"Saving Eliza" campaign helps another child

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Will Byers

Valerie Byers had long suspected that her son Will's diagnosis of autism was wrong. So when she saw a clip on the homepage of the Today Show about a little girl named Eliza, in late February, she knew instantly that 5-year-old Will had something far worse.

The clip featured the O'Neill family of Columbia, South Carolina. Last year, their "Saving Eliza" viral video raised awareness and funds to continue a gene therapy trial for their daughter's inherited brain disorder, Sanfilippo syndrome type A.

The O'Neills have been under self-imposed isolation in their home for more than a year, even keeping 8-year-old Beckham from going to school and seeing his friends. They're trying to protect 5-year-old Eliza from picking up a virus that could disqualify her from participating in a gene therapy clinical trial for the very rare disease at Nationwide Children's. (DNA Science covered Eliza's story here and here.)

Watching the Today Show clip, Valerie was transfixed. "Eliza resonated with me. Tears were falling down my face. As I saw Eliza's story and the symptoms, I knew that's what my son had," Valerie told me on a recent afternoon when she managed to get her two kids to nap at the same time.

When she read the factsheet that Cara O'Neill, Eliza's mom, who is a pediatrician, had put together, "my heart sank again. It just fit Will."

An odd collection of symptoms

Valerie and her husband Tim hadn't really thought anything was seriously wrong with their "happy, healthy little boy. Then when he was

3 years old we noticed a slow down in his development. He was still hitting milestones, but slower. We were told to watch for the next year. By age 4 he had missed a couple of milestones in speech and motor skills. So from that point, last summer, 2014, he was sent to a pediatric specialist. They diagnosed him with autism," she recalls.

But the diagnosis seemed off. Valerie knows kids with autism, and she has a master's degree in psychology. But mostly she knows her son.

"Autism didn't take into account everything that was different, quirky, about Will. And he was always social. He wanted to engage with people, he was just delayed in ways that didn't make any sense. He had potty issues. His facial features correspond to Sanfilippo, a large head with prominent eyebrows and widely-spaced teeth. He has joint stiffness and trouble writing and pedaling. And he's hyperactive."

The distraught mother called the pediatrician right away, asking about the urine test mentioned in the factsheet. The next morning, she brought in a urine sample from Will.

A shattering diagnosis

A week later, Valerie and her husband Tim's fears were confirmed. Will's urine had the telltale buildup of heparan sulfate, a consequence of an impaired or deficient enzyme. A genetic test on a blood sample then confirmed that Will has mucopolysaccharidosis type IIIB. It's a different form of Sanfilippo from Eliza's type A, slightly less severe and rarer. But still a relentless neurodegeneration that would drastically shorten life.

Sanfilippo is a lysosomal storage disease, described in my first post about Eliza. Mutations in any of four genes cause it. Will, with type B, is 1 in 200,000. Incidence for all types of Sanfilippo is one in 70,000. Although all four types lead to buildup of the same biochemical –

heparan sulfate – they require interventions targeted to the specific underlying genetic problem.

Valerie recalls learning the results of the genetic test. "It was a devastating day for us, to confirm that's what was happening, to find out your child who is perfectly healthy and happy is now having his future taken away from him. It crushes you."

Knowing helped. "We didn't understand why there were things Will wasn't getting, what we were doing wrong. Understanding what is really happening takes all that guilt away and you can focus on what's important," Valerie says.

But the family was lucky to get a diagnosis so quickly, because then an amazing thing happened.

Enzyme replacement therapy

"As we dove into the research and talked to the O'Neills and others we've connected with, we were able to get Will into a Sanfilippo syndrome type B clinical trial in Minnesota. He got the last spot! If we hadn't seen Eliza's story we wouldn't have had Will diagnosed until next year, because he wouldn't have regressed for another year," Valerie says. The trial is testing an enzyme replacement therapy. Will, who turned 5 in June, was the eleventh and final patient.

"It was a pretty incredible set of circumstances," says Glenn O'Neill. "We feel proud to know our supporters and awareness and early diagnosis forms helped this child get diagnosed and into a clinical trial, which could possibly save his life. If anyone asks about how can awareness help …. here you have a GREAT example!"

Will and Valerie travel from their home in Texas to Minnesota every

other week, for an initial 24 weeks, after which Will will be eligible to continue for up to 3 years. So far, after three intravenous infusions, he's doing well. And the parents are awaiting blood test results on their 20-month-old, Samantha, whose urine test was normal.

Linked silos

Now the Byers' want to pay it forward, by talking about their experience and fundraising. "We don't want other families struggling with this and deal with losing time. Not having a correct understanding isn't fair to any family. Thanks to the awareness the O'Neills raised we now know and we can value our time."

Genes may be silos in terms of therapeutics, which is why families funding research for subtypes of diseases must compete for media attention and funding. A gene therapy for one form of Batten disease, for example, won't help the other seven, caused by mutations in different genes. Hopefully, the 21st Century Cures Initiative, which recently passed in the House, will eventually lessen the competition. Meanwhile, Will's story dramatically shows that raising awareness of any rare disease can help other families in unexpected ways.

More information: For further information on Sanfilippo syndrome, see: www.curesff.org/

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