature me would have said run and run a lot! I'm a lot older and wiser now – what I would do is all the house chores, grocery shopping and all the boring things I haven't been able to do, for the rest of my life as a huge thank you and show of gratitude to my wife who has stuck by me all those years.

*** * * * *** To read more "LGMD Spotlight Interviews" or to volunteer to be featured in an upcoming interview, please visit our website at <http://lgmd-info.org/spotlight-interviews>

Karen

LGMD “Spotlight Interview”

Name: Karen **Age:** 54 Yrs. old



Country: USA

LGMD Sub-Type: LGMD 2I

At what age were you diagnosed?

I was diagnosed at the age of 33.

What were your first SYMPTOMS?

My first symptom was an overall weakness compared to other kids in gym class in grade school. High School gym class was even worse. I experienced tight hamstring and calf muscles. My calves were large. Next was the noticeable change in my gait.

Do you have other family members who have LGMD?

No, I am the only family member diagnosed with LGMD.

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

The greatest challenges of living with LGMD are completing daily personal and household tasks, managing activity and rest, being prepared to access public spaces, and planning for

future needs.

What is your greatest accomplishment:

With the love and support of my family and friends, I embrace and experience life in spite of ever changing limitations and obstacles. That is my greatest accomplishment.

How has LGMD influenced you into becoming the person you are TODAY?

Living with LGMD has formed me into an accepting, determined, resourceful, humble, and grateful person. I learned to accept my diagnosis and realize that, as it progresses, it provides me with the opportunity to grow as a person.

What do you want the world to know about LGMD?

LGMD is a progressive disease. It affects every thought that goes into every motion of every day. Continued research and development for a cure, treatment, and assistive devices are essential.

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

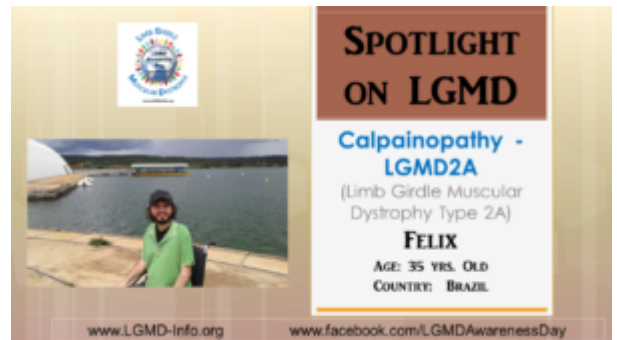
If I was cured tomorrow, I would Rollerblade!

* * * * * To read more “LGMD Spotlight Interviews” or to volunteer to be featured in an upcoming interview, please visit our website at:
<http://lgmd-info.org/spotlight-interviews>

Felix

LGMD "SPOTLIGHT INTERVIEW"

Name: Felix Age: 35 yrs. old



Country: Brazil

LGMD Sub-Type: LGMD2A / Calpainopathy

At what age were you diagnosed:

I was diagnosed at the age of 11.

What were your first symptoms:

My first symptoms included: falls, tiredness, walking on my tip toes. Plus, I could not keep up with other people my age in physical activities.

Do you have other family members who have LGMD:

Yes, I have a sister with LGMD2A

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

The biggest challenge was having to adapt to the changes of my body, developing forms of defense against probable falls, tactics when going up and down stairs, care not to tread lightly on some obstacles, and always seeking a chair for support – especially when getting up from a chair and the

floor. I am always trying to be careful of anything that could bring me down.

What is your greatest accomplishment:

I learned to play the guitar! This was a great challenge because the difficulty was enormous – especially when holding the instrument and making the movement with my arms to play the strings of the guitar. This can be very tiring but still, I can do it.

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

Life with LGMD has taught me and teaches me today to fight and to give more value to life. Surviving in such a difficult world takes a lot of effort and it makes me a better person especially when dealing with others. There are situations in which we live that only those who share in the same struggle actually know and understand. There is a purpose of God in all the lives of this world. I think LGMD may have given me a feeling in life, the biggest challenge I could have. We are opponents, and like every good fight, I intend to fight to the end – winning, of course!

What do you want the world to know about LGMD:

The world needs to know about LGMD, as the people are living with this disease. It is important to be aware of how to help the cause. Knowing that does not make us better or worse than other people who have physical limitations. And hopefully, in the near future, the world will know the greatest of all news – – that there is a cure for this disease!

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

If I was cured tomorrow, I would glorify the name of God. I

would do things like play football as well as climb stairs and sit and stand up without help from anyone. It is these simple day to day activities that most people do not really give much attention but are dreams to those of us in wheelchairs. A cure will be an epic victory, one that will be read in history books.

* * * * * To read more "LGMD Spotlight Interviews" or to volunteer to be featured in an upcoming interview, please visit our website at <http://lgmd-info.org/spotlight-interviews>