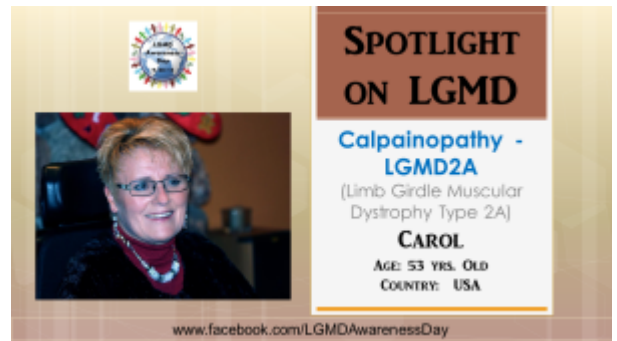


Carol

01/30/2015



NAME: Carol

AGE: 53

COUNTRY: United States

LGMD Sub-Type: LGMD 2A / Calpainopathy

AT WHAT AGE WERE YOU DIAGNOSED:

I was diagnosed with LGMD when I was 9 yrs. old but did not receive genetic confirmation of LGMD2A until I was 51 yrs. old

WHAT WERE YOUR FIRST SYMPTOMS:

As a child, I had a tendency to walk on my toes. By the time I was 9 yrs. old, I could no longer get my heels onto the ground when standing due to contractures. I began to develop somewhat of a "waddling" gait and was also experiencing difficulty in running and climbing stairs.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

No, I am the only one that we are aware of.

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

Continually dealing with the on-going progression of this disease is always a challenge. One of the greatest challenges

though is to be dependent on others for most activities of daily living. I need someone to help me get dressed, to go to the washroom, to bathe, etc. It is very difficult to lose your independence!

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

I was named the 2000-2001 Rehab Institute of Chicago (RIC) "Female Disabled Water Skier of the Year". But in all honesty, my greatest accomplishment is living a productive and active lifestyle despite the challenges that I encounter on a daily basis due to my LGMD diagnosis. I worked for 20+ years as a registered occupational therapist (OTR), I am happily married and I am very involved in my church & community.

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

LGMD has made me stronger as a person despite causing me to be physically weaker. Living with LGMD has help me to realize & accept the fact that everyone has challenges in their life. Unfortunately, my LGMD is more apparent or visible...but it has also helped me to realize that we all have special gifts or qualities that more than make up for the challenges. I've learned the importance of focusing on & appreciating the things that I CAN do versus dwelling on the things in which I cannot. It has also enabled me to find the blessings in my life and to appreciate even the smallest of gifts. Through this all, I have become an advocate and role model to many who struggle with a disability or challenge in their life...and it is in part due to my LGMD.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

Currently there is NO treatment or cure for LGMD. The unrelenting nature of this rare disease just keeps on making us weaker and weaker...even when you think it is impossible to get any weaker! Historically, LGMD attracts significantly fewer research dollars than other forms of muscular dystrophy

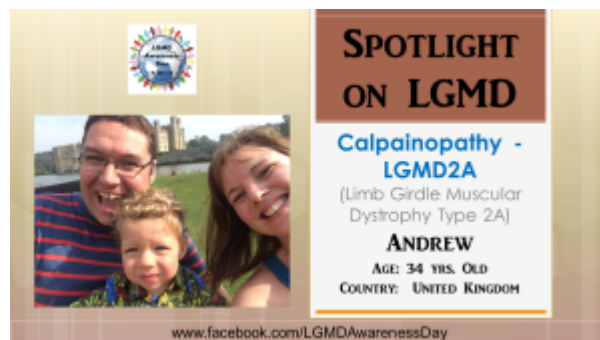
& thus fewer researchers working to understand the disease & discover a cure. This has to change! We need more funding and more research to help discover a cure and treatment for all forms of LGMD.

IF YOUR LGMD COULD BE "CURED" TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO: I would want to run up to my husband and wrap my arms tightly around him to give him a huge hug! Then, go running on a sandy beach.

Andrew

01/24/2015:

NAME: Andrew



AGE: 34

COUNTRY: United Kingdom

LGMD Sub-Type: LGMD 2A / Calpainopathy

AT WHAT AGE WERE YOU DIAGNOSED: 18 yrs. old

WHAT WERE YOUR FIRST SYMPTOMS:

I would fall over when trying to run & also struggling standing up from a chair or the ground.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

No , I'm the only one, no history on either parents side of

the family.

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

As a full time electric wheelchair user the biggest challenge I face is using the toilet. Being sat down so much I find I suffer from IBS from time to time and being unable to stand to transfer onto & off the toilet it makes traveling a bit of a challenge. Traveling by airplane is also very challenging.

We have an 18 month old baby (our first) & at the moment my biggest challenge is being a good dad, especially when I can't lift him up or get on the floor & play cars with him.

Having to rely on family & friends for simple DIY things such as putting up a curtain or a shelf. Getting used to having caregivers come into the family to help out

Feeling you should be able to help family & friends when they are going through tough situations (or just simply popping round for a cup of tea or helping people when they move house etc.).

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

Getting married and starting a family. Finding the right balance between work and home life. Developing a small student accommodation rental business. Becoming Managing Director of the family business (although this is far from accomplished as I've got great plans to take the business to new heights). Completing a Masters and PhD degrees.

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

It has taught me to be patient and if I focus my energies on something anything is possible. A well placed smile goes a long way and opens many doors (literally). You have to be positive and not dwell on what's not possible but concentrate on what you can do , "where there is a will there is a way."

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

Not to take their health for granted , if they see someone struggling , be they disabled or simply old ask if they can help. Put their own problems into perspective and not moan about things that are insignificant.

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

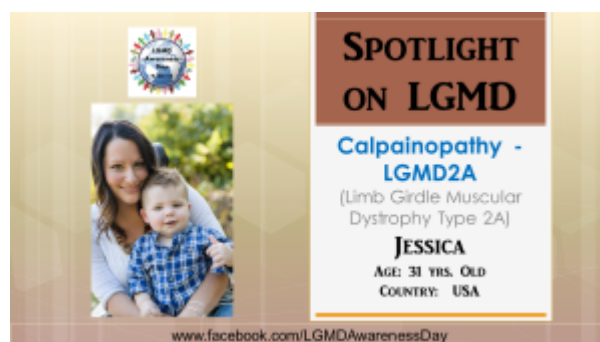
I like to think I would drive down to a beach , take my shoes off , walk at first and then run over the sand , go for a swim in the sea and then relax on the beach. Lay down on the grass in the sunshine on a warm summers day. In reality I would probably just sit down and then stand up and then sit down and stand up over and over because I could. It would be great to go skiing again and maybe scuba diving but really it would just be nice to be able to clean the house so my wife can take the weekend off while I shop , cook and generally spoil her.

It's a dream which I believe will be a reality for us one day but in the meantime I'm not holding my breath letting life pass me by , now is the time to achieve things despite the disability.

Jessica

01/18/2015:

NAME: Jessica



The graphic is a vertical rectangular poster with a light beige background. On the left side, there is a photograph of a woman with dark hair smiling and holding a young child in a blue plaid shirt. Above the photo is a small circular logo with a globe and the text 'Limb Girdle Muscular Dystrophy'. To the right of the photo, the text reads: 'SPOTLIGHT ON LGMD' in large, bold, black letters. Below that, 'Calpainopathy - LGMD2A' is written in blue, followed by '(Limb Girdle Muscular Dystrophy Type 2A)' in smaller black text. The name 'JESSICA' is printed in bold black letters, with 'AGE: 31 YRS. OLD' and 'COUNTRY: USA' below it. At the bottom of the graphic, the URL 'www.facebook.com/LGMDAwarenessDay' is written in small black text.

AGE: 31

COUNTRY: United States

LGMD Sub-Type: LGMD2A / Calpainopathy

AT WHAT AGE WERE YOU DIAGNOSED:

I was diagnosed with Limb Girdle Muscular Dystrophy at the age of 12, but I was not sub-typed with 2A until around age 25.

WHAT WERE YOUR FIRST SYMPTOMS:

I exhibited some toe-walking from my very first steps, but my parents were told that I would outgrow it. It became harder to stand and walk flat over time, but I did not exhibit any other symptoms until around age 8 when I began to have some difficulty walking up stairs, running and doing activity in gym class, and getting up off of the floor.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD:

No, I am the only person in my family known to live with a form of Muscular Dystrophy.

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

I am thankful that there are many aspects of living with LGMD that, by the grace of God, I have come to accept and adjust to over time, but one aspect that I still struggle with is the desire for greater independence and the extent to which I must rely upon others for help and to accomplish normal everyday tasks. The desire and inability to accomplish tasks is like having an itch that you can't scratch. It often requires a great deal of patience, which I tend to struggle with. To complicate things, add in to the mix the guilt that one can sometimes feel. There is both the reality of one's own constant need for help from others and the reality that those serving in a caregiving role have needs, desires, agendas and limitations of their own that I must be mindful and

understanding of.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

My greatest accomplishment is the completion of my doctorate program and receiving my Psy.D. in Clinical Psychology.

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

God has used my living with MD for good, in many ways. He used it to show me my need for Him, which changed my entire life. I now have a lasting peace and joy despite my circumstances. "The weaker we feel, the harder we lean. And the harder we lean, the stronger we grow spiritually, even while our bodies waste away." (J.I. Packer) God has shown me many other blessings through it. I have become creative in problem solving and learned to adapt to changes. It shifted my focus, as I got older, from physical activity to deepening relationships, exploring art, and working especially hard on my academics. I have made life-long friendships with so many special people who have disabilities. It also directed my educational and career pursuits towards hoping to aid others in coping with disability in healthy ways.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD: LGMD is a group of disorders primarily affecting the shoulder and pelvic girdle muscles. There are more than 20 different subtypes of LGMD that have been identified. LGMD may look different depending on various factors, including things like the specific genetic mutation(s), age of onset, rate of progression, etc. You may not even be able to tell that someone has LGMD. You may see another person and think they just walk differently or slower. Another person may need to make use of a wheelchair full time. Even two people with the same exact subtype can present entirely differently. Try not to make assumptions, especially just based on what you see. Most importantly, I want the world to know that LGMD is not a punishment or reason for pity or a reason to believe that someone's life is any less valuable or joyful. God can and

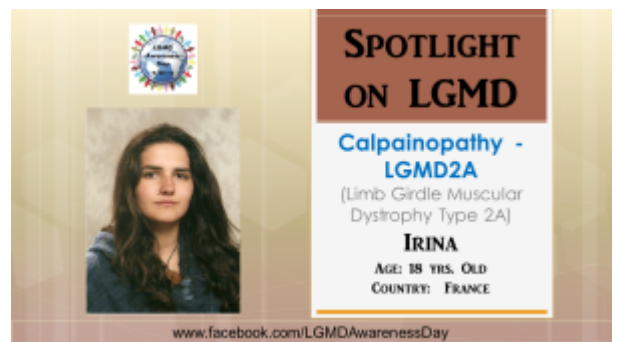
does use difficult experiences, trials, and suffering

IF YOUR LGMD COULD BE “CURED” TOMORROW, WHAT WOULD BE THE FIRST THING THAT YOU WOULD WANT TO DO: I would want to pick up my son and walk around holding him closely. I would want to dance with my husband and share in some sort of outdoorsy/sporty/exercise activity with him. I would want to run, jump, and skip...and praise God for it all!

Irina

01/12/2015:

NAME: Irina



AGE: 18

COUNTRY: France

LGMD Sub-Type: LGMD 2A / Calpainopathy

AT WHAT AGE WERE YOU DIAGNOSED: When I was 14 years old.

WHAT WERE YOUR FIRST SYMPTOMS:

The first symptom that I experienced was having difficulty running.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD: No.

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

Stairs...there are just so many of them! Also, not knowing if people feel sympathy or just pity.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

I was able to get to the top of the Cathedral Santa Maria del Fiore famously known as "The Duomo" in Italy. It has 465 grueling steps to the top! But better than that, the greatest accomplishment of mine is to stay happy and keep smiling no matter what.

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

Knowing that I have LGMD has made me grow up or to be more mature for my age. I have become calm and relaxed by not letting the little things bother me. I also find that I worry less – only about what is important. I don't have time to worry about trifles.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

I want people to know that the disease of LGMD exists! That just because someone has trouble running – don't call them lazy. Also, if a girl uses a walking stick it is probably because she needs it – it is not about looking good!

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

To run after the bus that I just missed!

Martijn

01/06/2015:

NAME: Martijn



AGE: 23

COUNTRY: The Netherlands

LGMD Sub-Type: LGMD 2A / Calpainopathy

AT WHAT AGE WERE YOU DIAGNOSED: Approximately around my 15th birthday.

WHAT WERE YOUR FIRST SYMPTOMS:

The first symptoms were visible since I was a baby. I used to creep like a crab and I was physically 'different' from my peers. I was active in a number of different sports, such as: gymnastics, volleyball, judo and tennis. But my performance was declining due to the on-going progression of muscle weakness in my body.

DO YOU HAVE OTHER FAMILY MEMBERS WHO HAVE LGMD: No.

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

The more practical problems are worse to me. For example walking by my own on the street with crooked sidewalk tiles is quite challenging. It requires a lot of attention and I get tired of this. In most cases I decide to use a wheelchair if I have to walk long distances. Also, explaining my muscle disease to strangers is not my favorite hobby. I am a little bit unsure on how much information I should provide. Some of them are asking about the 'whole process' and the more personal things. And some others feel frightened to ask me about any details. Talking about my disease is not a problem

for me, but determining how much information to share is the challenge.

WHAT IS YOUR GREATEST ACCOMPLISHMENT:

Do you have a few seconds? One of my greatest accomplishments are skydiving, studying and doing everything that healthy people consider as 'not possible' for disabled people. I do like to cross the borders in my life. I am always looking for adventures, like traveling with friends (actually we scheduled a trip to Iceland in February 2015) and so on. Besides that, a personal accomplishment to me is a presentation about my muscle disease and the way I am dealing with the disease to about 500 medical students.

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

LGMD has influenced my life for sure! It has made me more mature in many ways. The way I handle problems and enjoy my life has significantly changed since my diagnosis.

WHAT DO YOU WANT THE WORLD TO KNOW ABOUT LGMD:

People with LGMD or any other muscular disease are not different from healthy people, they just may need a little assistance, at times. Also, do not talk to the person behind the wheelchair, but speak directly to the person in the wheelchair.

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

I would want to go Sky Diving solo!