

# Amie

## LGMD “SPOTLIGHT INTERVIEW”

**Name:** Amie **Age:** 31 yrs old



**Country:** United States

**LGMD Sub-Type:** LGMD2J (Titinopathy)

### **At what age were you diagnosed:**

I was just diagnosed at the age of 31 with LGMD2J – also known as Titinopathy.

### **What were your first symptoms:**

I have always had problems with my legs and walking, but was always told that it was from short Achilles tendons. After having surgeries to lengthen them, my calves atrophied. My core muscles seemed weak & I always thought it was from having two C-sections. It wasn't until this year that my body took a drastic change in becoming weak and I couldn't ignore it anymore so I made it my mission to find answers.

### **Do you have other family members who have LGMD:**

No known family members suffer from this disease.

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

My LGMD has many challenges. I struggle every day with "normal" daily activities: walking across a room, standing up from a seated position, going up a step or uphill, climbing into a truck, bending down, balancing etc. It can be frustrating at times. I miss out on a lot because of the constant pain I feel. If I do too much (which doesn't take a lot), it will put me down for days. I have to pick and choose what is important and what is not. All of this affects my family's lives as well which I carry the burden for. LGMD has definitely taken some of my freedom which is hard to deal with, but I try to keep a positive mindset!

**What is your greatest accomplishment:**

My greatest accomplishment is being a mom, wife & sister! I have two beautiful daughters, an amazing husband & the most supportive family anybody could ask for. They are the reason I stay positive and "fight like a girl"! All my life all I ever wanted was to be a mom and have a loving family! I am definitely blessed!

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

Though my diagnosis is fairly new, I always knew something was wrong deep down. I actually self-diagnosed myself with LGMD before getting a diagnosis from my neurologist. So when the test results came back I felt an awkward sense of joy. I finally had a diagnosis! I feel like I have been given this disease for a reason that I haven't found an answer for yet. I truly believe everything happens for a reason and timing is everything. LGMD is such a rare disease that is definitely unknown in my area so I am going to research as much as possible and advocate to as many people as possible. I know that I am strong even though my muscles aren't. I enjoy the little things in life and live in the moments! I also seek the positive in every situation. Life is too short to focus on the negative!

**What do you want the world to know about LGMD:**

I want the world to know that 1 in 43,000 people have Limb Girdle Muscular Dystrophy. Unfortunately there is no cure for this disease. Due to its rarity, there is less funding for research which keeps me further from receiving a cure. LGMD can creep into any family & it effects lives on a daily basis. Eventually I foresee this disease taking my ability to walk, but I am going to fight it as long as I can!

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

Where do I start? I would take my daughters to Disney World and then hiking in Colorado! I would walk up a flight of stairs just because I could! I miss the little things I use to do like shopping, dancing or even taking a bath! You don't realize the freedom you lose until it's gone!

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# Anna

**LGMD SPOTLIGHT INTERVIEW**

**Name:** Anna    **Age:** 36 yrs. old



**Country:** Canada

**LGMD Sub-Type:** LGMD2I

**At what age were you diagnosed:**

I was initially diagnosed with a “non- specific muscle myopathy when I was a young child (sometime in grade school). Later in life, when I was 26, I was given a “working diagnosis” of LGMD. Finally, at the age of 36, I was formally / clinically diagnosed with LGMD 2I via genetic testing.

**What were your first symptoms:**

I’m not really sure what my first symptoms were because though I had this disease from birth my parents never talked to me about it. They felt that because my symptoms didn’t medically impact my life then there was no need to dwell on them. Looking back on it- if I had to guess- my earliest symptoms (that I can recall) were toe- walking, and achy calves. I also recall not being able to easily participate in some gym activities like cross- country running and gymnastics.

**Do you have other family members who have LGMD:**

I have three brothers. It is interesting, the two brothers closest in age to me are affected with LGMD but my youngest brother (who is seven years younger) is not affected at all and, in fact, is extremely athletic.

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

For me, the first greatest challenge is the ever changing needs of this disease- sometimes it's hard to keep up! For example, you get used to doing a certain task one way and suddenly, one day, you can no longer do it that way (or you might discover you can no longer do that task at all). Also, you buy a piece of equipment to assist you and it works for a while but then you reach a place where it is no longer helpful and have to either buy yet another piece of equipment or think of something else.

The second greatest challenge living with LGMD, for me, is not knowing how this disease will affect your future. Every one progresses differently, to varying degrees and rates so it is difficult to anticipate what your future needs will be or to plan any kind of future because physically you just don't know what kind of shape you will be in.

**What is your greatest accomplishment:**

I work very hard to live a full life so it's hard for me to name one accomplishment. Sometimes living with disease just getting out of bed feels like my greatest accomplishment J In all seriousness though, I am a mother to two children (one biological and one adopted) so watching them grow gives me tremendous joy and gives me an immense sense of accomplishment. I was also a parish priest for almost ten years and so I had the great honor of journeying with people through their most difficult times. Knowing that I may have helped them navigate that tough patch (even if it was to a small degree) gives me a sense of pride.

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

LGMD has influenced my life in many ways. It has taught me to be adaptive to my environment. I think, when living with this

disease, you naturally learn to think outside the box.

It has also taught me not to dwell because when you dwell you feel 'stuck' which isn't helpful or healthy. LGMD has also given me the gift to appreciate all the little things in life. Most importantly, LGMD has given me the ability to not take myself so seriously. My youngest daughter tells me regularly that I am "goofy" and I'm good with that- life is too short and too uncertain not to give pause and have a good laugh. One of my mottos in life is: "If plan 'A' doesn't work, there are twenty five more letters in the alphabet!" J

**What do you want the world to know about LGMD:**

This disease affects everyone differently and even with the same person it affects them differently depending on the day and surrounding environment. This means that sometimes there may be a lot of anxiety for the person with M.D. Sometimes a seemingly simple and mundane task to an able-bodied person can seem overwhelming to a person with M.D. because of the process and/ or physical barriers involved. All I can say to the world, friends, family and caregivers is that it's important to love and support those with M.D. not just physically but emotionally and psychologically too- even when you may not completely understand what it is they are feeling and why.

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

If I woke up today and had no symptoms I would, without a doubt, head directly to the beach!

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