

# Thomas

01/30/2016

## LGMD "Spotlight interview"

Name: Thomas

AGE: 31



COUNTRY: Switzerland

LGMD Sub-Type: LGMD 2B / Miyoshi Myopathy

### At what age were you diagnosed:

My first diagnosis (biopsy) was at 28, followed by the proof with genetic test one year later.

### What were your first symptoms:

That is not so easy to say. Now I know where I have to look at, so many examples out of my memories came up; in my childhood we had a steeply rising stair around our home, where I always had difficulties to climb because the steps are very short. In teen age I started snowboarding, there I was not able to give much pressure to the front edge of the board. As young adult I could not jump to the music in discos. At age 24 / 25 I realized that I was not able to stand on my toes and my calve-muscles are actually inexistent. In the last few years I saw that my leg and arm muscles are shortening.

At 18 my doctor told me that I have too high creatine kinase

values in my blood. It was around 250 – 300 somethings, too low for well-known diseases and too high to be normal. Now I know that this was the first medical indicator to Miyoshi.

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

Yes, my sister has LGMD too. Her symptoms (stand on toes, creatine kinase) are the same, but she seems to be less affected.

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

In my opinion, the greatest challenge is to accept this challenge! At the first time after diagnosis, it was hard to accept. You need psychological strength to deal with that. Also when you look at your course of disease, not to overrate or underrate the symptoms and effects. By looking into the future, you always have to plan things with LGMD in mind (maybe you need a wheel chair in 5, 7, or 10 years).

**What is your greatest accomplishment:**

To accept LGMD and enjoy doing things I won't be able to do in future.

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

My diagnosis came just at the beginning of my Master study. Beside my Bachelor and Master study (fulltime study) I was working at weekends and school holidays. Now I work part time and have a good reason to enjoy my leisure time with my wife, our dogs and our garden. I value more what I have now.

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

That it exists. I think there are many unrecorded cases of LGMD.

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

I have just lost my driver license for trucks and coaches because of Miyoshi. It was not only a job to finance my studies, it was also my hobby. So I would get it back and drive around.

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# Laurence

**“LGMD SPOTLIGHT INTERVIEW” – 01/18/2016**

**Name:** Laurence      **Age:** 49



**Country:** France

**LGMD Sub-Type:** LGMD2A / Calpainopathy

**At what age were you diagnosed:**

I was diagnosed at the age of 42.

**What were your first symptoms:**

The symptoms that I experienced included: great fatigue, difficulties climbing stairs, falling when walking on an uneven ground, and difficulty rising from a seated position.

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

No, I am the only family member with LGMD2A.

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

The evolution is rather slow but I must be quite careful to have a regular life otherwise it is worse and the evolution is then quicker when I'm tired and the atrophy is stronger. It's hard to conciliate it with private or professional activities.

**What is your greatest accomplishment:**

I try to keep working which is important not to think too much about the disease and keep going.

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

In fighting against pains and physical difficulties, the disease teaches me how to prioritize but also to be conscious that there are always people who are more ill than yourself so that you must be optimistic.

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

I want the world to know that LGMD is part of the orphan diseases therefore it is difficult to raise funds necessary to find an efficient treatment. It is important to talk about LGMD around us but most of all to the medical community and to general practitioners who can help their patients to go to appropriate medical centers. I have a very personal example to

illustrate this point – before being diagnosed, my GP told me that my symptoms were due to a nervous breakdown!!!

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

I would like to walk naturally without a stick and visit the whole world, be able to go abroad with my family, feeling free!

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# Janet

**LGMD “SPOTLIGHT INTERVIEW” – 01/07/2016**

**NAME:** Janet    **AGE:** 33 yrs. old



**COUNTRY:** United States

**LGMD Sub-Type:** LGMD2B

**AT WHAT AGE WERE YOU DIAGNOSED:**

I was originally misdiagnosed with Polymyositis at the age of 17. Years later at the age of 28, I was diagnosed with

LGMD2B.

**WHAT WERE YOUR FIRST SYMPTOMS:**

I started having difficulty climbing stairs, running, dancing and unable to play soccer as well as any other sports.

**DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:**

There is no family history of LGMD until me.

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

Losing my independence has been very difficult. I am no longer able to walk long distances. I now need a power chair to move around. I am unable to independently care for my two children, 2 years old and 3 months old. I need someone to assist me in taking care of them.

**WHAT IS YOUR GREATEST ACCOMPLISHMENT:**

My greatest accomplishment has been accepting my disease. I mentally prepared myself for my transition to a power chair, where now things can be a little easier when I'm outside my home.

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

I have become a much stronger person. Despite the obstacles that come along with my condition, life goes on and I have and will make the best of it for myself and for my family.

**WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:**

It's a rare disease, but it is out there and it's important to raise awareness that can help find treatments and a cure.

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

I would run towards my kids and lift them up into the air!

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