

**“I’VE OFTEN FELT MISUNDERSTOOD AND ISOLATED AS A PERSON WITH EPILEPSY, BUT THERE IS STRENGTH IN ACCEPTING THE MOST VULNERABLE PART OF WHO I AM, SO THAT IT DOES NOT DEFINE ME, BUT EMPOWERS ME TO BE MYSELF.”**

# Finding acceptance in epilepsy

One of the main drawbacks of epilepsy is the stigma that comes with having the disorder

*Diane Peters*

**I**n 2004, when Isabelle Siciliano was 13, she woke up in the middle of the night to a loud commotion and family members hovering over her bed. “I felt nauseous and very disoriented,” she recalls.

She’d had a seizure. After a series of medical tests, Isabelle received a life-changing diagnosis: She had tuberous sclerosis, a genetic condition that causes mostly benign tumours inside the body.

Isabelle, now 27, had one tumour on her heart, which has since gone away, and still has them on her kidneys. She can still develop growths on her lungs, and she risks passing the condition on to future children. But most troubling of all is that tumours in her brain cause nerves to misfire, creating seizures.

When someone has regular seizures like this, they’re considered to have epilepsy. Epilepsy can be caused by genetic factors, like in Isabelle’s case, or it can be caused by brain injury, cancer, stroke or other unknown reasons. While the other aspects of her original diagnosis worry her, it’s the epilepsy that impacts Isabelle’s life every day. She takes medication but can never drive. She goes to bed at 8 p.m., or risks the possibility of sleep deprivation causing more seizures. “Even stress itself can be a trigger,” says the Toronto-based theatre educator.

In her early 20s, Isabelle developed severe anxiety. That’s partly because of what happens in the brain when a seizure occurs – doctors think that the process of nerves misfiring can create anxiety in itself – but it’s also because of the stigma associated with epilepsy. According to a 2008 study from the *Canadian Journal of Neurological Sciences*, people with epilepsy are often “wrongly viewed as having mental health and antisocial >

We asked Isabelle Siciliano to sum up life with epilepsy. The phrase above her is how she feels, in her own words.

issues and as being potentially violent toward others,” and, say the authors of the report, “they fear rejection and often feel shame or loneliness from this diagnosis.”

While epilepsy has been around for centuries, few know it’s one of the most common neurological diseases in the world, impacting an estimated 350,000 Canadians, and 50 million people worldwide. “I’m sure everyone knows someone who has it, but they just don’t know who,” says Dr. Danielle Andrade, medical director of the Krembil Brain Institute’s Epilepsy Program.

Silence has led to a lack of understanding. “Every time I disclose that I have epilepsy, I am fast to say, ‘Don’t worry – you might be thinking I’m going to drop down on the ground and convulse, but I’m not going to do that.’” says Isabelle. These attitudes have made it harder for researchers to attract generous charitable donations and land big government research dollars.

At Krembil, though, work is underway to change the lives of people with the disorder, including their treatment and

how they’re viewed by the public. With a patient base of 2,000, one of the largest in the world, Dr. Andrade and her team are doing much-needed work on the genetics of the disease and using new cutting-edge technology to better understand seizures and improve brain surgery outcomes.

They’re also educating the public about this misunderstood disorder to push against stereotypes and encourage more interest in research. “We want to reduce the stigma, but at the same time we want people to remember this is a serious disease,” says Dr. Richard Wennberg, clinical neurophysiologist and researcher with Krembil and a member of the epilepsy team.

**THE ROOTS OF STIGMA**

Reactions to this disorder range from ignorance to prejudice. While Isabelle has found full support at work, doctors at Krembil often hear of their patients facing discrimination. Elsewhere, it’s worse. “In some countries, the stigma can affect the whole family and [epilepsy can] be considered a curse,”

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Isabelle Siciliano  
Krembil Brain Institute patient

says Dr. James Eubanks, a senior scientist at Krembil.

One misconception is that all seizures are like ones you see on TV. While Isabelle used to have tonic-clonic seizures – formerly called grand mal, the kind in which you convulse – she now has focal seizures, which are almost invisible. “I could be sitting here talking to you, but I could feel like I am being separated from my body.”

She sometimes forgets words or slurs her speech during these quick episodes. “You may look like you are intoxicated during or right after a seizure,” adds Dr. Andrade, who recommends that all her patients wear medical alert bracelets, which Isabelle does, so people won’t make false assumptions.

**BETTER TREATMENTS**

Patients with epilepsy want a better understanding of their disorder, not just by society, but by medical science. For starters, they need better treatment options. There are 14 drugs approved to treat epilepsy in Canada, but no way to know which one will work on any given patient. “Treating epilepsy with medications is sometimes hit-or-miss,” says Dr. Eubanks, as doctors can’t tell in advance which prescription will work on specific patients.

More research on genetics might help: About 70 per cent of epilepsies have a genetic cause, which could help with linking the right drugs to patients and assist in finding new medications. Over the last 10 years, researchers have identified more than 500 genes connected to the disorder. “We know there are many more to be found, and we are working on that,” says Dr. Andrade.

For instance, the Adult Epilepsy Genetics Program at Toronto Western Hospital (TWH) discovered a gene connected to sudden unexpected death in epilepsy (SUDEP), a rare phenomenon that happens to about one in 1,000 people with

the disorder during or after a seizure. Before finding this gene, doctors had no way of predicting who was at risk for SUDEP in patients with moderate seizure control. This is because SUDEP

often happens to people with frequent and severe seizures, but it can occur in some people who have relatively mild cases of epilepsy. Now that the gene has been identified, doctors at Krembil can screen epilepsy patients for it to find out if they are at risk and ensure that those people stick to their medication regime.

At a higher level, Dr. Eubanks wants to better understand how a genetic mutation impacts cells and leads to disease. He’s currently working with the gene CDKL5, mutations of which cause a very severe form of epilepsy. “We’re trying to fill in the missing parts of the map,” he says.

**A LEAP IN TESTING**

Another area in epilepsy with serious gaps is related to seizures themselves. Dr. Wennberg, the Krembil clinical researcher, is using TWH’s magnetoencephalography (MEG) machine – acquired thanks to philanthropic support from Mitchell Goldhar – to measure brain activity during seizures. Doctors test patients with epilepsy after they’ve been sleep-deprived so that they’ll nod off and have seizures. He says the hospital has measured more actual seizures in the MEG than any other organization in the world. Working with a mathematician, Dr. Luis Garcia Dominguez, the doctors can isolate brain activity using MEG and closely track a seizure’s signals to understand where and how they happen.

MEG has another practical function: It can reduce the number of surgeries people need to eliminate seizures. For the 30 per cent of epilepsy patients who don’t respond to medication, surgery can reduce or even get rid of seizures. Even some patients who can control their condition by medication will consider surgery, since they can then stop taking medications, many of which come with side effects.

Surgery, of course, is intrusive and involves removing the parts of the brain that cause seizures. For those who have a choice of surgery, the decision to go ahead with an operation is a difficult one. Currently, to assess whether someone is a good candidate for surgery, they may need to be monitored by electroencephalography (EEG),

→ **30% - 40%**

Percentage of people with intractable epilepsy – seizures that cannot be controlled by current treatments.

(National Institute of Neurological Disorders and Stroke)



(Top) Isabelle, at work with a friend, leads the same kind of life as everyone else. (Bottom) For her, the best medicine is to live normally, including spending time with her boyfriend, Scott Lacombe.



where electrodes are placed in the brain to pinpoint tissue that needs to be removed. That procedure is invasive, too. If they can have surgery, they need to assess whether the risks – memory problems, behavioural changes and vision issues are the big ones – are worth it.

While choosing to have brain surgery will always be a tough decision, MEG may help do away with EEG implants, which will then make the procedure that much less invasive, says Dr. Wennberg. Using MEG, he can overlay the neurophysiologic results with existing scans of the patient’s brain to create a more precise map. “We are pushing the envelope further with the accuracy we get,” he says.

In 2016, Isabelle went through a lengthy surgery assessment process that included

spending 12 days in TWH’s monitoring unit, where she found out she is a candidate for surgery. Using MEG, doctors were able to localize her epilepsy to the edge of one of her brain tumours.

For now, though, she has decided to put off surgery. She wants to focus on work, and she also hopes the cutting-edge research Krembil is doing will reduce the need for an operation. At some point, she may get access to better drugs with fewer side effects, for instance.

In the meantime, she has joined a surgery support group at TWH, because she wants to get as many perspectives as possible. But the priority for now is on living her life. “I am putting surgery aside for the moment,” she says. “I am focusing on work right now, as it’s hard to focus on both.”



Dr. Danielle Andrade, medical director of Krembil’s Epilepsy Program, is working with state-of-the-art technology to learn more about why seizures occur.