

MEET OUR TEAM

Endocrinology

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Focus on Advocacy

For families with children who have congenital hyperinsulinism, advocacy can take on many faces. Some parents have to advocate on behalf of their child to ensure timely treatment. Others advocate for all HI patients, working to create support for families facing the uncertainty of an HI diagnosis. Still others turn their advocacy toward helping to support research. This issue of *HI Hope* portrays families that are advocates of all types.

From Personal to Public Advocacy

Two days changed Ben Raskin-Gross' future. For his first two days of life, his congenital hyperinsulinism (HI) went undetected, and the resulting extremely low blood sugar levels added many challenges to Ben's life. Those two days also changed his mother forever. Ben's experience became Julie Raskin's motivation to tirelessly advocate on behalf of children with HI and to diligently spread the word about HI so other children will get immediate treatment and avoid the difficulties Ben lives with.

"While I was still in the hospital with him I had concerns, but the doctor said, 'He's a perfectly normal baby,'" Raskin says. When she brought him to an emergency room two days later, his sugar level was too low to be measured. It took 10 days to receive the diagnosis of HI, and Ben was transported to CHOP to be treated by the Congenital Hyperinsulinism Center. He had diffuse disease and needed three surgeries (the last to remove part of his pancreas that had regenerated). Ben, now 14 and diabetic, manages his disease with an insulin pump that keeps his blood sugars in check.



Ben

He's a freshman at Glen Ridge (N.J.) High School, which has been supportive of his special needs. Even though he's legally blind and has difficulty with motor skills — the result of his low blood sugar as a newborn — he's been in 16 plays, is a member of his school's elite madrigal choir and can rattle off the causes of the French Revolution. "He's a determined young man," says Raskin. "He has a can-do attitude about him and a wonderful disposition."

Over the years, Raskin turned her advocacy on behalf of Ben into a broader effort. She joined with other parents of children with HI, first to be a part of a listserv and then to found Congenital Hyperinsulinism International (CHI) in 2005. CHI's top priority is to advocate on behalf of those with HI. Through its website, congenitalhi.org, CHI provides a virtual vehicle for support and information to families. CHI also shares information with the broader medical community to increase timely diagnosis and improve outcomes for patients.

"Timely diagnosis is extremely important," says Raskin, executive director of CHI. "It can mean the difference between severe disability or living unscathed by developmental challenges."

Finding Help for their Son — and Helping HI Research

When Logan Green was diagnosed with congenital hyperinsulinism (HI) at the age of 5 months, his doctors in Boston were direct with his parents: The experts on HI are at The Children’s Hospital of Philadelphia.

While physicians close to home were able to stabilize Logan’s condition, about a year later, his family took a trip to the Congenital Hyperinsulinism Center at CHOP for additional testing and a second opinion.



Logan with his dad, Gordon Green; mom, Mason Smith; and sister, Phoebe

“We felt very comforted talking to the experts at CHOP who knew this disease so well,” says Logan’s mom, Mason Smith. “They gave us a lot of practical advice that has made dealing with HI a lot easier.”

And after learning that CHOP is the leading research center for HI, the family also decided to financially support the research being done by center director Charles Stanley, M.D., and his colleagues — something they have done every year since 2003.

By contributing to the research fund for HI, Mason Smith and her husband, Gordon Green, have helped support several promising studies under way at CHOP.

In one study, Stanley and his colleague, Diva De León-Crutchlow, M.D., are testing an investigational compound called exendin-(9-39). After showing that exendin-(9-39) successfully suppressed insulin release in mice, Stanley and De León-Crutchlow obtained FDA clearance to test the drug in adolescents and adults with diffuse HI. While the FDA has only allowed trial of the drug for a 12-hour window, exendin-(9-39) effectively prevented hypoglycemia in the nine adolescents and adults tested.

“We’ve done studies now in six newborns as well, infusing the investigational drug over a 12-hour period,” says Stanley. “The results have been very encouraging.”

De León-Crutchlow recently received funding from the National Institutes of Health to test the drug’s toxicity so that it can potentially be given for longer periods of time. If successful, exendin-(9-39) could allow some patients with HI to avoid surgery while also preventing the hypoglycemia that causes seizures, brain damage and developmental delay in many children with HI.

“If the drug works, it might have to be given by shots three or four times a day, but it would avoid surgery for these children,” says Stanley. “And because the disease tends to gradually get better over time, we might only have to treat them through the teenage years.”

In a separate study, Stanley and his colleagues are looking at genetic data for 400 HI patients, seeking to understand the genetic causes of the disease and how specific genetic mutations can predict its course. They expect to publish their findings shortly.

Today, Logan is a happy third grader who loves skiing, horseback riding, rock climbing, karate and, perhaps most of all, the Red Sox. His HI is well-controlled with medication and careful monitoring.

“We’re glad to be supporting such an excellent research center and hope that we can help other children have an easier time with the disease,” says his mother.

Connection to CHOP Came Just in Time

“She’ll get better; she’ll outgrow it.” Over and over, doctors back home in Montana said what should have been comforting words to Shauna and Kayle O’Brien as their newborn daughter’s blood sugar levels stayed dangerously low. When Ellianna was born in the tiny hospital in Plains, Mont., her glucose level was 14 (normal is 80 – 100), and she was soon sent to Community Medical Center in Missoula, Mont., which has a 16-bed neonatal intensive care unit.

“She kept getting worse and worse,” Shauna says, remembering those first days of Ellianna’s life. “The doctor increased the amount of glucose, and they tried diazoxide. She retained more fluid and looked awful.”

That’s when Kayle found an article about congenital hyperinsulinism (HI) by Charles Stanley, M.D., director of the Congenital Hyperinsulinism Center at CHOP, on the Internet. “This is exactly what’s going on,” Kayle told his wife. They took the article to the local doctor, who didn’t agree. After all, 99.99 percent of the time low blood sugar is not a result of HI, which is so rare it occurs in only one of 25,000 – 50,000 births. “It was frustrating; we felt no one would listen to us,” Kayles says.

“We kept praying,” says Shauna, “and our prayers were answered — just not the way we thought they were going to be.”

Through a distant relative by marriage, they were connected to a family whose son’s HI was treated at CHOP. The child’s mother, Rachel Riley, put the O’Briens in direct contact with CHOP’s HI Center. Endocrinologist Andrew Palladino, M.D., called the local doctor, and before long arrangements were made



Ellianna at 2 months old, about a month after surgery cured her HI

to transport Ellianna to Children’s Hospital for care.

Flight nurse Mike Glady, R.N, arrived in Missoula two days later to care for Ellianna on her cross-country journey. “It wasn’t scary,” Shauna says. “He had the protocol; he knew what to do.” Shauna’s confidence was reinforced once she arrived at CHOP. “Everyone was on the same page,” she says. “In the emergency room when we came in and in the N/IICU, every nurse here knows how to handle babies with HI.”

When a PET scan showed Ellianna had focal disease, surgery was set. Surgeon N. Scott Adzick, M.D., M.M.M., removed the large lesion in the body of her pancreas, and at 21 days old, Ellianna was cured.

“The whole time we were at CHOP, the doctors and nurses explained everything that was happening,” Kayle says. “They asked us what we thought, and they listened to what we told them. Dr. Palladino — he’s wonderful. We can’t thank everyone enough for everything they did.”

“We feel so blessed,” says Shauna. “Sometimes you need to be persistent and keep advocating for what you feel your baby needs. It was an emotional rollercoaster, but we ended up with the best result possible.”

If you’re ever in Plains, Mont., population 1,248 — make that 1,249 — be sure to stop in the Mangy Moose Mercantile and say hello to the O’Briens, proprietors. They’ll be the ones smiling with the now-healthy baby.

Childhood is
a *gift* and you
can *give it*.

*For information about giving to the
Congenital Hyperinsulinism Center, contact*

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CHOP Ranks No. 1 in Endocrine



The Children's Hospital of Philadelphia was rated No. 1 in treating endocrine disorders, of which hyperinsulinism is one, in *U.S. News & World Report's* Best Children's Hospitals issue. CHOP was one of only eight pediatric hospitals named to the magazine's elite Honor Roll.

Reminders for HI Parents

Even though you may live far away from Children's Hospital, we want to hear how your child is doing! Please ask your pediatrician and local endocrinologist to send us copies of your child's visit letters. And remember, because of the risks associated with hypoglycemia, we recommend that every child with HI undergo formal developmental assessments at 2 and 5 years of age. Your pediatrician should be able to recommend a developmental pediatrician for your family to see.



Have you watched *Hope Revealed*, our video about congenital hyperinsulinism?

[View it from hyperinsulinism.chop.edu](http://hyperinsulinism.chop.edu)

Keep the connection.

