The difficulty of getting a diagnosis

It's every parent's worst nightmare to have to take their child to hospital. And when a child with Down syndrome has difficulty communicating their symptoms, it can compound the difficulties faced by health professionals when making a diagnosis.

Leonie White shares her experience of what it was like navigating the health system when her daughter Caitlin started showing symptoms of a stroke, and highlights some of the challenges her family came up against.

Initial assessment

In May 2018, just short of Caitlin's 18th birthday, she collapsed. She couldn't speak and had a facial droop and weakness on her right side. I rang an ambulance explaining that I thought my daughter was having a stroke, and as soon I said she also had Down syndrome they were on their way.

At hospital, a stroke team assessed Caitlin and then admitted her to an acute stroke ward. Caitlin was understandably scared and resisted the first attempt at doing an MRI of her brain, making a diagnosis difficult.

After a few hours of observation and no obvious sign of lingering stroke symptoms, Caitlin was discharged. Her neurologist suggested it was more likely that she had a severe migraine and not a stroke, but before we left, they wanted to do an ultrasound of her carotid and intracranial arteries.

The ultrasound found she had a significantly narrowed artery in her brain that probably caused a transient ischemic attack, or a mini stroke. The sonographer who performed the ultrasound suggested she could have moyamoya syndrome, but Caitlin was still discharged from hospital that day.

The next month Caitlin had an MRI under some sedation, confirming she did have a mild stroke. She had already been placed on blood thinning medication (aspirin) and a statin to lower cholesterol. But no other further treatment was suggested at that time. Caitlin continued to have persistent stroke-like symptoms – mainly loss of speech, headaches and weakness on the right side. There were no obvious triggers for the symptoms, but they were occurring frequently.

Caitlin was in her final year of school and was traumatised by the whole experience. A week didn't go by without the school ringing me because she was continually complaining of symptoms.

The next steps

She was reviewed again in November and we agreed to have another MRI. I hoped this would show Caitlin was now okay. Through the hospital system we waited until February 2019 for that test.

My husband and I had anticipated that this consultation might have been a difficult one for Caitlin to manage, so we requested to speak with her neurologist first without Caitlin being present. Her neurologist agreed and we were able to have an open and frank conversation prior to Caitlin joining us. She was already so confused and overwhelmed by her recurring symptoms that we felt we needed to reduce her concerns and stress.

The results were not what we hoped for.

The MRI showed progression and narrowing of additional arteries in Caitlin's brain. Her neurologist felt that Caitlin may have moyamoya and referred her to see a neurosurgeon.

Prior to seeing the surgeon, a brain SPECT scan was needed. In preparation for the test, Caitlin created a playlist of her favourite songs on her phone to listen to. It was not a pleasant test as it brought on stroke-like symptoms, but listening to music and singing when she could, got her through it. The nurses and doctor also sang along too, making a huge difference. The test confirmed the need for brain bypass surgery to revascularise her brain and improve the blood flow.

After the initial consultation with the surgeon, Caitlin walked out thinking she was going to die. The surgeon did not really engage with her, which made her extremely reluctant to even see him again, let alone have him perform surgery on her.

I explained to the surgeon that Caitlin was scared, and she did not want to meet with him again. I suggested it would be great if he could try to talk to her directly about a few things that she was interested in. To his credit, he did this and their relationship improved significantly.

At this point we felt we needed a second opinion; brain bypass surgery is a difficult surgery that is rarely performed and therefore only a few experienced surgeons are capable of doing it. There were no guarantees that the surgery would improve Caitlin's conditions, and there was still a high risk that things could go wrong. A second surgeon proposed a slightly different approach, but surgery really was looking unavoidable.

After a huge amount of deliberation, we agreed that surgery was needed to improve Caitlin's quality of life. She had just finished school, and we were all working so hard for her to get a job and eventually live independently. In the end it felt like we didn't have a choice. Without surgery there would be an increased chance of further strokes, and that was not something we were prepared to risk.

In June 2019, Caitlin underwent eight hours of brain bypass surgery.

Recovery has been slow and there have been a few lingering symptoms, but further tests have confirmed that the blood flow in her brain has improved and her risk of stroke has reduced.

She is presumed to have moyamoya, but only time will tell. Continual testing will be required for the rest of her life to monitor her symptoms and check for any further narrowing of arteries in her brain.

Lesson learnt

Looking back over the past few years, we have all certainly learnt a lot. We learnt the power of music, and the ways that it could be used to diffuse anxiety, divert the focus from pain and help to fall asleep in a busy ward with lots of distractions. When we knew an IV line was needed, Caitlin would create a long play list in anticipation that finding a vein was going to be difficult and that it was probably going to be needed to be done under ultrasound.

Many social stories were written by her occupational therapist to help Caitlin prepare for hospital, surgery and recovery. These really helped reduce her concerns and armed her with tools to use when she needed them, like playing songs, hugging a soft toy, holding (or squeezing) my hand and singing.

One thing that I struggled with throughout our stay in hospital was the variety of ways that health professionals communicated with Caitlin. Most directed questions or instructions to her, but many couldn't speak to her in a way that she could understand, particularly when asking her about her pain levels.

This is a really difficult concept for most people with Down syndrome to monitor. When asked how much pain you have where one is low and 10 is really bad, often Caitlin's response would be a low number. She had no understanding of what they were asking, so trying to convince staff that pain relief was needed was challenging. I erred on the side of requesting regular pain relief to proactively get on top of pain before it became a problem.

The most important thing I learnt was how strong my daughter was. She endured continual testing, prodding and poking, gruelling surgery, hospital food, shaved hair, a long, slow recovery and the likelihood of more surgery in the future. For the most part, she did it all with a smile on her face and a song in her heart.

As I keep telling her, she is the bravest young woman I know.

What is moyamoya?

Moyamoya is a rare condition where the vessels supplying blood to the brain become narrowed. This limits the flow of blood to the brain, giving a higher risk for stroke. It is a progressive condition which gets worse over time if left untreated.

People with Down syndrome are at a higher risk of developing the syndrome, and may initially present with transient ischemic attack-like symptoms.

For more information, see: https://www.mayoclinic.org/diseases-conditions/moyamoya-disease/symptoms-causes/syc-20355586