

# “WE’LL NEVER GIVE UP ON BETHANY”

Five years ago the Toughill family didn't have much to be thankful for. They had just heard the crushing news that their baby had a rare and often fatal disease. This is their story.  
 BY MARGARET JAWORSKI

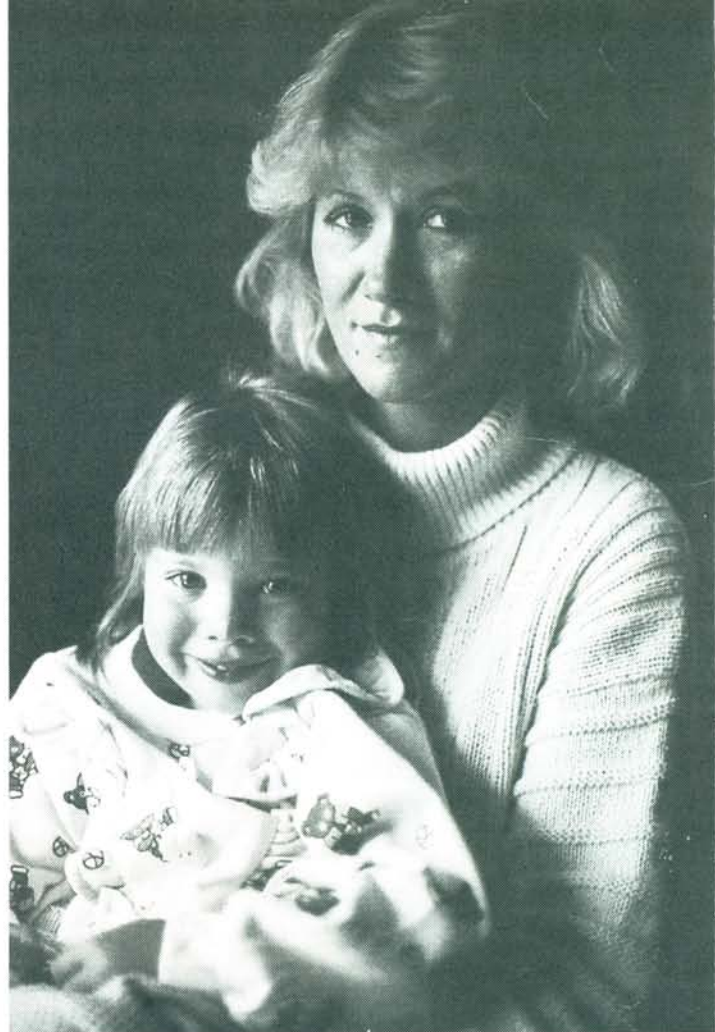


**I**n September 1985, 10-year-old Jennifer Toughill wrote an essay about her younger sister for her fourth-grade English class. It wasn't about sibling rivalry or any of the usual disagreements between sisters. It was about something far more serious: *"Bethany has a rare disease called histiocytosis-x. It is similar to cancer. She was 4 months old when we found out. She had to go on chemotherapy*

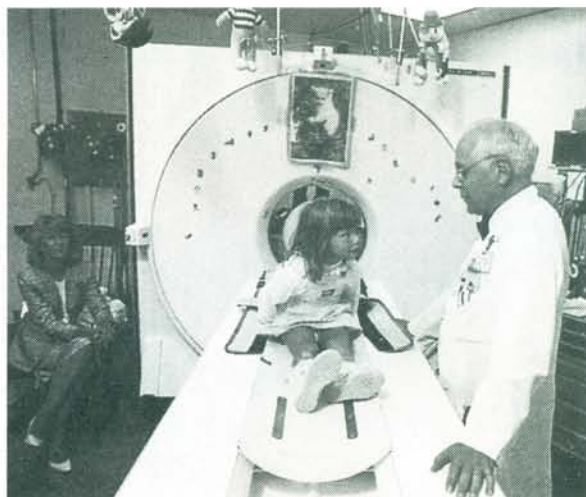
*for a year. She also had a tumor in her head and had to have radiation therapy. I prayed every night that she would be alive in the morning. God answered my prayers. She is now 3 and doing great."*

Two years later it seems the prayers are still working. Scampering around the dining room of her parents' home in Glassboro, New Jersey, Bethany looks like any other healthy 5-year-old. It's hard to believe that this rosy-cheeked child with sparkling hazel eyes—wearing miniature Frye boots and offering her view of "good" banks (the kind that give out lollipops) and "bad" banks (the kind that don't)—is sick at all. It's only when she points to the tiny blue dots above and below her left ear and says, "That's where they radiated me," that the reality hits. An invisible enemy lurks inside her body. It could attack at any time. It could kill her.

Histiocytosis-x is a mysterious and frequently fatal blood disease that affects the body's immune system,  
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*"I try not to dwell on what the future may bring,"* says Sally Toughill, *above*, cuddling her daughter Bethany. *"We cling to hope."* *Left:* *"I want my sister to grow up like any other kid,"* says 12-year-old Jennifer as she combs Bethany's hair. *Below:* *In the CAT scan room at Children's Hospital in Philadelphia, Bethany chats with Dr. Giulio D'Angio, director of the cancer center.*



allowing a type of white blood cell called a histiocyte to multiply wildly and attack vital body organs. Its cause is unknown, its progression unpredictable. One third of its victims die within two or three years after diagnosis.

The disease is so rare—affecting only about 2,000 people in this country (60% of them children)—that it is known in medical circles as an "orphan" disease, one too obscure to generate substantial research. That means that potentially life-saving treatments may never be available for Bethany.

### **The First Sign**

It started with a rash. "I thought it was just a case of prickly heat," says Bethany's mother, Sally, recalling the morning she first noticed the small crusty red scales clustered on her 6-week-old daughter's head. On that summer day in 1982 it seemed inconceivable that it could be anything more serious. Of Sally and Jeff Toughill's three children, Bethany was the healthiest and least troublesome. When the older two, Jennifer, then 7, and Jonathan, 5, were babies, they'd had a succession of maladies: "Colic, allergies, you name it," their mother recalls, "but Bethany was a dream baby from the first. She took to people right away and she was always smiling and giggling."

That August was especially hot and humid; so Sally wasn't alarmed when the rash persisted. But at Bethany's routine two-month checkup, her pediatrician suggested Sally consult Melvin Gruber, M.D., a local dermatologist.

"Dr. Gruber didn't seem overly concerned at first," Sally remembers. "He thought it was just a bad case of seborrheic dermatitis, a common skin disorder." He prescribed some medication, but the rash didn't respond. In fact, over the next two weeks it spread to Bethany's chest.

New, stronger medications didn't help either. And by October the smiling, bright-eyed baby had become listless and suffered from terrible attacks of itching. "Sometimes she would scratch until she bled," says Sally.

Increasingly concerned, Dr. Gruber did a skin biopsy. A week later he asked the Toughills to come to his office. The biopsy had confirmed his unspoken suspicions: "Your daughter has a disease called histiocytosis-x," he told them. "She needs immediate treatment. Anticancer drugs may help..."

The very next day Sally and Jeff found themselves carrying 18-week-old Bethany into the oncology clinic at Children's Hospital in nearby Philadelphia. In one of the small white examining rooms, pediatric oncologist R. Beverly Raney, M.D., a gentle and warmhearted man with a Southern drawl, examined her. The rash was severe, he said. They would have to run some tests in order to determine the extent of the internal damage.

X-rays revealed the urgency of the situation. Like tiny marauders, the histiocytes were waging war inside Bethany's body. Already the aberrant white blood cells had eaten away bone—in her skull over the left ear and in her left leg—and invaded the lymph nodes on both sides of her head. Dr. Raney explained they had only one choice: to begin chemotherapy at once.

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But the drugs, he said, could be administered at home. Bethany would not have to stay in the hospital.

Fourteen times a day Sally doled out the drugs. "The pills were bitter, and Bethany would spit them out," she remembers. "So I crushed them and mixed them with vanilla ice cream. I kept thinking how ironic it was: My healthy breast-fed baby was digesting these horrible chemicals.

"During those first few weeks, Jeff and I never acknowledged to each other that Bethany might die. Then one day he sat me down and said, 'We have to prepare ourselves. She might not make it.' Well, I didn't want to hear that. I could only deal with the present, and that was bad enough."

The uncertainty about Bethany's future affected the whole family. Where once the house had been full of laughter and activity—games, pajama parties—there was now sadness and fear. Jeff, an ex-cop turned security chief at Glassboro State College who had left the police department after six years because "I'd had my fill of tragedy," suddenly found himself facing one of his own as he watched his golden-haired daughter fight for her life.

"There were times when the fear became overwhelming," Sally confesses. "I was obsessed with the thought that Bethany could be gone at any moment. I was so wrapped up in her that even Jennifer and Jonathan took a backseat. Then one day I was sitting in the oncology clinic and heard a nurse call to a little girl named Jennifer. And it hit me—it could have been *my* Jennifer. This could happen to her too."

Five months into the Toughills' ordeal, X-rays revealed that Bethany was responding well to the chemotherapy. "It gave us hope that she'd make it," says Jeff. "We clung to that hope."

### **A New Year, A New Beginning**

On January 4, 1984, Bethany was taken off chemotherapy. The swelling in her face had begun to recede, and the sores in her mouth were gradually healing. "We felt as though we'd come through the worst of it," says Jeff. "God had given us a new beginning, a chance to get our family life back to normal."

During that year, life did get back to normal. Histiocytosis-x can sometimes go into spontaneous remission. There was a chance, doctors said, that Bethany's disease would not recur. Her energy returned, her eyes sparkled again.

But the respite was short-lived. In early 1985 Bethany's behavior underwent a noticeable change. The easy-going, even-tempered 2½-year-old began to experience abrupt mood swings. She also developed an insatiable thirst, and she drank glass after glass of water—only to have it pass through her system in minutes.

On February 3 Bethany was admitted to Children's Hospital for tests. A CAT scan showed the histiocytes had attacked again—she had a tumor behind her left eye, near the pituitary gland. Her bizarre symptoms were caused by damage to the pituitary—which, among other functions, regulates the body's production of urine. This time the doctors decided to try to shrink the tumor with minimal doses of radiation.

On February 19 at the Hospital of the University of Pennsylvania, Bethany underwent the first of three radiation treatments. Each session lasted about 30 minutes, but to Sally and Jeff that half hour seemed like "a century." They watched helplessly as their daughter,

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just a few months shy of her third birthday, disappeared into a pristine room dominated by a mammoth cone-shaped machine. As the door closed, they heard her cry out.

The radiation managed to shrink the tumor, but the mood swings and episodes of excessive thirst persisted. In March an endocrinologist at Children's Hospital diagnosed the problem. Bethany had developed diabetes insipidus, a complication that occurs in 25% of children with histiocytosis-x. In Bethany's case the condition was irreversible. For the rest of her life, she would need twice-daily doses of a synthetic antidiuretic hormone to regulate her water balance.

Soon after Bethany finished radiation therapy, Sally began volunteering one day a week in the intensive-care unit at Children's Hospital. "I needed to keep myself busy," she says. That's when she heard about the Candlelighters Childhood Cancer Foundation, a voluntary organization that provides support for cancer victims and their families. It gave her an idea: Why not start a support group for the victims of histiocytosis-x?

"Finding out that your child has a life-threatening illness is bad enough," Sally says, "but when you feel you're the only people in the world experiencing that particular anguish, it makes the ordeal that much worse. If we could help others, then perhaps Bethany's illness might have some meaning." She and Jeff placed a notice in the Candlelighters bulletin asking anyone who had been through their experience to write to them.

In August a letter arrived from Michael Osband, M.D., chief of the Division of Pediatric Hematology-Oncology at Boston University School of Medicine. Dr. Osband had been studying histiocytosis for 10 years and had recently received funding from the U.S. Food and Drug Administration to test the effectiveness of an experimental hormone treatment.

"We were heartened to learn that there was research going on," Jeff says. "Dr. Osband offered us his professional resources as well as his personal commitment." Soon families affected by histiocytosis also began writing.

The following August (1986) the Toughills mailed out the first monthly newsletter of their newly founded National Histiocytosis-x Association. It is

still a kitchen-table operation run by the Toughills and a few volunteers, but the newsletter now offers information, advice and comfort to almost 200 families. "We get calls and letters from all over the United States and from Europe and Africa," Jeff reports. "Each one is answered, sometimes with a letter, sometimes with a phone call. Sally usually makes the calls—I'm better at writing letters."

#### WHERE TO GET HELP FOR RARE DISEASES

An "orphan" disease is defined as any disorder that affects fewer than 200,000 people. In 1983 Congress passed the Orphan Drug Act, which authorized \$4 million in Federal funding to encourage clinical research and commercial development of therapies for rare disorders. According to experts, this law has made a difference, facilitating important new research and paving the way for a range of drugs previously unavailable in the U.S. The National Organization for Rare Disorders (NORD) was founded in 1980 to act as a clearinghouse for current information on such diseases. NORD refers patients to existing support groups and maintains a computerized Rare Disease Data Base, which offers medical advice and treatment information on 5,000 orphan diseases. Mail requests with a business-size self-addressed 39¢-stamped envelope to: NORD, P.O. Box 8923, New Fairfield, CT 06812. □

#### A "Normal" Life

"Bethany hasn't had a recurrence for a while now," says Sally with a sigh. "The doctors tell us her chances are good. I try not to dwell on what the future may bring, but sometimes it's all too hard to ignore. The slightest red spot on, her skin or white spot on her gums (in children, histiocytosis-x often attacks the gums) sends me into a panic. Those cells are still there..."

Every three months Bethany visits Children's Hospital for a checkup. She navigates the brightly painted halls like a pro, smiling at familiar faces, occasionally volunteering a shy hello. The routine is always the same: first a CAT scan, then blood tests, then a physical exam. It's a tedious full-day session.

At 5, she is now old enough to realize that she has a chronic disease. She talks

with uncanny authority about white-blood cells, endocrinologists, injections. She pronounces the name of her disease without the slightest hesitation. Even when she's told it's time to go to the hospital for another checkup, her face clouds over only for a split second. Then she catches herself and asks matter-of-factly, "When?"

Jennifer and Jonathan also display an understanding beyond their years. "Bethany and I fight just like anybody else," says Jennifer, now 12, "but I never stay mad at her for long. When I think about her being sick, I get all choked up. I just want her to grow up like any other kid."

Ten-year-old Jonathan agrees. "Sometimes I get jealous because she gets a lot of attention," he admits, "but I can understand why."

Meantime, Sally is speaking out for victims of other "orphan" diseases. In fact, last June she was cited by New Jersey Senator Bill Bradley, an honorary trustee of the National Histiocytosis-x Association, as one of the state's outstanding "women of the year."

Now 35, Sally is studying toward a nursing degree at Gloucester Community College—in between working part-time for a local doctor, mothering three active kids, fund-raising for the National Histiocytosis-x Association and helping with the newsletter. "Working keeps me busy—it's my escape, my therapy. And doing something for others helps me feel less helpless."

Jeff echoes the sentiment: "I'm not a doctor—I can't go out and find a cure for this disease. But I can contribute my energy and time to help get something done for Bethany and others like her. Just because Bethany has a disease that most people have never heard of doesn't mean that her pain is any less than that of a child with something better-known but equally horrible. We want the same chance for her that other children have. We need to know someone out there is looking for a cure." ■

**EDITOR'S NOTE:** For more information, write the National Histiocytosis-x Association, 609 New York Road, Glassboro, NJ 08028.

Margaret Jaworski, an editor-at-large of FAMILY CIRCLE, writes frequently about family, marriage and health.