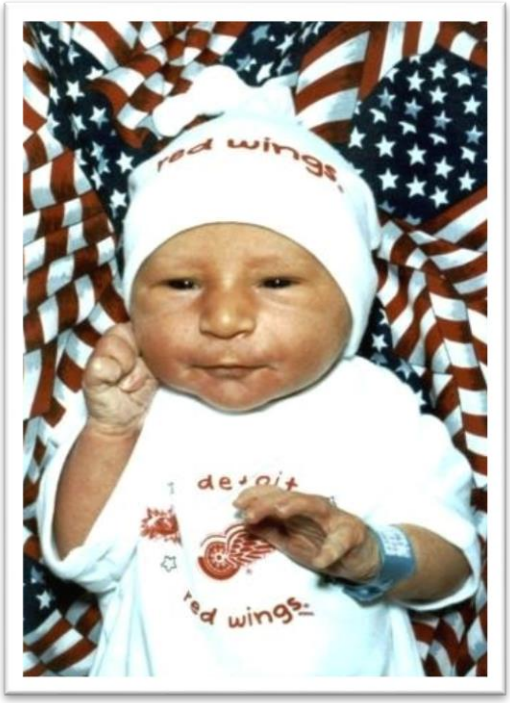
Barth Syndrome Foundation®

MEET NATHANIEL & DEVIN

A Personal Message from the Nicole Derusha-Mackey



My first son, Nathaniel Joseph, was born September 9, 2002. He appeared very healthy and we couldn't wait to get him home. At Nate's one-week doctor visit, we learned he had gained one pound in just a week on breast milk alone. Other than a little jaundice, he was given a clean bill of health.

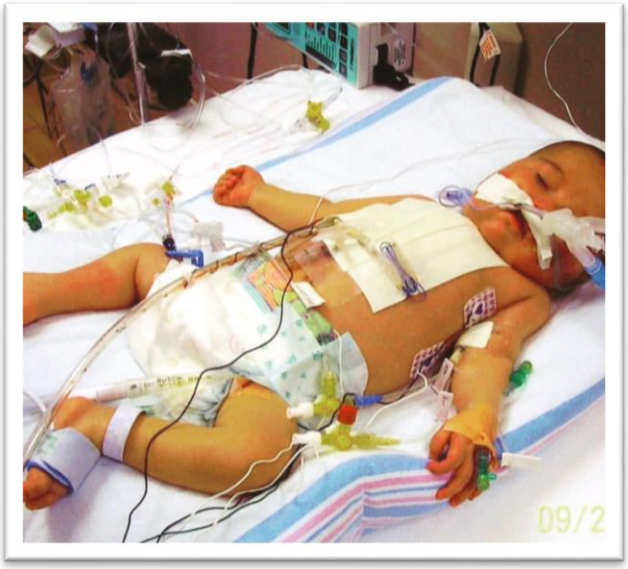
On September 22, 2002, we had a normal day with our new baby boy and when we went to bed around 10:30 pm, everything seemed fine. Around 1:20 am on September 23, I was awakened by Nate's cry, so I got up and gave him a bottle, but he appeared to fall asleep in the middle of his feeding. I tried waking him, but got no response, so I woke his father and called 911. As the EMTs arrived, Nate stopped breathing. They began CPR in the ambulance, told us they were taking him to Hurley Medical Center, and asked that we drive separately.

The short drive to Hurley seemed to take forever. Upon arrival, we were brought to a desk to answer insurance questions, and then led to a private room where we were told to wait for news on our son. When a nurse finally came in, we were asked to come into the treatment room, as Nate was not responding to any treatment. He explained that sometimes having a family member in the room can help. We entered the room to see about 20-30 doctors, nurses, EMTs, etc. surrounding our two week old son on a large gurney. His clothes had been cut open and docs were trying to revive our son with no success. After an exhaustive effort, the doctors decided to "call it" and noted the time of death at 2:56 am.

The next few hours could only be described as surreal. Our parents were called, and we were asked to choose a funeral home and stay at the hospital until some paperwork could be completed. After about two hours, we were told we were waiting for the police. We discovered that when a seemingly healthy child dies at home, an official police report must be filed and an investigation opened. I was floored. I could not believe this was happening! It turned out the hospital staff called the wrong police department, so we were allowed to go home.

Upon arriving home, we were met by police officers from the City of Burton and the Genesee County Sheriff's Department. They asked us questions and walked through our home, taking pictures of where our son slept, the bottles in our fridge, etc. They were very nice, but being under investigation is an unimaginable ordeal when you have just lost your child.

The preliminary autopsy results indicated Nate died of natural causes, possibly SIDS, but it would be 4-6 more weeks for the final results. Nate's official cause of death was determined to be "Neonatal Idiopathic Dilated Cardiomyopathy." In lay terms, this means a baby with an enlarged heart of unknown cause. We were told it was a fluke, a one in a billion thing, nothing could have been done, and it would never happen again.



Although we were told it was most likely NOT genetic, I insisted on seeing a Fetal Medicine Specialist when I discovered I was pregnant with Devin in November 2003. Numerous fetal echocardiograms were performed throughout the pregnancy, and we were told Devin had a healthy heart.

When Devin James Addington was born on July 22, 2004, he had significant difficulty breathing. He was rushed off to the NICU, where an X-ray revealed a severely enlarged heart (dilated cardiomyopathy). He was intubated and airlifted to University of Michigan's C.S. Mott Children's Hospital within six hours of his birth. The doctors in the Pediatric Cardio-Thoracic Unit (PCTU) initially told us their goal was to transition him to oral meds and send him home. But when an echocardiogram on day three revealed Devin's heart function had worsened, it became clear that a heart transplant would be Devin's only option. Unfortunately, the cause of Devin's cardiomyopathy was unknown, and there were many tests ahead before transplant could even be considered.

Devin went into cardiac arrest at three weeks of age, but thankfully he was revived after about five - ten minutes and seemed to come out unscathed, even though doctors insisted it was a sign of things to come. At 4 1/2 weeks, we were told that it appeared Devin had a genetic mutation that could make him ineligible for transplant. However, after five long, hard weeks, Devin finally made it on the transplant list. On September 20, 2004, Dr. Robert Gajarski (Devin's cardiologist) delivered the news we had been waiting for – they had accepted a heart for Devin! And in the early morning hours of September 21, 2004, Devin successfully underwent heart transplant surgery.

October 27, 2004 was a wonderful day – Devin finally came home. He still had an NG tube, due to his failure to bottle feed, and he was terribly jaundiced. But in the coming months, he was able to overcome those obstacles and managed to stay out of the hospital. Due to his "failure to thrive" (failure to gain weight), Devin had a g-tube placed in May 2005, and did wonderfully with it. After almost three years of having his tube in place, Devin had his hole surgically closed in 2008.

Barth syndrome had been suspected early on, but genetic testing performed prior to transplant revealed a mutation that had never been seen. Although Devin's symptoms and family history indicate Barth syndrome, it was not considered enough for a diagnosis, and Devin's medical records listed a possible diagnosis of Desminopathy. This never sat well with me, as I never felt this was the cause of Devin's issues.

In 2006, after watching an episode of Discovery Health Channel's "Mystery Diagnosis" that featured Barth syndrome, I immediately got in contact with the mother of the affected boys featured on the show, Shelley Bowen. She urged me to look further into genetic testing; upon receipt of the earlier results, a mutation had been found on the G4.5/TAZ Gene. Mutations in this gene are known to cause Barth syndrome, the leading genetic cause for infantile dilated cardiomyopathy in boys. Again, we were referred to a geneticist, and after six months of additional testing, Devin was finally diagnosed with Barth syndrome at the age of 27 months – more than two years after receiving the Gift of Life. This diagnosis is so important in many aspects of Devin's care, including nutrition, metabolism, growth, cardiac care, physical therapies, education, etc.

Devin turned four-years-old in 2008 while we were in Florida for the Barth Syndrome Foundation's bi-annual Scientific, Medical & Family Conference. We were able to learn so much about this incredibly rare condition (less than 200 diagnosed cases) and meet many other boys and their families affected by Barth syndrome. And we finally got to meet the Bowen Family, who, because of their participation in that television show, were the catalyst for getting Devin diagnosed.

Devin is now a relatively healthy, active twelve year old boy. I am so grateful for this and give much of the credit to the Barth Syndrome Foundation for the continued Barth syndrome research, family support, and advice we have received.

Not a day goes by that we don't think of the donor family. In their darkest hour, they were able to make a very tough decision and donate their child's organs. The Gift of Life is the most precious gift anyone can ever give, and we are forever thankful that they were able to allow a part of their child to live on in Devin. I am proud to say that I am in regular contact with the mother and father via Facebook and celebrated Devin's Gift of Life with them on the 10 year anniversary of his heart transplant.

Through Devin's entire journey, we have met many wonderful families. Some have lost their children, and our hearts go out to them, as we've shared in their pain. Other families continue to struggle with their children's health issues, and our hearts go out to them as well. This is definitely the road less traveled, but I like to see it as the scenic route.