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'Lilly's Law' inspiration hopes database kick-starts diabetes awareness

Researchers see state registry as valuable tool in study of disease, cause and push for cure



Laurie Jaffe helps her daughter Lilly Jaffe with her school homework at their home. (Abel Uribe / Chicago Tribune /September 1, 2009)

By Margaret RamirezTribune reporter

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Three years after she made medical history and was freed from painful insulin injections, 9-year-old Lilly Jaffe is just beginning to understand how much her story changed the course of [diabetes](#) research and treatment.

Since her breakthrough, 70 other children and several adults in the U.S. also have been able to switch from insulin shots to oral medication. And last month, her story inspired Illinois' adoption of "Lilly's Law," which established a registry in hopes of helping other children and gathering more genetic information on diabetes.

"When I first got off insulin, I was happy, a little nervous and confused," Lilly said at her North Shore home. "Now I know that if I hadn't shared my story, then none of those children would have

known about this. I want even more people to know."

Lilly's story began when researchers at the [University of Chicago](#) found she had a rare genetic mutation known as monogenic diabetes. Although she had been taking insulin injections since she was a baby, the discovery allowed her to take pills often used to treat the milder Type 2 diabetes.

A September 2006 story by Tribune science reporter Peter Gerner about Lilly's diagnosis triggered hundreds of inquiries from families across the U.S. who believed their children might also have the genetic mutation. Some of those were among the 70 who, indeed, had the same mutation as Lilly; others possessed a different genetic variant that formed the basis for another groundbreaking research paper published in 2007.

This summer, Lilly was able to share her remarkable tale with dozens of other children affected by diabetes when she traveled to London. Along with her mother, Laurie Jaffe, she attended a neonatal diabetes conference with the two British researchers who discovered her mutation, Dr. Andrew Hattersley and Dr. Frances Ashcroft.

"That was incredible for Lilly because she got to meet and make friends with some children that were directly affected by her story," her mother said.

After Lilly's life-changing switch from injections to pills, Laurie Jaffe began moderating an e-mail discussion among parents with children who have monogenic diabetes. Initially, it was little more than an online support group, but through the discussions the families stumbled onto links between the mutation and neurological issues and began gathering informal data on a new field of diabetes research.

Dr. Louis Philipson, medical director of the Kovler Diabetes Center at the University of Chicago, said it was well-known that some diabetes patients with mutations also have neurological problems. Therefore, he and his colleagues follow the family discussion group closely.

Philipson and his team have begun working with neurodevelopmental experts to better understand the connection between diabetes and the brain.

"These [genes](#) that cause diabetes are not only expressed in the cells that make insulin, but are playing a role in the brain and elsewhere," Philipson said. "It is a relatively new [study] area ... less than five years. It points out how exciting a time this is for medicine and biology, to better understand the role of genes in disease."

Lilly was diagnosed with Type 1 diabetes, the more serious form, at 1 month. Also known as juvenile diabetes, Type 1 is most often diagnosed in childhood and adolescence, but it strikes adults as well. About 15,000 children are diagnosed with diabetes in the U.S. every year, according to the Juvenile Diabetes Research Foundation.

Lilly started with insulin shots to stabilize her blood sugar, but after two frightening nighttime [seizures](#), she was switched to an insulin pump that was attached to her hip. With the pump, Lilly's mother still had to monitor her glucose levels about 10 times a day and move the painful infusion sites from [leg](#) to leg to avoid irritation and infection.

In June 2006, her parents learned from Philipson about cutting-edge diabetes research that had been done in Britain. He told them of a new study that showed some children diagnosed with Type 1 diabetes in the first six months of life actually have an unusual mutation of Type 1 that can be treated with pills. Soon, Lilly was found to have the rare genetic mutation. A couple of months later, Lilly was able to start the first grade completely insulin-free, taking five glyburide pills twice a day.

With Lilly now in fourth grade and leading a more active life, her mother said, the goal now is to identify more people, advance research and eventually find a cure. Researchers estimate there are about 2,000 people in the U.S. with Lilly's genetic mutation who could benefit from the breakthrough.

"There are so many more people out there that can be helped, but it's [a matter of] identifying them," Laurie Jaffe said. "And it's not just children. ... The adults that have been found are those that have had children with monogenic diabetes."

Hoping to find more patients and gather more genetic information to study, University of Chicago doctors proposed the creation of the first state-mandated diabetes registry. They crafted legislation after joining forces with [Rep. Tom Cross](#), R-Oswego, whose 16-year-old daughter, Reynolds, has Type 1 diabetes, though not the mutation.

The bill officially became Lilly's Law when signed by [Gov. Pat Quinn](#) last month. The law requires Illinois physicians to register all children with diabetes onset before 12 months of age with the state Department of Public Health. Its backers hope the registry, which was approved as a three-year pilot program, leads to further advances in understanding the genetic cause of diabetes.

"Many of the genes that cause special forms of diabetes are not yet known," Philipson said. "This is not only for the knowledge, but for specific treatment and for the family members to understand the genetic risks to future generations."

After Lilly stopped taking insulin, her mother said, one of her first questions was whether her two cousins with Type 1 diabetes could get off insulin too. She was disappointed to learn they didn't have the mutation so they could not.

"Now, we're committed to finding cures for our nieces, cousins and all our friends," Laurie Jaffe said.

maramirez@tribune.com

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