



## Dr. Erik Lontok Joins BSF as Director of Research



*Dr. Lontok*

BSF welcomed Erik Lontok, PhD, as the foundation's new Director of Research in August 2019. Lontok succeeds BSF's prior Director of Science, Matthew Toth, PhD, in a new role that has been established to leverage the past two decades of research partnerships and achievements toward the development of viable therapies for people with Barth syndrome. Trained as a biochemist at the University of California, San Francisco, Lontok joins BSF after serving as the Chief Science Officer of the Lipedema Foundation where he successfully engaged world-class researchers, key clinical leaders, and patient advocates to launch the first lipedema research program.

Lontok brings direct experience and enthusiasm to his role and focus on advancing scientific and medical research in support of patients and rare disease therapies. Through appointments at FasterCures, the Center for Strategic Philanthropy, and the Forum for Collaborative Research, Lontok has led and managed efforts to accelerate the pace of scientific discovery in lipedema, inflammatory bowel disease (IBD), chronic myelomonocytic leukemia, as well as advanced regulatory science for hepatitis C virus (HCV) and human immunodeficiency virus-1 (HIV) infections. Alongside the BSF Science and Medical Advisory Board, Lontok will be responsible for guiding BSF's scientific strategy, overseeing the peer-reviewed grant-making program, advancing the Barth Syndrome Registry, and driving the collaborations needed to develop treatments for Barth syndrome.

## FDA Stakeholder Workshop

BSF participated in the U.S. Food and Drug Administration (FDA) stakeholder workshop on Sept. 6 to address recent advancements and key challenges facing mitochondrial diseases. BSF Executive Director Emily Milligan rounded out the patient advocacy perspective of the discussions alongside Mindy Leffler (Casimir Trials) and Phil Yeske (UMDF). BSF's central takeaway from the discussion is the importance of understanding the natural history of Barth syndrome, which we continue to address by supporting the Barth Syndrome Patient Registry, on-site research conducted during the International BSF conference, and Barth syndrome clinics led by Hilary Vernon (JHMI), Germaine Pierre, and Effie Chronopoulou (NHS, Bristol UK).

**“Central to the success of BSF is the sense of community and partnership demonstrated by patients, families, clinicians, and researchers.”**

**- Dr. Erik Lontok**

# BARTH 2020: BSF's International Conference

## Collaboration, Community Building Blocks of Biennial BSF Conference

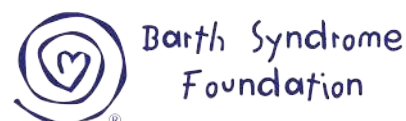
Registration is now open for #BARTH2020! Join Barth Syndrome Foundation (BSF) in its 10th biennial conference, a multi-track event that brings together researchers, clinicians, and families from around the world to help advance the potential for new therapies for Barth syndrome.

The first gathering was 20 years ago at a time when most families caring for children with the condition were being advised that they were "the only ones" with the rare disorder. Defined by and named after Dr. Peter Barth of the Netherlands, Barth syndrome and BSF were little more than words on paper and passionate intentions of parents and doctors back in 2000.

Today, the Barth syndrome community is global and the conference attendees represent more than 12 countries, 250 family members, and nearly 100 scientists and clinicians.

Celebrating the 20th anniversary of the organization and coinciding with the year 2020, BSF is launching a new decade of research for Barth syndrome with the theme "For a new GENERation."

This year, five parallel tracks are planned:



- Track 1: Family and Youth Educational Sessions
- Track 2: IRB-Approved Human Subjects Research
- Track 3: Updates in Research, Science, and Medicine Related to Barth Syndrome for Researchers, Clinicians, and Scientific Professionals
- Track 4: "We are Stronger Together": Community-Building Program
- Track 5: Symptom and Disease Management Forum for Healthcare Providers

Learn more and register: [barthsyndrome.org/conference](http://barthsyndrome.org/conference)

### New Family Scholarship Program Established to Allow More Families to Attend #Barth2020



New for the 2020 BSF conference, a pilot Family Travel Scholarship program has been established to help individuals with Barth syndrome and their families offset costs of attending the conference. Unique to BSF's conference and central to our mission, registration is free for all families and affected individuals, as well as most of the food and entertainment. However, as we recognize that travel and lodging costs associated with the conference may be unrealistic or cause extreme hardship for some in our community, BSF has pledged to offer up to 10 scholarships for #Barth2020.

Support from sponsors and generous donors makes this program possible. Please consider learning more and sponsoring a family at [bit.ly/Barth2020gift](http://bit.ly/Barth2020gift).

Learn more and apply at [barthsyndrome.org/conference](http://barthsyndrome.org/conference)



# RESEARCH

## BSF Conducts First Research Portfolio Review

Members of BSF's Board of Directors and the Scientific and Medical Advisory Board (SMAB), as well as four carefully selected outside experts, gathered in June 2019 to strategically and tactically evaluate potential therapies for Barth syndrome nearing clinical development.

The review meeting was intended to identify specific ways BSF can continue to advance the respective therapeutic ideas it has helped to fund over the years through its Research Grant program. This was the first of what will be a continuing series of research review meetings.

Three promising potential new therapies currently in pre-clinical development by three separate research groups were reviewed. Each of these theories has been supported by BSF's Research Grant program to get to this stage:

- Gene Therapy (Drs. Barry Byrne, Christina Pacak, and Todd Cade);
- Enzyme Replacement Therapy (Dr. Michael Chin); and
- Modifier Gene ALCAT1 (Dr. Roger Shi).

Of the three therapeutic ideas presented, gene therapy has been advanced the furthest. The consensus on this potential therapy was quite favorable, and the external reviewers encouraged BSF to continue to move this idea toward a clinical trial. BSF started funding gene therapy projects nearly a decade ago when Dr. Byrne received a BSF research grant to investigate "Gene therapy in mouse model in Barth Syndrome."

*"After nearly 20 years of directed strategic effort, BSF and the Barth syndrome community have finally reached the threshold of new therapies,"* said Kate McCurdy, BSF Scientific and Medical Advisory Board member, Emerita. *"Not only are the first two clinical trials for potential treatments underway, but the development of a possible cure in the form of gene therapy is in active planning. There is still much to do but, together, we are making exciting and distinct progress!"*



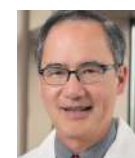
Dr. Byrne



Dr. Pacak



Dr. Cade



Dr. Chin



Dr. Shi

## 2020 BSF Research Grant Cycle Begins with 15 Novel Applications

Submissions for BSF's 2019/2020 grant cycle ended on October 31, with research applications spanning cellular and biochemical biology, systems biology, as well as continued exploration of Barth syndrome animal models and potential therapeutic avenues.

Funded projects will be announced in Spring 2020.

Since 2002, BSF's research grant program has been the only mechanism that uniquely provides seed grants to advance basic and pre-clinical research in Barth syndrome around the world. BSF encourages applications from young and established investigators, with the goal of attracting the best and the brightest to apply their research interests and talents to Barth syndrome.

The goal of the program is to fund the testing of initial hypotheses and collection of the preliminary data required for successful long-term funding by the National Institutes of Health (NIH) and other major granting institutions around the world. As a result of this program, other organizations have contributed more than \$21 million for follow-on funding.

Two funding categories are available: IDEA grants with a maximum budget of \$50,000 (over 1-2 years) and DEVELOPMENT grants for up to \$100,000 (over 2-3 years). Submitted applications are assessed by expert technical reviewers based on their relevance to Barth syndrome, feasibility of research plan, and the prospects of long-term funding.

# Piecing Together the Puzzle: Planning Gene Therapy Program in Barth

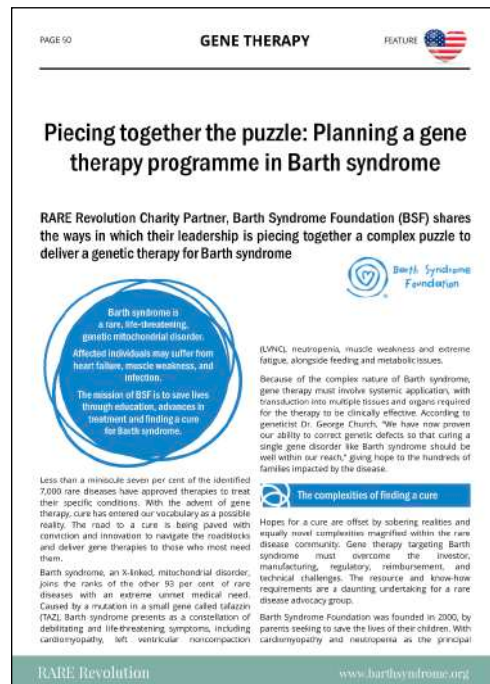
**“We are where we are today because we planned to be here 20 years ago. We have been plotting our course ever since. Our community is on board because we have shown the roadmap. They know where we are going and have been engaged throughout the process.”**

**- Shelley Bowen**  
*Director of Family Services, BSF*

BSF’s article was featured in the autumn 2019 Gene Therapy edition of Rare Revolution Magazine, describing our navigation of regulatory, financial, manufacturing challenges and steadfast efforts building a clinical trial-ready population. Driven by a community committed to finding a cure for Barth syndrome and two decades of research, the potential for realization of gene therapy for Barth syndrome -- and the complexities of making it work -- are at the forefront of the organization's mission today.

Because of the complex nature of Barth syndrome, gene therapy must involve systemic application, with transduction into multiple tissues and organs required for the therapy to be clinically effective. *“We have now proven our ability to correct genetic defects, so that curing a single gene disorder like Barth syndrome should be well within our reach,”* according to geneticist Dr. George Church.

To read the full article, visit the November BSF Research Brief: [bit.ly/BSFNov19](http://bit.ly/BSFNov19).



## CLINICAL TRIALS AND PUBLICATIONS

### CARDIOMAN

BSF and Barth Syndrome UK announced the CARDIOMAN clinical trial, the second trial in Barth syndrome and first in Europe. The Phase 2 trial aims to investigate the efficacy of bezafibrate on lipid metabolism and subsequent heart function in boys and young men with Barth syndrome.



Bezafibrate is a lipid-lowering drug that has been safely and broadly used to treat hypercholesterolemia in Europe and Canada since approval in

1978. Many patients with Barth syndrome experience metabolic consequences related to impaired fat metabolism at the cellular level.



**“It’s an equation of collaboration. UHB and UoB brought the academic experts; we found the families. We believe that clinical trials in rare diseases such as Barth syndrome require cross-sector partnership.”**

**- Michaela Damin, Barth Syndrome UK founder**

## TAZPOWER

Stealth BioTherapeutics' Phase 2/3 TAZPOWER study shows that elamipretide may be associated with improvements in cardiac function in Barth syndrome patients, in addition to possible improvements in muscle function.

*"Based on the data presented showing an increase in stroke volume (the amount of blood that the heart's left ventricle pumps with each beat), treatment with elamipretide appears to have improved heart function, which might indicate cardiac remodeling,"* said Dr. W. Reid Thompson, Associate Professor of Pediatrics at the Johns Hopkins University School of Medicine.

Stealth announced the findings from the open-label extension portion of the study at the American

Society of Human Genetics (ASHG) 2019 Annual Meeting in Houston, Texas. The study showed that treatment with elamipretide resulted in a 27% increase in average cardiac stroke volume.

*"Most patients with Barth syndrome have underlying heart disease, so this cardiac effect would be an important outcome in this setting that warrants further investigation,"* Thompson said.

BSF co-hosted an informational webinar with Stealth BioTherapeutics and Dr. Hilary Vernon, MD PhD in August to provide the community an update on the TAZPOWER clinical trial results and ongoing data from the open-label extension arm of the study.

**Keep up to date on the latest clinical trials: [barthsyndrome.org/clinicaltrials](http://barthsyndrome.org/clinicaltrials)**

## The Glucose/Exercise Connection in People with Barth Syndrome

During exercise, individuals with Barth syndrome are limited in their ability to generate energy from fat and instead rely on glucose metabolism to meet their energy needs, according to recent study results.

With the participation of 29 Barth patients, Dr. Todd Cade and colleagues used tools that included the bod pod (right), heart function monitoring, blood draws, and graded exercise tests to better understand the rate of glucose and fat consumption and conversion into energy.

The limited ability to generate energy from fat and instead rely on glucose not only associates with impaired muscle and heart function, but also aligns with anecdotal reports of younger patients consuming cornstarch (a complex carbohydrate source for glucose) to prevent nocturnal hypoglycemia. These results further expand our physiological understanding of Barth syndrome and provide potential outcome measures for future therapeutic opportunities for people with Barth syndrome. Read more in the October BSF Research Brief: [bit.ly/BSFOct19](http://bit.ly/BSFOct19).



## COLLABORATIONS

Collaboration is a core value for BSF. We rely on meaningful ongoing collaborations with many cross-sector partners:



**NORD**<sup>®</sup>  
National Organization  
for Rare Disorders



Stealth  
BIOTHERAPEUTICS



UNITED  
MITOCHONDRIAL  
DISEASE  
FOUNDATION.



Global Genes<sup>™</sup>  
Allies in Rare Disease



American  
Heart  
Association.



SOCIETY FOR  
LEUKOCYTE  
BIOLOGY

## Sponsor BSF's Mission to Save Lives Through ...

### EDUCATION

Please make a donation to assist with family scholarships, free attendance for our community, family and youth programs, and more.

### ADVANCING TREATMENTS

Support BSF bringing together more than 75 researchers from around the world to disseminate findings and form new collaborations.

### FINDING A CURE

Give the hope of therapies and treatments to the hundreds of families searching for a cure to Barth syndrome.

Show your support: [barthsyndrome.org/donate](http://barthsyndrome.org/donate)

## FAMILY SERVICES AND ADVOCACY

### Inspiring Young Men Living with Barth Syndrome



#### Cameron

"I am studying as a secondary education teacher in food and textiles technologies, which is a fancy name for home economics teacher. I believe the things I have learned through my own life experiences will help me to be a better teacher. As long as you are happy, who cares where happiness comes from."



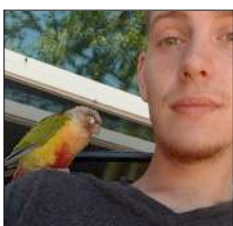
#### Aldo

(On having energy after transplant): "The best metaphor is like when you fill your gas tank up. There's a little buffer above full. That's what it feels like. You have all your energy back plus that little extra but eventually the gauge begins to go down."



#### Darryl

"I'm not taking anything for pain. It's still there. It's me. I don't know how to explain it. It's a part of life. It's just there. It's a part of living. If I'm hurting, I know I'm living. That's how I get through it. I honestly think I have become immune to it."



#### Travis

"My advice now to my younger self would be this: Stay active. Move, just move; even if it's just to walk for two minutes or lift 2 pounds, keep moving. Barth syndrome is like that sneaky little devil on your shoulder that taps to remind you it's still there. It's not just one thing; it's the constant reminders that you need to learn how to handle."

Read their full stories: [bit.ly/BSFcommunity](http://bit.ly/BSFcommunity)

## Care Management Toolbox

Published after years of cultivation, BSF's new Care Management Toolbox is a comprehensive family resource designed to help compile, organize, and use important medical information. The toolbox includes materials and tips to help our Barth syndrome families consolidate and organize complex medical records, such as:

- Suggestions for Organizing the Medical Binder
- Emergency Department Guidance
- Medical Forms
- Lab Results

Find the toolbox: [barthsyndrome.org/toolbox](http://barthsyndrome.org/toolbox)

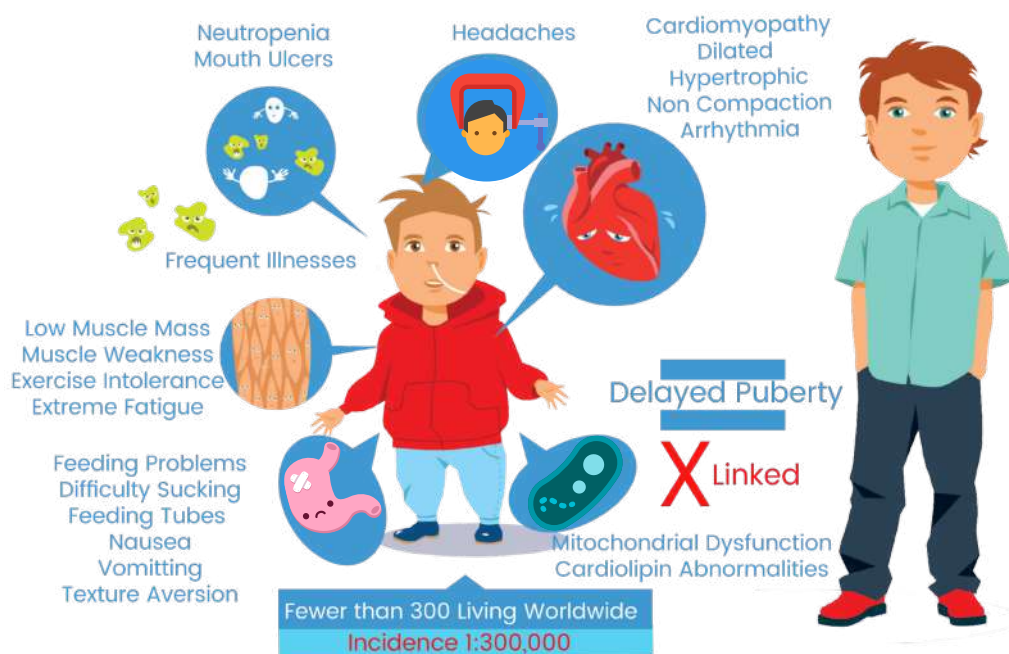
## School Support

"A Parent's Guide to Strategies for Educational Advocacy" and the corresponding resource for teachers are living documents that have been updated and developed over the last 15 years and are offered now as a robust educational resource to help parents, students, and teachers make school a success for those who have Barth syndrome. The guide contains multiple sections, including:

- Developmental Issues
- Protections of Students with Disabilities
- Choices of Educational Environments
- Preparing the Master File and Communicating with Schools
- Additional Resources

Visit [barthsyndrome.org/toolbox](http://barthsyndrome.org/toolbox) → Tools for School.

## What is Barth Syndrome



#Breakthroughs4Barth  
 YOU DID IT!  
 \$30,450 Raised to Power More  
 BREAKTHROUGHS for  
 Barth syndrome  
 THANK YOU #TeamBarth!!  
[barthsyndrome.org](http://barthsyndrome.org)

## Breakthroughs for Barth

Your generosity during our Breakthroughs for Barth campaign in August raised \$30,450 to help improve lives through family support programs and propel potential therapies. Because of your support, BSF continues to bring breakthroughs to our community by:

- Expanding support programs and resources for families.
- Attracting researchers and clinicians to advance science around gene therapy, enzyme replacement, and the use of existing drugs to treat Barth syndrome.
- Collaborating with FDA and industry to bring about more clinical trials in Barth syndrome.
- Improving tools to further a positive experience for families, researchers, and healthcare providers at the 2020 BSF International Conference.



**Barth Syndrome Foundation**  
**2005 Palmer Ave., #1033**  
**Larchmont, NY 10538**  
[www.barthsyndrome.org](http://www.barthsyndrome.org)

A promotional banner for the Barth Syndrome Foundation's 2020 campaign. On the left, a photograph shows Santa Claus sitting on a chair with a young child on his lap. The child is wearing green pajamas with a Santa Claus pattern. The background is a brick wall with a Christmas wreath. On the right, a blue background with white polka dots contains the text "Help BSF achieve #MoreIn2020" in blue and red. Below the text are four icons representing the foundation's focus areas: a microscope for "RESEARCH", a family of three for "FAMILY SERVICES", a megaphone for "ADVOCACY", and a syringe for "TREATMENTS".

Your gift makes a difference: [barthsyndrome.org/donate](http://barthsyndrome.org/donate)

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