Featured Story: Meet Connor

Connor

On November 25, 2012 our son, Connor, took his first steps. He was 2.5 years old. There were times when we didn't think that would ever happen. Thankfully, we were wrong.

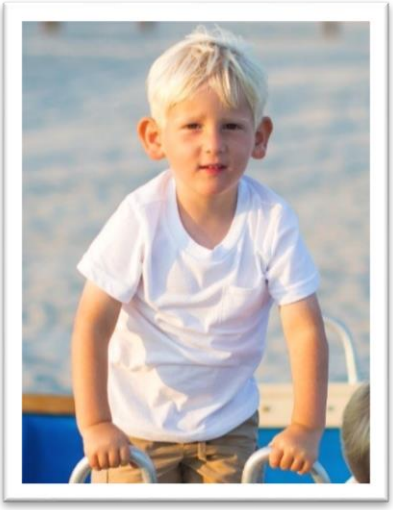
Rewind to June 2, 2010. My wife, Stacey, was entering her seventeenth hour of induced labor, and not progressing. It was decided that a C-section was the best course of action, and at 4:23am, Connor Grant was born into this world. The doctors immediately realized something was wrong when Connor decided he wasn't ready to begin breathing on his own. Several tense minutes passed as they worked on him and they finally successfully intubated him in the delivery room. They brought him around the other side of the curtain to show him to us, and then they quickly whisked him off to the NICU. They had saved his life.

The doctors were worried that those minutes in which Connor was not breathing might result in brain damage, so we agreed to have him transported via ambulance to the NICU at Johns Hopkins Hospital in Baltimore, MD. There he would undergo a specialized hypothermic treatment regimen only offered there, in which his core body temp would be lowered to 92 degrees for 72 hours. The idea was that cooling his internal organs, and specifically his brain, might stop or slow down any damage suffered as a result of him not breathing and not receiving oxygen during his traumatic birth. He would also be further stabilized and closely monitored for any other issues that might arise. We agreed, and at four hours old, we watched him get carefully placed into an ambulance and drive away.

Connor

Connor completed the hypothermic treatment without any major issues – it seemed as if his brain was fine. However, a problem with his heart was discovered as a result of all the testing he underwent, and when he was about a week old, a confirmed diagnosis of congenital dilated cardiomyopathy was delivered. Connor

had heart disease and nobody knew why or how. It was very much a mystery. As it turned out, Connor would spend 26 days in the NICU. During those weeks, Stacey spent nearly every waking minute by his side. Thankfully, he did relatively well there. We were thrilled when he was finally discharged and we brought him home, without any oxygen supplementation. That was just the beginning of our journey though.



Connor

Over the next 17 months, there were many more questions than there were answers. He was followed very closely by many different doctors and specialists. His heart function never really got worse, but it didn't improve either, and we still had no explanation as to why. Luckily, it was good enough that treatment with medication seemed to be enough to keep it stable.

Stacey and I both underwent echo cardiograms and everything was normal. Still no answers. Additionally, Connor had extremely low muscle tone, and he missed all of the main physical milestones that normal kids reach. We started physical therapy at nine months of age when he wasn't even close to being able to sit up on his own, let alone crawl. As it turns out, Connor would never learn to crawl. He also barely ate for months, yet he gained modest weight and had a ton of "baby fat". As for his size, he was barely on the growth charts. Thankfully, from a cognitive standpoint though, everything did seem rather normal.

In December of 2011, a genetics team of doctors at Johns Hopkins persuaded us to submit some of Connor's blood for a specialized genetics test to try and discover a cause for his heart disease. We agreed without giving it much thought. Connor gave blood for testing all the time, and to us, this one seemed no different. To be perfectly honest, I forgot all about it. Then on February 14, 2012 – yes, Valentine's Day - our telephone rang and the doctors informed us that Connor had Barth syndrome, a rare genetic disease with no cure, whose chief symptom is often cardiomyopathy. We were grief stricken as we began to read about it, yet at the same time, there was a strange sense of relief in finally understanding why.

Fast forward to present day, late in 2015. Connor is now 5 years old and doing quite well, all things considered. He began kindergarten last month in a traditional, public elementary school so we are very excited about that. While it is very clear that Connor is physically different from other normal children, he has adjusted very well and seems to be doing fine. From a medical standpoint, he has not exhibited symptoms of being chronically neutropenic, another common and dangerous symptom of Barth syndrome. His heart function has not gotten significantly worse either. His muscles continue to be extremely weak, and we know Connor is going to be physically challenged for his entire life. He receives physical and occupational therapy weekly, and he takes a lot of daily medications and supplements, something he will also do for the rest of his life.

(L-R) Kevin, Connor, Stacey & Ryan

Connor is also a big brother. His little brother, Ryan, is 2 years old and is not affected with Barth syndrome, so we are very excited about that.

We have been members of the Barth Syndrome Foundation since the day after Connor was diagnosed. Kevin also now sits on the Board of Directors. We wouldn't be the people we are today without the support of this wonderful foundation. Dealing with such a rare and deadly disease is difficult to say the least, and knowing there is a community of knowledgeable experts and other supportive families out there that we can meet and spend time with is invaluable.

Kevin & Stacey
Parents to Connor (5, BTHS) and Ryan (2, Unaffected)