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Focus on Post-operative Diffuse HI Patients

For hyperinsulinism patients diagnosed with diffuse disease who cannot be managed with diet or medication (approximately 50 percent of severe HI patients), the best treatment option is frequently a near-total pancreatectomy. What happens after surgery varies from patient to patient. For some, the 2 or 3 percent of remaining pancreas still produces so much insulin the child requires medication to manage low blood sugars. Other patients manage their glucose through diet. The remaining children need insulin to control high blood sugars, as a diabetic does. But even those on insulin have a trickier task than a typical child with type 1 diabetes because the pancreas may produce some insulin now and then, making managing the disease unpredictable. Often, children go through some or all three scenarios as they grow. This issue is devoted to those patients with diffuse HI who needed surgery and how they successfully cope with chronic blood sugar problems.

'So Much Better Now' After HI Surgery



Max is active in sports.

Frustration and fear led Max Aronoff-Sher's parents to bring him to The Children's Hospital of Philadelphia's Congenital Hyperinsulinism (HI) Center. "We couldn't get answers in California," says Max's mom, Jessica. "We knew we'd be in good hands at CHOP."

Max arrived at CHOP with extremely low glucose levels. Medication didn't raise them, and when a PET scan indicated Max had diffuse disease, his parents knew a pancreatectomy was the best option. When he was 5 weeks old, 97 percent of his pancreas was removed.

After a difficult few months, during which Jessica and Larry fed Max every three hours day and night, they began giving him continuous dextrose through a G-tube at night. "Then

we were all able to sleep through the night for the first time," she says. Max was also taking octreotide. Little by little, they lowered the dose of dextrose. He stopped taking octreotide. Today, at 5, his HI is managed completely by diet. He's active in sports and is already reading and doing arithmetic.

"This is the best I could have hoped for in the scary days," his mother says. "His sugar levels are still unpredictable. He'll eat the same meal and his body will react differently" because what's left of his pancreas still produces varying amounts of insulin. "You can't really let your guard down fully."

Down the road, Max may require insulin. "I don't worry about it," Jessica says. "It's so much better now than it was. And in five or 10 years, who knows what advancements in treatment they'll find?"

Notes on Nutrition: The Diffuse Disease Rollercoaster

By Susan Becker, B.S.N., R.N.; Amanda J. Lee, M.S.N., C.R.N.P.; and Linda Steinkrauss, M.S.N., C.R.N.P.

Children with diffuse KATP hyperinsulinism present a unique nutritional challenge. As babies, they often require a subtotal pancreatectomy, yet 57 percent still have hypoglycemia. Parents naturally focus on preventing low blood sugars. Babies may be on medication and may depend on overnight dextrose through a feeding tube. This can lead to the infant always feeling full and never establishing a normal sense of hunger. Nutritional deficiencies are possible. Many babies will require speech and feeding therapy, which should be started as soon as a problem is found.

As children grow, their HI tends to stabilize, requiring less medical support. For a time, the remaining pancreas may secrete enough insulin for the child's needs. They can also tell you when they feel hypoglycemic. Many parents are able to check blood sugars far less frequently.

Children with diffuse KATP can eventually progress to diabetes. For about one-third of post-operative patients, this trend to hyperglycemia, or high blood sugars,

happens shortly after surgery, and patients go home on insulin therapy. For others, it can occur when their growth and nutritional needs outpace what insulin their bit of pancreas can provide; often this is at puberty.

Feeding a child with recessive diffuse HI can be an ongoing rollercoaster. Parents go from feeling that their child **MUST EAT** to prevent blood sugars from being too low to feeling that they must limit their child's food, counting every gram of carbohydrates, to prevent high blood sugars.

One of CHOP's guiding principles is that all patients should eat as normally as possible — whatever their age, stage or type of HI. With newborns prior to surgery, we manage blood sugars with intravenous dextrose and let them feed on demand by mouth, like any patient. This can stave off feeding issues after surgery.

Therapy at any stage may require a fasting test and treatment adjustments based on labs. A nutrition consult is a critical part of these visits, to assess the child's food preferences and to create an individualized diet plan that works for the child and family.

We know this can be challenging, and we are here to help. As we see more and more children with HI grow into adulthood, we learn more about the natural patterns of each therapy choice.

Hope on the Horizon: Research News

The Congenital Hyperinsulinism Center has numerous promising research studies under way. One example:

Research Continues on How HI Is Inherited

All patients referred to the Congenital Hyperinsulinism Center at The Children's Hospital of Philadelphia and their parents are invited to participate in continuing research into the mechanisms of how HI is inherited. There are several ways genetic defects causing HI are known to be passed from parents to children. These include recessive, dominant and sporadic inheritance patterns of four common genes. There is also an unusual phenomenon called loss of heterozygosity (LOH) which is associated with focal KATP HI. These can now be tested for in commercial labs. But approximately 40 to 50 percent of the time, those tests can be negative for any mutations. As part of ongoing research by Charles Stanley, M.D., we continue to look for additional genetic changes that may be associated with the conditions that cause hypoglycemia.

At least five additional genes have been reported in the literature in recent years which may be associated with HI. We continue to try to collect blood samples, and if we cannot get blood, other types of DNA samples such as saliva, to be able to categorize the cause of HI in as many patients in our care as possible. When positive results are found in our research lab, the institution's research review board (IRB) requires that the test be confirmed in a CLIA-certified clinical lab before the results are used for clinical or family-planning purposes. The results of genetic testing done under research are kept confidential; they are not part of the clinical chart, and are shared only with the affected individuals or their parent/legal guardian(s) in the case of a child. If you have questions about our genetics research, please contact Dr. Stanley or one of the research coordinators, Susan Becker, B.S.N., R.N., (beckers@email.chop.edu) or Stephanie Givler (givler@email.chop.edu) for more information.

HI Hope: Family Story

Navigating the Peaks and Valleys of HI



For Rianna Sommers, 17 years of tracking her blood sugars has been like a trek through the mountains: the highest highs and the lowest lows, with plateaus in between. That's how it is when you have congenital hyperinsulinism (HI) and need a near-total pancreatectomy to control the disease.

Rianna Sommers

"Sometimes I stumble," says Rianna, who sees her glucose

levels range from 30 to 400 on a normal day. "But I embrace my disease. I can't imagine myself without it. It's an active part of my life."

Rianna, from Plainview, N.Y., on Long Island, was diagnosed with hyperinsulinism at 10 days old and, when traditional drug therapies didn't control it, had 98 percent of her pancreas removed at The Children's Hospital of Philadelphia. The surgery made her HI much more manageable, and a controlled diet — supplemented by GI tube feedings — kept her from suffering seizures and other complications that could have affected her development.

Even when she needed to add medications to control her sugar levels — Precose® for highs — the 2 percent of her pancreas that remained kicked out enough natural insulin that she avoided insulin injections until she turned 16.

"I liked having normal HI," says Rianna. "That's what I was raised with. Now I'm more like a type 1 diabetic, and I treat myself with shots of insulin. I preferred it when I didn't have to give myself injections all the time."

But, as she has done since she was small, she's taken control of her care as it entered its latest phase. She administers the basal (long acting) shot of insulin each evening and bolus (short acting) shots before each meal. She counts carbs and monitors her glucose. "I've been pretty independent since I was 11, when I started e-mailing my doctors directly and making my own appointments," she says. "My mom always taught me, 'This is your life, your health. You need to take care of yourself.'"

"I'm a firm believer that kids need to understand the basic fundamentals of their disease," says Amy Sommers, her mother. "By 3, she was testing her own blood. By 7 or 8, she was setting up the pump for her tube feeds at night. I felt she needed to be as independent as possible."

That's the message Rianna shares with younger HI patients through a Yahoo group she started to open a discussion among kids with HI. "We have about 15 members from all over. I provide mentoring and try to answer questions from my own experiences," she says.

Rianna was also well prepared for her transition to Muhlenberg College this fall. She plans to study pre-medicine and become a pediatric endocrinologist. "It's not just because I have HI," she says. "I took an advanced placement honors class in biology and I loved learning all about the endocrine system. What can I say? I'm a nerd."

Muhlenberg, in Allentown, Pa., is a lot closer to CHOP than Plainview — whether Rianna is coming to Philadelphia for her twice-a-year checkups or if she were to need immediate care. "I love it at CHOP," she says. "When I tell my friends I'm going to the hospital, they say, 'That's too bad.' I tell them, 'No, it's great there. I always have such a good time.' Since I've been coming to CHOP for so many years, I know so many people, and it's always good to see them again."

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

*For information about giving to the
Hyperinsulinism Program, contact*

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Family Conference Draws HI Experts

On June 18 and 19, 2010, a group of 39 HI family members met with physicians and researchers from around the world in San Diego, Calif. The conference, sponsored this year by Congenital Hyperinsulinism International (CHI), a non-profit organized in 2005 to support, advocate and educate on behalf of HI families, was the third of its kind. CHOP sponsored the 2003 conference and co-sponsored the 2006 conference in Philadelphia.

Among the speakers this year were leaders in HI from around the world, including representatives from Children's Hospital: Charles Stanley, M.D., Amanda Lee, M.S.N., C.R.N.P., and Susan Becker, B.S.N., R.N. Others included Paul Thornton, M.D., from Cook County Children's Hospital, Fort Worth, Texas; Morey Haymond, M.D., from Texas Children's Hospital, Houston; Khalid Hussain, M.D., M.Sc., and Dunia Ismail from Great Ormond Street Hospital for Children and UCL Institute of Child Health, London; Jean-Baptiste Arnoux, P.H.T.P.A., from Hôpital Necker-Enfants Malades, Paris; and Kusiel Perlman, M.D., F.R.C.P.C., from Hospital for Sick Children, Toronto, Canada. David Zangen, M.D., Hadassah-Hebrew University Medical School, Jerusalem, Israel, was unable to attend in person, but sent a video sharing some of his recent research.

Highlights of the meeting included a session where several young adults shared their stories with other families and a session presented by CHI vice president Julie Raskin and Melanie Cohen from the CHOP Foundation about ways to advocate and fundraise for HI research and family support.

There was also plenty of time for informal discussion among families, as well as with medical professionals. Plans for future conference dates and locations were also discussed. Watch for more information in later newsletters.

CHOP Ranks No. 1 in Endocrine

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



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