Journal of Inherited Metabolic Disease (2018) 41:905–907
https://doi.org/10.1007/s10545-018-0183-2

EDITORIAL

View from inside

Living with mitochondrial disease

Faye Wylie[1]

Published online: 24 September 2018

My name is Faye Wylie. I am 28 years old and I have a condition called Leigh syndrome, a rare mitochondrial disease which affects my entire body, particularly my muscular system. I now run a small organisation called Leigh Network, which supports and brings together families affected by mitochondrial diseases, as well as supporting mitochondrial research in Newcastle.

Here is my story.

I was born in East Yorkshire and grew up in a lovely countryside village with my parents and baby brother, Sam. In 1993, when Sam was a year old, he began to have a wavering eye. After tests, doctors discovered this was neurological. Further tests showed it was likely to be Leigh syndrome. He lost his fight at 19 months old.

By the time I was aged 7, I was riding my bike without stabilisers and I used to love doing this with my dad. We would go for long bike rides through the village, passing by friends' houses, fields and farmlands. Despite being able to ride for miles, when I walked, I would have to stop for a few moments. When walking to school, which wasn't far, I would have to stop up to ten times to rest, as it felt as though my muscles were fizzing like a shaken up bottle of pop. Amazingly, I could run long distances, but not walk very far.

Aged 10, my teachers noticed I was struggling in school. When teachers would dictate in class, I would be a few sentences behind because my hands were too slow to keep up. My friends often had to help write for me.

I also remember when I was 11, whilst doing PE, I could not jump at all. I felt confused as I was watching the rest of the class jumping and doing hops, skips and star jumps, while I could not. My muscles could not 'spring'. A few weeks later, I was able to jump again and I spent the whole lesson just jumping because I could. This was the last time I jumped.

Whilst this was happening, I was being seen by a doctor at the local hospital who had been my brother's doctor too. He had implied that my walking and jumping issues may have been my way of trying to gain attention, because when he asked me to walk up and down his small office to show him how I had to stop to rest, I could walk seemingly fine. It was such a short distance though, and I did not advise him of this

✉ Faye Wylie
leighnetwork@hotmail.co.uk

[1] Leigh Network, The Wirral, UK

as, contrary to his perception, I was not the type of child to seek attention.

In 2001, when I was 11, my parents and I moved across the country to the Wirral and set up home to be near family, as we are all very close. I moved to a new GP surgery and my mum mentioned to the doctor about losing Sam and my trouble walking. The following summer, when I was 12, I was referred to Alder Hey Children's Hospital due to my declining mobility, where I had many blood tests, a kidney functions test, ultrasounds, CT scans, MRI scans, ECG tests, muscle biopsy and a lumbar puncture, to name but a few! The muscle biopsy and lumbar puncture where the worst tests to endure and had the worst after-effects. I was able to walk into Alder Hey but left in a wheelchair and I have not been able to ride my bike since.

Following these tests, I was extremely unwell for six months. I was in bed all the time—even to just sit up was an effort. I was vomiting every day, which caused dehydration and weight loss. At one point, I thought I would die. Nobody told me the implications of having all the tests done and the trauma it would cause to my body. The dehydration from vomiting caused me to suffer with headaches, which lasted ten years. These only stopped after my second lumbar puncture and acupuncture. During this time, I found solace in writing, reading and artwork. When I returned to high school, I found it hard to connect with people and make friends, as I was in a new area and had spent so much time away from school.

When we look back at photographs from my mid-teenage years when I left high school and started college, it is noticeable that my hands started becoming clenched, into tight fists. I realise now that this was the start of dystonia. When it's bad, it feels like the muscles in my hands and legs are so tight that they never relax. It's as if my muscles are having a game of tug of war with each other. The pain is not like a sharp, acute pain, but more like a continuous, humming pain. Despite the dystonia affecting my hands, I was still able to achieve an A level in fine arts and was fortunate enough to be selected by a local artist and be featured in her art exhibition at the Williamson Art Gallery.

As I got older, it was becoming apparent that my speech was getting worse. I became shyer and, unless I knew people very well and could trust they would not feel embarrassed to ask what I was saying, I would be reluctant to speak. When I was 13, I was able to walk, pushing my wheelchair for support. However, by around the age of 16, I had to be pushed around in my wheelchair, as I was too tired to push it myself and my mobility had worsened. I hated losing this independence.

I'd heard the name 'Leigh syndrome' a few years ago, when all those tests were done, but I had never pressed my parents or doctors for more information. They had always described my condition as being due to my body's cells not producing the energy I needed. I was often told that, as there has been little research carried out on this particular illness, nobody could say for certain what would happen to me or when. But, as I aged and as my symptoms failed to improve, I decided to research Leigh syndrome, the disease my brother had died of, for myself. As I trawled through articles, blogs and medical websites, it became apparent that Leigh syndrome symptoms matched my own, including low muscle tone, poor weight gain and dystonia. Seeing the familiar names of pills prescribed to Leigh patients, cold comprehension crept through me as I realised I had the same disease that had claimed my brother's life.

I asked my paediatrician for an appointment with a specialist and they referred me to a doctor from Manchester, who I met at Alder Hey. He was very knowledgeable, which was good as I had lots of questions! We met on New Year's Eve, which was Sam's birthday. The specialist explained to me the scientific elements of mitochondrial disease and he answered my questions. In these four hours, my life changed completely. I now knew that it was an inherited disease that is passed on maternally and any children I may have will also have it, but to what degree, nobody could say. The specialist finally told me that my health will probably get progressively worse—Happy New Year, indeed!

After being told this devastating news and having no support offered (there simply wasn't any available), I left the hospital feeling numb and cried for most of the night. It took me almost two years to get used to the idea of having Leigh syndrome. My method of coping at the time was to focus on other things, as I was doing my A levels and helping to look after my wonderful Nan, who had dementia.

Around aged 20, I learned that there were many others who had mitochondrial disease, but I had no way of contacting them. I wanted to help other people like me, as I had felt so isolated since my diagnosis, but there was nowhere that could support me to accept and cope with my illness. I started studying counselling skills in college. Although my teacher was unable to understand or appreciate my condition and the difficulties I suffered because of it, I became a fully trained listener and used this skill to help others with mitochondrial disease. I began to reach out to those affected by mitochondrial diseases by approaching hospitals, hospices and charities across the country. I arranged the first mitochondrial meeting in Liverpool, where a specialist doctor gave an informative talk about mitochondrial illnesses. As soon as I went to the venue, I felt an instant connection to the families there. We were able to find and support each other and, from this, Leigh Network was born.

Due to the overwhelmingly positive response, I decided to organise more meetings and each one got bigger and better. Families with children, teenagers and adults affected by mitochondrial diseases attended and we would try to arrange days where everyone could have fun, share information and feel

supported. Aged 22, I got the idea to host a Fun Day to raise funds for our fourth meeting, with the support of the local press and the charity Leonard Cheshire Disability.

This took eight months to organise and involved arranging stall holders, volunteers and publicising it. Fortunately, on the day, the sun was shining and we had lots of visitors, including the Leigh Network families. We also had a local group display their gymnastics skills and this was extra special, as one of the gymnasts also had Leigh syndrome. The Fun Day was a great success, as we raised awareness and met new people. All proceedings went towards organising the fourth meeting of Leigh Network, held in Peterborough. Also that year, I was delighted to win the Local Campaigner of the Year Award from Leonard Cheshire Disability.

In August 2012, I achieved my lifelong dream of driving. I knew how lucky I was to be able to do this in an adapted car, and I cherished every hill start, junction, traffic light and dual carriageway. I particularly enjoyed driving in the ice and snow! Just as I was preparing to take my driving test, my eyesight started to fail and I could not see the traffic lights ahead of me. This meant I could no longer get behind the wheel, which left me devastated.

In April 2013, I was rushed to hospital after an eye test showed I had a swollen optic nerve—here began the worst time of my life, as I had another lumbar puncture, ECG, MRI, blood tests and a CT scan. The lumbar puncture was clear and tests showed that the swelling on the optic nerve was not, as feared, an indicator of a brain tumour or stroke, so, luckily, I was OK. A few days later, after I discharged myself, I was back in hospital after suffering with severe dehydration from vomiting following a migraine. I felt scared as the tests on my eyes began again, but, optimistically, I had envisaged that I would get better and be able to see properly again soon.

One summer's day a few months later, I was still having ongoing tests and I was sat in the garden reading a book. It was a childhood favourite of mine, and as I turned the page and tried to focus my eyes on the words, the words blurred into nothing and I could no longer see to read. As someone who has loved reading and had always found it a wonderful escape from my problems, to lose this freedom and escapism was devastating.

Two weeks later, I went back to hospital to have more tests done on my eyes. At about ten past three in the afternoon on 18th July, I looked at the ophthalmologist and asked him, "What can you do for me?" His blunt answer was, "There is nothing I can do for you. If you believe in miracles, get hoping." With that, the appointment was over. My confidence, plans for my future, hopes and dreams had all been damaged and, once again, I was offered no support.

That summer, I sank in to a deep depression and hit lows I never felt possible. In October that year, I went back to hospital to be officially registered as severely sight impaired. I left hospital, discharged from their care, as there was nothing more they could do.

Despite my failing eyesight however, I managed to host the fifth family meeting for Leigh Network, which was held in Legoland in Windsor, with help from Merlin's Magic Wand Charity. I was in contact with a fantastic researcher from Great Ormond Street Hospital who spoke to those of our families who were well enough to attend. After Windsor, we revisited Liverpool, Cadbury World, Chelmsford and then Glasgow for Leigh Network meetings.

Also that year, I was nominated for the Cheshire Woman of the Year Award and went to a grand reception at Eaton Park in Cheshire. Despite not winning, I had an amazing time and even met a few celebrities.

In 2016, I tried to turn my failing eyesight in to a positive and undertook some training in the use of my blind cane, which I passed. This gave me a new sense of confidence in accessing my community and I hope to be able to start going out and about again independently.

To date, Leigh Network has hosted nine meetings across the UK. We have brought together countless families and built many friendships. I am incredibly proud of all of my achievements—I may have a mitochondrial disease, but I am not defined by it.