After the birth of their son, Steve and Kate McCurdy found themselves...

Fighting a Rare Illness—Together

By Meg Massey
PARADE Magazine
Publication Date:
02/13/2009

For more on the McCurdys and the Barth Syndrome Foundation, watch NBC's TODAY show which aired on Monday, February 16, 2009.

The McCurdy Family
(clockwise from left): Steve, Will, Kate, and Eliza
(photo courtesy of the McCurdy Family)

From the day he was born in 1986, Will McCurdy was a bright, engaging, and alert child. But when he reached the age at which other babies began to roll over by themselves, he did not. Then, as his peers progressed to sitting up, he also didn't reach that milestone. His parents, Steve and Kate of Westchester County, N.Y., took him to physicians up and down the East Coast to find out what was wrong, but none could identify the exact problem.

Two years later, Will came down with what the McCurdys thought was a bad chest cold; what's more, they noticed that his heart was racing. When they took him to their pediatrician, he sent them to the ER because he was concerned that the toddler might have an infection. At the hospital, they received some shocking news: Will was in heart failure and might need a heart transplant.

"I remember wondering in the ER whether all this might lead to something bigger and broader," says Kate. She was right.

After many medical tests, it was revealed that Will also had a white-blood-cell irregularity. One of his physicians decided to input that finding, along with Will's other primary symptoms--muscular weakness and heart problems--and conduct a search of the medical literature. A single journal article came up, by Peter Barth, a Dutch neurologist. In the paper, Dr. Barth discussed the case history of a family in which more than 20 males were affected by the same problems Will had. All of them--except for one young boy--had died.

"It was devastating when we learned this," says Steve. "We thought we had a healthy 2-year-old who had weak muscles. Suddenly he had a life-threatening illness."

The McCurdys wrote to the Dutch physician about Will, and he replied telling them that he thought their son did indeed have the same disorder he had written about--which eventually would be called Barth syndrome, after the doctor who first identified it. He also gently informed them that the five-year-old boy whom he had been treating had passed away while playing at home.

"On the one hand, we were glad we finally knew what Will had," says Kate. "But now we knew nobody else in the world who also had it."

Right now, there are a few hundred people, mostly males, living with Barth syndrome worldwide. However, it's known to be under-diagnosed since all of the symptoms do not necessarily show up simultaneously. (A genetic test for the disorder does exist.)

In Barth syndrome, the body's cellular metabolism is impaired, thus afflicting all systems. People with Barth have difficulty absorbing and utilizing nourishment to build tissue, especially muscle, and as a result, they suffer tremendous physical weakness and fatigue.

In the decade after his diagnosis, Will and his care absorbed the McCurdys. Since his symptoms were so varied, he needed to be seen by an ever-expanding array of specialists. So rare was Barth syndrome that Kate brought along Dr. Barth's journal article as an explanation each time they went to see a new physician.

Like all kids his age, Will was often on the Internet. Every once in a while, he would do a search for the phrase "Barth syndrome" just to see if anything appeared. Nothing did--until one day in November 1999. Will decided to try a different search engine, and after he pressed "Enter," the names and e-mail addresses of three women showed up on the screen. The McCurdys were all stunned. Could it be that they weren't alone? With his parents' permission, Will e-mailed the women and found out that they were all mothers of children with Barth syndrome. They were planning the first-ever gathering of families affected by the disease at Johns Hopkins University in Baltimore, Md., in the summer of 2000.

Those e-mails--and the following get-together--changed the McCurdys' lives. They met more than a dozen other families from around the world who were all fighting the same disease.

"From that meeting, it became so clear that we all needed each other," Steve says. Not only did they give each other some badly-needed support and empathy, they realized that, as a united force, they could advance scientific understanding and treatment of the rare genetic illness. The following year, the McCurdys and three other parents from the Johns Hopkins meeting set up the nonprofit Barth Syndrome Foundation (BSF).

Will McCurdy, age 3
(photo courtesy of the McCurdy Family)

Today, the BSF holds international conferences every two years with physicians, scientists, and families, and raises money in order to fund research. So far, the foundation has given out more than $1.3 million in seed grants. To further study, BSF has started a centralized registry--with blood, DNA, and tissue samples--of people with Barth syndrome. Another priority of the foundation has been to ensure that the entire medical community knows about the disease and its many manifestations.

"Most doctors will see one patient with Barth syndrome in their entire professional lives," says Steve. He adds that children who receive a diagnosis of Barth early in life have much better chance of surviving into adulthood; without an early diagnosis, the survival rate drops dramatically.

Will, now 23, has graduated from high school. Because of his health, he is unable to attend college or take on a full-time job, but he is continuing his studies at home. He takes 31 pills a day, has an implanted defibrillator to jump-start his heart if it stops, and is fed intravenously.

In 2007, he spent about one-third of the year--114 days over 8 separate admissions--in the hospital. He was better in 2008, logging 54 days as an in-patient. "For now we focus on the day to day," says Steve. "Not long ago, almost all kids with Barth syndrome died before the age of 3 so the fact that most of them now are living into adulthood shows great progress and gives us hope."

Will is doing his part to educate people about the disease. He has given presentations on Barth syndrome to students at nearby Sarah Lawrence College who are studying to be genetic counselors.

"He's very smart and articulate and would love to go to college but can't attend himself," says Kate. "So instead, he's teaching graduate biochemical genetics! It's pretty incredible."

To other families who are raising children with rare, chronic diseases, the McCurdys say that the most crucial thing is support. "Find doctors with whom you can work, because it really is a team effort," says Kate. "You need people who will listen and think outside the box."

The McCurdys also cannot stress enough the importance of connecting with other families who are dealing with the same illness. There is strength in numbers; by sharing medical information, families can help doctors gain medical knowledge. "You also have to find a place to feel positive and be positive," says Steve. "One of the parents said that our group is the only place she feels comfortable laughing about things, because everyone else expects you to be sad." Adds Kate, "We know that we are on a long journey, a marathon. We could never do this alone."