Barth Syndrome Foundation®

BSF BEAT

Saving lives through education, advances in treatment, and finding a cure for Barth syndrome

News and updates from the Barth Syndrome Foundation 4th Quarter 2022

We Are Stronger Together!

The Stronger Together World Tour has wrapped up, and what an amazing experience it was! With regional gatherings across the globe, including the United States (US), The Netherlands, Canada, and the United Kingdom (UK), affected individuals, families, friends, and professionals gathered to develop and strengthen bonds with opportunities for shared learning. The feedback we received from attendees was overwhelmingly positive and they left feeling connected and excited for the next in-person gatherings. "Words cannot describe how amazing it was to see some of our Barth family at the Stronger Together World Tour 2022 in Gurnee, Illinois," said Kelsey B. of her experience. "Thank you to our Barth family for being our friends but most importantly a part of our family. We missed you as soon as we left Gurnee and are counting down the minutes until next time."

Historically, the Barth community gathered every two years for the International Scientific, Medical, and Family Conference for five

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or six days of meetings, gatherings, and comaradery. However, the global pandemic necessitated the postponement of the 2020 conference to 2024. The Stronger Together World Tour was designed to bridge this gap in a meaningful and safer format. These smaller gatherings were strategically located to reduce the amount of travel exposure for attendees while still providing activities and learning opportunities.

The US Tour had stops in Fitchburg, MA, Gurnee, IL, LaGrange, GA, and Scottsdale, AZ with a collective 142 attendees. “With just 300 individuals living with Barth syndrome in the world, it’s always powerful when you can be with others who share the same struggles and triumphs,” said Tracy B. “Sharing our stories, being supported, encouraging one another is what I love most. We are definitely stronger together. We NEED each other!”

Each of the US stops included a storytelling workshop, facilitated by Kevin Freiert of Salem Oaks Consulting. Affected individuals and families learned how to effectively share their stories of living with Barth syndrome as they navigate their daily lives and develop connections and support. “Though it can be difficult and emotional to share one’s real story, it also felt cathartic as well as useful,” said Kate McCurdy, BSF’s Board Chair. “It was wonderful to have that kind of sincere conversation one-on-one with others in our Barth family.”

At the Gurnee event cardiac pediatric critical care specialist, Dr. Conrad Epting spoke to the attendees about the importance of biorepositories in research and development of therapies. The Scottsdale event featured clinical geneticist, Dr. Kristin Lindstrom who spoke to the attendees about the importance of coordination of care. She offered tips on building a multidisciplinary healthcare team to care for the medically complex Barth syndrome patient.

The tour also provided an opportunity for some individuals and their families to meet in-person with others living with Barth syndrome for the first time. These twelve US families were able to spend time surrounded by support from our community, a true gift for those in any ultra-rare community. “It was so much fun and meant so much to finally meet in-person the people who have been family for years,” said Ayana P. “We love you all!”

Melissa Huang joined BSF as Clinical Research & Registry Coordinator at the beginning of the year when the pandemic was still in full swing, so she hadn’t met any affected families in-person before STWT. These regional gatherings allowed her to hear the stories of the Barth syndrome community from multiple walks of life and gain new perspectives about how the disease impacts their daily living. “The resilience of the families motivates me to work hard with BSF on the research front to positively impact the lives of those affected.”

Much like the US gatherings, the three international stops shared similar experiences and sentiments. The Netherlands event began with a presentation by researchers from the Amsterdam Medical Center and a pharmaceutical company about their plans for future research. “People who attended found this really interesting and promising, and of course it was great seeing each other again after more than three years,” Peter van Loo, said. “We’re already looking forward to, and in the process of, planning another meeting in May 2023.”

The UK event in October was an “out of this world experience at the UK National Space Centre in Leicester,” according to Michaela Damin, CEO of Barth Syndrome UK. “We held informal workshops as we worked together to plan what we want and need from both our Bristol NHS Barth Syndrome Service and from Barth Syndrome UK. The Bristol Service also provided resuscitation training, a very useful skill to learn!” With 100% of attendees rating the overall weekend, the venue, the food and the activities as “good” or “excellent,” it was clear that everyone had a great time. The next STWT UK weekend will be held on 14th – 16th April 2023 at the Penny Brohn Centre near Bristol.

Canada’s August gathering brought together four families, including a set of grandparents, and Dr. Hilary Vernon, Barth specialist from Maryland and BSF Scientific and Medical Advisory Board Member, who “gave an excellent presentation on The Barth Syndrome Clinic at Kennedy Krieger Institute with great audience participation,” said Susan Hone, President of Barth Syndrome Foundation of Canada. Activities were a mix of science and recreation, with trips to historic Heritage Park, a museum, and Calgary Children’s Hospital to meet with a geneticist. “We discussed many issues related to being an ultra-rare disease with fewer than a dozen patients in Canada. She gave us advice on how to make our voice stronger in the rare disease community and who to try and get to advocate for us in the medical field.”

Shelley Bowen, BSF’s Director of Family Services and Advocacy, had this to say about the STWT: “A great deal of planning goes into these events. Often something wonderful happens that can’t be described in words. It’s a beautiful experience to witness what happens when people are in the presence of others they had previously only known in a virtual space-- the energy is palpable. These events are the bridge to connect people with one another and ultimately those connections are what defines us as a community. There isn’t one person in our community who hasn’t been overwhelmed by the complexity and rarity of Barth syndrome. But through it all they know they are not alone. These events are medicine for the soul and remind them of just that.”

Since over 80% of this year’s attendees indicated they would attend another BSF World Tour, plans are moving forward for another Stronger Together World Tour in 2023 in addition to the full International Scientific, Medical, and Family Conference in 2024. However, we can’t accomplish either of the endeavors without support. If you would like to volunteer to help with either of these events, please email shelley.bowen@barthsyndrome.org. If you would like to contribute financially, whether through a donation or sponsorship, please email emily.madalinski@barthsyndrome.org for more information. Thank you to all of our volunteers, attendees, and healthcare/research professionals who made the 2022 Stronger Together World Tour possible. We are truly stronger together!

Thank you to everyone who attended the Stronger Together World Tour! See you next tour!

On November 5, 2022, with his wife Ginger cheering him on, Ghent Lummis proudly competed for TEAM WILL in Ironman Florida. He completed the 2.4 miles of swimming, 112 miles cycling, and 26.2 miles of running, raising almost $4,000 for BSF!

Ghent dedicated the race to the memory of his cousin Will McCurdy who passed away in 2014 at the age of 28 from Barth syndrome. Will led an extraordinary life, and his legacy lives in the extraordinary physical feats of TEAM WILL members, in and outside of competitions. Will's determination, wisdom, and ability to overcome countless medical challenges helped shape Ghent's decisions, not only to take on the Ironman, but to earn his medical degree at the tender age of 62!
"When Kate and Steve McCurdy asked me to join this effort in memory of their son, Will, I readily agreed," Ghent said. "The first time I met Will, and although it was a very brief meeting due to the state of his health, I was impressed with his kind heart and intellect."

Thank you and congratulations, Ghent, for championing BSF's mission!

Giving Tuesday: Together We Gave!

Can you believe it? We raised more than $123,000 this Giving Tuesday! These mission-critical dollars fund research grants, support advocacy programs, and help families and people with Barth syndrome.

Over 50 people raised funds and more than 600 individuals donated, making Giving Tuesday 2022 a record-breaking year for BSF. These numbers demonstrate that, together, it was truly a global community effort. We could not have done it without your help! Thank YOU!

We would like to extend a thank you to our fundraisers:

Diane S., Chandler L., Michelle F., Kelsey B., Kathryn M., Elizabeth H., Marykate B., Shelia M., Derek S., Anne and Bill Z., Jay R., Alanna B., Eliza M., Darryl B., Ned K., Rebecca H., Leslie B., Laura N., BJ D., Michael T., Anna B., Kristi P., Kellye F., Laura D., Susan M., Christie B., Bryan D., Amy W., Steven G., Shelley B., Melissa H., Wilkens and Wiederspan Families, John and Megan B., Steve M., Stacey W., Erik L., Brett S., Emily Mad., Emily Mil., K. Todd H., Jeremiah L. Gale C., Pat R., Tiffany D., Wesley B., Kim K.

And thank you to others who raised money for BSF this giving season to commemorate birthdays: Diego L., Jessie R., Dawn H., and Teri B.

Barth Syndrome: Psychosocial Impact and Quality of Life Assessment

Barth Syndrome: Psychosocial Impact and Quality of Life Assessment" has been published in the Journal of Cardiovascular Development and Disease (JCDD). This studay was funded through an award to principle investigator John L. Jeffries, this study was funded during our 2014 Grant Cycle by Barth Syndrome Foundation and Barth Syndrome UK/Barth Syndrome Trust.

"**Background**: Barth syndrome (BTHS) is a rare X-linked genetic disease that affects multiple systems and leads to complex clinical manifestations. Although a considerable amount of research has focused on the physical aspects of the disease, less has focused on the psychosocial impact and quality of life (QoL) in BTHS.

Methods: The current study investigated caregiver- (n = 10) and self-reported (n = 16) psychological well-being and QoL in a cohort of BTHS-affected patients and families. Participants completed the depression and anxiety components of the Patient-Reported Outcomes Information System (PROMIS) Short Form 8A and Health-related quality of life (HRQoL) surveys at enrollment and again during a follow-up period ranging from 6 to 36 months after baseline.

Results: Quality of life changed significantly over time and the various domains with some improvement and some decline. Among the available caregiver-patient dyad data, there was a trend toward discordance between caregiver and self-reported outcomes. Most notably, patients reported improvement in HRQoL, while caregivers reported declines. This suggests that there may be differences in perceived quality of life between the patients and parents, though our study is limited by small sample size.

Conclusion: Our study provides valuable insights into the impacts of psychosocial and mental health aspects of BTHS. Implications of these findings include incorporating longitudinal assessment of QoL and screening for psychological symptoms in BTHS care to identify interventions that may drastically impact health status and the course of the disease." (JCDD, 12/9/22)

Access the full article at:
www.mdpi.com/2308-3425/9/12/448

About BSF's Research Grant Program

BSF and our International Affiliates welcome innovative applications that address the basic, translational, and clinical research challenges of Barth syndrome. Through this program, BSF seeks to provide seed grant funding to young and established investigators in order to generate the preliminary data required for successful follow-on funding available from major grant-making institutions such as the National Institutes of Health (NIH).

Learn more at www.barthsyndrome.org/research

Little Book For Children Now Available in Spanish

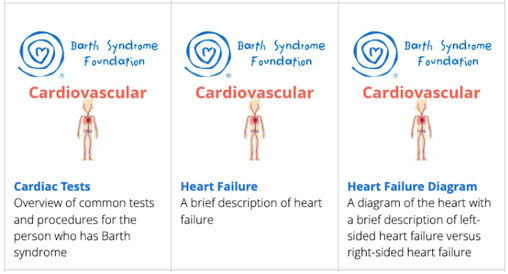
"The Barth Syndrome Little Book for Children" is now available in Spanish! This book, also available in English and Icelandic, is a resource designed to help affected children and their support network talk about Bath syndrome with peers, educators, and other community members.

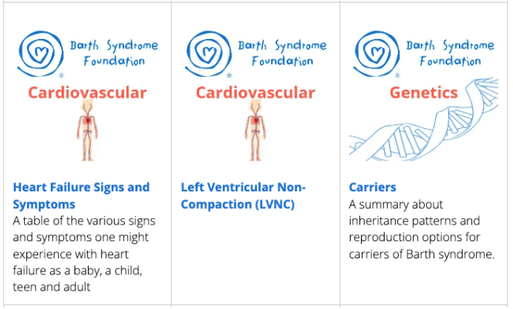
The original book was written by Florence Mannes, a Barth mother in France, and illustrated by Zoé Viot. This Spanish translation was made possible by community member and mother of a Barth syndrome affected son, Eliana Perez. As we continue to expand our Barth syndrome resource library, this was a much needed and welcomed addition.

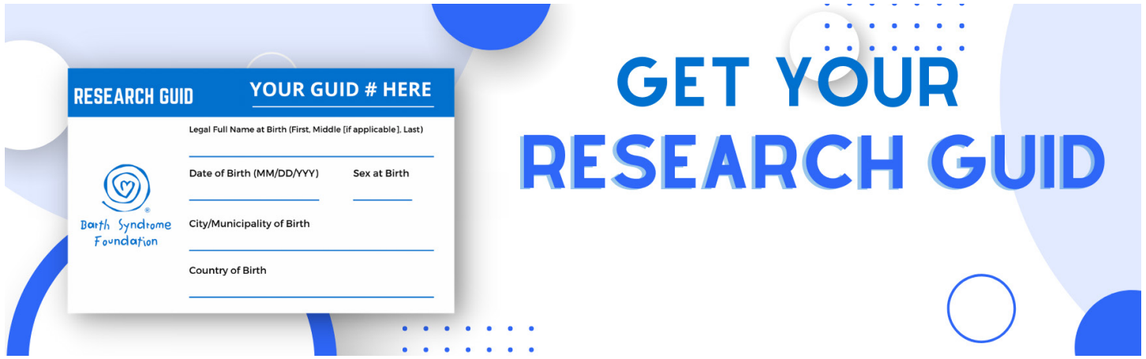
For those who are in or near their teenage years, "The Barth Syndrome Little Booklet for Teenagers" is also available. To access our full resource library, including our detailed fact sheets, please visit www.barthsyndrome.org.

Thank you again to Eliana for providing your expertise!

Visit the BSF Resource Library to access these Fact Sheets and more! www.barthsyndrome.org







Barth syndrome is an ultra- rare disease, so each piece of data generated by participating in research is of outsized importance, and its careful curation is key.

Our Research GUID (Global Unique Identifier) Program aims to:

- Efficiently connect datasets with all other Barth syndrome research efforts
- Promote data sharing and maximize the utility of each dataset
- Ensure the fidelity and confidentiality of research participant's personal identifiable information (PII)

For Affected Individuals and Families:

When you participate in a research study, researchers typically assign you an ID number that links to your data within that study. Each study uses its own ID number system, making it difficult to connect a participant's data across various studies.

The Research GUID program aims to tackle this obstacle by employing an ID numbering system that can be used to link and share your data across many studies without reveraling who you are. Affected individuals and their family members can obtain a Research GUID number and card by enrolling in the Barth Syndrome Registry and Repository. To get your own Research GUID card, please contact us.

For Researchers:

A major challenge for Barth syndrome research is its rarity and the availability of data has a huge impact on scientific progress and treatment development. Our Research GUID is a secure mechanism that can efficiently connect datasets with various other Barth syndrome clinical research efforts by employing a common participant identifier within and across research studies and repositories. By using Research GUIDs, you will be able to link your dataset to the larger Barth syndrome dataset and identify matches, thus maximizing the utility of your data. If you are interested in employing the Research GUID in your research, please contact us.

For How the Research GUID Works:

A participant shows their Research GUID card so that researchers collect the necessary PII and store that data in a local database that is not made available outside their institution.

The 9 fields required to generate a Research GUID are all listed on the card:

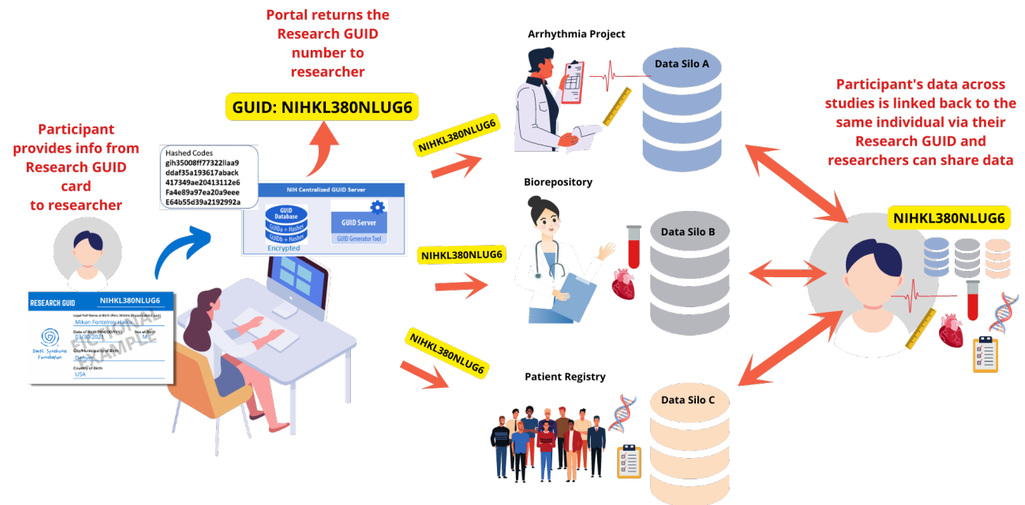
- Complete legal given (first) name of the participant at birth
- If the participant has a middle name, the complete middle name of subject at birth
- Complete legal family (last) name of participant at birth
- Day of birth
- Month of birth
- Year of birth
- Name of city/municipality in which participant was born
- Country of birth
- Sex at birth
- Advantages of the Research GUID program

As you can imagine, it is absolutely critical that each piece of this information is given and entered exactly the same way each time so the system does not think you are a different person. BSF equips members of our community with Research GUIDs to ensure the PII given to researchers remains consistent across studies. PII never leaves the organization that is inputting the data. Potential research collaborators will only need to communicate once through this universal, yet unique de-identified participant coding system for data.

Once a researcher enters the necessary PII accurately into a secure NIH-hosted server, it will generate either a new GUID or match to the participant if they already exist in the system. This GUID will stay with that research participant forever, so the process only needs to be done once. Researchers can match their list of Research GUIDs to BSF's master list and determine if there are matches across other studies and initiate the data sharing process with the other researchers/ institutions.

Contact Information:

If you have questions about our Research GUID program, want to get your own Research GUID card, or are a researcher who wishes to implement the Research GUID for your study, please reach out to Melissa Huang, Clinical Research Coordinator at melissa.huang@barthsyndrome.org



Adapted from (Nesbitt & Murphy, 2022) NIH GUID Tool

BSF Emergency Relief Program

BSF recognizes the tremendous financial burden that a hospital stay or passing of a loved one with Barth syndrome places on families. These life-altering moments often mean time away from work while simultaneously generating expenses difficult to absorb into household budgets. When families are confronting some of the most emotionally challenging moments of their lives, finances should be the last of their worries.

For this reason, BSF has funded the Barth Syndrome Emergency Relief Program through the National Organization for Rare Disorders (NORD). In 2022, BSF established the fund through NORD to support approximately 20 families in the inaugural year of the program. This program cannot directly pay medical expenses (because each family has a different insurance situation), but it can pay other bills that a family finds difficult to cover as the result of paying medical bills. Families who meet the eligibility criteria can apply for short-term assistance to cover the cost of unexpected or emergency non-medical expenses.

BSF recognizes that asking for help is sometimes hard to do. Partnering with NORD protects families' privacy and allows families to seek assistance with dignity and support offered by their specialized team of patient service representatives.

You can learn more about whether you or your family may qualify for the Barth Syndrome Emergency Relief Program by clicking here. You can also call a NORD Patient Services Representative at 1.203.292.0293 (Monday – Thursday 8:30am – 7pm ET or Friday 830am – 6pm ET) or email NORD at barthsyndrome_assist@rarediseases.org.

Help Support BSF

Since 2000, BSF has been a lifeline for those who suffer from Barth syndrome, offering 24/7 support, pioneering standards of care and diagnosis, creating collaborations between clinicians, researchers and patients, and most importantly, making sure no person with Barth syndrome is ever alone.

Your support is vital to the success of our mission, so please consider making a gift today.

Donate at www.barthsyndrome.org/donate

Welcome Emily Madalinski

Please help us welcome BSF's new Development & Stewardship Manager, Emily Madalinski! Emily is a nonprofit enthusiast with a passion for making the world a better place. She has a background in donor relations and stewardship with specialized communications and relationship building skills. Emily received her bachelor's degree in Communication with a focus in Public Relations from Florida Gulf Coast University. She also holds a master's in Communication with an Emphasis in Education from Grand Canyon University.

Prior to joining BSF, Emily held several interesting roles. First, she interned with the Make-A-Wish foundation as a special events intern. The internship thelped her land the role of Donor Relations Coordinator with the David Lawrence Center, a mental health and rehabilitation facility. Most recently, Emily held a donor relations coordinator role with the Krieger School of Arts & Sciences at Johns Hopkins University.

When not drafting donor acknowledgements, Emily likes to spend time outside and going on adventures with her corgi, Maisy. A Florida native, you can also find Emily frequently escaping the cold of Maryland and visiting family in Naples, Florida

Welcome to the team, Emily!

Barth Syndrome Community Voices

"My name is Quentin and I have been living with Barth since birth. I'll be 11 in December [2022]. I didn't understand it much when I was younger but as I've advanced in school I understand how different I am to the kids in my class. They call me small, or say I can't run fast, and sometimes it is hard to make friends. I really wish I didn't have this condition. I hope a cure is discovered someday." "The Barth Syndrome Foundation helps my mom educate my teachers, schools, and local doctors."

Advocacy: Legislation to Watch

21st Century Cures 2.0 Act

The draft of the 21st Century Cures 2.0 Act H.R. 6000 builds on the successes of the 21st Century Cures Act, passed in 2016, to advance biomedical research, regulatory science, public health, and payment policy innovation so critical for rare disease patients and families. This builds on the framework of the Cures Act and aims to further modernize the nation's healthcare pipeline in the hopes of avoiding some of the burdens that the system has faced during the COVID-19 pandemic. Some of the proposed areas for policy include public health and pandemic preparedness, healthcare delivery systems, patient engagement in healthcare decision-making, caregiver integration into the care team, modernizing CMS, and increasing diversity in clinical trials.

Additional resources available on RDLA website at:
https://everylifefoundation.org/rare-advocates

The BENEFIT Act

Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act S. 373/H.R. 4472 requires the Food and Drug Administration (FDA) to consider relevant patient-focused drug development data, such as data from patient preference studies and patient-reported outcome data, in the risk-benefit assessment framework used in the process for approving new drugs.

After a new drug application has been approved, the FDA's public statement about how it used patient experience data shall include a description of how such data was considered in the risk-benefit assessment framework. This action would send an important signal to all stakeholders that patient experience and PFDD data will be fully incorporated into the agency's review process and will encourage such entities to develop scientifically rigorous and meaningful tools and data.

We at BSF know how critically important patient information and perspective can be in therapy development, and this act would require that it is considered in development deliberations. Take Action at www.parentprojectmd.org

To learn more about the legislation that we are watching and to get involved, please visit www.barthsyndrome.org/advocacy.

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