



Barth Syndrome  
Foundation

#### Quote of the Day:

**“Cardiomyopathy, neutropenia, delayed growth and hypotonia are symptoms, not a diagnosis. Any combination of these and other lesser-known symptoms are the characteristics that define Barth syndrome. Doing something about it is what defines the Barth Syndrome Foundation.”** ~ Steve, Father of Diagnosed Son; Chairman, Board of Directors, Barth Syndrome Foundation, New York

## Featured Story: Fighting a Rare Illness - Together

The Barth Syndrome Foundation (through one Barth family -- Steve, Kate and Will) was profiled in the on-line version of PARADE Magazine on Saturday, February 14, 2009 and featured on the Today Show on Monday, February 16, 2009. BSF is grateful to PARADE, NBC, and it is a tribute to all families, physicians, scientists, donors, and volunteers who make up this wonderful community. Thank you for your support!! (By Meg Massey, PARADE Magazine, Publication Date: 02/13/2009)

#### Today Show Episode: [Turning anguish into action](#)



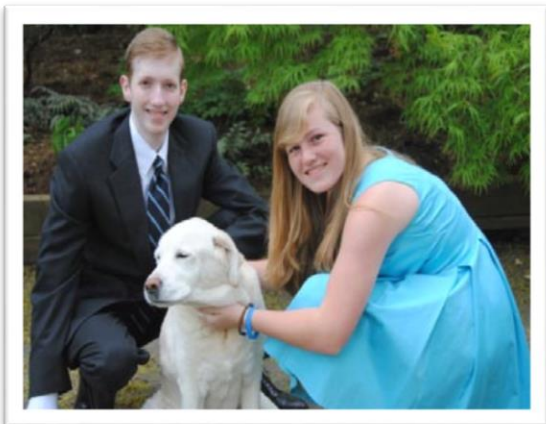
Will, age 12  
(01/23/1986 – 10/25/2014)

From the day he was born in 1986, Will 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, NY, 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 his parents 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.

Barth Syndrome Foundation  
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Will, Eliza, & Cally <sup>2011</sup>

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." Steve and Kate 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 syndrome have difficulty absorbing and utilizing nourishment to build tissue, especially muscle, and as a result, they suffer tremendous physical weakness and fatigue.

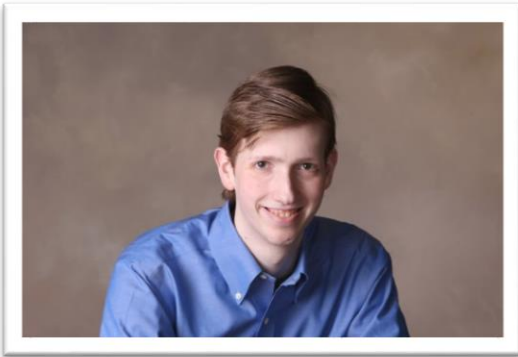
In the decade after his diagnosis, Will and his care absorbed his family. Since his symptoms were so varied, he needed to be seen by an ever-expanding array of specialists. So rare was Barth syndrome that Kate brought along Dr. Barth's journal article as an explanation each time they went to see a new 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. Will's family were all stunned. Could it be that they weren't alone? With his parents' permission, Will e-mailed the women



(L-R) Kate, Will, Eliza, & Steve

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.



Will, age 26

Those e-mails--and the following get-together--changed the lives of Will's family. 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, Steve and Kate, together with three other parents from the Johns Hopkins meeting, set up the nonprofit Barth Syndrome Foundation (BSF).

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 [Update note: this figure has now increased to \$2.3 million in 2012]. 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 [Update note: in 2012, he is now 26], 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 [Update note: and he has a J-G-tube now as well].



Will & Steve

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. [Update note: Will has been in the hospital a great deal recently as well; he spent more than half of the days from November 2010 through February 2011 in the hospital, including Thanksgiving, Christmas and New Year's Day. But he is doing better and now has been home for the last 6 weeks!] "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."



Kate & Will

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 gives an annual presentation about a subject he knows a lot about to graduate biochemical genetics students! It's pretty incredible."

To other families who are raising children with rare, chronic diseases, Will's parents 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."

Kate and Steve 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."



(L-R) Eliza, Steve, Will, & Kate <sup>2012</sup>