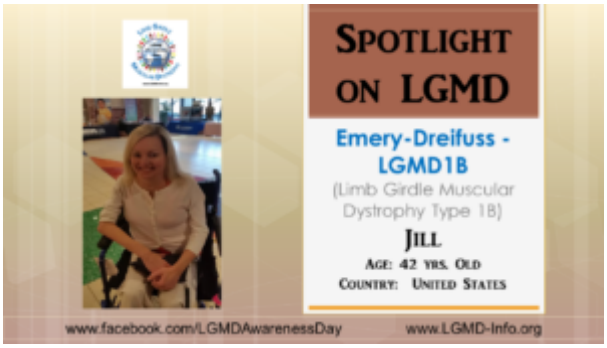
Jill

LGMD "Spotlight Interview"

NAME: Jill AGE: 42
COUNTRY: USA
LGMD Sub-Type: autosomal dominant Emery-Dreifuss muscular dystrophy (LMNA mutation/LGMD1B)



AT WHAT AGE WERE YOU DIAGNOSED:
I was diagnosed when I was 4 years old.

WHAT WERE YOUR FIRST SYMPTOMS:
I was slight at birth, and my aunts both recall thinking there was not enough fat and muscles on my bottom and legs. My early milestones were unremarkable; I was able to sit, crawl, walk, etc. at appropriate ages. As I entered pre-school, it was noted that I fell much more often than the other children, and I had difficulty keeping up with others. I remember having great difficulty remaining upright in a "bouncy house" at a carnival, and also I didn't enjoy bumper cars because I had difficulty with neck weakness.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:
My father and two brothers and one sister all have EDMD, but with a wide variety in severity. One sister is unaffected. It is suspected that my paternal grandmother and great-grandmother also inherited this disorder, but it was so mild they never knew this in their lifetime.

WHAT DO YOU FIND TO BE THE GREATEST CHALLENGES IN LIVING WITH LGMD:
I get frustrated because I want to do the things someone my age should be able to do without significant difficulty. For example, keeping up with housework is very challenging, and I

don’t end up with the house looking the way I imagine it should after working hard.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:
My greatest accomplishment was uncovering my own diagnosis as a teenager and informing my father’s cardiologist that our genetic disorder required implantation of a pacemaker. Perhaps this saved his life when he was only 44 years of age. It gave us the chance to love him for another 18 years.

HOW HAS LGMD INFLUENCED YOU INTO BECOMING THE PERSON YOU ARE TODAY:
It made me learn to rely on myself rather than looking to others to solve my problems. I learned quite early in life that if you have a very rare medical condition, you are going to have to become your own advocate. Also, having a muscle disease made me very dedicated to academic pursuits. I studied genetics as an undergraduate, and creative writing in graduate school. Without my achievements in higher education, I might struggle with uncovering my purpose in life.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:
In terms of what I would want people to know about Emery-Dreifuss MD (EDMD), probably I would want people to know I’m just the same as any other person on the inside. I feel the same hurts, have the same dreams, and most of all, just want to be included in all there is to experience in life. I have been happily married for nearly 12 years, and I’ve raised a son to the age of 11, and most of all I would want people to understand that these life goals can be met by a person even if they are unable to walk.

IF YOUR LGMD COULD BE “CURED” TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO:
I would want to advocate for EDMD to be included in the genetic testing offered to newborns. My greatest wish is that I don’t want a future generation to go through the difficulties I went through, and the best time to intervene is

at birth.

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Karen

11/19/2015

Name: Karen AGE: 61 yrs. old

Country: United States

LGMD Sub-Type: LGMD1B / Laminopathy

At what age were you diagnosed:

I was diagnosed when I was 54 yrs. old.

What were your first symptoms:

My first symptoms included: falling, inability to climb stairs and difficulty lifting.

Do you have other family members who have LGMD:

My sister, her son and his daughter, her daughter, and a brother all have the same diagnosis. There have been 2 that

were also tested and do not have it. LGMD1B has an autosomal dominant inheritance pattern which means that typically this disease appears in every generation without skips.

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

The greatest challenges for me include: getting around, being able to get up when I sit, bathroom problems, lifting, being able to reach items, loading walker in car alone, walking, shopping, and getting dressed.

What is your greatest accomplishment:

For me, my greatest accomplishments include raising my kids and working with school children for 18 years.

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

It makes me more sympathetic to others and their issues. I am feeling blessed to have family members around. I appreciate what I have left.

What do you want the world to know about LGMD

Not all disabilities are immediately visible. Some of us might not look like anything is wrong and yet people judge us. I get plenty of stares when I park in a handicap parking space. I get the look…until I have to get a cart to even walk. I wish that more people knew how to help us – when we fall, etc.

It is frustrating that a lot of people do not know and understand the difference between MD and MS. I say I have MD and they still say MS.

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

I would want to be able to hold as well as sit down and play with my granddaughter.

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Andrea

05/21/2015:

Name: Andrea **Age**: 17 yrs. old

Country: Norway

LGMD Sub-Type: LGMD 1B / Laminopathy

At what age were you diagnosed:

I was about 6 months old when they discovered that something was wrong, but it wasn’t until I was about 2 years old that they decided it was muscle dystrophy. I have probably had the disease my whole life. I was 7 years old when I got the diagnosis LGMD 1B.

What were your first symptoms:

I couldn’t hold my head up when I started sitting on my own.

Do you have other family members who have LGMD:

No, I am the only one with LGMD in my family. Both my parents have been tested and the results showed that I didn’t inherit it, it was from a mutation.

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

The greatest challenges are that I can’t do anything on my own. I need help with practically everything, like getting dressed, getting in and out of bed, and picking up stuff.

What is your greatest accomplishment:

My greatest accomplishment is my mind. How I have a positive look on everything.

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

It has made me appreciate the small pleasures and made me a very positive person.

What do you want the world to know about LGMD:

I want the world to know that I am not chained to my wheelchair, the wheelchair is the most helpful aid I have. Without it I would be chained, it is my freedom. When some people see me they only see the wheelchair, the noise around me, and not me. The disease only affects my muscles and not my brain, I am a normal 17-year-old girl.

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

I would jump, run up and down the stairs, pick up things, do all the normal things I never have been able to do!

Marjolein

05/15/2015

Name: Marjolein

Age: 27 yrs. old

Country: The Netherlands

LGMD Sub-Type: LGMD1B / Laminopathy

At what age were you diagnosed:

I was diagnosed at the age of 4 with LGMD but the sub-type was unknown. At the age of 23 I had a DNA test done which was relatively new in Holland, and that is when they found out that I had LGMD type 1B.

What were your first symptoms:

My mother noticed that I had trouble climbing the stairs. My sister is 1 year and 9 months younger and she could already climb stairs but I still had to use my arms to help me climb the stairs. I was also born with heterochromia iridum (two colored eyes (with one blind eye)) and a palato cisis (an opening in the palate) so my parents figured I had something more.

Do you have other family members who have LGMD:

No, I am the only one so far. We looked in to our history but couldn't find anything that said that others in the family had LGMD.

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

The greatest challenge is having to accept all of the losses regarding the physical things that I cannot do anymore. Sometimes it goes so that my head can't keep up.

What is your greatest accomplishment:

My greatest accomplishments are finishing college, finding my soulmate and buying a house together.

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

I don't really know because I don't know how it is to not have LGMD. I do have a lot of patience. Maybe that is because of all the waiting we have to do to get the things we need to function in our lives….you know, things like wheelchairs and other stuff.

What do you want the world to know about LGMD:

LGMD is a very frustrating disease. People living with LGMD deal with losses day in and day out.

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, I would dance with my boyfriend in a club all night! And, I would go on a hiking vacation.