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HOPING FOR A CURE

DILLON PAPIER AND HIS PARENTS ARE FIGHTING AGAINST NIEMANN-PICK DISEASE

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URBANA—

“Is it time to go yet? We need to go to my game!”

It's a Wednesday afternoon, and 5-year-old Dillon Papier can't wait to get to his Little League game. He loves baseball and is rarely seen without a ball in his hand, and as game time approaches, he peppers his parents with departure time questions.

Some parents tire of such inquisition. But Darrile and Mark Papier are thankful for every bit of excitement Dillon shows, since they don't know how many more games they'll watch him play.

Dillon has Niemann-Pick disease Type C, one of only 500 cases worldwide, and only about 150 in the U.S. Those with the disease cannot metabolize cholesterol and other lipids properly, causing the material to accumulate in the liver, spleen and brain. The disease is always fatal, and the majority of affected children die before age 10.

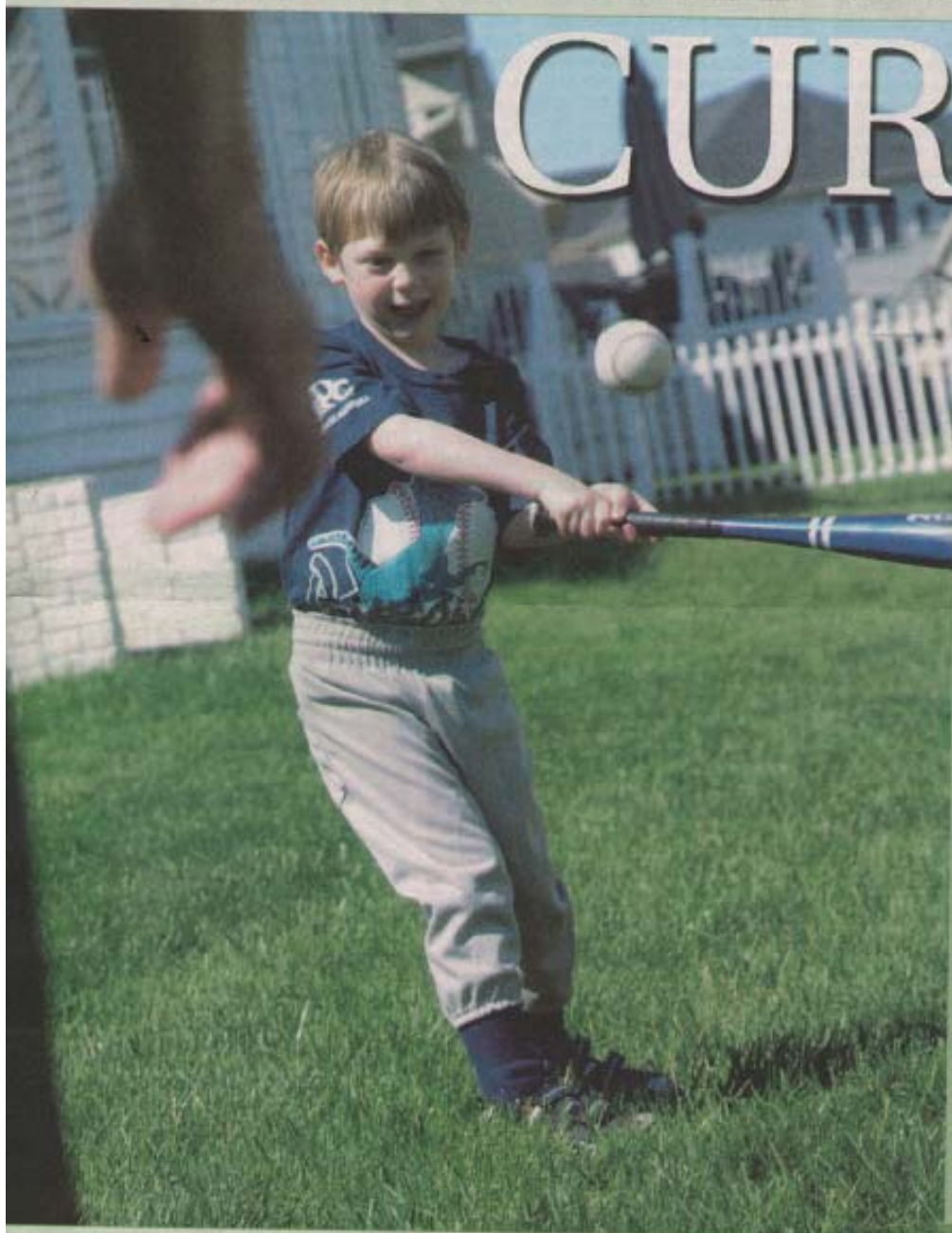
The disease causes Dillon's "sea legs," as Darrile says, causing him to fall at random times. He also has trouble climbing stairs and can't move his eyes without moving his head. While experimental drugs may slow the disease's progression, the Papiers know these problems will only get worse as he gets older.

Dillon's diagnosis came at age three, after two and a half years of testing.

Doctors first diagnosed 6-month-old Dillon with neonatal hepatitis and prescribed several medications. To their relief, the Papiers were told that 85 percent of children with the condition recover with minimal liver problems.

Over the next two years, Dillon's health

(See CURE C-2)



Staff photo by Skip Lawrence

DILLON PAPIER LOVES BASEBALL AND IS SEEN HERE PRACTICING WITH HIS DAD, MARK PAPIER.

“““ YOU LOOK FOR HOPE ANY WAY YOU CAN. THERE HAS TO BE A FIRST CHILD TO SURVIVE THIS; WHY CAN'T IT BE DILLON?”

DARRILE PAPIER

Cure

(Continued from C-1)

improved and he was at what was expected to be his last office visit focusing on the condition when doctors found his spleen was enlarged. They began testing for other conditions: leukemia, cancers. Everything came back negative.

When doctors said they wanted to test Dillon's bone marrow three days before his third birthday, Darrile said she knew something was wrong. The results indicated what's known as a lysosome storage disease, a condition that affects how the cells absorb certain molecules. Doctors suspected one of three types of Niemann-Pick disease, and did a skin biopsy to determine which one Dillon had.

Darrile said she and her husband immediately began researching the disease. If Dillon did have Niemann-Pick, they hoped it was Type B, which affects internal organs but has no neurological effects. They ruled out Type A, which they learned nearly always takes a child's life before age 2, and hoped against Type C, which affects both internal organs and the brain.

Meanwhile, Dillon's coordination was deteriorating.

"He couldn't walk down the sidewalk without falling down," Darrile said. "He'd fall every 15 steps."

When the biopsy results showed Dillon had Niemann-Pick Type C, Darrile and Mark contacted one of the only doctors in the nation focused on the disease and enrolled Dillon in a clinical study of Zavesca, a drug that researchers hoped would slow the disease's progression.

All three major types of Niemann-Pick are part of a group of about 60 genetic enzyme deficiency diseases that affect how the body breaks down certain molecules, said Dr. Marc Patterson, who led the Zavesca study and now works at Mayo Clinic. For some of these, like Gaucher disease, enzyme replacement therapy

is an option, but for Type C, there's no known effective treatment other than Zavesca. Researchers are currently studying the effectiveness of enzyme replacement therapy in Type B, and since it's lethal by age 2, there's no treatment for Type A.

While it's not a cure, Zavesca has been shown to slow the onset of Niemann-Pick Type C and extend the affected's lifespan. "This is the first treatment to have any kind of effect on the disease in humans," Patterson said. "We want to improve someone's quality of life for as long as possible, that's our goal."

After a month and a half on Zavesca, Dillon could run and walk more easily, even on grass or other uneven surfaces. Darrile said neither she nor his doctors are totally sure if it's because of the drug or whether Dillon had adjusted to his own clumsiness.

Zavesca's side effects can be debilitating, and several children in the study had to quit taking the drug because of severe diarrhea or tremors. In other children, the drug simply didn't work.

"Even when it's not working in other kids, you just start wishing, and you think maybe it works differently for each child," Darrile said. Dillon experienced some side effects, but the Papiers' all-natural, high-fiber diet helped control their severity, she said.

The Papiers also allowed Dillon to become part of a National Institutes of Health study group of a handful of children with the disease. The research is grueling,

WAYS TO HELP DILLON

■ **THE FIRST MONDAY** of each month, Foster's Grille, 8925 Fingerboard Road, Urbana, will donate 25 percent of all purchases to the National Niemann-Pick Disease Foundation and will match any amount raised through purchases.

■ **JUNE 22, SUBWAY** at 3531 John Simmons Street, Urbana, will donate 50 percent of its total sales to NNPDF. Donation jars are on display at Subway stores in Frederick and Montgomery counties.

■ **AT 6 P.M. JULY 13**, the Frederick Keys will play a benefit game for Dillon's sixth birthday. Tickets are \$10.

For details, call Darrile Papier at 301-607-4439 or e-mail papierfamily@gmail.com. For more of Dillon's story, visit www.dillons-fight.org.

as the doctors collect data through spinal taps, MRIs, and blood draws. But the parents of children in the study were encouraged by the thought that information collected from their kids could help families down the road.

"The kids are basically being used as guinea pigs," Darrile said. "Everyone knows they're doing this for the future."

Even while undergoing testing and taking Zavesca, the Papiers wanted to ensure Dillon's life was as normal as possible. Though other parents encouraged them to wait, the Papiers enrolled Dillon in kindergarten last year. The school has been helpful, Darrile said, allowing him a rest period each day since he loses his balance if he gets tired. Classroom aides help him climb stairs and hold books and papers where he can best see them, to accommodate his lack of eye coordination. While his first year went well, Darrile said the disease will eventually cause Dillon to lose his ability to retain information.

The family has also been able to count on the generosity of family and friends to raise money for research. Since Niemann-Pick Type C is so rare, it's known as an orphan disease, and grant requests for research are many times denied, Darrile said. Families affected by Niemann-Pick see grassroots fundraising as the only hope they have in someday beating the disease, and in the past year, the Papiers' friends, neighbors and classmates have raised more than \$95,000 for Type

C research. Their support gives the Papiers hope that a cure might be found in time to save Dillon's life.

"We have no control over this disease, but we have control over fighting it," Darrile said.

The Papiers and others with Niemann-Pick children hope to spark interest in the disease among well-known research facilities. Preliminary findings show Niemann-Pick's effects on the brain are similar to those of Alzheimer's disease, and the hope is that researchers will be willing to tackle the former as a way to find a cure for the latter.

As Type C progresses, problems increasingly affect the brain. Affected children eventually have trouble swallowing and speaking, about half have seizures, and about a quarter experience sudden loss of muscle tone when they're amused, Patterson said. Dillon's parents have already seen him collapse while laughing.

Some diagnosed with the condition during childhood live into their 20s and 30s, but with severe neurological problems, Patterson said. Others who are diagnosed in adulthood with what's known as Niemann-Pick Type E can live into their 60s, he said.

But while they've accepted the fact that their time with their son is most likely limited, the Papiers aren't hopeless.

"You look for hope any way you can," Darrile said. "There has to be a first child to survive this; why can't it be Dillon?"