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At what cost?

Controversial blood transplant offers hope to 2 local girls with fatal Tay-Sachs disease

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Julie Bihn watched helplessly as her 6-year-old daughter, Dakota, lay in her hospital bed, repeatedly digging her nails deep into her skin. Dakota was covered head to toe with a rash that triggered an unbearable itch. It was the latest side effect to erupt from an unorthodox treatment for her fatal disease.

Three months earlier, the kindergartner had been skipping down the halls of Falls-Lenox Primary School, her pink "Dora the Explorer" backpack and her long, blond pigtails bouncing, giggling with friends and repeating funny lines from the movie "Ice Age." Now, Julie couldn't remember the last time she had seen her daughter's blue eyes open.

It was Day 51 on a calendar that started when Dakota began treatment in January at Duke University Medical Center in Durham, N.C. It was how Julie, 37, and her husband, Ken, 41, had begun to mark time. The therapy had taken Dakota's hair and sapped her strength. Her parents suspected it had taken more of her brain function, too. She could no longer eat, walk or say more than a word or two. Now, Julie watched her daughter squirm from the rash and wondered, again, what she was doing to her child.

Dakota has Tay-Sachs disease, a genetic disorder in which harmful amounts of fat accumulate in her brain cells. The enzyme that is supposed to clean the cells is missing. As they fill with fat, the disease destroys Dakota's central nervous system and rapidly steals her mental and physical abilities. Certain populations, such as descendants of eastern European Jews, are at higher risk. But it's extremely rare, occurring once every 4 million births worldwide.

Rarer still is finding two girls living 25 minutes apart with the same diagnosis. In Amherst, 4-year-old Alexis Markowich also was diagnosed with Tay-Sachs. Her parents, Craig and Nichole, both 31, found out about her disease a month before the Bihns learned of Dakota's condition.

There is no treatment. That's what every medical Web site said. That's what neurologists at the Cleveland Clinic and a geneticist at University Hospitals Case Medical Center told them.

The families found each other by chance. Then they found Dr. Joanne Kurtzberg.

Kurtzberg, a research hematologist at Duke, said she would treat Dakota and Alexis. Transplanting blood from umbilical cords had helped babies with Tay-Sachs, the doctor said. She said data showed cord-blood transplants had helped kids with similar diseases.

Most doctors say transplants don't work for Tay-Sachs. Dakota's and Alexis' doctors advised the parents to enjoy the time they had left with their kids rather than subject them to the transplant, a brutal, high-risk procedure that would mean months in a hospital far from home. The children could live years without the transplant, some doctors said.

Ken got the impression from one local physician that he thought what Kurtzberg was doing was out of the mainstream. Another said it might accelerate the girls' disease and their deaths.

A pediatrician who examined Dakota and Alexis and who collaborates with Kurtzberg told the parents she didn't think a transplant would help. She said the girls would still be severely impaired.

But the parents knew they had to do something. Do nothing, Dakota and Alexis die. Do this, they might die, too. With those odds, what were they really risking?

Within the field of bone marrow and umbilical cord-blood transplants, Kurtzberg is known as a maverick, pushing the limits of cord-blood treatments for Tay-Sachs and other metabolic storage diseases -- inherited disorders characterized by a missing enzyme required for proper cell function. She's an avid proponent of using cord blood, and a cord-blood collection and processing bank operates out of her Duke office.

Kurtzberg considers such transplants to be the standard of care for Tay-Sachs, therapeutic rather than experimental.

Critics say her treatment is anything but established. They say it offers her young patients and their desperate families scant hope of benefit.

Kurtzberg said she had treated five babies with the disease. Two are dead. The others were alive but severely disabled as of July.

The doctor told the Bihns she had never done a transplant on an older child with juvenile Tay-Sachs, a different form of the disease than babies'.

Julie and Ken knew their other doctors believed no medical evidence supported doing a transplant. It didn't matter.

They just wanted to save Dakota.

Now, six months after her diagnosis, Dakota lay in a Duke hospital bed. She wouldn't let Julie hold her because she was in so much pain.

Julie wondered what her daughter was thinking: Did she remember home? The mural her grandfather painted of Beauty and the Beast and Cinderella in her playroom? Did she wonder when this would end?

For the first time since she had come to Duke, Julie wasn't sure Dakota would survive.

When Dakota, her second child, was born on May 30, 1999, Julie felt her life was perfect. Two beautiful little girls, a new house. All she ever wanted to be was a mom. She loved keeping house on the quiet cul-de-sac in Olmsted Falls. Ken's work in his family's automotive supply business allowed them to take frequent trips to the beach and Disney World. Charmed life. Bright future.

Julie first sensed something was wrong when Dakota was 18 months old. She stuttered and was clumsy. Julie tried not to compare Dakota to her older daughter, Bailey, but it was hard not to. Bailey talked like crazy before she was 2 and excelled in school, Irish dance and soccer. Julie told herself Dakota was just developing at a different speed.

In fall 2003, when Dakota entered preschool, she lost her ability to add single digits and could no longer remember her friends' names or do simple tasks like putting on socks or brushing her teeth. She suddenly feared climbing stairs and street curbs and taking a shower. Loud noises startled her. She stared, as though struggling to process what she saw.

Julie wasn't that worried. Dakota could still skip and run, though she fell a lot. And Julie thought the blank stares were just Dakota daydreaming.

When Dakota started dragging her right foot in the summer of 2004, Julie called a pediatric neurologist, who did some tests and referred her to the Cleveland Clinic.

Tay-Sachs is so rare that most neurologists will never see a case their entire career. For a year, Dakota saw the Clinic neurologist every three months. Each visit, the doctor ordered tests. The Markowiches followed a parallel track.

Between the Clinic visits, Dakota worsened. In August 2005, the neurologist told the Bihns something "strange" had turned up in Dakota's urine test, and he referred them to Dr. Marvin Natowicz, a Clinic neurologist who specializes in inherited disorders like Tay-Sachs.

The date of the Bihns' appointment with Natowicz is etched in Ken's memory: Sept. 1. Natowicz walked into the exam room, his eyes to the floor, holding a slip of paper -- Dakota's latest test results.

Natowicz started with a lesson in cellular biology. The disease would wreak havoc on her central nervous system. Her speech, swallowing, walking, brain function would diminish and eventually cease. First she'd be in a wheelchair, then go blind and deaf, and then dementia would set in. She'd eventually need a helmet because her seizures would be so severe, Natowicz said.

"Will she live until she's 30?" Ken asked.

Natowicz shook his head no. Julie choked back a sob.

Some make it to 15, he said. Some, he emphasized.

"I don't know the pace or how long she has," Natowicz said.

That evening, after the Bihns put their girls to bed, Ken sat in the dark in their family room, staring at the TV. He was numb. How could he have missed that his daughter was this sick?

Julie walked in and began speaking, fast. "We need to have a family picture taken we need to buy Dakota a new bed for her room because she has a loft bed with a little slide on it and I'm going to have to be able to crawl into bed with her and I can't do that with her now and I don't mind the wheelchair or the ventilator but I don't ever want Dakota to wear a helmet."

That her daughter wearing a helmet concerned Julie made Ken smile. Ken stood and hugged his wife. He held her in the dark as she cried.

"OK, Jules, no helmet," he assured her.

"Oh God, Ken, what are we going to do?" Julie moaned. "Oh God, what are we going to do?"

Serendipity. A friend handed Julie an article about Alexis Markowich. She was going to Duke for treatment. Julie read it halfheartedly. It had been only a week since Dakota's diagnosis. She couldn't care less about anyone else.

Two days later, another friend handed her a flier about a fund-raiser for Alexis. Julie called the National Tay-Sachs and Allied Diseases Association, and the nearby family came up again.

Because the disease is rare, it was very unusual to have two kids with Tay-Sachs so close, the woman there said. Julie called the number on the flier.

When the families met, they were struck by how much alike the girls looked. They walked stiffly. Their long blond hair, blue eyes, disease-frozen faces and smiles -- they were identical. So were their blank stares.

Alexis' disease had progressed further. She had problems breathing and eating, often choking on food. Nichole told Julie about Kurtzberg, whom she found on the Internet. The Markowiches already had been to Duke.

"The docs at Duke really know this disease," Nichole said. She told them Kurtzberg was taking care of the insurance paperwork. On Craig's steelworker salary and her part-time job at an optical store, there was no way they could afford the \$600,000 their insurance company said it would cost.

The Markowiches would leave for Duke in a few weeks, if insurance came through.

Ken was pumped. The next day, he e-mailed Kurtzberg. He wondered why no doctor had mentioned anything about the transplants at Duke. Was it because they didn't know about it? Maybe professional jealousy?

As he clicked "Send" on the e-mail, he thought Dakota's prognosis wasn't as dire as he had feared.

Things were looking up.

A few weeks later, Ken phoned Natowicz. Ken couldn't find much information about the Duke program and wanted Natowicz's opinion.

Natowicz said he couldn't find any evidence that cord-blood transplants for Tay-Sachs were effective. He recommended not doing the procedure.

That wasn't what Ken wanted to hear.

"If this were your child, what would you do?" Ken asked.

Natowicz paused and said he would try to make her remaining years comfortable. He told Ken that the complications caused by a transplant were a painful way to die.

"Is it as bad as dying from Tay-Sachs?" Ken asked.

"No," Natowicz said.

That did it. Ken had to do this.

Ken hung up and told Julie that Natowicz was against the transplant and ticked off the reasons: Because it would separate their family for long stretches. Because Dakota would be in the hospital for at least four months and have to stay near Duke for months after. Because heavy doses of toxic chemotherapy before the transplant would make her sick and weaken her so much she might never recover. Because, even if she survived, it might not work.

Julie didn't want to put Dakota through chemo. She stomped away.

Ken couldn't believe Julie was being so uncharacteristically stubborn.

"What are we really risking here, Julie?" Ken asked.

Julie was doing the math: If you had only so much time to spend with your child, do you spend those last precious months isolated in a hospital far from home, watching your daughter lose her hair, vomit, writhe in pain and then possibly die anyway?

Julie couldn't go through with something that might do Dakota more harm than good. Dakota was so happy, smiling and giggling all the time. Natowicz told Ken that with no treatment, it could be years before she was bed-bound.

It had been two weeks since Ken met with Natowicz. Ken tried to get Julie to talk about going to Duke. Julie wouldn't.

Ken started to resent her. He worried that if he persuaded her, she'd never forgive him if Dakota died because of the treatment. He knew he'd have a hard time forgiving her if they didn't do it and Alexis lived and Dakota didn't.

Julie was sick of fighting. She agreed to let Ken take Dakota to Duke.

"Only for the testing, Ken," Julie insisted.

Days before Ken was scheduled to leave for Duke, Julie decided to go along. She had to meet this doctor and hear what she had to say. She was afraid Ken would give it a positive spin.

Kurtzberg had told Ken in e-mails that for her to treat Dakota, the damage from the disease couldn't have put her developmental age more than two years behind. Their first appointment, on Oct. 25, was with Dr. Maria Escolar, a pediatrician who specialized in development at the University of North Carolina in nearby Chapel Hill. She would evaluate Dakota.

During the testing, Dakota made a lot of mistakes. Julie was scared Dakota would flunk.

Kurtzberg's criterion played in her head. It wasn't that Julie had changed her mind; she just didn't want to be told that her daughter was sicker than she thought.

Escolar told them their 6-year-old had declined four years developmentally. She pointed out that Dakota's arms were turning inward, and she used them to demonstrate how eventually her wrists would lock and she would be unable to use her hands. She showed Julie and Ken Dakota's feet and pointed out her toes, which also turned inward. That meant she'd probably be unable to walk within nine months, Escolar said.

Julie felt she was punched in the gut with every word.

All of this was impossible for Julie to imagine. Dakota still took the bus to school and found her way to class. She could read a few words, write her name, color and laugh.

Escolar said there was no good reason to do this transplant.

She said her and Kurtzberg's own research supported not doing Dakota's transplant. The brain damage was already too advanced.

During chemotherapy, the transplant and the heavy drug regimen that followed, the disease would continue to progress. Dakota would slip even more. Ken later remembered Escolar saying she thought the procedure might kill Dakota.

The doctor said the treatment didn't guarantee Dakota would be able to learn new skills. Dakota's brain damage was too extensive for new cord-blood cells to repair, Escolar said. In fact, she solemnly informed them, more than likely, if she survived, Dakota would be in a vegetative state.

"What are you really saving?" Ken remembered Escolar asking.

Ken and Julie were stunned. Ken worried this doctor would stand in the way of Dakota getting a transplant. Julie was terrified. Dakota was really sick. She had less time left than they thought.

Ken and Julie debated whether to cancel more tests scheduled at Duke the next day. The Bihns were certain Kurtzberg would not accept Dakota for a transplant. But they decided to keep the appointments so they could get a full assessment of Dakota's condition.

As they waited, Kurtzberg came to greet them.

She wore wire-rimmed glasses and baggy, tan corduroy overalls with an aqua-striped turtleneck, just like one Julie wore in high school. She was smaller than Julie expected, coming only to Julie's shoulder.

The doctor plopped down, sitting cross-legged on the floor in front of Dakota. She leaned forward to talk. Dakota smiled big. She seemed to really like this doctor.

Julie trusted Kurtzberg immediately.

Two days later, the Bihns met with Kurtzberg again. She seemed to know the most about metabolic storage diseases of all of Dakota's doctors. She explained the cord-blood transplant in terms they understood.

Kurtzberg told them a transplant might prolong their daughter's life and, if they were lucky, she'd be able to learn new skills, Ken recalled.

The doctor made no guarantees. She said that with juvenile Tay-Sachs, she had no data to support whether this would help.

Ken told her that if his daughter could smile and maybe even giggle a little after the transplant, he would be satisfied.

Kurtzberg watched Dakota walk. She listened to her talk. She played with her.

"She's much better than I thought," Julie later recalled the doctor telling them.

Even with the tests showing how far Dakota's disease had progressed, Kurtzberg agreed to do the transplant. She told them she thought Dakota knew and understood far more than she could communicate, Julie remembered. Julie felt her shoulders relax.

Julie said Kurtzberg asked them if they were happy with what Dakota was able to do right now. They nodded yes. More than likely that's where she's going to be afterward, Julie recalled Kurtzberg saying.

Ken looked at Julie. He was about to suggest they go home to Ohio and think about it.

Julie didn't give him the chance. "Let's do it," she said.

"Like, now?" Ken asked.

Julie turned to Kurtzberg and said, "We'll do it, we're in."

She was determined to prove Escolar wrong.

"How soon can Dakota start?" Julie asked.

Monday: Ken and Julie arrived at Duke, confident. Most experts weren't. Doing transplants for Tay-Sachs is not widely accepted.

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