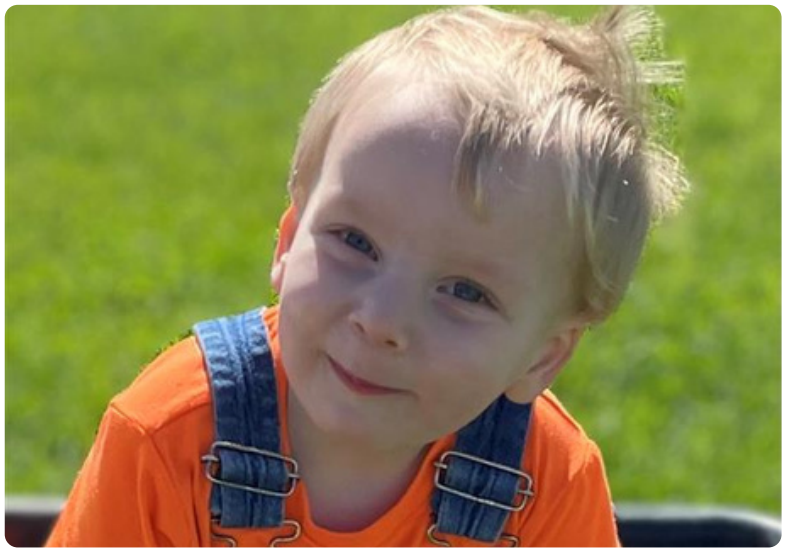
BARTH SYNDROME FOUNDATION
2005 Palmer Avenue #1033
Larchmont, New York 10538
Phone (914) 303-6323
Fax (914) 303-6323
barthsyndrome.org

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
2024 | Issue 01



PART OF EVERYTHING

"Grayson is definitely the apple of our eye," said Lisa, "We never miss the opportunity to be with him." Grayson is the youngest of Lisa's four grandchildren. He enjoys being outside, riding in a tractor, fishing, and playing with his big sister, Avery.

Grayson was born premature, weighing only three pounds, and spent the first 40 days of his life in the hospital. He wasn't properly diagnosed with Barth syndrome until he was around two years old when Grayson's pediatrician grew concerned about him not gaining weight. After multiple blood tests, they received the diagnosis that he had cyclical neutropenia* as well as Barth.

*Neutropenia occurs when you have too few neutrophils, a type of white blood cells. Neutrophils are important when fighting certain infections.

(story continued on page 3)

Executive Director's Note

Emily Milligan

As we step into the new year, I am thrilled to share with you the exciting developments happening at the Barth Syndrome Foundation. Despite the challenges we faced in the past, our community's resilience and dedication have driven us forward, and 2024 promises to be a year of significant progress and achievements.

Our advocacy for elamipretide remains steadfast. While the FDA has yet to review the NDA (New Drug Application), the collective efforts of 293 advocates, including dedicated letters, showcase our commitment. We press on, emphasizing the urgency of this matter.

In research, our Funding Year 2023 saw remarkable engagement. Twelve applications for the Research Grant Program and two for the Iris L. Gonzalez Prize highlight a global and diverse interest. Review by our Scientific and Medical Advisory Board is set for January's end, with grant awardees to be announced this spring.

I'm thrilled to announce our 2024 Barth Syndrome International conference—a momentous gathering marking our first in-person community event since the COVID shutdowns.

As we redefine our strategic plan in 2024, your insights are crucial. We will invite you to join us in shaping initiatives that align with our evolving community needs.

Thank you for your continued support. Here's to a year of progress, unity, and positive impact.

Emily Milligan
Executive Director
Barth Syndrome Foundation

(continued from cover)

Grayson's genetics doctor informed his parent's about the Barth Syndrome Foundation. They have been to Baltimore's Kennedy Krieger Institute twice, but the real insight into navigation life with Barth syndrome was being connected to the Barth Syndrome Foundation community. The family is provided with resources and support from those who have lived with Barth or are a caretaker of someone living with Barth. Those in the Barth Syndrome Foundation community have seen the good and the bad firsthand and can offer guidance based on their experience.

"I think it makes Grayson's parents feel a little better when they know other people have been going through what they have, and they've learned a lot."

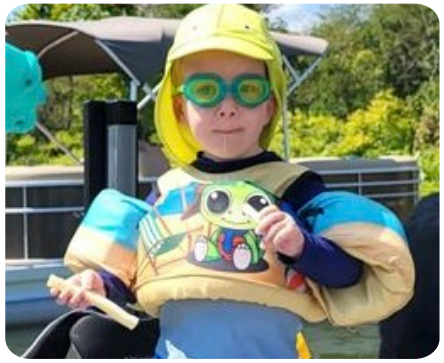
"Grayson gets one good week a month," said Lisa, "His numbers start to go down [due to the neutropenia] and the process from start to finish is usually about three weeks. They go down, then he bottoms out, then he starts to come back up and then he has a good week."

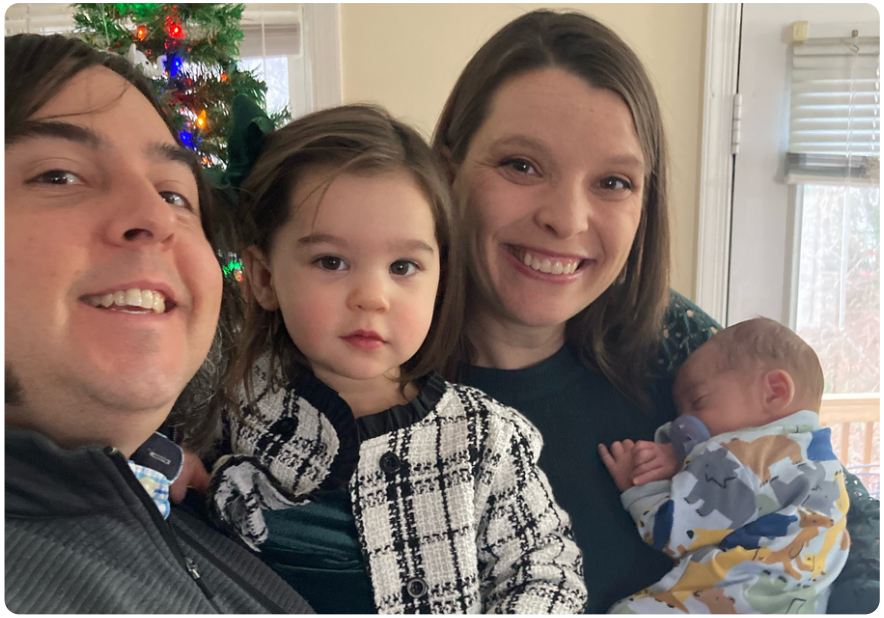
When his numbers are down, he'll often have a hard time getting the energy to eat. He'll eat a little bit and then ask to be fed. When he gets too tired, his risk for getting sick increases.

Grayson maintains high spirits despite being exhausted. His mom, Jamie, came up with a jingle while waiting for his fever to go down: "while we wait, we hydrate" to which Grayson happily joins in on.

His family does their best to take advantage of his good weeks and ensure that he feels part of everything despite his Barth syndrome diagnosis. His sister, Avery is careful to make sure that, when he needs a break, she includes him in activities that don't require a lot of energy. Grayson often joins the family out by the lake with his floppy hat, canopy, and personal fan. He loves tractors and even has his own little tractor he can ride in and chase his other siblings around in.

Lisa continues to be a cheerleader for Grayson's health and well-being. She hopes that sharing her story will raise awareness for other's living with or caring for someone with Barth syndrome.





LIFE RAFT

Owen is a sweet, two-month-old boy who is adored by his family (especially his big sister, Frankie). His mom, Amy, says that he is becoming more alert and interactive. The family enjoys being outdoors, going to local breweries, and travel.

Owen was born on November 16th. Two weeks later, his parents learned he had Barth syndrome. With their new baby being impacted by this ultra-rare disease and living in the Neonatal Intensive Care Unit (NICU), it was some time before Amy connected with the Barth Syndrome Foundation.

Amy's brother-in law was the first to make a move to connect with the Barth Syndrome Foundation. Then Amy and her mom met with Shelley, the Director of Family Services, to discuss what Barth syndrome is and the support that the Barth Syndrome Foundation offers.

"We're going to become best friends" Shelley said.

Within the first call, Shelley connected Amy to the Kennedy Krieger Institute, which is home to an interdisciplinary clinic dedicated to the diagnosis and treatment of Barth syndrome.

The Barth Syndrome Foundation was also able to put Amy in touch with Dr. Stacey Reynolds, an occupational therapist professor at Virginia Commonwealth University who studies and researches Barth.

After the initial call, Owen's family was able to meet some of the team at Kennedy Krieger in advance of the Barth Clinic in March. The occupational therapist at Virginia Commonwealth University offered guidance on signs to look for around Owen's muscle tone and mobility function.

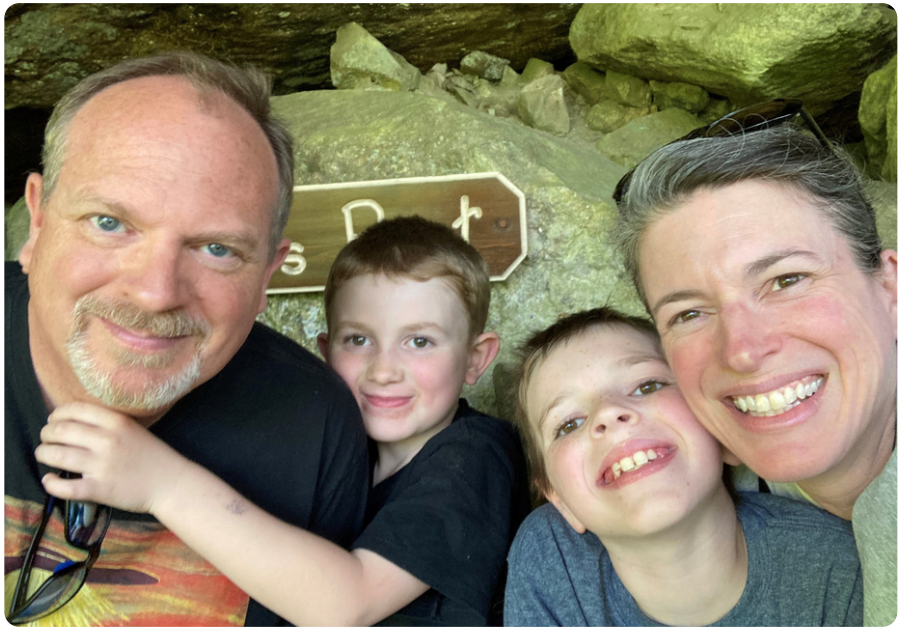
"We feel very grateful for the organization... this is like the very, very beginning. We have no idea where this relationship is going to go. It's possible that multiple people in my family will become more involved with the Barth Syndrome Foundation. This has triggered genetic testing for a lot of family members."

Amy's two cousins were diagnosed with congestive heart failure in the first 12 hours of life. Both are now in their late 30's, early 40's, and suspect they will receive a Barth syndrome diagnosis. One is living with a pacemaker defibrillator. One of Amy's sisters with no children is a carrier, another has two boys and waiting to hear her results before getting her boys tested.

"Barth Syndrome Foundation is like a life raft to the hope because they're going to connect us to people that give us hope," said Amy, "And that I think is something that we are grateful for and looking forward to is being a part of this community."

The family plans on attending the 2024 Barth Syndrome International Conference in July. They look forward to connecting with people in the community to gain more insight into Barth syndrome as well as hear stories of hope for Owen's future.

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



REMEMBERED

Kate and Sandt's story begins with the arrival of their son Rhys, a small but apparently healthy baby who brought joy to his family. Despite his small size, Rhys hit all the expected milestones and his doctors had no concerns. However, tragedy struck when Rhys was taken to the emergency room, misdiagnosed with a severe chest cold, and passed away suddenly and unexpectedly from dilated cardiomyopathy.

One doctor who was there that day had seen Barth syndrome once before and suspected Rhys may have Barth. This doctor prompted genetic testing, which became an emotional and logistical nightmare for the family.

Kate and Sandt's second born came shortly after the loss of Rhys. Bryn was born small with a thickening of the heart muscle, slight cardiomyopathy, and methlglutonic acid in his urine. The test results for both Rhys and Bryn came back around the same time, and it was confirmed that both boys had the same genetic defect on the 2000 genome indicating Barth syndrome. Subsequent testing revealed that Kate was a carrier.

Kate, Sandt, and six-month old Bryn attended their first Barth syndrome conference in 2012. Attending the conference became a pivotal chapter for their ability to navigate Bryn's diagnosis, connecting them with a supportive community that understood their struggles. It was at the conference that Bryn surpassed his late brother in age – a bittersweet moment for the family.

"The community, even though it is remote, remains a support network of people who know what we are going through," Kate said. "The group answered questions that we had that the doctors didn't know about."

Kate and Sandt not only guide their own family through their challenges, but also remain dedicated in the pursuit for the equitable evaluation of elamipretide, the sole medication in advanced stages of development for Barth syndrome, Kate has actively engaged in numerous discussions with legislators. Through heartfelt storytelling, she encourages the FDA to conduct a thorough and impartial review.

"It's good to get the information out there," Kate said, "It makes me feel like Rhys will be remembered. Particularly considering how underdiagnosed our syndrome is, it's good to get the word out. It helps us, but it also helps other people."

The family's journey is marked by both heartache and resilience. They will always carry both the loss of Rhys and hope for Bryn's future, and have become strong advocates for awareness, education, and support for Barth syndrome. Through their advocacy, they help others navigating the complexities of raising a child with a rare genetic disorder and leave an indelible mark on the story of Barth syndrome awareness.

With all of this, they still make time for Bryn and his younger brother, Rory, to be kids. They find joy in shared activities, like ice fishing adventures in winter, visits to theme parks during the warmer months, and serene lake activities. The adaptation to Bryn's limitations fostered a closeness that transcended the challenges they faced, even as they recognize the need to accommodate the energy of his younger brother, Rory.