

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.