

Michele

LGMD "SPOTLIGHT INTERVIEW"

NAME: Michele AGE: 52 Yrs. Old



COUNTRY: United States

LGMD Sub-Type: LGMD2A – Calpainopathy

AT WHAT AGE WERE YOU DIAGNOSED:

I was initially diagnosed with an unspecified form of muscular dystrophy at the age of 25 years old. I received confirmation of my LGMD2A diagnosis at the age of 43.

WHAT WERE YOUR FIRST SYMPTOMS:

In my late teens I developed an awkward waddle when walking and in my early 20's began limping.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

Yes, one sibling has LGMD2A but the other does not.

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

To be blunt, the worst part is dealing with public restrooms because the transfer to/from a wheelchair is physically

difficult and the restrooms are not always adequately set-up so I am always worried about drinking water or coffee while I'm out. Sometimes it feels as though my life revolves around fluid management!

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

Professionally, my greatest accomplishment is my involvement in the creation and management of Coalition to Cure Calpain 3 (C3) which is a non-profit group focused on funding high potential research to cure LGMD2A (also known as Calpainopathy), and educate the global community about the disease.

Personally, my latest accomplishment is solving a New York Times Mini Crossword puzzle in 15 seconds,

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

On the plus side, it has made me more empathetic and given me the opportunity to show my children that it is important to take action when you are frustrated with the state of things. Don't complain; be an agent for change.

On the down side, it has made me adjust my daily life as my mobility has worsened, and while I certainly make the most of my life and am generally happy and productive, it certainly presents its challenges, and that can get tiring physically and emotionally.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

We need more funding for research and more scientists engaged in finding a treatment or cure.

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

Ride a bike (preferably a pink Schwinn with a basket on the handlebars and a thermos of coffee) to the beach and walk

along the jetty with my family.

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Kelci

LGMD “Spotlight interview”

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



Country: Australia

LGMD Sub-Type (if known): LGMD2J – Titinopathy

At what age were you diagnosed:

I was just recently diagnosed at the age of 23.

What were your first symptoms:

At first I noticed having to consciously think about stepping up on to pavement or ledges. I then noticed that climbing stairs was getting more difficult as well as walking distances.

Do you have other family members who have LGMD:

Yes, one sibling does.

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

The greatest challenge about living with LGMD is finding daily activities such as walking distances or climbing stairs difficult, as these were things that I once didn't think twice about. Another challenge for me is having a rare form of Muscular Dystrophy, as there is hardly any information for me to relate to or predict what may happen.

What is your greatest accomplishment:

LGMD is still very new to me as I was only diagnosed 3 months ago, I am still trying to adjust and figure out what living with LGMD is going to mean for my future. My biggest accomplishment is just simply learning how to stay positive and incorporate Muscular Dystrophy into my life.

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

I believe I have learned not to take anything for granted and to appreciate what you have, while you have it. It's so important to stay positive!

What do you want the world to know about LGMD:

I think it's important to realize that we are still the same person we once were. People should also know that not every day is the same for someone with LGMD, some days we will be better than others and what we can do one day we may not be able to do the next.

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Tony

LGMD “Spotlight Interview”

Name: Tony **Age:** 32



Country: United States

LGMD Sub-Type: LGMD2i

AT WHAT AGE WERE YOU DIAGNOSED:

I was diagnosed with LGMD around the age of 26 at the Cleveland Clinic and then officially diagnosed with 2i last year through genetic testing.

WHAT WERE YOUR FIRST SYMPTOMS:

Looking back, I was always struggled to keep up a bit with other kids while playing sports, hiking, running, etc. My first noticeable “something might be up” symptoms started showing while I was touring the country with bands and loading gear every night about 10 years ago. I began to struggle a bit doing things I’d previously never had problems with (going up

stairs without using a handrail, carrying large gear, etc.) I continued to notice small things getting more difficult through college but (of course) ignored it for years.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

Nope. Just me.

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

Adjusting and adapting and giving up things I've greatly enjoyed. Having to be extremely careful with seemingly every single movement. I greatly miss things like going for a run (even if it wasn't quite graceful), walking to get groceries, and just getting from point A to point B uninhibited.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

Finishing college after putting it on hold to play music and travel all over with my friends. Writing comedy for an independently released movie, web series, and book. Moving to Los Angeles and pursuing/accomplishing a career in entertainment/media and marrying the most perfect person for me in the world.

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

It's greatly improved my problem solving skills; making me more resourceful, creative, and patient. I've been able to apply that to my career and other parts of my life that have definitely made me a better teammate, friend, husband, and creative person. At least I think it has and I hope others would agree!

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

A good start would be getting the entire world to know it actually exists. That it's a major struggle for those affected

but so many of us are living happy/productive lives, and there are treatments and cures on the horizon that we'd love everyone's help and understanding in pushing them to the finish line.

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

Man...I think about this constantly so I already have the day planned: I'd take my dog for a nice, long walk (maybe run?). I'd carry all the groceries in instead of needing my small wife to deal with it. I'd play hacky sack with some college dudes and maybe try this "hiking" thing that people in Los Angeles seem to love so much.

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Scott

LGMD "SPOTLIGHT INTERVIEW"

Name: Scott **Age:** 48 yrs. old



Country: United States

LGMD Sub-Type: LGMD 2L – also known as Anoctaminopathy

At what age were you diagnosed:

Although my MD began about 15 years ago, I was finally diagnosed recently with limb-girdle MD Type 2L. Until now, I was only informed by doctors that it was LGMD.

What were your first symptoms:

I began having difficulty climbing stairs and running.

Do you have other family members who have LGMD:

No

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

I have always been an independent person. Since I am an only child, I always did things on my own. Lately, I have had to rely more and more on family and friends. Also, it drives me nuts as to how long it takes me to do things now.

What is your greatest accomplishment:

I am proud to have completed college and earned my degree.

Also, I am happy to have worked 30 years for the same company before having to resign due my struggling with MD.

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

I am still trying to develop patience. I take pride in accomplishing tasks that once I took for granted.

What do you want the world to know about LGMD:

Please continue to support local and national MD charities

whether it be financially or volunteering time to those in need. It is greatly appreciated.

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

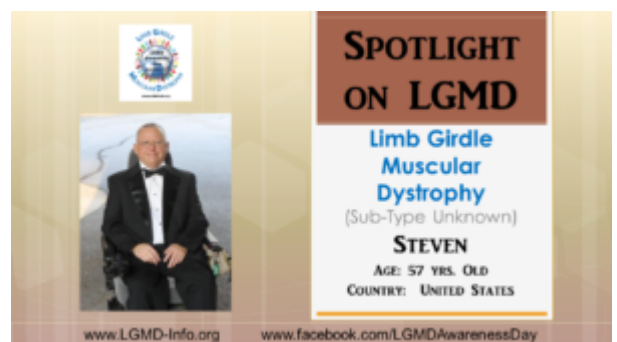
Walk unassisted down by the ocean at one of my local beaches.

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Steven

LGMD Spotlight interview:

Name: Steven **Age:** 57 yrs. old



Country: United States

LGMD Sub-Type : LGMD (sub-type unknown)

At what age were you diagnosed:

I was diagnosed at the age of 43.

What were your first symptoms:

Vague symptoms have been present since birth. I had a waddling gait, could not climb, had difficulty with stairs but the doctors could not diagnose the cause. My mother always knew something was wrong...but what? I was finally diagnosed with LGMD when heart problems began at the age of 43.

Do you have other family members who have LGMD:

Yes, most likely. A second cousin (female) and my Grandpa Stone possibly had a form of MD because he was crippled from youth but back then they called it "rheumatism". I also had another second cousin (male) who was totally helpless and died young.

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

The greatest challenge are falls! I was in Boy Scouts since the age of 11 and had difficulty keeping up with other boys in activities. I have experienced numerous falls. I require the rescue squad and neighbors to help me up as my wife is usually at work when I fall.

What is your greatest accomplishment:

My greatest accomplishments include fathering 4 children – 3 of which are still living. I was able to hold a job for 25 years and volunteered with the local rescue squad for 38 years; beginning at the age of 16. I continue to volunteer with the rescue squad but only answer the phone.

I served as a Boy Scout Leader (assistant scout Master for 25 years.) Currently, I am now an organizational representative. I was an Eagle Scout in 1978. For my Eagle project, I did a church cassette tape ministry. I continue to serve as the sound man at church today,

How has LGMD influenced you into becoming the person you are

today:

All of my life – everything that I do is a challenge! But with work and determination, I have ‘Kept on Keeping on.’ I am determined to stay as active as I can be despite my LGMD.

What do you want the world to know about LGMD:

LGMD makes every accomplishment a real challenge. But work and the desire to succeed can make many things possible. My whole life has been hard but it could be worse...especially if I had a different type of muscular dystrophy or I had a more severe form of LGMD.

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

If my LGMD could be cured tomorrow, I would want to be recertified as an EMT and then volunteer on active duty with the rescue squad.

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Melanie

LGMD Spotlight interview – 09/02/2016

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



Country: Germany

LGMD Sub-Type : LGMD2i (heterozygous type)

At what age were you diagnosed:

I was diagnosed at the age of 4 by having a muscle biopsy.

What were your first symptoms:

I always fell when I tried to run and to take the stairs. This resulted in many bruises and scars! In addition, I couldn't get up from the floor by myself. These symptoms already showed up around the age of 2.

Do you have other family members who have LGMD:

No, I am the only one in my family with LGMD.

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

The greatest challenge is to make people understand that I'm not too lazy to do everyday tasks but I am too weak to do them on my own. Another challenge for many of us is accepting a wheelchair as your best friend!

What is your greatest accomplishment:

There are far too many accomplishments! Living a good life with LGMD is an accomplishment in itself! On my blog

(www.littlemissturtle.com) I write about all of my accomplishments and adventures on wheels.

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

I grew up with LGMD. It made me a very strong person. Dealing with a progressive disease that causes muscle weakness and atrophy while being fully conscious about it is a demanding challenge I appreciate every day of my life! I stopped working recently to have more time to do all the things I love. LGMD taught me to take things the easy way. Enjoy more, worry less, stay positive and spend as much time as possible with loved ones. That's my way!

What do you want the world to know about LGMD:

LGMD is a rare and tricky disease. It cannot be compared to any other disease like Multiple Sclerosis (MS), etc. It is important to fully listen to a person who has LGMD when he or she asks for assistance – to avoid hurting him or her in the process.

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

I would get up from the bed by myself, dress and then go for the walk of my life! I would walk for as long as my feet would carry me.

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