



Barth Syndrome
Foundation

Quote of the Day:

"...We contacted BSF immediately after receiving the results. If we could help in any way with the research to find a treatment or a cure we wanted to get started straight away. One boy could have all of the answers, and maybe that one boy was Henry." ~ Tiffini, Mother of Diagnosed Son, Indianapolis, IN

Featured Story: Meet Henry



Henry

Henry Michael was born on June 30, 2008, just in time to watch his first Chicago Cubs game. Unfortunately, Henry missed the game. His sugar levels were low, warranting a night in the care unit where his nurses could keep a closer eye on him.

Fortunately, the stay in the care unit led to an early diagnosis of dilated cardiomyopathy. Henry was three days old when an echocardiogram revealed that his heart was severely dilated and his ejection fraction was merely 10-15%. That day he was whisked off to the NICU, and after only nine days, his ejection fraction was at 25-30%. He was doing so great that his team of doctors released him to go home to sleep in his cozy crib.

After a couple of months out of the hospital, Henry's cardiologist suggested we seek a second opinion. After a review of Henry's echos, he was diagnosed with left ventricular noncompaction of the heart, and it was suggested that a muscle biopsy be performed to rule out a mitochondrial disorder, just in case Henry would need to be placed on the transplant waiting list. That was shocking to hear, and we needed to educate ourselves. After a little on-line research, we suspected Henry had Barth syndrome (BTSH). We mentioned this to one of Henry's former doctors who dismissed the idea. While we lack medical degrees and have the utmost respect for all those who have cared for Henry, we wish that particular doctor hadn't been so dismissive, and, in hindsight, we should have insisted on a genetic test.



Henry

Barth syndrome is such a rare disorder, and we just couldn't believe Henry could have it. Although we also had that moment of thinking someone has to have it, so why not Henry? Henry was four months old when his muscle biopsy came back positive for abnormal mitochondria, which made BTBS even more realistic. He had many of the symptoms: left ventricular noncompaction, abnormal mitochondria, and he was below the 3rd percentile in height and weight.

In February 2010, Henry was admitted to Riley Hospital for Children due to a viral infection. His neutrophil count was zero, making any bacterial infection very dangerous. The hematologist said that he thought Henry had something called "neutropenia." We said, "If he has neutropenia then he must have BTBS." The hematologist said they'd draw blood immediately and send it off to the diagnostic laboratory at Baylor College of Medicine for genetic testing. We'd read about neutropenia during our research, but we learned what it really meant after watching a segment on the Today Show that featured the Barth Syndrome Foundation (BSF).

As we suspected, on March 10, 2010, Henry's genetic test came back positive for BTBS. We were grateful that we finally had an answer. A weight had been lifted from our shoulders, and although we felt very educated about BTBS through the research we had done, especially from the information gained from BSF's website, we couldn't wait to learn more. We contacted BSF immediately after receiving the results. If we could help in any way with the research to find a treatment or a cure we wanted to get started straight away. One boy could have all of the answers, and maybe that one boy was Henry.

Shelley Bowen, President of the Foundation, called us that evening, and we talked for hours. She told us about the 2010 International Conference, and we said that we'd be there. The conference was phenomenal! Researchers, doctors and families from all over the world gathered in Florida to help find treatments and a possible cure. It was amazing to us that so many doctors and researchers were interested in a disorder that only affects approximately 150 families worldwide (although BTBS is believed to be under-diagnosed by many physicians).

The information we came home with is indispensable. We realized that the research the scientists are doing is not only helping our boys with BTBS, but it is also helping kids and adults with other diseases such as cancer, diabetes and mitochondrial disorders. We knew the researchers and doctors were not just people that we would meet once and never hear from again. They are now our friends. We are also very fortunate to have a fantastic team of doctors at home that are more than willing to work with the doctors and researchers we met at the Conference which adds a lot of comfort to our lives. Henry could not be in better hands!

We created invaluable friendships with the other families attending the conference that will last a lifetime. We've realized that if Henry didn't have BTHS, our life would lack the richness these relationships have brought! On behalf of Henry and our entire family, we thank everyone who has become a part of our extended BTHS family.



(L-R) Kevin, Henry & Tiffini