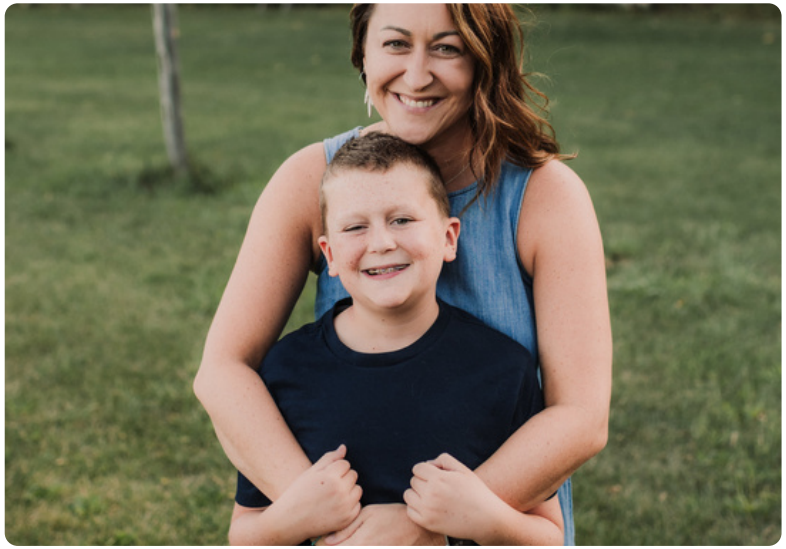
BARTH SYNDROME FOUNDATION
2005 Palmer Avenue #1033
Larchmont, New York 10538
Phone (914) 303-6323
Fax (914) 303-6323
barthsyndrome.org

In an effort to be good stewards, we ask for your help maintaining accurate records to minimize unnecessary postage and printing.

Make corrections on the label. Mail it back to us or send an image to Emily.Madalinski@barthsyndrome.org

- ◯ Name change
- ◯ I received more than one copy
- ◯ I'd like to receive this publication by email:_____
- ◯ Please remove me from the mailing list

Stories from the Heart
Barth Syndrome Foundation Newsletter
Fall 2023 | Issue 03



BEAUTIFUL DREAM

Levi is a happy kid with a ton of life in him. He plays baseball, likes to ride bikes, and keeps up as best as he can with his younger brother and sister. Despite having Barth syndrome, he lives every day to the fullest and has tremendous support from those around him.

Levi was the first child born in the family; his mother was adamant about ensuring the health of her baby during pregnancy. She ate well, took prenatal vitamins, and took any necessary precautions. After being two days past her due date with very little movement, Levi's mother knew something wasn't right.

After Levi was born, they started to notice a lack of crying; he wasn't latching to eat, and a heart saturation test read 80% when it should be 100%. A few more tests showed his heart was only functioning at 20%.

(story continued on page 3)

Executive Director's Note

Emily Milligan

In the last few months, our team has been working tirelessly to advance our mission of saving lives through education, advances in treatments, and finding a cure for Barth syndrome. One significant milestone has been initiating our advocacy efforts urging the FDA to give elamipretide a fair and appropriate review. We've conducted over 60 meetings with state legislatures, with more on the horizon. These efforts are crucial steps toward bringing us one step closer to a viable treatment for Barth syndrome.

You - the community - make this work possible. Your generosity provides the funding needed to expand upon the knowledge already discovered about Barth syndrome. With each contribution, we move closer to our goal and make a real impact on the lives of those affected. As we approach the end of the year, we reflect on the progress we've made and the challenges that lie ahead. Your continued support is instrumental in fueling our efforts and turning hope into reality. Your belief in our cause inspires us to push boundaries and strive for breakthroughs.

We understand the importance of the upcoming holiday season, and we want to take this opportunity to wish you a happy and safe time with your loved ones. May your holidays be filled with joy and warmth. Once again, thank you for being a vital part of our community. Your commitment to our cause makes a lasting difference, and we are truly grateful.

Emily Milligan
Executive Director
Barth Syndrome Foundation

(continued from cover)

April 1st, only a few days after his birth, the family was transported to a children's hospital to better monitor his health. "This has got to be the worst April Fool's joke possible," Levi's mother said. Their stay lasted around a month before genetic testing, and multiple different doctors' opinions revealed it was Barth syndrome.

"When you have your first child, it's like everything will be sunshine, roses, and butterflies... It was the opposite for us. It wasn't like that dream, beautiful dream. It was more like a nightmare. I never thought I'd bring home a kid on oxygen and medication with syringes."

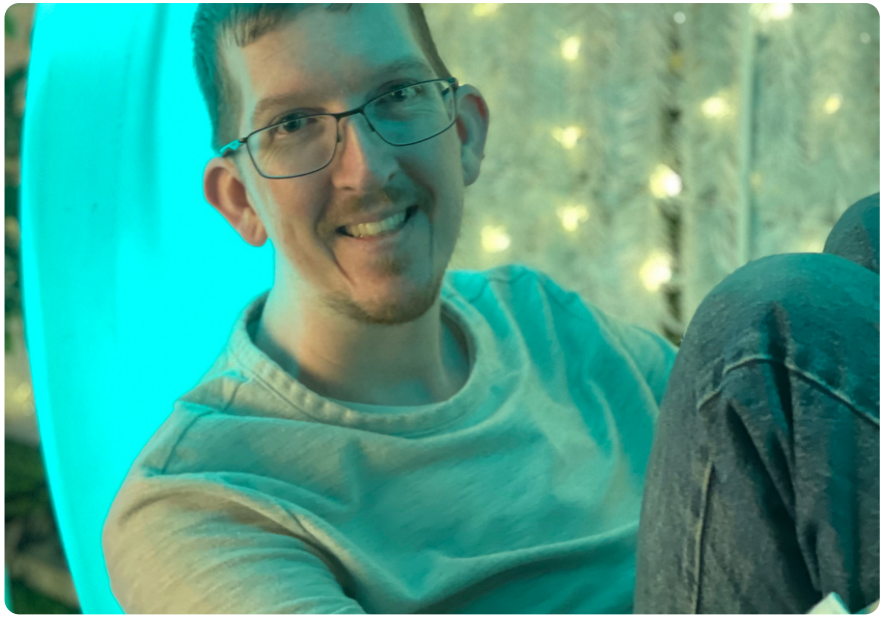
It was a tough pill to swallow for the family. It was hard to accept the fact this little boy had this ultra-rare disease that would impact his life significantly. Levi's mother wanted to ensure that he was seen as Levi, not Levi with Barth syndrome, so she decided to keep his diagnosis private.

At the age of two, Levi started to show improvements, and his mother decided to attend the Barth Syndrome Foundation's biennially conference to finally meet the individuals that were there for her from the beginning of Levi's disagnosis.

Barth Syndrome Foundation gave her the facts she needed to finally feel comfortable discussing Levi's diagnosis with family and friends. Not only did Barth Syndrome Foundation provide Levi's mother with the education about Barth syndrome she needed, but it also provided her with a community of others who know exactly what she is going through.

"I recall one time I felt hesitant to leave Levi to go to my best friend's wedding. He was only one at the time and couldn't talk. I turned to the Barth Syndrome Foundation family chat Facebook group and asked if I should go or not. It still makes me cry to this day, but I received an email from another Barth boy that said if Levi could talk, he would want you to go."

After many doctors' visits, insurance battles for medications, and physical and speech therapy, Levi is doing well. He still has a hard time doing the things he wants to do, but he does his best to push past his limitations. This boy, filled with life and happiness, won't let Barth syndrome put him down, and he has a mother who goes to the ends of the Earth to ensure the best life possible.



BREAKTHROUGH

Steve is a 37-year-old Business Analyst for a healthcare system, a proud graduate of Northern Kentucky University, and an avid sports fan.

Steve's journey began with a series of health challenges that surfaced shortly after his birth. He faced muscle weakness, cardiomyopathy, failure to thrive, and neutropenia. These conditions cast a shadow over his early years, making his health a central focus for his family.

In those initial stages, Steve was under the care of Cincinnati Children's Hospital Medical Center. The medical team worked diligently to understand and diagnose his condition. At that time, he was diagnosed with an undefined muscular dystrophy and cardiomyopathy. This uncertainty made for a challenging time for Steve and his family, adding an element of mystery to his health struggles.

It wasn't until Steve reached his thirties that advances in genetic testing brought about a breakthrough. He went back to Cincinnati Children's to meet with his genetic counselor. It was decided they would run a cardiomyopathy gene panel.

This panel revealed he was one of the few individuals in the world with Barth syndrome. This diagnosis marked a turning point in Steve's understanding of his health, providing clarity and setting him on a path of discovery.

This revelation ignited a passion in Steve to understand more about his condition. He learned about the Barth Syndrome Foundation through a search on the internet and discovered a supportive community. He quickly became acquainted with the older individuals who are also affected by this disorder.

"As I started to learn more about my condition, my participation in the Foundation increased." Steve was asked to participate in fundraising and awareness campaigns. He spoke with Kentucky state legislatures for advocacy efforts. He even met with another affected family that lived only 15 minutes from his hometown.

"Over time, the Barth Syndrome Foundation became less like an organization and more like a family," Steve said. "I often would ask myself what my goal was. What my purpose was. Why was I given these challenges growing up? I found my goals and my purpose through the Barth Syndrome Foundation - to raise awareness, to raise funds to support the efforts of finding treatments, and to finding a cure."

Steve's journey is a testament to resilience and the importance of advances in medical science. The struggles he faced in his early years have not only shaped his personal narrative but have also fueled his commitment to raising awareness and supporting research for Barth syndrome. Through his experiences, Steve has become not just a survivor but a beacon of hope for others facing rare and complex health conditions.

HEART DAY

Julia holds the cherished roles of being both a sister to Walker and a beloved aunt to Jackson. Despite the distinct moments in time that brought Jackson and Walker into the world and the uniqueness of their personalities, they are bound together by more than just family ties—they share the unspoken connection of battling an ultra-rare disease.

Julia grew up with her brother, Walker, going to the hospital frequently. "I didn't understand why he always had to go to the doctor or take a bunch of medicine all the time. I attended appointments with him and did not really understand what was being discussed."

Julia realized something was different about her brother when she noticed he was very small and had low muscle tone. Walker loves baseball and was on a little league team. He was slower than the other players and had difficulty making it to first base.

Julia, three years younger, didn't know the extent of what Walker had gone through when he was born. He had cardiomyopathy, but there weren't a lot of options for heart transplants around that time.

It wasn't until 2009 that Walker's cardiologist called and said that he may have Barth syndrome.

In October 2009, Jackson was born. Julia noticed in the first picture she received of her nephew his feet were blue, and she knew something was wrong. Jackson was airlifted to a hospital in Atlanta, where he stayed for 172 days. Julia met her nephew in the NICU and didn't know if that would be the last time she would ever see him.

"I just remember thinking he was perfect. Like, he was the cutest kid I have ever seen. Now, looking back at the pictures, he did not look good. That was a scary picture."

Jackson received a heart transplant on February 1st, and from that day forward, the family declared that "Heart Day." When Jackson was around 3, his mom started the tradition of bringing cupcakes to Jackson's school so his friends and teachers could celebrate with him. The class was curious about the cupcakes. "It's because you're my friends," Jackson would say, "and I love you."

Today, Jackson is 13 years old, and his family describes him as joyous and very friendly. Much like Walker, Jackson found a love for baseball. He plays in a league that is specifically for children with special needs. He also has an interest in music and plays the drums and piano. Jackson and Walker have a special bond, and their relationship continues to grow as time goes on.

Jackson's mom discovered the Barth Syndrome Foundation after traveling to Maryland to meet with a doctor to learn more about Barth syndrome. That's when they realized there were others out there with the same ultra-rare disease, and they were not alone.

Since then, the family has been involved with Barth Syndrome Foundation's advocacy efforts. They work hard to make sure new therapies can be available. New therapies will give both Jackson and Walker opportunities for happier lives doing what they love.

If you are interested in sharing your story about how Barth Syndrome Foundation has impacted you or your family please contact Emily Emily.Madalinski@barthsyndrome.org