Quote of the Day:

"The information from the Barth Syndrome Foundation's website has given me, as a mother, a sense of empowerment. I am able to provide quality information to my son's doctors and teachers, and this allows Isaiah to have a customized plan of care that fits his unique needs." *~ Leah, Mother of Affected Son, Arizona*

Featured Story: Meet Isaiah

Isaiah

When Isaiah was born in November 2012, I knew becoming a mother would change my life, but I had no idea just how much this little boy would impact me. From the very beginning, Isaiah was a good baby: he ate well, slept well, and fussed very little. Not only was he a good baby, he was also a very healthy baby; each time I would take Isaiah to the doctor for his checkups, he would measure on the small side of the spectrum, but he was growing consistently and he was thriving. Life seemed to be off to a wonderful start for my new family, and we were excited to watch our little boy grow and learn new things.

The day Isaiah turned six months old is a day that is so clear in my mind. It was a Friday, and I had scheduled Isaiah's six-month checkup for that day. We arrived at his pediatrician's office and went through the usual routine of measurements, and then we were sent into a room to wait for the doctor. It took longer than usual for the doctor to enter the room, and when he finally did, he explained to me that Isaiah had gained almost no weight in the last two months. He also expressed concern with the way Isaiah was breathing. He listened to Isaiah's heart for a long time and told me everything sounded fine, but he ordered a chest x-ray, just to be certain. The following Monday we took Isaiah to have his chest x-ray, and we waited to hear from the pediatrician.

The next day, I received a phone call from Isaiah's pediatrician, and my world was turned upside down. Isaiah's heart was severely enlarged, and he needed an echocardiogram immediately. The echocardiogram showed that his heart was extremely enlarged and was barely squeezing. Isaiah was admitted to the cardiac ICU at Phoenix Children's Hospital, where we were told he had severe dilated cardiomyopathy. Tests and medications began immediately, and we watched our little boy's health deteriorate very rapidly. His heart's ejection fraction was 13%, and his doctors began to discuss the possibility of a heart transplant.

Isaiah soon was unable to eat because it caused too much strain on his heart. He was losing weight rapidly and relied on a feeding tube for nutrition. The doctors also expressed concern about Isaiah's muscles, stating he had low muscle tone. The doctors ran countless tests and asked us what felt like millions of questions, but nothing seemed to give us any answers. Isaiah's cardiomyopathy was labeled as "idiopathic," or no known cause. All we knew was our boy was extremely sick, he was quite honestly knocking on death's door, and we didn't know why. All we could do was treat his symptoms, trust his doctors, and pray for God to give us a miracle.

Years went by, and we received our miracle. Isaiah's heart improved, progress was slow but steady. Isaiah has spent years recovering his ability to eat by mouth, and he has worked hard in physical therapy. Although he was significantly behind, Isaiah eventually reached each of his milestones. We are thankful for his progress, but one piece has always felt like it was missing. Why did my baby get sick? What caused his heart to fail, and, if I had another baby, would that child be at risk for the same kind of health problems? It seemed we might never have answers to these questions, and as a mother, that was something that weighed heavily on my heart.

Dr. Lindstrom & Isaiah

Our answers came nearly three years after Isaiah was initially diagnosed with dilated cardiomyopathy. In December of 2015, we met with one of the geneticists at Phoenix Children's Hospital, Dr. Kristin Lindstrom. She reviewed Isaiah's medical records and looked over our family history. Dr. Lindstrom felt strongly about the possibility that Isaiah's condition was caused by a genetic disorder, and she found some abnormalities on biochemical blood and urine studies that suggested Isaiah could possibly have Barth syndrome. This led to genetic testing of the TAZ gene in February of 2016, which found a mutation but one that had never been seen before in other people with Barth syndrome. Since all of Isaiah's symptoms seemed so consistent with Barth syndrome, she felt that this was still probably the correct diagnosis. To be absolutely certain, however, she sent a special blood test for cardiolipins, which was abnormal and definitively confirmed that Isaiah had Barth syndrome.

Dr. Lindstrom not only found a diagnosis for Isaiah, but she sought out further information to help us learn what a diagnosis of Barth syndrome really meant for our boy and his future. Dr. Lindstrom found the Barth Syndrome Foundation's website and shared it with us. She used information from this site to help us learn how to deal with Barth syndrome. She was able to find a cornstarch dosing chart as well as a growth chart that has been created for children with Barth syndrome. All of this information has made a huge impact on the quality of my son's life. Isaiah will probably never grow according to standard charts, and he may never have the same nutritional needs as other children. When asked about the Barth Syndrome Foundation's website, Dr. Lindstrom said, "I feel fortunate to have such an easily accessible and well maintained website to turn to for questions or family resources. Many genetic conditions do not have such a helpful foundation, and it can make it more difficult to get reliable and trustworthy information for parents and families." The information from the Barth Syndrome

Foundation's website has given me, as a mother, a sense of empowerment. I am able to provide quality information to my son's doctors and teachers, and this allows Isaiah to have a customized plan of care that fits his unique needs.

No parent ever wants to hear there is something "wrong" with their child, but, throughout our journey with Isaiah, I have found that knowledge really is power. If we don't know what is wrong or why things are happening, we can't be properly equipped to deal with the road ahead of us. Dr. Lindstrom certainly seems to share the same way of thinking as we do, and we feel extremely fortunate to have the opportunity to work with her. Dr. Lindstrom didn't stop at just finding a diagnosis. She gave us a wealth of information by recommending the Barth Syndrome Foundation website to us, and she found us an amazing support system in the Barth Syndrome Foundation itself. Dr. Lindstrom did so much more than give my son a diagnosis, she changed our lives, and I know my son's quality of life will be so much greater because of her.