Barth Syndrome Month of May Awareness Campaign

Quote of the Day:

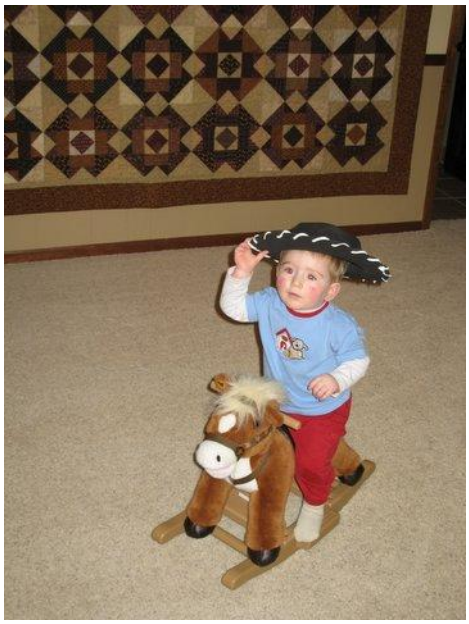
"I remember how impressed Peter was by this experience; meeting all those families, feeling the strength and involvement of you all, seeing the way you all join together to fight Barth syndrome. Being a researcher and not a medical doctor, the experience of meeting the patients and their families had a big impact on Peter. He was very touched by all the warmth he saw within your group. This motivated him even more to spend as much time as possible on his research for Barth syndrome." *~ Annelies Vreken on behalf of her late husband Peter Vreken, PhD*

Featured Story:

A Personal Message from the Brody Family

My husband, D.W., and I (Tracy) joyfully welcomed our first child, Bly, into our lives on February 16, 2004. He weighed 6lbs 5oz, measuring 19 ½ inches long. Things went well for Bly the first 4-6 weeks, just a little jaundice the first few days and a slight, occasional cough. He gained weight and ate well. Then, it seems suddenly, that he didn't want to nurse as long, with his feeds being cut in half, and he also began vomiting occasionally. We also noticed at this time that he would sometimes become very uncomfortable, especially when he needed to have a bowel movement. It seemed odd and we called and went to Bly's pediatrician several times, but nothing resolved our son's problems over the next month or so. We were told he was colicky, he was allergic to my milk, he had acid reflux, he was lactose intolerant, and he had too small of an anus. Our concerns never seemed to concern the pediatrician much, though, so we continued on, thinking it was a phase and he'd surely grow out of it.

After Bly's feeds reduced to only a few minutes at a time, and his vomiting increased we sought out the help of different medical personnel because there was clearly something wrong with our little baby boy. She was a breath of fresh air and seemed to take the matter more seriously. After seeing her several times in the following weeks, we noticed no weight gain with Bly and he was also anemic. She felt that she should refer us to a larger clinic within the state for further advice. She felt that Bly had acid reflux, but wanted another opinion to make sure there wasn't some underlying cause. We were thankful that she was humble enough to seek additional help.



Later that week, D.W. and I took our precious son to Ames, IA to see a pediatric doctor, who looked Bly over carefully. His only thought was acid reflux as well so he ordered a barium swallow for Bly. I remember standing in the x-ray room as Bly screamed through the swallow test and looking at the computer screen, telling my husband that Bly sure had a big heart. I had no medical background whatsoever and didn't have a clue that it was abnormal.

Needless to say, things took quite a different turn at that point as we were told that Bly's problems were the result of having an enlarged heart. Our doctor told us, "I need you to go directly to Mercy Hospital in Des Moines, IA. They are waiting for you. Do not stop anywhere on the way." We felt nearly immobilized, with a million concerns and fears racing through our minds. We knew things were very serious, imagining the worst, but praying for a miracle!

After a very quiet car ride, we arrived in Des Moines and Bly was started on all kinds of medications. Bly's ejection fraction (EF) was at 8%, he had severe mitral valve leakage, his heart was severely enlarged, and D.W. and I were in a whirlwind of emotions. We were told if Bly's heart didn't improve within one week that he would need a heart transplant. The cardiologists believed that Bly's heart failure could have been the result of an infection that attacked his heart.

Thankfully, we saw some improvement by the next morning with his EF up around 20%, but it didn't improve any more in the days to come.

D.W. and I would take a break from the hospital room from time to time and I remember us saying the most difficult prayer we ever thought was possible one sunny, warm, June afternoon outside the hospital. "Heavenly Father, we want you to bring healing upon Bly's heart, but if Bly needs to come home to you we trust in your decision. We don't want him to have a life of suffering. Lord, we don't understand this, but we know that you work together for the good of those who love you, in Jesus' name, Amen." We surrendered completely at that moment and Bly was 100% in the Lord's hands.

After we spent one week in Des Moines, they wanted Bly to be transferred to Iowa City, IA for a transplant, but we wanted to get a second opinion and decided to go to Mayo Clinic in Rochester, MN instead. The medications were adjusted and changed upon our arrival in Rochester and Bly's EF remained stable in the low to mid 20's.

After spending a week in Rochester, we heard the words we didn't think were possible; ALL three of us were able to go home! We didn't know if Bly would ever come home to Arcadia, IA again. What a joy! What a blessing! What an answer to prayer! We were ecstatic!

Our stay at home didn't last long due to Bly becoming dehydrated and having digoxin toxicity. Bly was 5 months old. Another week at Mayo got him stabilized once again. We were homeward bound. Thankfully, that has been Bly's last hospital stay!

After several months of no more improvement, our cardiologist at Mayo thought it would be a good idea talk to the transplant team at our next visit so Bly had a better chance of making it to his 2nd or 3rd birthday. Our hearts sank with hearing this, but we continued to pray and trust God. The cardiologist said

that he was going to discuss Bly's medical concerns with the team of cardiologists at Mayo that week. Their decision was to increase Bly's four heart medications and give him more time to hopefully improve. With Bly having left ventricular non-compaction of his heart (weak, sponge-like muscle), we were told that he would do one of three things: improve, stay the same or get worse. Deep in my heart I always held the hope that he would be healed one day, desperately wanting a miracle. We couldn't get discouraged when we saw no improvements, yet some moments were very stressful.

When we returned for a follow-up appointment at 13 months old we heard miraculous news! The cardiologist said he that didn't think he was looking at the same heart as he was giving Bly an echo. His EF improved to 35%, his heart size shrunk by 10%, and his mitral valve leakage went from severe to moderate. Wow, praise God!

His heart continued to improve over the next several months and his heart was soon considered to be within normal pumping percentages, although his heart muscle wasn't normal.

At about 17 months old, Bly's cardiologist noticed that Bly's neutrophil counts had been low and wanted us to see if Bly could have Barth syndrome (BTHS). We were shocked that this could be genetically possible, but when we read about BTHS it fit Bly perfectly. Reading about BTHS on paper was extremely concerning and it seemed like our problems grew greatly, if this is what Bly had? It wasn't just his heart now, but his immune system, muscles, blood, his entire body. I didn't want Bly to have BTHS for a number of reasons; however, I thought it would be so nice to be able to explain what was wrong with our son.

After genetic testing, at 23 months, we received a diagnosis of Bly having Barth syndrome. We found the Barth Syndrome Foundation website and contacted them immediately, hoping that perhaps Bly could be the missing link that could lead to a cure one day. Although every one of these boys/men are necessary for finding a cure, we found so much more than just a place to share medical information about BTHS. We found an incredible group of loving people who encourage and support in joyful times and in very trying times. Most importantly, 'they just get *it*!' Words don't need to be spoken, because they understand. God has blessed us with a group of people from all walks of life, from around the world. If Bly didn't have BTHS, we wouldn't have the opportunity of knowing any of this great and mighty group.

I had thoughts in the beginning weeks of the diagnosis of feeling guilty that I carried this genetic disorder and passed it on to Bly, but later realized that this is how I was suppose to be made. I have had many heart-breaking times of wanting another child, but what would having another child mean? I honestly don't know? Am I being fearful and not trusting, or am I being selfish? I've had about every question run through my head of what it would mean. I had become a slave to the idea over the years, of wanting to have another child, but I have so much freedom now in that area. It's been a rough road, but I've learned so much on the way. I have surrendered and it's in the Lord's hands now, not mine. I'm 100% happy and content with one child, yet, if DW and I would be blessed with another precious child (BTHS or not), we would welcome that gift wholeheartedly!

Bly had a couple therapies from 1 to 3 years old to help with his physical delay and also to help with his poor eating. It was great seeing him accomplish milestones as the therapies were implemented. It seemed as though feeding him was extremely difficult from 1.5 months to about 2.5 years old. As a baby he'd want to eat only 1-2 ounces every 1-2 hours. It was exhausting. He refused nearly every food as a toddler, but would occasionally take a few bites or licks of something. We were so excited when he did taste something, but so many times he'd start gagging and it would all come up. After an hour of encouraging him to eat at one meal, it all ended up on his highchair tray. However, with age and patient

trying, Bly's eating is wonderful now, compared to what is used to be. Maybe not normal for an average child, but perhaps normal for a child with BTHS! Knowing little things like this makes me very thankful having a diagnosis.

Looking back on all our trials, all credit goes to the Lord for us getting through them. He carried us through our tears of joy and in our darkest hours and we know that He will continue to do so. I remember numerous sleepless nights, tireless attempts to get Bly to eat, terrifying thoughts of losing Bly and we could endure all of it by God's amazing grace. It hasn't always been easy, but it's always been worth it.

Bly is now a delightful 7 year old and is in home school 1st grade. He enjoys trains, 4-wheeling, catching butterflies, Curious George, playing with his golden retriever, playing Wii, socializing with others around him and especially playing and spending time with his Daddy.

Bly's heart remains stable in the low normal ranges, his heart is borderline enlarged, with no mitral valve leakage. He swallows two heart pills, carvedilol and enalapril, 2x a day and receives a daily injection of G-CSF (Granulocyte Colony Stimulating Factor) to boost his neutrophils (a white blood cell that fights against bacterial infections). He takes his meds like a champ! He may physically tire sooner than other kids his age or may never win a high jump contest, but he's still wonderfully made. To look at him, you'd probably never know that he has a life-threatening disorder. It certainly keeps D.W. and I humble and thankful.

One of the more difficult things that we've had to juggle around a bit over the years is being around germs and illnesses, especially during flu season. I'm sure we seem paranoid and obsessive to most people when we arm ourselves in public and at home with anti-bacterial wipes and hand sanitizer or when we have to ask people if they are healthy before we meet. Perhaps we go over board at times, but unless you're in the situation 1st hand, you may never understand. Most times we feel it is good health practices and we're simply using discretion. We have been able to do much more with Bly publicly since the start of G-CSF, however, we remain cautious. Recently, Bly has been handling colds very well and it's becoming much easier with age to make more outings.

We couldn't imagine life without Bly! He brings so much laughter and joy into our lives. We have been taught so many life lessons through this precious gift from God. Just like any of our lives, we don't know what our tomorrow holds. We know that we will face trials in the future, with or without BTHS in our lives. We must be thankful for the things that teach us to become better people.

We are grateful for our past and what it has taught us….we are thankful for today as we enjoy it together….and we are trusting God for our futures as we pray that He'd teach us His will. May God bless each person touched by Barth syndrome, the affected boys/men, their families, friends, the doctors in their care, and everyone who has been blessed by one of these amazing BTHS boys and men.