From Seattle Children’s “InHouse”

Epilepsy Program: Stella's Story and the Secrets to Suppressing Seizures

Stella had her first seizure when she was 6 months old in connection with a fever, which is relatively common in childhood. But she had two more seizures a few months later, and at the age of 2 she ended up in Seattle Children’s Emergency Department (ED) with a two-hour seizure. Dr. Russ Saneto, an epilepsy expert, saved her life that day… and after four years of tests and studies, he determined that she had a rare type of genetic epilepsy called Dravet syndrome.

“Before we found a diagnosis and the right treatment, Stella would often have upwards of 100 seizures a day,” says her mom, Amanda Stansfield. “Dr. Saneto prescribed nearly every epilepsy medication available in the United States. Sometimes she was taking as many as five different medications at one time, which meant 20 pills in a day, but she continued to have seizures.”

Epileptic seizures – sometimes described as “brainstorms” – occur when there is a dramatic change in the electrical activity in the brain.

Stella had different kinds of seizures, including atonic seizures, which caused her to fall; tonic-clonic seizures, which involved convulsions; and non-convulsive status seizures, which resulted in long-lasting stupor and unresponsiveness that sometimes lasted for many days.

“Stella had to wear a helmet because she frequently had seizures where she would fall suddenly and with force to the ground,” Amanda recalls. “She was like a little old granny because she was really shaky when she walked. In addition,
she could have tonic-clonic seizures that would last anywhere from two minutes to over two hours.”

At age 4, Stella tried the ketogenic diet – a special high-fat diet managed by a dietician – with no success. At age 5, neurosurgeon Dr. Jeff Ojemann implanted the Vagus Nerve Stimulator, a device that can control seizures in some patients, to no avail.

Doctors had found early on that Stella was not a likely surgery candidate because her seizures were happening throughout her brain with no single origin – called a focal lesion or seizure onset location – that could be removed by surgery.

However, they started to consider surgery again because none of the medications were effective in suppressing her seizures. After performing more imaging tests of her brain, including nuclear magnetic resonance and magnetic resonance imaging (MRI) scans, they determined that surgery was not an option.

Finally, when Stella was 6, Russ ran a blood test that analyzes the genetic material found in white blood cells to find defects in particular genes. The test found an altered gene that causes Dravet syndrome, a severe type of epilepsy.

“Many neurologists never would have thought of running that test, but Dr. Saneto never gave up hope,” Amanda says. “Having access to specialists is so important. When Stella was diagnosed, there were approximately 20 kids in the country with Dravet syndrome.”

Now Stella is 11 years old and she is finally learning to read. Thanks to some new medications, she has had only three seizures in the past three years – and none at all for the past 18 months.

**Putting the pieces together to reach a diagnosis**

Stella was in a coma for three weeks and spent a month in the PICU.
Stella’s story has a happy ending, but it was a long battle fraught with constant uncertainties, including ED visits and a three-week coma where Stella spent a full month in the PICU.

That’s why the experts in Children’s Epilepsy Program are striving for earlier diagnosis and treatment.

Reaching a specific epilepsy diagnosis isn’t easy. Genetic testing is used to identify rare disorders in patients like Stella, but imaging of the brain is the key for most patients.

All patients who have had more than one seizure have an electroencephalogram (EEG), which involves placing electrodes on a child’s head in order to record the brain’s electrical activity.

In patients who appear to have seizures with a single origin, the epilepsy team uses high-tech imaging to analyze brain structure and activity.

“The new technologies are critical because they help us see if there is a reason for the seizure, which is critical to diagnosis and treatment,” says Russ. “If I were to look at the moon when it’s full from my back porch, it’s just this thing in the sky that could be Swiss cheese. With a telescope, I could see more, and at the observatory I could see even more, down to the crevasses. But if you could walk on the moon, you could even see the dust in Neil Armstrong’s footsteps. The technology gets us closer to walking on the moon.”

The technology continues to improve, and Children’s has acquired state-of-the-art equipment. A 3-tesla MRI scanner achieves higher resolution images than ever before, while a technique called functional MRI enables the team to identify areas of the brain that are used for different purposes, such as speech or hand movement, in order to avoid important areas during neurosurgery.

Children’s also has positron emission tomography (PET) scanning equipment onsite, whereas we used to send patients to the University of Washington Medical Center – where it wasn’t always possible to successfully sedate children for the procedure.
Stella recently received an award at school. She has been seizure-free for the past 18 months!

“Imaging techniques like MRI and PET help us identify the underlying cause of the seizures,” says Dr. Edward “Rusty” Novotny, director of the Epilepsy Program, who joined Children’s last year. “Understanding the cause and pinpointing the origin are critical to determine the treatment – whether we’re going to use medications, try the ketogenic diet or consider neurosurgery.”

An important next step involves drawing on Rusty’s expertise in integrating the images generated through different techniques.

“A child can now get EEG, PET and functional MRI on different equipment, and we can use new software to put the different images together to get a better sense of the whole picture,” says Rusty. “Conventional images, like MRI, only give us an image of the structure of the brain, but some of these new techniques can be used to look at brain function.”

Epilepsy specialists also use imaging to decide if neurosurgery is an option for children who continue to have seizures after trying different medications.

“Imaging allows us to identify types of epilepsy where surgery can be a cure,” says Rusty. “Surgery is the only way to actually ‘cure’ epilepsy, but it only works for some epilepsy patients. Imaging helps determine what operations can be done safely and minimize the risks for children as a result of the surgery.”

Read the second article in this series in the next edition of InHouse to learn about neurosurgery options for children whose seizures just won’t stop… and the guild that Stella’s mom, Amanda, is organizing to raise money to help children with genetic epilepsies.

– Kim Arthur