

His life was saved because Wisconsin required testing babies for 'bubble boy disease.' Now that's standard in 50 states.

Mark Johnson, Milwaukee Journal Sentinel Published 5:00 a.m. CT Feb. 7, 2020



Dawson Bornheimer, 11, plays basketball in the kitchen at his home in Edgar as his mother, Melissa, watches. When Dawson was an infant, he tested positive for severe combined immunodeficiency and was cured with a transplant of umbilical cord blood, which is similar to a bone marrow transplant. (Photo: Mark Hoffman / Milwaukee Journal Sentinel)

Dawson Bornheimer is an 11-year-old who plays football just about every day, likes mac and cheese and shooting video game zombies and is such a picture of health that he can tell you with authority, "I haven't puked in two years."

"He probably can tell you the date," says his mother, Melissa Bornheimer.

Dawson's claim to fame is that in his first year of life he tested positive for a rare disease, launching a movement in newborn screening that has since spread across all 50 states and 20 countries around the world.

The long struggle to win widespread acceptance for the lifesaving test brought together a remarkable group of parents, children, doctors and health officials.

It began here in Wisconsin, in the small village of Edgar near Wausau, with a phone call.

Melissa Bornheimer was weeding in the family garden. Dawson, hazel-eyed and just 12 days old, lay sleeping in the stroller beside her, his baptism set to take place later that day.

On the phone was the baby's pediatrician. Dawson had tested positive for one of the almost 50 diseases for which all newborns in the state were screened.

The disease Dawson had was one that had just been added to the list. No one in the state had ever failed the new test.

In January 2008, six months before his birth, Wisconsin had become the first place in the world to screen all babies for a life-threatening illness called severe combined immunodeficiency, or SCID.

Most Americans, if they've ever heard of the illness, likely know it by a different name: "bubble boy disease." The colloquial moniker comes from a movie, "The Boy in the Plastic Bubble," which starred John Travolta playing a child whose name was David Vetter. Born in 1971, Vetter had a rare, usually fatal disease that crippled his immune system, leaving him so vulnerable to severe infections that he was forced to live in a sterile environment, or "plastic bubble."

He died at the age of 12.

As soon as she got off the phone, Bornheimer Googled the disease to see what Dawson was up against. What she read terrified her. SCID died before their first birthday.

The baby's baptism went ahead that afternoon at St. John the Baptist Catholic Church. "I cried all the way through the ceremony," Bornheimer recalls.

What she and her husband, Michael, did not realize then, what they would come to appreciate later, was that the test given to their son was actually good news.

It would save Dawson's life.

A long campaign to add screening

Another couple, Vicki and Fred Modell, had not been so fortunate with their son.

Jeffrey Modell was born on Sept. 25, 1970. When he was 10 months old, he began suffering from a high fever. Doctors discovered the baby had inherited an immune system disease called primary immune deficiency. His illness, though similar to Dawson's, was not as perilous in the short term.

Jeffrey's body would not manufacture white blood cells. While these cells normally make up only about 1% of all the cells in the blood, they are the main line of defense against viruses, bacteria and other foreign invaders.

The Modells were told that their son's condition was a time bomb. Any day, he could catch an illness, and, without white blood cells to fight it off, a common disease could easily turn fatal.

The couple decided not to keep their son sequestered from the outside world. Instead, Jeffrey grew up in Manhattan playing with other children, swimming, participating in softball and squash and sailing.

For years, his parents took him to see doctors all over the world. They had the money to do so because Fred Modell owned a successful wholesale diamond jewelry company for many years. None of the doctors who saw their son, though, was able to cure him.

A few days after attending a Princeton University football game, Jeffrey became very ill with pneumonia. He was placed on oxygen, then on a respirator.

"I am sure you are afraid," Vicki remembers telling her son, "but don't be, because we have each other and everything will be OK."

She remembers the tear that ran down his face. Soon after, Jeffrey drifted into a coma. He died on Jan. 7, 1986. He was 15.

Knowing the risk that loomed over Jeffrey's life had still not prepared the Modells for his death. For a long time, they could not say his name to each other.

In 1987, though, they found a way to say his name and to honor it. They founded the Jeffrey Modell Foundation to boost research and awareness of primary immune deficiency, a group of diseases that includes SCID.

The Modell Foundation produced posters showing the 10 warning signs of primary immune deficiency. They also began campaigning to have SCID included on the panel of diseases for which millions of American newborns are screened.

To the Modells, the decision seemed obvious. If SCID is detected early enough, it can be cured with a bone marrow transplant, which provides the recipient with a brand new immune system. At the time, however, the condition was thought to be very rare — affecting one in 100,000 births or fewer.

From 2001 to 2007, the Modells lobbied health officials, seeking to get SCID added to newborn screening.

"We failed in 2001. We failed in 2002. We failed in 2003 and on and on," Fred Modell recalled years later.

Better news came in January 2004 when a paper in the Centers for Disease Control and Prevention journal suggested SCID was a promising candidate for newborn screening. The potentially fatal disease could be treated effectively if detected soon after birth.

In 2006, during a meeting with state and national health officials, the couple ran into more resistance. One official told the parents the disease was very rare and the newborn screening decision was simply a matter "of the numerator and the denominator."

"Wait a second. Let's look at the numerator," Fred recalls snapping. "How many dead babies shall I bring to you? One? Two? Five? Ten? Tell me how many."

Because no one tracked the number of deaths from SCID, health authorities did not know how many lives they could save by screening for the disease.

There was only one way to find out. Someone would have to start testing babies.

Mother's death shapes medical career

Jack Routes was drawn to medicine by a family tragedy.

He was growing up in Indiana with five brothers and a sister when their mother died from an aneurysm, a ballooning blood vessel that burst. Routes was 11 years old.

"It blew up the world," he said. "My mom was the glue."

Her death was the reason he became a doctor. He could not shake this gut feeling that more could have been done to save her.

Routes focused on science in school and went on to earn his medical degree from Indiana University School of Medicine in 1981. He trained in internal medicine and conducted research in mice, examining basic immune responses to human adenovirus and human papillomaviruses.

Like many scientists who work with mice, Routes wanted to shift his research to humans.

In February 2005, while working at National Jewish Health in Denver, Routes read a journal paper that would change the course of his medical career.

The paper, in *The Journal of Allergy and Clinical Immunology*, discussed a test that could be used to diagnose children with SCID. The test worked by discovering when important cells in the immune system have not matured enough to do their job.

A healthy immune system depends on T-cells, which are made by the thymus gland. When these cells have matured, they contain small circles of DNA; absence of these circles of DNA is a hallmark of SCID.

After reading the paper, Routes remembers thinking, "I bet I could run with this."

He had never treated a child with SCID, but he saw the kind of opportunity that comes along only once or twice in a career.

In 2006, Routes was recruited to Children's Hospital and the Medical College of Wisconsin. Soon after arriving, he mentioned the idea of screening babies for SCID to Robert Kliegman, chairman of the department of pediatrics at Children's.

Wisconsin had a reputation as an early adopter of newborn testing. In 1985, the state became the second after Colorado to screen for cystic fibrosis, which damages the lungs, digestive system and other organs.

Why not take the lead on SCID?

\$250,000 pledge sets stage

In 2007, Vicki and Fred Modell traveled to a meeting in Atlanta. They were still smarting from their disappointment the previous year and still looking for someone willing to begin testing babies for SCID.

At the meeting, Fred Modell noticed two doctors he'd never seen before. After a discussion of the test for SCID described in the 2005 paper, one of the doctors stood up.

"I think I've seen enough," Fred Modell recalls the doctor saying.

The doctor was Ronald Laessig, who had directed Wisconsin's State Laboratory of Hygiene for more than 25 years. Laessig said he was confident the test would work, "but we won't know if we can do it unless we do a pilot study."

That's when the second doctor stood up. It was Jack Routes.

"I don't know how we're going to do it," Routes said, "but we're going to do it in Wisconsin."

Enthusiastic as he was, Routes was also honest. He told those in attendance that he did not know where money for the pilot test would come from, only that he would find it somehow.

Fred Modell raised his hand.

"We'll go half," he said, referring to the foundation named for his son.

The total cost would be about \$500,000; Routes would have to find the remaining \$250,000.

Children's Hospital joins effort

The search led him to Cindy Christensen, president and chief operating officer of Children's Hospital. Christensen had an "open door" policy. In 2007, Routes walked into her office.

He had just joined the hospital. Now he'd come to ask for \$250,000.

In her job, Christensen was accustomed to hearing from doctors with requests for money. There's never a shortage of promising research and life-saving equipment to invest in.

Her job often involved saying "no," and it was especially so in years like 2007 and 2008, the start of the worst economic crisis since the Great Depression.

"It was a time when we did have our belts pretty tight," Christensen said.

Still, when Routes told her about the idea of launching a pilot study to screen newborns for SCID she listened and did not reject the idea.

"He was a very vibrant individual with some far-reaching ideas about how to make things better for children," Christensen said. "He pitched this in such a way that you really believed this can happen."

Routes was back in her office just weeks later, this time with Fred and Vicki Modell. They told Christensen their story. Routes stressed that one test at birth could help families avoid the Modells' anguish.

"This is going to be really important," he said.

Christensen made her decision. The hospital would proceed with the project.

With money no longer an issue, Laessig at the Laboratory of Hygiene went to state officials and gained permission to conduct the pilot study.

Fine-tuning the test

Securing money and approval for the study didn't mean the state could start screening newborns right away. The test, as described in the 2005 paper, was not ready for use.

"It's like cooking," Routes said. "You have a recipe. You need to do troubleshooting in the lab."

The test produced some false positives, a problem that had to be corrected to avoid needlessly alarming families. Also, the test had to be adapted for use with technology that allows scientists to conduct millions of tests rapidly.

For six to eight months in 2007, Routes worked with two colleagues, James Verbsky and Bill Grossman, to adjust and prepare the test for use.

Screening of babies in Wisconsin began in January 2008.

Born on June 12, 2008, at Aspirus Wausau Hospital, Dawson Bornheimer became the state's first infant to test positive for SCID.

A transplant for Dawson

Dawson's family had returned from the baby's baptism when Melissa Bornheimer's sister, Kristine, took her outside to ask why she was crying. The new mother explained about the call from the pediatrician and the scary disease her baby had inherited.

Kristine hugged her. They would get through this, she said.

Later that day, Dawson began to run a fever. His umbilical cord, which had yet to fall off, reddened and appeared sore. The baby howled.

He was taken to Aspirus hospital where he was kept in isolation for three days. The baby was then placed on antibiotics and released.

On the first weekend in July, the Bornheimers drove north where they planned to celebrate the Fourth. But the soreness in Dawson's belly button worsened. The surrounding tissue turned black. A redness spread across his abdomen. When they called Aspirus, they were told that an ambulance would take the baby to American Family Children's Hospital in Madison.

Dawson had omphalitis, an infection of the umbilical stump; it was not an infection typically seen in children with SCID.

At the Madison hospital, Dawson had surgery to remove the infected tissue. The procedure left him without a bellybutton, just a tiny dimple where it had been. He was recovering after the surgery; the Bornheimers were packing their bags to leave the hospital.

Suddenly, they were face to face with a team of doctors, all gowned in protective clothing and masks. Something was wrong with Dawson's immune system. They needed to keep him in isolation while they ran more tests.



This was Dawson on the weekend of July Fourth. You can see the umbilical cord had not fallen off yet. It was very hot to the touch and looked infected (Photo: Family photo)

The baby stayed in the Madison hospital for more than two weeks, receiving antibiotics. He was referred to Verbsky, one of the Children's Hospital colleagues Routes had worked with on the SCID test.

In July and August, Verbsky examined Dawson, working to confirm the SCID test and reconcile it with the infection the baby had acquired. Verbsky also treated the child for anemia, a deficiency of red blood cells. The doctor was able to pinpoint the genetic cause of the baby's SCID to a single mutation on one of the 20,000 or so human genes: RAC2. Verbsky determined that Dawson could be treated with a transplant.

The Bornheimers learned their son would receive a transplant of umbilical cord blood, which is very similar to a bone marrow transplant. The procedure was set for Sept. 25, 2008.

Routes called the Modells to tell them what was happening. The campaign they started more than 20 years ago had led to a test that now promised to save the life of an infant in Wisconsin. The doctor invited them to Children's Hospital for the historic moment.

Vicki and Fred realized immediately the significance of the date. It was their late son Jeffrey's birthday. He would have turned 38.

The couple liked to stay close to home on Jeffrey's birthday. All these years later, the wound still felt fresh.

"Please," Routes said. "You've got to come out here."

A harrowing journey

The Modells flew to Milwaukee.

On the morning of the transplant, they were ushered into a meeting with doctors and state newborn screening officials. Then Melissa and Michael Bornheimer entered the room, both dressed in blue T-shirts imprinted with the words, "Dawson has big dreams."

Later a courier entered Dawson's hospital room carrying a cooler with the umbilical cord blood. The cord blood determined to be the best match had come to Wisconsin all the way from Germany.

As an intravenous line allowed the blood to seep into Dawson's thigh, the baby's mother and father took turns rocking him gently. Melissa prayed constantly. She prayed that the baby's tiny body would not reject the new immune system. If his body could just hold on for a week, it would give the cord blood time to start working.

Her fears were well-grounded. Bone marrow and cord blood transplants take place after a patient has been blasted with chemotherapy to wipe out their old immune system. Only then does the patient receive the bone marrow or cord blood that will build the new immune system.

This means the patient must pass through a dangerous period in which the old immune system is gone and the new one has yet to establish itself. During this time, the patient is vulnerable to all kinds of viruses and bacteria.

A week after the transplant, Dawson began suffering severe complications.

The Bornheimers were told that his organs were shutting down. He was sent to the pediatric intensive care unit. On his second night in the unit, doctors told the parents to begin calling relatives. Dawson wouldn't make it through the night.

"It was terrifying," Melissa said. "I didn't sleep the entire time he was in ICU."

Dawson lived through that second night, then a third.

On his fourth day in intensive care, tests showed new white blood cells. The cord blood was building his new immune system. By the day's end, Dawson was back in a regular unit.

The baby still endured bouts of vomiting. Blood showed up in his stool. But in January 2009, after four months in the hospital, the Bornheimers took Dawson home to Edgar.

The pilot study was over. Wisconsin had adopted SCID as a permanent component of newborn screening.

RELATED: (<http://archive.jsonline.com/watchdog/watchdogreports/Deadly-Delays-Watchdog-Report-Delays-at-hospitals-across-the-country-undermine-newborn-screening-programs-putting-babies-at-risk-of-disability-and-death-228832111.html>) The nation's newborn screening programs depend on speed and science to save babies from rare diseases. But thousands of hospitals fall short. (<http://archive.jsonline.com/watchdog/watchdogreports/Deadly-Delays-Watchdog-Report-Delays-at-hospitals-across-the-country-undermine-newborn-screening-programs-putting-babies-at-risk-of-disability-and-death-228832111.html>)

A plea to follow Wisconsin's lead

A year later the couple flew to Washington, D.C. So did the Modells. By then Massachusetts had followed Wisconsin's lead and started screening babies for SCID.

Melissa Bornheimer was in the nation's capital to testify before an advisory committee to the secretary of the U.S. Department of Health and Human Services. The committee recommends changes in newborn screening.

"Today," she said, "Dawson is the first baby in the world born with severe combined immunodeficiency who was cured as a result of newborn screening..."

"Our only wish is that young families, like ours, in Minnesota, Michigan, Iowa, Illinois, and all of the states can feel secure that if any one of them gets the call that we did from the pediatrician, a program of newborn screening can turn a devastating tragedy into the kind of joy Dawson gives us every single day."

While Melissa spoke, Vicki Modell held Dawson on her lap. She held up a little container of french fries and ketchup; the toddler's face was smeared with ketchup. Vicki felt glad that members of the committee could see him and realize: Here's a child doing what every child does.

The committee voted 26-0 in favor of adding SCID to the uniform newborn screening panel recommended to every state.

The Modells and Bornheimers were overjoyed.

Then a member of the committee clarified what had taken place: a recommendation, and no more.

Each state, not the federal government, decides what diseases to include in newborn screening.

"You have to go to every single state," the committee member told the Modells.

Their work wasn't done; it was just beginning.

Leading a crusade

If they had to go to every state, that's what they would do.

Over the next decade, the Modells traveled more than 100,000 miles campaigning for SCID testing.

The resistance was always the same. The state has no money in the budget. The disease is too rare to be a priority. The state lab doesn't have the expensive equipment needed to confirm a positive test.

Vicki and Fred did their best to stay calm. They always mentioned Wisconsin. Now, there was a state that had the courage to take a chance and conduct a pilot study. There was a state unwilling to let babies die from this disease.

Often, they stressed the risk of losing a baby to a condition that could have been treated. Sometimes families affected by SCID joined the fight, speaking to the media and public officials.

In 2010, California and New York added SCID to their newborn screening tests. In 2011, Delaware and Michigan followed; 2012, six more states, including Florida and Texas. In 2014, 11 more states. More followed.

Finally, in 2018, the last of the 50 states added the SCID test: Nevada, Alabama, Indiana and Louisiana.

Between 2016 and 2017, the first countries outside the U.S. began screening for SCID: Israel, Norway and Taiwan. Pilot programs began in Spain, Sweden, Great Britain, France and Germany.

"We've trained the scientists," says Routes at the Medical College. "They come out here and we show them how to do the test."

As more places began testing, scientists learned something surprising about the disease.

Once believed to affect only one in 100,000 babies or fewer, SCID appears to be more common. Routes said the current estimate is that the disease occurs in 1 out of every 40,000 or 50,000 births.



Fred and Vicki Modell, whose son, Jeffrey, died from primary immune deficiency, hold Dawson Bornheimer at a gala in New York where Dawson received the Man of the Year award in 2009. The Modells were instrumental in efforts to boost research and awareness of primary immune deficiency, a group of diseases that includes SCID. A newborn screening test showed Dawson had SCID. (Photo: Family photo)

A bond between families

The Modells have remained close to the Bornheimer family.

When Dawson left the hospital in 2009, the Modells bought his family a laptop computer so that they could all stay in touch. The Modells buy Dawson and his three brothers presents at Christmas. The two families talk on the phone and through email.

Melissa Bornheimer feels a special thrill when she watches Dawson score a touchdown or a basket at one of his games. Sometimes she and Dawson take out the CaringBridge journal she kept when he was in the hospital.

"I think he is starting to understand how very important his life has been," she said.

Dawson tells his friends sometimes that he almost died when he was a baby. He's even told them that one of the surgeries left him without a belly button.

"They all seem to think it's really neat," Melissa said.



Mark Johnson has written in-depth stories about health, science and research for the *Journal Sentinel* since 2000. He is a three-time Pulitzer Prize finalist and, in addition, was part of a team that won the 2011 Pulitzer Prize in Explanatory Reporting for a series of reports on the groundbreaking use of genetic technology to save a 4-year-old boy.

Email him at mark.johnson@jrn.com (<mailto:mark.johnson@jrn.com>); follow him on Twitter: [@majohnso](https://twitter.com/majohnso) (<https://twitter.com/majohnso>).