

“Together We Are Strong”



Before we get started, what is Pompe? Pompe disease is a rare lysosomal storage disease, that affects approximately 1 in 40,000 births. This disease is caused by a mutation in the gene that is responsible for making an enzyme called acid alpha-glucosidase (GAA). Ordinarily this enzyme would use GAA to breakdown a substance called glycogen (a form of stored sugar the body uses for energy). The enzyme stores GAA in compartments called lysosomes. In Pompe disease, the mutation in the GAA gene is reduced or completely eliminated causing excessive amounts of glycogen to build in the lysosome of the cell and cause muscle weakness. The severity of the disease varies based on the degree of the enzyme deficiency.

Firstly, I would like to reflect on my own journey of being diagnosed with Pompe disease. Hopefully this will give others who have been diagnosed or are in the process of being diagnosed hope; hope that there are doctors and clinicians out there that know about this disease, and hope that these medical professionals want to help and have the expertise to do so. Secondly, knowledge is power. Knowing your illness will not only strengthen you as a person, but also as a patient. Finally, I have learned that my strength in coping with this disease is building relationships with others who have Pompe and the relationships that are build with our doctors and medical teams.

My journey of getting a diagnosis...

Why would a healthy teenage girl develop an obscure habit of lifting her legs to get into a vehicle?

This is a question that my parents struggled to find an answer for.

My Name is Erin Gosselin, I was born and raised in a mining town called Sudbury, approximately 5 hours north of Toronto, Ontario. As a child, I would say I was fairly normal, I was as active as all the other kids my age. In high school, people close to me started noticing that I was developing a strange weakness in my legs which became more pronounced in my mid-20's. It was not until after I had my third child (age 29) that the real trouble began. At first, I felt that I was not bouncing back from my pregnancy and as time went on my strength continued to decrease. My symptoms were very characteristic of someone with severe anorexia. I had lost a lot of weight and could not eat properly due to a swallowing issue. Finally, after several trips to my family physician (who was quite certain that I was just suffering from severe postpartum depression) my husband decided it was time to go to the emergency department. Here, they admitted me to the psychiatric floor to be treated for Anorexia. I continued to tell the nurses that I did want to eat - it was my throat was the problem; however, they continued to treat me as if I was Anorexic. All of my meals were monitored by a nurse.

After four days on the psychiatric floor the reality of my situation became clear and I went into acute respiratory failure. I was then moved to the ICU where I was stabilized and then air lifted to London Health Sciences (LHS) in London, Ontario to a neurological department. After much testing, I was diagnosed with Pompe disease.

This diagnosis was a major shock for my family. When I returned to Sudbury it was even more difficult because my doctors were completely unfamiliar with Pompe and were unable to get me the help I needed. I was told that my days were numbered, my baby was only 5 months old, we were devastated. My husband and I decided not to let my diagnosis stop us from trying to seek out a solution. We knew there had to be someone, somewhere that knew about Pompe disease. Through our research, we found Dr. Mark Tarnopolsky of McMaster Hospital in Hamilton, Ontario. Dr. Tarnopolsky completely changed me grim situation. He assured me that he had a treatment that could save me. I left his office feeling much more hopeful about my future.

I have been on Myosyme, Enzyme Replacement Therapy (ERT) for four years and I am doing extremely well. At the time I received my diagnosis Dr. Tarnopolsky told me, at the rate I was regressing, he figured that without treatment, I would have been in a wheelchair and possibly on a respirator

within 6 months to a year. Today I am back to work full-time as a hairstylist and can do most activities with my children. I have been so blessed to have gotten the help I required before it was too late.

My message to everyone that is struggling with a grim diagnosis, is to not let it the first health care provider you see be the final word, especially if they are not familiar with your diagnosis. I challenge you to seek out a physician that knows about your condition and knows the treatments available to help you. By doing this you may find that there is hope for your situation and be able to return to the things that matter the most.

Education goes a long way

When you have been given a confirmed diagnosis for a rare disease it can be an overwhelming struggle of emotions, leaving you not knowing where the road ahead is taking you. In my experience, learning about my condition and how it affects me has served me well. I would encourage anyone that has a rare disease to educate themselves on their condition, by doing so you become empowered to make informed decisions. Through the process of educating yourself, you become transformed into a reflective individual that is able to make active decisions regarding their own health objectives. I believe that most physicians would encourage their patients to become more knowledgeable about their condition. Having good knowledge and information about my condition has helped me with communication and to develop a more collaborative relationship between myself and the physicians involved with my care.

In the healthcare world it is important that patients realize the importance of their own participatory action. This is fundamental in the effectiveness of the treatment. In order to participate and be involved in the decision-making process the patient needs to be educated about their disease. If you understand your condition properly, the better equipped you will be to know what's happening to you and why. Take responsibility for your own care. The best way to do this is to listen to your body and track any changes, by doing this type of home monitoring you will be able to spot potentially harmful changes before they turn into real trouble.

The ongoing relationship between you and your physician is a very important part of your overall healthcare experience. This relationship establishes long-term goals for your health and is key to getting great care. A doctor who not only knows your health history, but also what you value as important will be the resource you need most when you are facing tough healthcare decisions.

“Together we are strong”

If you are a person that struggles with a chronic illness you are more sensitive than you would otherwise be. This is because of all that goes on within our bodies, both physically and emotionally. Whether you realize it or not we have a greater need for consideration and caring from the people close to us. Often because of our health, family members, spouses and friends do not always respond to us in an “understanding” manner. Often times, this hurts our feelings because we feel the people close to us are not concerned or do not care about us. It is to be expected that you will feel more vulnerable and possibly alienated from your friends and family. I feel it is important to remember that there is no way a person without a chronic illness can understand what it is like to live with one. Even with the best intentions, it is difficult for anyone to understand what they have never experienced. It is your responsibility to help others understand how your illness affects you emotionally and physically. Also, it is equally important to strengthen your inner ability for coping with your illness. To do this you need to discover and develop value within yourself as a person even though you have a rare disease. From this will come life-sustaining purpose and you will have less need for emotional support from others.

In order for a person to strengthen themselves it is important to join together with other people that are experiencing the same challenges. By doing this you will eliminate the feelings of isolation and loneliness that tend to build with having a rare disease. Joining patients together that have the same illness is a great way for people to share experiences, get information, and advice in their own individual situations and simply build friendships. This also gives people the opportunity to express their feelings with people who will understand and will be a source of support.

I believe that the patient/researcher partnerships that developed will assist researchers to continue to discover ways to lessen the affects of Pompe for the patients, and together we can bring hope for a cure.

I have been truly blessed by meeting other patients with Pompe disease and hearing their stories. Whether we have had a diagnosis for a month or 10 years we all have the same underlying problem and need to come together and encourage one another, whatever our severity - there is strength in unity.

My journey to diagnosis of Pompe and education about the disease has strengthened me. However, what has brought me to a new level of strength is the relationships I have formed with other Pompe patients. Having the support and friendships of other Pompe patients encourages me daily - because *together we are strong.*



I would like to extend a big thanks for the ongoing from my support I receive from Dr. Mark Tarnopolsky and his clinical team at McMaster Hospital, Also to Dr. Christopher Bourdon for overseeing my Enzyme Replacement Therapy (ERT) at Sudbury Health Sciences North. I would like to recognize the wonderful team of nurses that administer my treatment for their great care and support with my health. Lastly, but certainly, not the least, I would like to thank my husband and family for their daily support and love.

Thanks so much!

Sincerely,
Erin Gosselin