Our personal journey: Learning About Primary Lymphedema

Photo: Nate & Nicole Photography

By Joanne Small-Hopkins

In 2007, my husband and I were excited to have our first child, a beautiful baby boy. During that same time, my husband found a small lump on his forehead over his eye. However upon examination, he was told it was not cancerous and that it was nothing to worry about.

Two years later, I gave birth to a beautiful baby girl. She was born with a droopy eyelid, otherwise known as "ptosis." It was congenital and hereditary as my husband also displayed this condition when he was born. My husband's ptosis was bilateral; however, our daughter's ptosis was only in the left eyelid. We knew her ptosis was more noticeable than my husband's and understood plastic surgery was in her future. In her first year of life, I struggled to understand why both of them had this congenital condition of the eye. The doctors said sometimes these are hereditary, but there is rarely a father-to-daughter hereditary transmission with ptosis, rather it may be further down the bloodline.

Photo: Wikipedia.org

We pushed ahead treating my daughter's ptosis by patching her eye and clearing the tear ducts to maintain her eye health.

In the spring of 2010, my husband came home from hockey after being hit in the leg with a puck. There was a large bruise and the leg was tender. Over the coming weeks, he experienced swelling of the right ankle.

As time passed, the swelling did not recede and started in his other leg and ankle. For the next few months, doctors ran a series of tests but nothing showed up. In the meantime, his legs were swollen and it was very uncomfortable for him to be standing for long periods.

Our frustration grew as time passed, and we could not get an answer regarding his symptoms. Almost every night, I was reading international publications and circulations about various conditions and diseases. I kept thinking about my daughter's ptosis and how I felt it was related. What I found stunned me: As I was searching the genetics of a condition called lymphedema, I found many case studies where ptosis was an anomaly. Lymphedema occurs when your lymph vessels are unable to adequately drain lymph fluid. I found that there is a gene called FOXC2—considered the master switch for your body; this gene is also responsible for lymphedema distichiasis syndrome.

The same physician, who was seeing my daughter for her ptosis, was also consulted for my husband's leg edema. I presented him with research I found concerning primary vascular failure in the lower limbs and the FOXC2 gene. I noted that with lymphedema distichiasis syndrome 50% of patients have deep vascular insufficiencies. I spoke to him about my concerns that my daughter was at risk for the same swelling my husband was experiencing. The doctor's facial expression changed as he stated "your husband does not have ptosis."

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I confirmed he did but it was not documented formally in his medical history. In the fall of 2010, my husband's vascular testing showed insufficiencies, while the lympho-scintography test showed normal. Later, my husband was referred to a local geneticist, and in May 2011 blood tests returned from a lab in the United States confirmed that my husband tested positive for a mutation in the FOXC2 gene causative for lymphedema distichiasis syndrome. Currently there are 50 documented mutations in this gene in

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the scientific literature and his particular variant had never been recorded. When I received that phone call my heart broke into a million pieces. I was devastated but also relieved that we finally had an answer. I knew that my entire family was affected and I was not sure how to process this news and how I would cope. My daughter had a consultation visit with her plastic surgeon at Sick Kids that same month, followed by a ptosis repair surgery in June 2011 in Toronto. The surgery failed and we were devastated to know that we would have to bring her back.

In July 2011, my husband and I flew to Toronto to attend an International Lymphedema Conference to see what exactly we are facing and make contacts with international specialists. We found the trip very upsetting but at the same time useful and educational.

When we returned in July, we discovered that both my children aged 4 and 2 had inherited the exact gene mutation found in their father and are at high risk for developing lymphedema. It seemed that my world had turned upside down. In October 2011, we drove our daughter back to Sick Kids Hospital for a second attempt at repairing the ptosis in her left eye. The surgery went well. The eyelid healed nicely, however, the results dropped slightly over the coming weeks. On September 17, 2013, our daughter had a Levator Resection Surgery at SickKids to repair her ptosis, and it seems thus far that the third time is a success!

In summary, 2011 was the most stressful year of our lives—my entire family was diagnosed with a rare lymphatic disorder and my husband with an arteriovenous malformation on his forehead outside the brain. This was investigated further in 2011 and though never documented with this disorder was thought to be related somehow to his vascular insufficiency. I could not comprehend how this could happen with no family history of disease and the probability of all these things happening to us. Today we take one day at a time. I have a strong desire to educate others and myself on lymphedema and rare diseases. I believe the correct diagnosis is extremely important for your family and your generations to come. I urge more lymphedema specialists, physicians and researchers to read about genetic diseases that include lymphedema so other patients are not left to suffer physically and mentally. Currently there are at least 45 known syndromes that include lymphedema. Let's educate our fellow Canadians! LP