2023 Annual Report

Our Mission

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

Our Vision

A world in which Barth syndrome no longer causes suffering or loss of life

Barth Syndrome Foundation®

Contents

FAMILY SUPPORT

Supporting Barth families comes first. Last year, we added fresh educational content to our website and engaged diverse speakers for our monthly webinars, partnered with a new medical support company, and grew our online community to span affected families living in 38 countries. *See pages 5-7 to read about how we support Barth families.*

ADVOCACY

Advocacy played an outstanding role in 2023. We focused on educating members of Congress about inequities in the U.S. drug review process to influence the Food and Drug Administration (FDA) and find partnerships for solutions toward equitable and appropriate review of a new treatment for Barth syndrome.
See pages 8-11 for more about our advocacy work.

RESEARCH & DEVELOPMENT

BSF is seeding innovation and strategically supporting scientific research with the potential to improve treatments for our affected individuals while we strive for a cure. Our grant-supported researchers are working on initiatives to identify and test Barth syndrome-specific nutritive supplements, uncover new therapeutic targets, improve immunity against infection, and more. *See pages 12-19 to learn about our commitment to and updates in research and development.*

Barth Syndrome Foundation®

Barth Syndrome Foundation (BSF) is the only global network of families, healthcare providers, and researchers solely driven by the mission to save lives through education, advances in treatments, and finding a cure for Barth syndrome.

OUR CORE VALUES

Credibility | Integrity | Inclusion | Professionalism | Compassion

A MESSAGE TO OUR COMMUNITY

Dear Barth Families, Friends, and Partners,


Emily Milligan

BSF had an extraordinary year. We have been in the public eye in a way that our 23-year-old organization never has been before. We celebrated our first-ever worldwide Barth Syndrome Awareness Day on April 5th, 2023, and filled our other days with FDA-focused advocacy and meetings with congressional leaders, all towards ensuring that our population – and others living with rare diseases – get the chance for fair and equitable access to potential therapies.

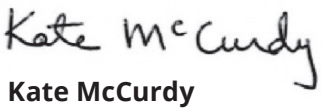
2023 demonstrated that results alone aren't sufficient to move the needle. While we have seen first-hand that our first potential FDA-approved therapy, elamipretide, works for many people, the FDA continued to resist review of a New Drug Application (NDA) for our ultra-rare disease and tested both our commitment and fortitude and that of the potential drug's sponsor, Stealth BioTherapeutics. However, our community is strong as well as persistent, and we taught ourselves to advocate even better for our cause -- showing up, speaking out, and paving a path forward. We have been proactive along many fronts and ended 2023 with nearly 20,000 signatures on our petition to the FDA seeking a fair, equitable and appropriate review of the NDA that has been submitted to them. We have rallied together, we are being heard, and we are harnessing that momentum for the work that still lies ahead. To all who joined in this effort, we thank you!


Kate McCurdy

In this annual report, we share with you what has stood out to us in 2023. We include big moments from our advocacy and research work as well as ways BSF remains an indelible presence, by providing community and healthcare support, in the lives of our families. We share notable research updates from many of the outstanding scientists who strive to better understand Barth syndrome and find new and life-improving therapies, and we celebrate all of you who have put so much work toward our goal of saving lives through education, advances in treatments, and finding a cure for Barth syndrome.

Our commitment, strength, and remarkable capabilities were on display in 2023, and we are so proud of this community. We give thanks for the individuals who make BSF the extraordinary organization that it is, for the donors who support our critical initiatives, for our families for their dedication and engagement, and for the volunteers who give their time, talents, and other resources in support of our mission. We are grateful to all of you and look forward to working together towards a world where people with Barth syndrome lead healthier, longer lives.

Emily Milligan
Executive Director



Kate McCurdy
Board Chair

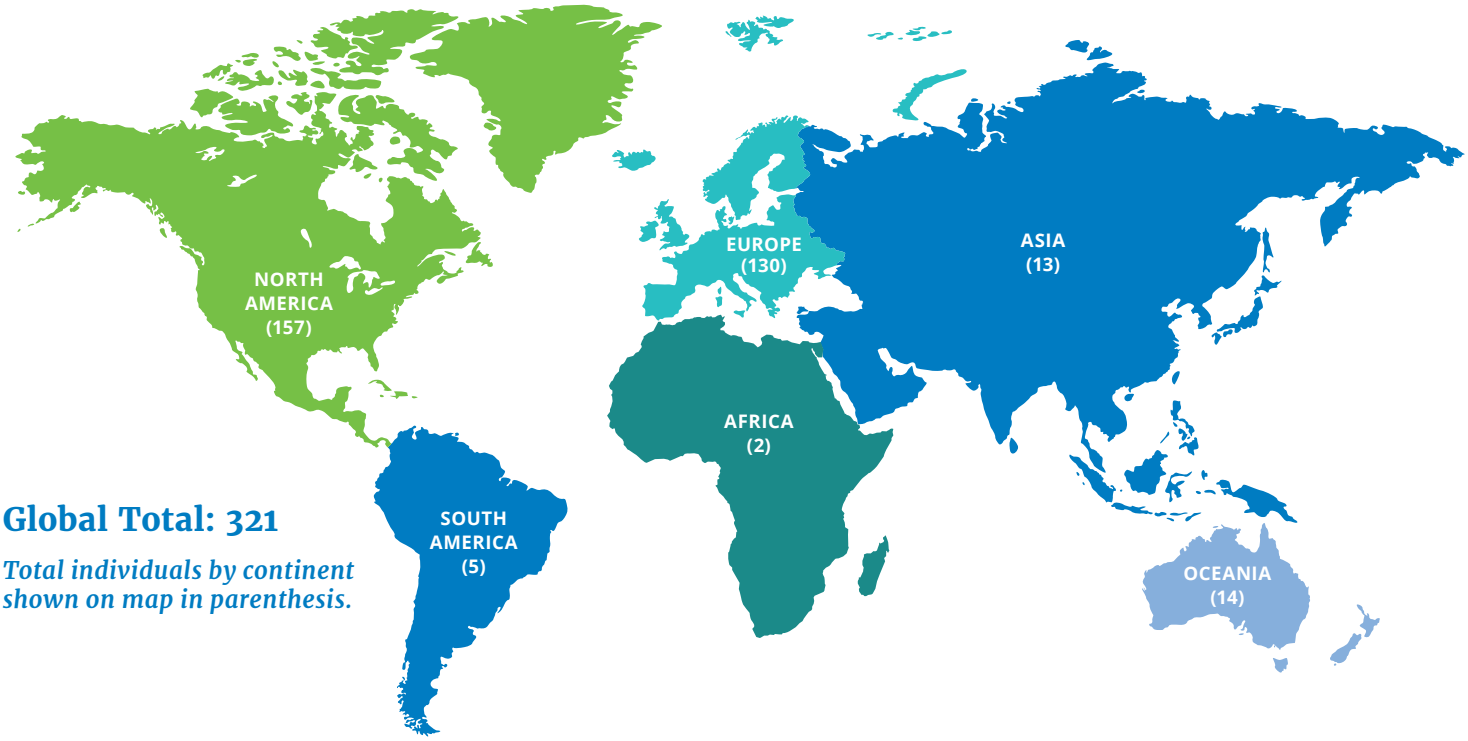
GLOBAL STATISTICS

321 individuals are living with Barth syndrome worldwide*

*as of December 2023

"The foundation is a lifeline to us and so many others."

— **Megan B.**, mother to Henry



Global Total: 321

Total individuals by continent shown on map in parenthesis.

By Continent & Country

AFRICA	ASIA		EUROPE			NORTH AM.	SOUTH AM.	OCEANIA
Egypt (2)	China (2)	Israel (3)	Austria (1)	Germany (11)	Slovakia (1)	Canada (16)	Argentina (2)	Australia (13)
	Taiwan (2)	Korea (1)	Belgium (6)	Ireland (4)	Scotland (3)	Mexico (1)	Bolivia (1)	New Zealand (1)
	Vietnam (2)	Kuwait (1)	Bulgaria (2)	Italy (16)	Spain (6)	USA (140)	Brazil (2)	
	India (1)	Saudi Arabia (1)	Czechia (6)	Netherlands (11)	Switzerland (1)			
			Denmark (1)	Poland (6)	Turkey (1)			
			England (26)	Portugal (2)	Ukraine (1)			
			France (19)	Russia (4)	Wales (2)			

Source: Barth Syndrome Foundation Inc. and Affiliates

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OUR COMMUNITY

Supporting Our Barth Families

BSF is proud of its active role in supporting our families through up-to-date educational content shared through our website, webinars, research roundtables, various family support programs, and online communities. BSF is for Barth families.

FAMILY SUPPORT GOALS

- Offer easier access to critical information about Barth syndrome for families
- Provide outside experts who can help affected individuals and families better understand the disease
- Support affected individuals and families when they are scared, unsure, or in the middle of a crisis
- Connect families across the world through our conference, outreach events, and online communities
- Include affected individuals in opportunities to help steer our future

Scan code to learn more about BSF

April 5th Declared as Barth Syndrome Awareness Day!

The first-ever Barth Syndrome Awareness Day was celebrated in 2023 around the world on April 5th (4.5) to bring together all members of our ultra-rare disease community for a single cause: to increase awareness of Barth syndrome so all affected individuals can access safe and effective treatments. We couldn't be prouder of this formal recognition through a Congressional resolution introduced by Representative Tonko of New York.

Why 4.5? Because Barth syndrome is caused by a mutation in the *TAFAZZIN (G4.5)* gene.

Educating to Understand Barth Syndrome and Improve Treatments

BSF has developed 25 fact sheets on topics from how to swallow pills to anesthesia considerations, all available on our website. Last year, in response to inquiries from the community, we added two new fact sheets on neutropenia in infants and heat intolerance.

Throughout the year BSF hosted 13 webinars with invited guest speakers with the goal to educate our community about ongoing research, their potential role in advocacy, and management of Barth syndrome.

Facilitating Partnerships For Barth Families

While we hold hope for future treatments and a cure, we understand that families need assistance now. The Barth Syndrome Emergency Relief Program administered through the National Organization for Rare Disorders (NORD) aims to ensure families can focus on caring for loved ones without the added stress of financial instability. In 2023 alone, BSF extended critical aid to 6 families experiencing financial hardship. This program, entirely funded by generous donations to BSF, provides funds that go directly to individuals and families in need.

> *"The community, even though it is remote, remains a support network of people who know what we are going through."*
>
> — **Kate M.,** mother of an affected individual

MedicAlert Partnership

In 2023, BSF established a partnership with MedicAlert for individuals with Barth syndrome in the U.S. MedicAlert provides services to relay critical medical information to first responders during an emergency. This includes a digital health profile of the affected individual's critical health information, 24/7 emergency response team, emergency contact notification, and more.

> *"He has not had to use it, but he wears it every day!"*
>
> — **Mother of Luke F.,** a Barth syndrome teen with a medic Alert bracelet

Connecting With Each Other Around the World

With today's latest technologies, BSF serves as the nexus for Barth families to stay informed, remain connected and offer support to one another irrespective of geographic and language barriers.

CONNECTIONS BY THE NUMBERS

2,700
members in our Facebook group

461
members in our Family Chat

162
members in our Family BAND group

Events & Fundraisers:

YOUR EFFORTS ENABLE OUR PROGRESS

By supporting Barth Syndrome Foundation, you not only contribute to ongoing research and initiatives but also play a crucial role in raising awareness for this cause.

Celebrating Happy Heart Week in 2023

Happy Heart Week is an annual awareness and fundraising campaign championed by BSF Board Member Megan Branagh. The week-long celebration is in honor of her son, Henry, who is living with Barth syndrome.

Raised: $123,000

Hockey With a Heart Benefitting BSF in 2023

Each year, a child with Barth syndrome is honored by the New York Islanders hockey team for "Hockey with a Heart" benefitting BSF. In 2023, Henry N. performed the ceremonial puck drop at the March 25th game.

Raised: $117,000

Steven Woodward Memorial

The Woodward family launched fundraising efforts in memory of Steven David Woodward (1974-2023), uncle of Connor who is affected by Barth syndrome. These donations, nicknamed the "31 Fund" after Steve's lifetime hockey number of 31, are part of the BSF Family Services Fund which, among other things, helps mitigate financial challenges that would prevent certain Barth syndrome-affected individuals and families from fully participating in BSF programs.

Raised: $31,000

Giving Tuesday 2023

Through our peer-to-peer campaign, individuals within our community passionately reached out to their personal networks, sharing the importance of supporting those affected by Barth syndrome. In 2023, our efforts surpassed expectations and we exceeded our fundraising goal. Together, we make a meaningful impact, empowering progress, support, and hope for those living with Barth syndrome.

Raised: $109,000

Henry B.


Henry N. & Family


Steven & Connor

Luke's Make-A-Wish Gift

Luke F., a remarkable 17-year-old with Barth syndrome, was asked by the Make-A-Wish Foundation about his one true wish. In a selfless act of compassion, Luke chose, "To give it to BSF to help find a cure," and was presented a check by Dr. John Jefferies, BSF SMAB member, underscoring the profound impact of Luke's extraordinary gesture.

Raised: Priceless

Fighting For a World Where Barth Syndrome No Longer Causes Suffering or Loss of Life

BSF's journey with Stealth BioTherapeutics and elamipretide began in 2014 when BSF asked Stealth to include people with Barth syndrome in their clinical trial. In 2021, Stealth submitted the elamipretide New Drug Application (NDA) for treatment of Barth syndrome to the U.S. Food and Drug Administration (FDA). Despite compelling evidence that elamipretide was well-tolerated, demonstrated meaningful clinical benefit in Barth syndrome, and ongoing efforts by BSF and other advocates to engage with regulators, the FDA refused to accept the NDA. BSF and our community continued our efforts in 2023 to encourage the FDA to review a new NDA submission for elamipretide and allow people with Barth syndrome the possibility to access this potential new therapy.

Declan C. and his mother Jamie D.

(Update: *In early 2024, due in large part to BSF's exhaustive efforts, the FDA agreed to fully review an NDA for elamipretide.)*

STRATEGIC ADVOCACY GOALS

- Promote the interests of our community with state and federal legislators
- Give members of our community the tools they need to champion our cause
- Generate external interest by sharing community stories and the science of Barth syndrome
- Involve ourselves in product development and regulatory processes when we believe it will help
- Plan a health impact study and educate payors to improve access to care

2023 BSF ADVOCACY EFFORTS

19,374	16,000+	334	291	86
signatures on our petition to FDA	views of *Not Too Rare to Care* website	advocates trained through virtual 3-part series workshop	letters sent to the FDA	Congressional meetings with Barth constituents

THE CHALLENGE:

Researchers have reported that many Barth syndrome patients taking elamipretide have experienced remarkable improvements: over 40% in heart function, more than 25% in exercise tolerance, and over 40% in muscle function—none of which are expected in the natural progression of Barth syndrome. These improvements have led to transformational and meaningful changes in their lives.

However, the ultra-rare nature of Barth syndrome makes it difficult to conform to the rigid standards applied to larger clinical trials that the FDA typically reviews. Recognizing this disproportionate burden, and acknowledging the urgent and unmet clinical needs of people with devastating diseases, the U.S. Congress granted the FDA "regulatory flexibility" to adopt appropriate approaches for determining the efficacy of therapies for small patient populations with serious diseases and no specific treatments like those with Barth syndrome. Unfortunately, regulatory flexibility is subject to the discretion of regulators, which leads to its inconsistent application.

BSF and our patient population have been working tirelessly to encourage the FDA to exercise its regulatory flexibility and review an NDA of elamipretide, the first potential treatment for Barth syndrome. In doing so, we aim to afford patients with Barth syndrome the same opportunities for new therapies as have been granted to other patient groups faced with similar constraints.

Challenging the FDA to Address Our Treatment Needs

Kate McCurdy, board chair of BSF, hand delivered our petition requesting review of the NDA for elamipretide at the FDA White Oak Campus. Our petition amassed nearly 20,000 signatures within 2 months from individuals spanning all 50 U.S. states; Washington, DC; Puerto Rico; the U.S. Virgin Islands; deployed military personnel; and from almost 60 countries – a major advocacy initiative on behalf of our community and all rare diseases.

Kate McCurdy

> *"The potential of this drug — this isn't just a 'give you some energy' type drug. This is a 'give you a new life' type drug. This is a 'live what it's like to be a real human', for once..."*
>
> — **Tyler G.,** affected individual, quoted from a *Boston Business Journal* article


Steve G.

Teaching Our Own How to Advocate and Share Our Stories

BSF organized and hosted a 3-part webinar series to educate and mobilize the community around advocacy efforts to influence the FDA.

> *"Over time, the Barth Syndrome Foundation became less like an organization and more like a family. 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 G.**

Steve, who lives with Barth syndrome, spoke with representatives from the Kentucky state legislature on behalf of BSF's advocacy efforts.

BSF spearheaded a grassroots advocacy campaign called *"Not Too Rare to Care"* with an accompanying new website to raise awareness about Barth syndrome and the community's advocacy.

Scan code to visit *Not Too Rare to Care* website

Involving Legislators to Join Our Cause

The BSF team held 65 congressional meetings via Zoom and 21 in person with legislators across the country. These meetings educated congressional leaders about inequities in the U.S. drug review process and requested their partnership on solutions for fair, equitable, and appropriate review of potential new treatments for Barth syndrome.

65 Congressional meetings via Zoom

21 in-person Congressional meetings

BSF had the honor of participating in a Senate special hearing on the unmet medical needs faced by individuals with rare, progressive, and serious diseases led by Senator Mike Braun (R-IN), who serves as the Ranking Member of the Senate Special Committee on Aging, and Kirsten Gillibrand (D-NY).

> *"This was a unifying experience to be part of something bigger and join in solidarity, working together to fix parts of the FDA that aren't serving us or our loved ones well."*
>
> — **Emily Milligan,** Executive Director of BSF

Dr. Hilary Vernon

Senators Braun and Gillibrand photographed with Emily Milligan, Dr. Hilary Vernon, and other rare disease advocates.

Representative Tonko of New York's 20th District met with constituent Lynda S. to express his solidarity with the Barth syndrome community and sponsor H.Res.276 for Barth Syndrome Awareness Day.

Watch Emily Milligan and Dr. Hilary Vernon's Senate Speeches.

Barth families and advocates in Florida met with District 12 Representative Bilirakis to garner support for improvements to regulatory reviews of new drug applications for Barth syndrome therapies.

Partnering With Peers and Publishing for Biopharma Attention

BSF worked to galvanize support for the Barth syndrome community and mobilize the broader rare disease network to advocate for equitable regulatory solutions. This included partnering with peer organizations (UMDF, MitoAction, Global Genes, EveryLife Foundation) on several podcasts, participating together in speaking opportunities, and being highlighted in these organizations' communications.

Our efforts pushing for the FDA to review elamipretide continue into 2024. **Elamipretide is the only medicine currently in late-stage development for our life-threatening, ultra-rare disease.** We cannot thank our community enough for all the hard work and dedication they have put into advocacy efforts. Your continued support is indispensable. Together, we can make a lasting impact and bring about positive change.

not too rare to care ♡

Eliza McCurdy Colón, English Mann, John Wilkins, Marykate Baffa, and Alanna Boozer

Visit nottooraretocare.org/get-involved where you can write to the FDA, record your story, or let us know that you are willing to share your story with the media or attend an advocacy meeting.

Strategically Supporting Research

We are proud that in 2023 we expanded our investment in research to include a coordinated strategy of three different funding streams designed with complementary end goals, including our Seed Grants for early research, and two new funding mechanisms, the Dr. Iris L. Gonzalez Prize and our Strategic Initiatives Program.

BSF'S THREE STRATEGIC STREAMS OF FUNDING FOR 2023

1. Seed Grants

Seed grants are designed to identify novel, early-stage ideas. This is the only vehicle that exists for innovative, high-risk, high-reward projects to obtain non-dilutive funding and develop into fully fledged projects that can meaningfully change Barth lives. It also assists researchers in establishing proof-of-concept data prior to applying to larger funding institutions like the National Institutes of Health (NIH).

2. Dr. Iris L. Gonzalez Prize

The Dr. Iris L. Gonzalez Prize honors the top 2023 applicant advancing our collective understanding of genetic variants in *TAFAZZIN*, the gene responsible for Barth syndrome, through the use of BSF's meticulously curated database. Twenty-three years ago, Dr. Gonzalez launched the Human *TAFAZZIN* Variants Database, diligently aggregating variants (also known as mutations) of *TAFAZZIN* from genetic reports. This prize, named in recognition of Dr. Gonzalez's unprecedented contributions to Barth syndrome research, aims to accelerate BSF's mission by promoting expanded scientific exploration of this invaluable database.

3. Strategic Initiatives Program

The newly created Strategic Initiatives Program aims to advance potential therapies and interventions for our population that otherwise would not be possible or would experience significant development delays without the Foundation's support. The goal is simple: support the delivery of safe, effective treatments and a cure to our population faster. Importantly, this program extends beyond traditional grant funding mechanisms, enabling BSF to operate as a strategic partner in the R&D process.

In 2023, two mission-critical projects were funded through the Strategic Initiatives Program: 1) Optimization of Barth Syndrome Gene Therapy and 2) the Arrhythmia Project. Read more about these exciting projects on pages 18 and 19.

Since 2002, BSF has invested...

$6.6M
in research which has translated into...

$37.7M
in follow-on funding from other agencies and catalyzed...

$75M+
in additional funds for clinical trials.

To achieve these goals, BSF has invested $6.6M U.S. Dollars (USD) in research since 2002, which has translated into $37.7M USD in follow-on funding from other agencies and catalyzed more than $75M USD in additional funds for clinical trials. We are proudly at the forefront of seeding cutting-edge research that will lead to a world in which people with Barth syndrome are able to live healthier, longer lives.

Saving Lives Through Advances in Treatments and a Cure for Barth Syndrome

BSF is the only organization in the world uniquely focused on funding research and development (R&D) to deliver treatment options and eventually a cure for people living with Barth syndrome. Because Barth syndrome affects the cardiac, immune, musculoskeletal, and other systems in the body, we seek out and support researchers from an array of specializations who demonstrate scientific excellence and innovation.

RESEARCH & DEVELOPMENT GOALS

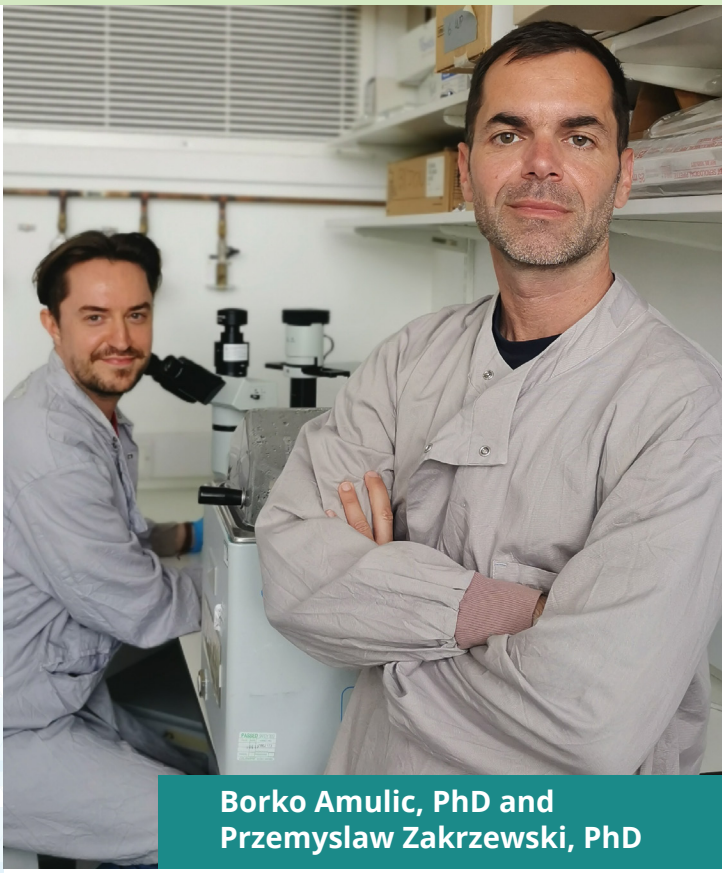
- Deepen our knowledge of the underlying biology of disease
- Pursue scientific hypotheses that may lead to new drug targets and therapeutic ideas
- Fund discovery research and tools to improve our disease understanding
- De-risk research programs to increase the potential of downstream partnering opportunities
- Assist in collaborative therapy development from bench to bedside

The Barth Syndrome Foundation Enriches R&D Globally

In 2023, over 25 papers on Barth syndrome were published in peer-reviewed journals including *Nature Metabolism, PLoS One, EMBO Molecular Medicine* and *Human Molecular Genetics.*

In 50% of the papers published on Barth syndrome in 2023, the senior authors received previous support from BSF, demonstrating the consistent role the Foundation plays in catalyzing research advancements.

Over 30% of the papers published in 2023 include BSF's Scientific and Medical Advisory Board (SMAB) members in the authorship.



Borko Amulic, PhD and Przemyslaw Zakrzewski, PhD

NOTEWORTHY EXCERPTS & ARTICLES

Altering disease onset and developing new treatments require researchers to first understand precisely the ways a mutation in the *TAFAZZIN* gene leads to the clinical symptoms of Barth syndrome. In their paradigm-shifting work, Miriam Greenberg, PhD (SMAB member and Scientific Liaison to the Board of Directors) and colleagues published in *Nature Metabolism* demonstrating the loss of the functional protein *TAFAZZIN* leads to structural and functional defects in the mitochondria and inappropriate interactions with a different protein in the mitochondria.

RESEARCH SPOTLIGHT

KAGAN ET AL., *NATURE METABOLISM* 2023

For the first time, researchers have demonstrated that high levels of the cardiolipin precursor, mono-lyso-cardiolipin, that results from loss of *TAFAZZIN,* lead to dysfunctional organization of the mitochondrial lipid membrane and drive an abnormal interaction of mono-lyso-cardiolipin with cytochrome c. This interaction causes cytochrome c to generate toxic lipid products that lead to reduced energy production and other mitochondrial dysfunctions. Imidazole oleic acid, a compound that blocks the toxic activities of cytochrome c, can lead to improvements in fatigue in a fruit fly model of Barth syndrome. In future studies, the authors will try to identify the most effective inhibitors of the peroxidation driven by cytochrome c and test them in a mouse model of Barth syndrome. These future studies have the potential to generate a new class of therapeutics for the treatment of Barth syndrome.

Innovative Research That Improves Our Understanding of Barth Syndrome

SMAB member Todd Cade, PT, PhD and collaborator Christina Pacak, PhD demonstrated that exercise before administration of gene therapy improved uptake, opening a new door into possible improvements to gene therapy studies. Pacak and Cade have applied for follow-on funding from NIH as a result of this exciting work.

RESEARCH SPOTLIGHT

PACAK ET AL., *JOURNAL OF TRANSLATIONAL MEDICINE* 2023

Pacak, Cade et al., found that four weeks after receiving AAV, the Barth syndrome mice that exercised before receiving the gene therapy showed increased *TAFAZZIN* gene expression in the heart and skeletal muscles, key targets for the treatment of Barth syndrome. One of the most important findings in this paper is the demonstration of improved mitochondrial function (energy source for cells) in the exercise group compared to mice that received AAV but did not exercise. These results mean that fatigue, one of the key features of Barth syndrome that impact quality of life, may also be similarly improved. Future work from this team will look at how different exercise routines or different versions of AAV impact *TAFAZZIN* gene expression and mitochondrial function.

Robin Duncan, PhD, who was funded by BSF in 2016 and 2020 with financial support from our affiliates in Canada and the UK, published two papers in 2023 characterizing *TAFAZZIN* knockout and female carrier mice. Read more about their work in the Research Spotlight below.

John Chan, Michelle Tomczewski, and Alex Cocco

RESEARCH SPOTLIGHT

TOMCZEWSKI ET AL., *BIOMEDICINES* AND *BIOLOGY* ARTICLES, 2023

The Duncan lab characterized a new mouse model of Barth syndrome where the *TAFAZZIN* gene is genetically removed (Taz-KO). They evaluated anatomy, metabolism, physical activity levels and exercise capacity in male mice at different ages. The team found many commonalities between the Taz-KO mice and human Barth syndrome, supporting the use of the Taz-KO mouse model as a new resource for understanding the pathophysiology and potential treatments for Barth syndrome. In a second mouse study, they found that female carriers were subtly different from unaffected females. At an age corresponding to human "middle age," female carriers reached exhaustion faster than unaffected females and had better blood sugar regulation. Their results suggest that female carriers endure an energetic cost and may have some protection from diseases like diabetes.

The Next Generation of Scientific Advancements

The BSF peer-reviewed research grant program provides seed funding to young and established investigators to generate preliminary data required for successful follow-on funding available from larger federal and private funding institutions.

Rigorously vetted by BSF's Scientific and Medical Advisory Board (SMAB) and external subject-matter experts, grantees demonstrate our unrelenting commitment to contribute to the advancement of disease biology with the goal to identify potential treatments.

In 2023, BSF granted upwards of $250,000 to four grantees

Over 75% of grant applicants had not previously received funding from BSF, broadening and diversifying the wealth of experts working in our discipline

35% of applicants were young investigators and 64% applied from outside the United States

Funding Young Investigators To Grow Barth Syndrome

By attracting emerging scientists at the beginning of their careers, BSF is seeding the next generation of dedicated researchers and physicians to focus professionally on finding treatments for Barth syndrome.

Through a partnership established with the American Heart Association, BSF has expanded our grant opportunities to include funding of trainee research.

2023 Seed Grant Program Awardees

Elucidating Cardiolipin Immune Dysfunction in Barth Syndrome

Kate Schroder, PhD, University of Queensland; *co-funded with BSF of Canada*

Dr. Schroder's project investigates a possible new drug target which could expand treatment options to prevent individuals' susceptibility to infection, thereby improving people's health and extending lifespans.

Induced Pluripotent Stem Cells to Study Neutrophil Development in Barth Syndrome

Taco Kuijpers, MD, PhD, Amsterdam University Medical Center

Dr. Kuijpers' research uses human induced pluripotent stem cells to make neutrophils and investigate whether a new compound, which appears to improve neutrophil formation in an animal model of Barth syndrome, may provide therapeutic benefit for people with Barth syndrome who contend with specific immune deficiencies.

Improving Physical Performance and Cardiac Function of TAFAZZIN Deficient Mice with Nicotinamide Riboside

Riekelt Houtkooper, PhD, Amsterdam University Medical Center

Dr. Houtkooper's lab found that nicotinamide riboside may make an essential molecule for energy production in the body's mitochondria. Through this grant, the team assesses its effectiveness in a mouse model to understand whether the molecule could represent a therapeutic option for people with Barth syndrome.

Optimizing Cardiac Energetics in Barth Syndrome

John Ussher, PhD, University of Alberta

Dr. Ussher's research explores whether using liver-produced ketone bodies as an alternative energy source can improve heart metabolism and reduce the risk for heart failure in a mouse model of Barth syndrome.

Foundation's R&D Community

Function of TAFAZZIN in Antibacterial Immunity

Mack Reynolds, PhD Student, University of Michigan

Winner of the 2023 American Heart Association/BSF Predoctoral Fellowship, Mr. Reynolds examines the ways specialized immune cells called macrophages behave during bacterial infections in patients with Barth syndrome. This study analyzes macrophage metabolism (i.e., how cells make energy) in the context of bacterial infections by studying the impact of different levels of protein production which is commonly impaired in Barth syndrome. This research will help understand specific ways macrophages contribute to inflammation and infection in Barth syndrome and may inform potential drug targets.

Strategic Investments Propel R&D Forward

Since the launch of our peer-reviewed seed grant program in 2002, we have supported research projects that span a range from discovery bench science, to physiological and psychosocial characterization of our population, to innovative cellular and animal models of disease. As the understanding of underlying Barth syndrome science has evolved, so too has our approach.

In 2023 BSF launched the Strategic Initiatives Program to accelerate targeted approaches and advance potential therapies and interventions that otherwise would not have been possible without the Foundation's support. This program extends beyond traditional grant funding mechanisms so the Foundation can operate as a strategic partner in the R&D process.

STRATEGIC INITIATIVES PROGRAM

GOAL: Accelerate targeted approaches to advance potential therapies and interventions through the R&D continuum

Translational science

Translate basic bench science into therapeutic and/or clinical applications.

Community input

Integrate consolidated perspectives from the Barth syndrome community.

Early-stage outcomes

Capitalize on outcomes of early-stage research that may inform future products or interventions.

De-risking development

Empower research team through collaborative iteration, partnership opportunities, and BSF support.

Cardiac Natural History Study Advances Collective Understanding and Standards of Care in Barth Syndrome

BSF focuses on curative therapies of tomorrow, and treatment options that improve life for people today.

Irregular heartbeats, known as cardiac arrhythmias, are a major cause of mortality in people with Barth syndrome. To better predict arrhythmias and prevent further loss of life, an initial team of researchers under the leadership of Drs. Reina Tan and Colin Phoon from New York University and collaborators from Johns Hopkins University and Children's Hospital of Pittsburgh launched a multi-site effort to retrospectively compile cardiac data from people with Barth syndrome. With a more complete picture of the cardiac natural history of Barth syndrome, the team intends to eventually expand this initiative to prospectively collect cardiac imaging and extended cardiac rhythm monitoring data to generate near-term, life-saving care management guidance.

This important strategic initiative has been made possible, in great part, by support from BSF affiliate the Association de Barth France.

Optimization of a Barth Syndrome Gene Therapy

BSF has pioneered the funding of gene therapy research in Barth syndrome for over 10 years. The very first grant for gene therapy was awarded in 2011 to Barry Byrne, MD, PhD (University of Florida). Since then, two additional gene therapy grants have been awarded to Christina Pacak, PhD (University of Minnesota) in collaboration with Dr. Byrne and Todd Cade, PT, PhD (Duke University).

Beginning in 2022, BSF expanded our pursuit of a gene therapy candidate and made strategic investments in Dr. William Pu's lab (Boston Children's Hospital) to catalyze the discovery of novel gene therapy capsid candidates. With safety being one of BSF's priorities, we are exploring next generation gene therapy approaches that could provide higher therapeutic benefit to patients at a lower dose and lower risk.

In parallel to the work that BSF funds in academic labs, Drs. Byrne, Cade, Pacak and Pu collaborated last year in applying for a Bespoke Gene Therapy Consortium grant from the National Institutes of Health (NIH). Although not selected, this powerhouse of investigators was one of a handful of applications that impressively advanced to the final round of funding consideration.

BSF has already invested over a quarter of a million dollars in gene therapy research, including acquiring a bench of project management, regulatory, and technical external consultants to guide our research for maximum rigor and efficiency. These initiatives, however, are just the tip of the iceberg—championing a gene therapy from the lab bench through clinical trials will take several more years and will cost millions of dollars more but may ultimately offer the most transformative therapeutic option for our community.

Young Wally B.

January 1, 2023 – December 31, 2023

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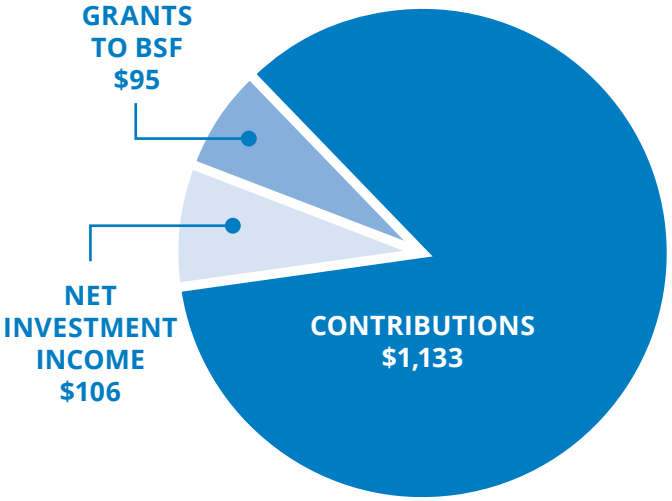
Our operations are driven by our unwavering commitment to our mission. We judiciously invest a portion of our accumulated funds to bolster therapy development and enhance family services. We extend our heartfelt gratitude to our donors for enabling these critical investments and supporting our ongoing operations. As of December 31, 2023, BSF maintained a robust financial position, with over $5 million in net assets.

2023 Revenues
TOTAL REVENUE $1,335
(in $ Thousands)



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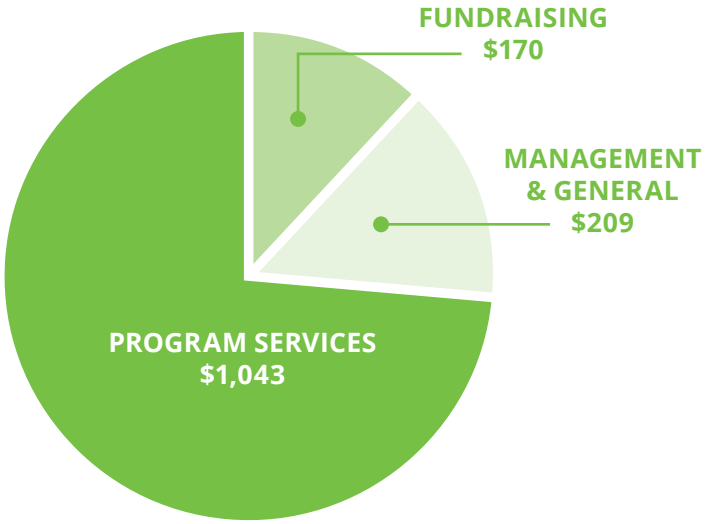
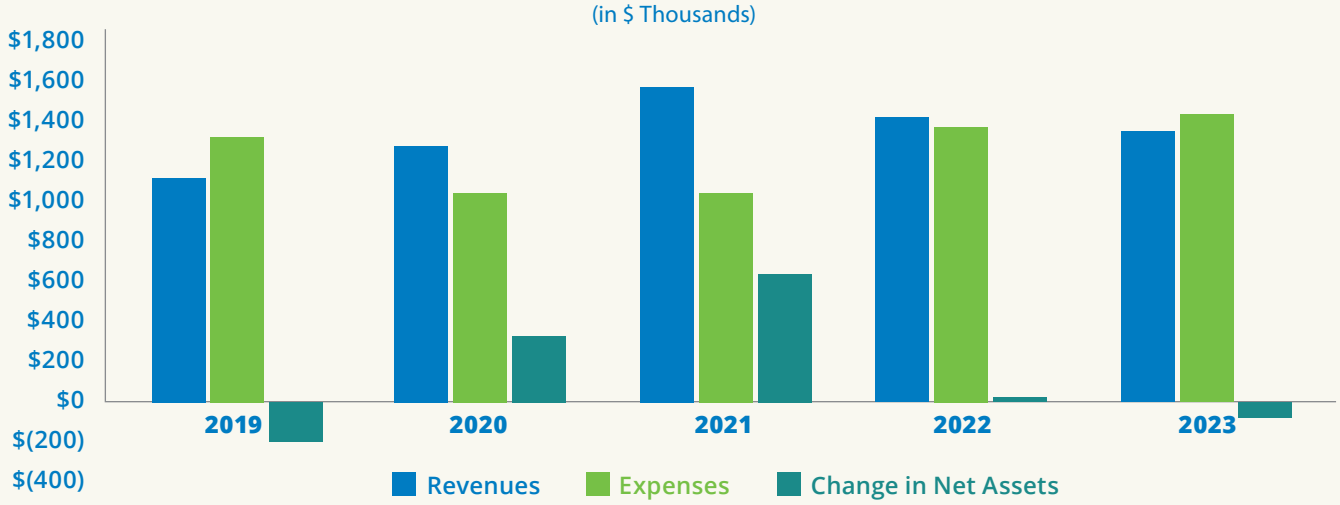
2023 Expenses
TOTAL EXPENSES $1,422
(in $ Thousands)



PROGRAM SERVICES BREAKDOWN

Research & Therapy Development	$626
Advocacy & Family Services	$265
Community Building	$151

Five-Year Trends in Revenues, Expenses, and Net Assets
(in $ Thousands)



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