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BROTHERS BORN WITH A BURDEN
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Dana Jobst loves watching her 3-year-old son Braden take out his plastic toy people and play pretend.

"Ashtin never did any of that," Jobst said of Braden's 7-year-old brother. "I am reminded all the time of how lucky we are."

Both boys have been diagnosed with Hunter syndrome, a rare and deadly genetic disease with no known cure. Left untreated, most boys with a severe case of the disease develop normally for their first few years and then regress until they end up in a vegetative state before dying, usually before reaching their teens.

The diagnosis came too late for Ashtin to be eligible for an experimental but potentially life-saving umbilical cord blood transplant. But because of his brother's diagnosis, the family tested Braden for the disease before any symptoms appeared. As a result, in 2005 Braden became just the fourth boy to receive the surgery at the Duke Medical Center in Durham, N.C.

It's been one year since Braden returned home after months recovering from the surgery in Durham. Since then, Braden has continued to develop and grow, while Ashtin continues to lose ground as the disease progresses.

Braden walks and runs like a healthy 3-year-old. Ashtin needs to wear hard plastic braces on his feet and ankles to help him walk properly.

Braden has a vocabulary of thousands of words and is picking up new ones and making sentences every day. Ashtin has lost many of the words he once knew and now uses just four or five.

Braden takes naps and sleeps through the night. At least once a week Ashtin is up all night, eating, watching TV and bounding around from room to room as though it was the middle of the day.

Ashtin's symptoms are common to Hunter syndrome. Braden's lack of symptoms is evidence his transplant has been a success.

"He's doing everything that a typical 3-year-old should do," Jobst said.

The brothers are among the fewer than one in 100,000 children worldwide diagnosed with mucopolysaccharidoses type II, commonly called MPS II or Hunter syndrome. They lack a critical enzyme needed to break down and remove complex sugar molecules produced by the body. Instead the molecules build up, causing progressive and permanent damage to body organs, including the brain, bones, liver and spleen.

Changing the diet doesn't change the prognosis, because the complex sugars made by the body aren't related to the sugars that we eat daily.

Instead, parents of children with Hunter syndrome have few options.

Children like Braden, diagnosed early and before symptoms begin to appear, might be eligible for an umbilical cord blood transplant now being done at Duke. The hope is that killing the patient's bone marrow and replacing it with stem cells found in healthy cord blood will trick the body to make the necessary enzyme.

So far it seems to be working for Braden. Before the surgery Braden had none of the enzyme needed by the body to break down and remove complex sugars.

Tests taken several months ago showed he now has a small amount -- 11 percent of what a healthy person has -- of the enzyme in his blood, which means his body is now making the enzyme for itself.

"Eleven is OK," said Dr. Vinod Prasad, of the pediatric bone marrow and stem cell transplant program at Duke. "I would like it to be as high as we can get it, but you really don't need to be at the 100 percent level."

Prasad said the key is that the transplant is functioning "absolutely normally" and his bone marrow is completely composed of donor cells.

"I could not expect anything better at this stage," the physician said.

In fact, Braden is doing so well that Prasad said they plan to wean him off the last of the transplant-related drugs he is taking, and within two to three months they hope to have him off all the medications.

Overall, Prasad said Braden is doing "wonderfully well."

"He was talking to me for the first time," Prasad said. "He never talked to me before. I was very impressed."

Because so few children have received a cord blood transplant, there's still no way to know whether it will have the desired effect: long-term production of the enzyme and

a reduction in sugars stored up in the body.

Children such as Braden's brother Ashtin, diagnosed after the onset of symptoms, are not eligible for the transplant. Instead their parents pin their hopes on treating the symptoms. Last year the Food and Drug Administration approved a new enzyme replacement therapy - - also known as ERT -- to treat Hunter syndrome. Ashtin started receiving the weekly infusions, which can take anywhere from three to 13 hours to administer, in October.

ERT won't cure the disease. But studies conducted before the drug was approved showed it might help boys with Hunter syndrome live longer.

It hasn't always been easy for the family.

During the first few infusions, Ashtin developed hives and his heart rate soared, forcing doctors to stop the procedure. Today, after more than 30 infusions, it seems his body is getting used to the drug and tolerating it better.

Jobst said it appears the synthetic enzyme is helping her son. Before he started getting the drug, Ashtin's liver and spleen were both enlarged, a typical symptom of Hunter syndrome. Today his liver and spleen are normal.

She also noticed that his skin seemed to be getting softer.

However, the drug can't pass into the brain, and, as a result, deterioration in mental capacity continues and the long-term life expectancy remains just into the teens.

The family hopes that for Braden, with a successful transplant behind him, the life expectancy ahead of him will be much closer to normal.

"We've come a long way," Jobst said. "But we still have a long way to go." *

WHAT IS MUCOPOLYSACCHAIDOSIS? (MPS)

Pronounced "Mew-ko-poly-sak-a-ride-dos-es," MPS includes seven diseases caused by a deficiency of a critical enzyme in the body.

WHEN ARE MOST CASES DIAGNOSED?

The severe form of the disease is usually diagnosed when boys are between the ages of 2 and 4. Mild cases may not be diagnosed until adulthood.

WHAT ABOUT TREATMENTS?

Children diagnosed early -- before stored sugars have been able to do too much damage to the body -- might be candidates for an umbilical cord blood transplant now

being done at Duke University Medical Center in Durham, N.C. For those who don't qualify, last year the U.S. Food and Drug Administration approved an enzyme replacement therapy developed by Shire Human Genetic Therapies of Cambridge, Mass.

HOW LONG CAN SOMEONE LIVE WITH HUNTER SYNDROME?

Most children with a severe case of the disease die before their 15th birthdays.

IS THERE A CURE?

No.

* More Hunter syndrome questions answered and how you can help on Page A8

ABOUT HUNTER SYNDROME

What is MPS II, also known as Hunter syndrome?

A rare condition affecting about one in 100,000 boys, it's named for Charles Hunter, a Canadian professor of medicine who first described two brothers with the disease in 1917.

WHY DON'T GIRLS GET IT?

Women carry the gene and might pass it on to their sons, but they don't get the disease. That's because the abnormal gene that causes Hunter syndrome is on the X chromosome. Because girls inherit an X chromosome from their mother and an X chromosome from their father, if one is abnormal there's a second one to compensate. Boys inherit one X chromosome from their mother and one Y chromosome from their father, so there is no second X chromosome.

WHAT HAPPENS?

Mucopolysaccarides are long chains of sugar molecules used to build connective tissues in the body. Usually the body continuously replaces used materials and breaks them down to be removed.

Children with Hunter syndrome are missing an enzyme called iduronate sulfatase, which is essential to breaking up sugar molecules. As a result, these molecules can't be removed from the body and instead build up in the cells, causing progressive and permanent damage.

WHERE CAN I GET MORE INFORMATION?

Check out the National MPS Society Web site at www.mpssociety.org.

HOW YOU CAN HELP

Medical bills continue to pile up for Ashtin and Braden Jobst. To make a donation to help the family:

Online: Go to the Children's Organ Transplant Association's Web site at www.cota.org. Click on "patient campaigns" and scroll down to Braden Jobst.

In person: At any Bank of America branch. Use account number 004351075816

By mail: Children's Organ Transplant Association, 2501 COTA Drive, Bloomington, IN 47403. Make checks payable to COTA for Braden Jobst.

PHOTO: Dana Jobst holds her son, Braden, at their home in Hampton. Braden and his brother, Ashtin, suffer from Hunter syndrome, an incurable genetic disease that kills most sufferers by their early teens.

Ashtin Jobst, 7, left, and his younger brother, Braden Jobst, 3, watch television at their home in Hampton. The two boys suffer from an incurable genetic disease that kills most sufferers by their early teens.

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