

Girl's rare form of diabetes leads to injection-free treatment for some

September 23 2009, By Margaret Ramirez

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 during an interview at her North Shore Chicago 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 usually used to treat a milder [Type 2 diabetes](#) instead.

A September 2006 story about Lilly's diagnosis by Tribune science writer Peter Gerner 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 that, 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 brand 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 the age of 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 about Philipson's cutting-edge diabetes research. 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, although not the mutation.

The bill officially became "Lilly's Law" when signed by Gov. Pat Quinn last month. Lilly's Law requires Illinois physicians to register all [children](#) with diabetes onset before 12 months of age to the state Department of Public Health. Its backers hope the registry, which was approved as a 3-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 that 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.

(c) 2009, Chicago Tribune.

*Visit the Chicago Tribune on the Internet at www.chicagotribune.com/
Distributed by McClatchy-Tribune Information Services.*

Citation: Girl's rare form of diabetes leads to injection-free treatment for some (2009, September 23) retrieved 25 April 2024 from <https://medicalxpress.com/news/2009-09-girl-rare-diabetes-injection-free-treatment.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.