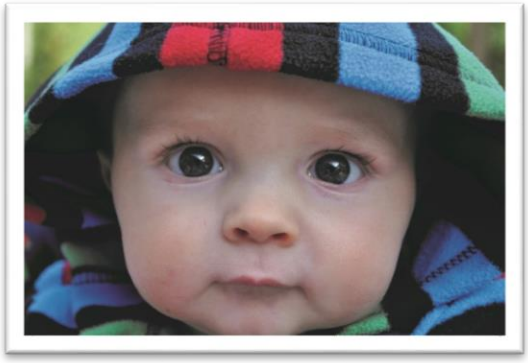
Featured Story: Bryn and Rhys
A Story of Hope after Tragedy



Bryn, age 9 months

Our first son, Rhys, died very unexpectedly of dilated cardiomyopathy on February 10, 2011. The autopsy revealed no indication of bacterial or viral infection, so a genetic cause was suspected. The pediatric hospitalist who delivered the report to us suspected Barth syndrome (BTHS) and encouraged us to see a genetic counselor. We had already made the difficult decision to get pregnant again by this time so she pulled some strings to get us seen right away. The counselor we saw went over our entire family history with us and came up with a short list of possibilities. BTHS was at the top of the list, however it was deemed only moderately likely as there had been no sign of skeletal muscle abnormalities in Rhys' autopsy report. She told us to let her know if we wanted to send tissue samples to Amsterdam (Emma Children's Hospital/Academic Medical Center) for analysis.

About a month later, we learned we were pregnant again, and I was put into the high-risk category and scheduled for many fetal echos. After the first two came back normal, we felt we might be able to breathe again, but I kept having nagging doubts and decided it was time to rule out BTHS. In September, I asked that tissue samples be sent to Amsterdam for genetic testing. Months passed with no news. Evidently there was some sort of logistical issue with getting the samples flown over. We were told it would happen but would be a waiting game.

The first indication of trouble with the pregnancy showed on the last scheduled fetal echo. All of the measurements were normal but the heart "looked thick." A repeat echo ended with the same frustrating result, so an echo was suggested during the first week of life. Bryn was born on December 15, 2011, and, although he was tiny, he was deemed "gestationally appropriate" and large enough not to be put on any watch lists. He also scored a 9-9 Apgar! He latched well and was a sweet, quiet, ever-watchful baby. We were hopeful that the echo would prove he was healthy — for how can a child with a cardiac problem score a 9-9?! However, the echo showed a decline in his heart function from "normal" in the fetal echo to 33% ejection fraction (EF) at day two. He also suddenly started showing signs of

lethargy, and tests revealed thickened blood and a possible kidney infection. He was transferred to the NICU at Dartmouth, New Hampshire where he spent the next 11 days.

Bryn, age 16 months

Our world was rocked again. The heart issue appeared different, but how could both our boys have different cardiac problems? Because BTHS was still a suspect, Bryn was referred to Dr. Gerald Cox at Boston Children's immediately. A cardiomyopathy genetic panel was drawn, and the waiting game began again. Biochemical urine tests were done and roughly one year after Rhys' passing, we got the call from our genetic counselor. The lab results from Amsterdam were back and they indicated BTHS for Rhys, urine tests for Bryn indicated the same. They were 99.9% sure...the tafazzin (TAZ) mutation was really just a formality. We got the final genetic results in early April.

Rhys' and Bryn's godparents's sprang into action after the biochemical diagnosis. They discovered the Barth Syndrome Foundation (BSF) and even invited themselves to dinner with the McCurdys! I lurked on the listserv for a month or so before finally getting the courage to introduce myself. We found out about the BSF conference that was happening in just a few short months, and my parents helped us with plane tickets.

We had been struggling with the issue that, while Bryn looked so good, he had a disease that had claimed his brother. Friends and family alike could not fully comprehend our anxiety and would, understandably, get frustrated with our reclusiveness. But here was an entire community of people who not only got the “chronic illness” thing but understood BTHS as well. Our experience at BSF’s 2012 Conference was nothing short of amazing. To be able to interact with other families and meet affected boys and men showed us that these boys could thrive. Meeting the people who are treating and researching BTHS helped us to understand what Bryn was going through. The Foundation and the conference gave us the hope we needed — that Bryn would not have to follow in his brother’s footsteps — that he could grow up.

Kate, Bryn, and Sandt