



Barth Syndrome
Foundation

www.barthsyndrome.org

Barth Syndrome Journal

Volume 18, Issue 1 ~ Spring 2018

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

FDA Gives BSF Green Light for Patient Focused Drug Development Meeting

By Susan McCormack, Chair, Barth Syndrome Foundation

The Barth Syndrome Foundation (BSF) is excited to announce that our organization was approved by the United States Food & Drug Administration (FDA) to hold an Externally-led Patient-Focused Drug Development (ELPFDD) meeting during our conference in July. This is a unique and critically important opportunity for our affected individuals and their families to create the Patient Voice for Barth Syndrome for regulators and drug developers. They want to know what it is really like to have Barth syndrome or to care for someone who does, as well as what we each most dream of in terms of specific symptoms that we hope might be alleviated by future treatments. The meeting will take place on Wednesday, July 18 beginning with a luncheon and ending at approximately 5pm. All in attendance at the conference are strongly encouraged to join in this crucial meeting. Broad participation by as many families as possible is so vital, that we have made it possible for those who cannot actually be in Florida to instead "attend virtually" via live streaming and on-line polling to register answers to questions.

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About PFDD Meetings

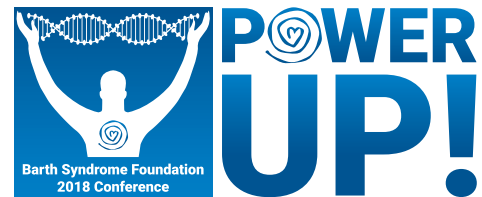
Patient-Focused Drug Development (PFDD) meetings bring together patients and their caregivers, doctors who are experts in the particular disease, representatives from the Food and Drug Administration, and pharmaceutical companies interested in developing medicines

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9th International Scientific, Medical & Family Conference

By Matthew J. Toth, PhD, Science Director, Barth Syndrome Foundation

The **Science and Medicine** portion of the 9th International Scientific, Medical & Family Conference on Barth syndrome, scheduled for July 16—21, 2018 in Clearwater Beach, Florida, will showcase recent advances in understanding Barth syndrome and the latest results of clinical studies. Finding specific treatments for Barth syndrome has been the major focus for BSF, and the Science and Medicine sessions of the previous international conferences have provided a convenient way to chart the progress being made. In early 2018 a subcommittee of members of the Barth Syndrome Foundation's (BSF) Scientific and Medical Advisory Board invited researchers to submit abstracts describing their work on Barth syndrome. Because of the time limitations of any conference, we could not accommodate all the researchers who wished to speak. The preliminary agenda provided below shows the speakers and the titles of their presentations. The 2018 Science and Medicine sessions promise to be the most interesting ever.



Clearwater Beach, FL ~ July 2018

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Research, University of Manitoba,
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Michio Hirano, MD, Neurology &
Mitochondrial Disease Research, Columbia
University, New York, New York

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FAHA,** Pediatric Cardiology, The Heart
Institute; Genetics, Cincinnati Children's
Hospital, Cincinnati, Ohio

William T. Pu, MD, Pediatric Cardiology,
Boston Children's Hospital; Harvard Stem
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Mindong Ren, PhD, Anesthesiology & Cell
Biology, New York University School of
Medicine, New York, New York

Colin G. Steward, PhD, FRCP, FRCPC,
Pediatric Stem Cell Transplantation,
University of Bristol, Bristol, England;
Medical Advisor, Barth Syndrome Trust (UK)

Arnold W. Strauss, MD, Pediatric
Cardiology and Research, Cincinnati
Children's Research Foundation, Cincinnati,
Ohio

Hilary J. Vernon, MD, PhD, Pediatrics,
Johns Hopkins University; Director, Barth
Syndrome Clinic at Kennedy Krieger
Institute, Baltimore, Maryland

Ronald J. A. Wanders, PhD, Genetic
Metabolic Diseases, Academic Medical
Center, University of Amsterdam,
Amsterdam, The Netherlands

Katherine R. McCurdy, Emerita
Barth Syndrome Foundation

Catharine L. Ritter, RN - ex-officio
Board Member, Barth Syndrome
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Mission and Vision Guide BSF During Time of Change

By Susan McCormack, Chair, Barth Syndrome Foundation



Susan McCormack
(Photo courtesy of
Amanda Clark 2016)

As most of you know, the past six months has been a period of transformation for the leadership of Barth Syndrome Foundation (BSF). We've experienced changes at both the Board and staff levels. And yet, BSF has continued to thrive and progress toward its goals. You may be wondering how BSF is able to manage such disruption. The answer lies in the mission, goals and values that were laid out almost 20 years ago by our founders.

Two members of BSF's Board of Directors were required to step down after reaching term limits — our Chairperson, Marc Sernel, and the head of our Board Development Committee, Susan Osnos. Marc's time leading BSF was marked by significant achievements: agreements with two new affiliates (Association Syndrome de Barth France and Barth Italia Onlus), the launch of our carrier studies program and our first pharmaceutical clinical trial for a treatment for Barth syndrome. The Board valued his courage, keen insights, and passion for our mission. We continue to grieve with Marc and his family over the tragic loss of their son, Ryan in March. Marc eloquently describes Ryan's legacy on pages 20-22. Susan Osnos has been responsible for the continued strength and depth of BSF's Board for many years. Her quiet grace, substantial experience in nonprofits and communications, and people skills were much appreciated and will be missed. Happily, she will continue her service to the Barth community by joining our Publications Team. I have taken over as Chairperson of the Board following service as Secretary for several years. I aspire to lead BSF forward with strength, kindness and wisdom. John Wilkins has been elected as our new Secretary. As the first Barth individual on our Board, John has brought us a much-needed voice from our community and we are excited he has accepted this new role.

BSF has also seen a material change at its staff level. In early May, we announced the hiring of our new Executive Director, Emily Milligan, after an extensive search process. Emily has dedicated her career to the betterment of families and children. She has served many nonprofits, most recently working toward a cure for Type 1 Diabetes at JDRF. Emily joins our other loyal staff members — Shelley Bowen, Lynda Sedefian and Matt Toth — who performed yeoman's work during the time BSF was without an Executive Director. We are thrilled to welcome Emily to #TeamBarth and look forward to the skills, knowledge and dedication she will bring to BSF. Emily is introduced on the following page.

As I stated above, BSF has weathered these transformations skillfully. We are fortunate that our organization was founded by a group that grounded the Foundation while remaining forward-thinking. The mission, vision, goals and values they generated 18 years ago continue to guide us today. We have a Board, staff and volunteers who are committed to these principles and devoted to the success of BSF and our community. In the future, we can continue to rely on these guidelines and resources no matter what may come our way.

As I look forward, I see numerous exciting opportunities:

- As mentioned above, we are in the midst of our first clinical drug trial in the USA, and we are poised to embark on another later this year in the UK. Barth researchers are actively working on possible gene therapy treatments for Barth syndrome, and we are hopeful for a therapy trial within the coming few years.
- Our biennial international conference is upon us, and it will be our largest conference to date. We expect over 50 affected individuals to attend; that's almost 25% of the population known to BSF! As always, the conference will be packed with research presentations, family information sessions, educational workshops and, of course, socializing (see pages 1/5-10).

Mission and Vision Guide BSF During Time of Change

(Cont'd from page 2)

- This year's conference will present our community with an exciting, once-in-a-lifetime opportunity to inform the U.S. Food & Drug Administration during a Patient-Focused Drug Development meeting (see pages 1/4) This meeting, which will last five hours, will give Barth individuals and their caregivers the chance to tell the FDA directly what life is really like and what is most important to them regarding treatments for Barth syndrome. We are elated to provide our community with this crucial opportunity.

Barth Syndrome Foundation has once again not only weathered change but has grown from it. We can anticipate much progress as we move forward, with new Board and staff members energizing our organization, propelling our mission: Saving lives through education, advances in treatment, and finding a cure for Barth syndrome!

Letter from Executive Director

By Emily Milligan, Executive Director, Barth Syndrome Foundation



Emily Milligan
(Photo courtesy of
Emily Milligan 2018)

I am elated to have joined the Barth Syndrome Foundation (BSF) as the new Executive Director on May 21st, and I thank you, the community, for the warm welcome. As a brief introduction to my background, programs for children and underserved populations have always been front and center in my life. I earned a BA from the University of Florida in Gainesville, FL and then received an MPH (Masters of Public Health) from Columbia University. I began my career with grassroot efforts in marginalized communities in Brazil. I went on to consult for the United Nations on programs to avert maternal mortality in Nicaragua. Returning home to the U.S., I managed multiple research studies at Columbia University to understand the correlation between social inequalities and health outcomes. I was later recruited to JDRF (formerly known as the Juvenile Diabetes Research Foundation) to manage a multi-million-dollar

research portfolio aimed at finding a cure, prevention, and treatments for type 1 diabetes. Most recently, I launched an \$80M philanthropic venture fund in Boston to finance companies developing innovative biomedical technologies and therapies for type 1 diabetes.

"I" is actually a story of "we." None of the accomplishments cited above would have been possible without the friends I have made and the "family" I have gained through my professional journeys. Indeed, community singularly united in one mission across geographies, languages, and demographics is a prominent attribute in the best-of-the-best mission-driven organizations. I found these qualities in BSF, and your "family" overwhelmingly influenced my decision to call BSF my new home.

In my first 120 days as BSF's Executive Director, it is my goal to meet each of you and become acquainted with your loved ones and their stories. It is my hope that you will be attending the conference in July, in which case I would like to meet you and to introduce you to my family as well – my dear husband, Daniel, and at least one of the four children we are blessed to have in our lives (Camilo, Laura, Leonardo, and Anaclara). If by chance you are not able to join us in Florida, I welcome the opportunity to meet by phone and look forward to our correspondence.

Transitions into a new organization always come with a learning curve. I extend my gratitude to my colleagues on the staff and the community at large for support as I begin my tenure. I am humble in that I have much to learn and welcome your input so we are jointly poised for success to achieve the BSF mission. Together we will do great things for those affected by Barth syndrome.

Barth Syndrome Foundation (BSF)

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Emily Milligan, Executive Director, *ex-officio*
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Catharine L. Ritter, RN, Board Member
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Emily Milligan, Executive Director
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Lynda M. Sedefian, Executive Assistant
Matthew J. Toth, PhD, Science Director

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Kate McCurdy, BSF
Lorna Moore, Barth Syndrome Trust
Nigel Moore, Barth Syndrome Trust
Susan Osnos, BSF



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meeting all accreditation standards



Member of the Genetic Alliance



Member of the GuideStar Exchange
Program



Member of the National Health Council



Member of the Rare Foundation Alliance

FDA Gives BSF Green Light for Patient Focused Drug Development Meeting

(Cont'd from page 1)

for the disease — all to hear from patients about the disease in question. In these meetings, the patient's experience is brought to the forefront for the FDA and pharmaceutical companies to understand.

There are currently two types of PFDD meetings: (1) FDA-hosted and (2) externally-led. FDA-hosted meetings were the first type of PFDD meeting established by the FDA in 2012 to learn about twenty specific disorders (Barth syndrome was not included). In December 2015, the FDA indicated that a second type of PFDD meeting would be available where stakeholders could hold their own PFDD meetings and invite the FDA to engage in the meeting through a formal process. This second type of PFDD meeting is referred to as an externally-led PFDD meeting — meaning that stakeholders outside of the FDA are leading the meeting. This is the type of meeting that BSF will be holding at our conference.

Benefits of an ELPFDD Meeting

ELPFDD meetings are an important mechanism for the FDA to understand the disease state and treatment needs from the patient perspective. At the ELPFDD meeting, individuals impacted by a specific disease convene in an organized fashion to share their personal experiences of living with the disease, and to share insights on treatment impacts that would be most important or beneficial. Importantly, the meetings provide an opportunity for the patient's voice to be heard and incorporated into the medical product development and evaluation process.

Holding an ELPFDD meeting focused on Barth syndrome will:

- Benefit patients and the FDA, and help pharmaceutical companies develop potential new medicines
- Aid the FDA in advising a company while it is developing a potential medicine
- Aid the FDA in its role to review and approve new medicines
- Focus the patient community on more fully describing Barth syndrome in a way that is useful in drug trials
- Create a document from the patients' perspective about Barth syndrome that can be used by anyone developing treatments for the condition or regulating this process around the world

By doing all of the above, the ELPFDD meeting will help the FDA to bring new medicines to Barth syndrome patients.

Our ELPFDD meeting can support making medicines available to patients by helping to identify:

- What symptoms, if addressed, would alleviate the greatest burdens on patients
- Certain disease symptoms or side effects that are/not tolerable by the patient
- Areas in which current approaches to treatment aren't satisfactory
- Criteria for drug-discovery scientists to choose chemicals with the greatest potential to become medicines
- Criteria that can be used to measure the benefit of potential medicines

Timing of BSF's ELPFDD Meeting

BSF's Board of Directors and staff determined that the optimal time to hold an ELPFDD meeting was at the 2018 International Barth Syndrome Conference. Reasons for this decision included the following:

- Last year, BSF's patient community entered the realm of pharmaceutical clinical trials. In the United States, the FDA must approve all new drugs prior to patient use. As such, it has become crucial for the FDA to understand the treatments most desired by Barth syndrome patients and their caregivers.
- We expect the largest gathering of affected Barth individuals to date at this year's conference. These individuals represent approximately 25% of the population living with Barth syndrome known to BSF. This provides a rare opportunity for the FDA, scientists and pharmaceutical companies to hear from our community.

Format of the Meeting

We expect the meeting will entail a combination of means by which patient feedback can be provided to the FDA. These will most likely include: videotaped interviews, testimony from panels of those in attendance at the conference, facilitated/interactive audience discussion, and live polling of audience members as well as anyone who participates in the meeting via live streaming video. BSF has invited FDA representatives to provide opening and/or closing remarks as well.

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FDA Gives BSF Green Light for Patient Focused Drug Development Meeting

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Meeting Work Product

After the meeting, BSF will produce a video recording of the meeting including slides and visual aids shared during the meeting, and a Voice of the Patient report. These resources will be posted on the BSF website. The Voice of the Patient report will provide a summary of perspectives of disease impact and preferences for future treatment shared during the meeting. The Voice of the Patient document will help to guide the FDA in their future decisions regarding potential approvals for drugs to treat Barth syndrome. In addition, it will be made available to anyone interested in learning more about what is most important to those who are living with Barth syndrome, such as doctors, researchers, and other drug approval agencies worldwide.

We Need YOU!

In order to make this ELPFDD meeting a success, we need YOUR input! The best way to help in this effort to further facilitate the development of treatments is to participate in the meeting itself either by attending in person or by “attending” via the live stream. Although we haven’t finalized meeting preparations with the FDA yet, we anticipate that we will ask for various inputs from our community. We expect to conduct live polling whereby questions can be answered by the larger group (both in the room and on-line) via a phone app. We also anticipate pulling together one or two panels of Barth individuals and/or caregivers, as well as siblings, other family members and/or carriers as well. If we call on you in the coming weeks to help with this meeting, please seriously consider contributing. And, in any case, PLEASE consider participating on July 18. Your involvement is critical; we simply can’t do this without you, our #TeamBarth!

9th International Scientific, Medical & Family Conference

(Cont'd from page 1)

SESSION 1—PATHOMECHANISMS OF BARTH SYNDROME *(Thursday, July 19, 2018)*

Chair—Arnold W. Strauss, MD, Cincinnati Children's Research Foundation, Cincinnati, OH

Knockin and Knockout Mouse Models of Barth Syndrome

William T. Pu, MD, Boston Children's Hospital, Boston, MA

A Novel Mechanism to Explain Cardiac Oxidative Stress and Energy Depletion in Barth Syndrome

Christoph Maack, MD, University Clinic Würzburg, Würzburg, Germany

Activation of the Mitochondrial Stress Response Underlies a Specific Heart Phenotype in Barth Syndrome

Douglas Strathdee, PhD, Cancer Research UK Beatson Institute, Glasgow, United Kingdom

Nicotinamide Replacement Improves Mitochondrial Function in Preclinical Models of Barth Syndrome

Christian Reynolds, PhD, Wayne State University, Detroit, MI

Increased ROS-mediated CaMKII Activation Contributes to BTHS iPS-CMs Pathogenesis

Xujie Liu, PhD, Boston Children's Hospital, Boston, MA

Altered Islet Function May Promote a Lean Phenotype in *Tafazzin* Deficient Mice

Laura Cole, PhD, University of Manitoba, Winnipeg, Canada

Defective Mitochondrial Cardiolipin Remodeling Causes Alterations in Cellular Signaling Pathways

Jan Dudek, PhD, University Medical Clinic Würzburg, Würzburg, Germany

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9th International Scientific, Medical & Family Conference

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SESSION 2—POTENTIAL THERAPIES FOR BARTH SYNDROME (Thursday, July 19, 2018)

Chair—W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO

Gene Therapy Vector Optimization and Testing for Barth Syndrome

Christina Pacak, PhD, University of Florida, Gainesville, FL

Elamipretide (MTP-131) in Subjects with Genetically Confirmed Barth Syndrome (TAZPOWER): A Phase 2 Randomized, Double-blind, Placebo-controlled Crossover Trial Followed by an Open-label Treatment Extension

Hilary J. Vernon, MD, PhD, Johns Hopkins University, Baltimore, MD

Evaluating Antioxidant Therapies in a *Tafazzin*-knockdown Mouse Model of Barth Syndrome

Colin Phoon, MPhil, MD, New York University School of Medicine, New York, NY

Identification of Novel Mitochondrial Targeting Peptides in *Tafazzin* and Long-term Efficacy of Enzyme Replacement Therapy in a Mouse Model of Barth Syndrome

Michael Chin, MD, PhD, Tufts Medical Center, Boston, MA

Cross-species Omics Integration Identifies New Potential Treatment Targets for Barth Syndrome

Riekelt Houtkooper, PhD, Academic Medical Center, Amsterdam, The Netherlands

Can Elamipretide, the First Cardiolipin-protective Compound, Benefit Barth Patients?

Hazel Szeto, MD, PhD, Weill Cornell Medical College, New York, NY

POSTER SESSION

Chair—Hilary J. Vernon, MD, PhD, Johns Hopkins University, Baltimore, MD

SESSION 3—CLINICAL CHARACTERISTICS OF BARTH SYNDROME (Friday, July 20, 2018)

Chair—Grant M. Hatch, PhD, University of Manitoba, Winnipeg, Canada

Barth Syndrome: Natural History of Cardiomyopathy and Cardiac Conduction

Carolyn Taylor, MD, Medical University of South Carolina, Charleston, SC

Functional Exercise Capacity, Strength, Balance and Motion Reaction Time in Barth Syndrome: Outcomes from the 2016 Barth Syndrome Foundation Scientific, Medical & Family Conference

Brittany DeCroes Hornby, PT, DPT, PCS, Kennedy Krieger Institute, Baltimore, MD

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Dr. Todd Cade presents at BSF 2016 conference



Dr. Yuguang (Roger) Shi presents at BSF 2016 conference

9th International Scientific, Medical & Family Conference

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Characterization of the Metabolic Phenotype in Individuals with Barth Syndrome With and Without Cardiac Transplantation

W. Todd Cade, PT, PhD, Washington University School of Medicine, St. Louis, MO

Summarized Findings from the Barth Registry Interviews: Key Points for Moving Forward

Anthony Aiudi, PharmD, Stealth Biotherapeutics, Newton, MA

2 Poster Presenters

Females with Barth Syndrome

Colin Steward, PhD, FRCP, FRCPCH, University of Bristol, United Kingdom

SESSION 4—CARDIOLIPIN AND BARTH SYNDROME (Friday, July 20, 2018)

Chair—Michael Schlame, MD, New York University School of Medicine, New York, NY

The Composition and Dynamics of the Mitochondrial Complexome

Ulrich Brandt, PhD, Radboud University Medical Center, Nijmegen, The Netherlands

Mechanisms Underlying TCA Cycle Defects in *Tafazzin* deficient Cells – Potential New Targets for Treatment of Barth Syndrome

Miriam L. Greenberg, PhD, Wayne State University, Detroit, MI

2 Poster Presenters

Targeting ALCAT1 for the Treatment of Barth Syndrome

Yuguang Shi, PhD, University of Texas Health Sciences Center, San Antonio, TX

Biophysical Approaches Toward Understanding the Molecular Mechanism of Action of SS-31 (Elamipretide)

Nathan Alder, PhD, University of Connecticut, Storrs, CT

The Structural Molecular Diversity of Cardiolipins

Markus Keller, PhD, Medical University Innsbruck, Innsbruck, Austria

FAMILY SESSIONS

The Barth Syndrome Foundation International Scientific, Medical and Family conferences are truly one of the greatest tools to deliver a better understanding of Barth syndrome to families, physicians, scientists and researchers. Each topic provides affected individuals and their families with crucial information about this complex disorder.

In an effort to give our affected families the tools to become the most effective advocates for their Barth individual, the Family Sessions will include expert presentations and panel discussions which focus on providing the most up-to-date information on current research advances, the clinical aspects of Barth syndrome (BTHS), day-to-day aspects of BTHS, and current resources and practical information about BTHS.

INDIVIDUAL CONSULTATIONS AND CLINICS (Monday, July 16, 2018)

Barth Syndrome Registry and Repository

Full details about the registry and assistance with enrollment or updating of entries.

General Barth Syndrome Questions

Information regarding biochemical, diagnostic, natural history, general medical or treatment strategy questions.

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9th International Scientific, Medical & Family Conference

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Genetics

Information about all aspects of genetics and how family members may be affected; collection of family pedigrees.

Investigation into Clinical, Metabolic and Molecular Factors in Barth Syndrome

Six-minute walk test and grip strength

Longitudinal Evaluation of Cardiomyopathy and Outcome in Barth Syndrome

Echocardiogram, EKG and questionnaire — for affected individuals who have participated in previous cardiology study led by Dr. Carolyn Taylor.

Pill Swallowing Workshop

Techniques and equipment for pill swallowing — creating a personalized program in a stress-free environment.

Vitals

Height, weight and blood pressure

Portraits

Family and individual portraits by Amanda Clark

Registration

Pick up badges, t-shirts/drop items for goody bags

SMALL GROUP MEETINGS *(Monday, July 16, 2018)*

First Time Family Session

General orientation and walkthrough meeting for new families and mentors

Barth 101

Small group meeting for parents, caregivers and adult individuals with Barth syndrome. Informal session to help prepare attendees for more complex information presented later in the week. Topics: What causes Barth syndrome; how to understand clinic letters, echo and lab results; a practical guide to symptoms and how to manage them; basic cardiology, hematology, and genetic information; muscle weakness; feeding issues; infant milestones.

Welcome Event

Overview of conference; family introductions

Heart Transplant Meeting

Informal get-together of families with heart transplanted individuals.

SMALL GROUP MEETING, CLINICS & CONSULTATIONS *(Tuesday, July 17, 2018)*

Life as a Carrier or Potential carrier

This small group is open to all mothers, daughters, grandmothers, and sisters 15 years old and over who are invested in the life and care of an individual with Barth syndrome.

Pre-test Carrier Discussion

This is for potential carriers 15 years old and over. This small group will focus on how to make the decision about genetic carrier testing, what is involved with testing and what to think about before testing.

Post-test Carrier Session

This small group will focus on exploring the impact of learning that you are a carrier for Barth syndrome. How this might impact relationships, future plans, reproductive options, etc.

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9th International Scientific, Medical & Family Conference

(Cont'd from page 8)

Carrier Session 4

Feedback from carrier study; creation of resource network; where do we go from here; what can we do to support each other.

Transplants

How to prepare for the possibility of needing a heart transplant; transplanted patient perspectives; questions and answers.

SMALL GROUP MEETING, CLINICS & CONSULTATIONS *(Wednesday, July 18, 2018)*

Where is My Cure/New Therapies

A simplified summary of potential therapies for Barth syndrome.

Practical Workshops and Question and Answer Sessions to Promote Independence, Coping with Daily Life

Separate sessions for parents of affected individuals (ages 0-4; 4-8; 8-12; 18+).

Externally-led Patient Focused Drug Development Meeting on Barth Syndrome

FORMAL LECTURES *(Thursday, July 19, 2018)*

Cardiology

Topics: PumpKIN (LVAD) for babies; transplants; longitudinal data; arrhythmia risks; electrophysiology, data on Barth individuals with ICDs; new drugs and their potential impact on taking part in clinical trials.

Barth Syndrome on a Daily Basis

Topics: Importance of exercise, OT and PT; pill swallowing; depression; anxiety; results of previous surveys; on-going quality of life research; energy conservation; pain management; peer-to-peer strategies.

Potential Therapies for Barth Syndrome

Merged with Science and Medicine Sessions (see page 6)

POSTER SESSION

Luminaries on the Beach

(Cont'd on page 10)



Dr. Arnold Strauss presents at BSF 2016 conference



Prof. Colin Steward presents at BSF 2016 conference

9th International Scientific, Medical & Family Conference

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FORMAL LECTURES (Friday, July 20, 2018)

Neutropenia

Clinicians' experience — Feedback from USA, France and the UK.

Genetics and Carriers

Topics: Genetic testing kits; strategies on talking to your children; what came out of the small group discussions.

Research and Survey Updates and Clinical Trial Explanation

Topics: Older affected individual survey results; upcoming trials; why and how to participate.

Small Group Sessions

Friday Night Social

FORMAL LECTURES (Saturday, July 21, 2018)

Topics: Wrap up — this is what we learnt this week; next steps; practical resources



Dr. Carolyn Taylor, RJ (age 20), Dr. Barry Byrne, and Dr. Todd Cade



Andrew (age 29) and Jack (age 25) speak at BSF 2016 conference



#TeamBarth entering Friday Night Social at BSF 2016 conference



Doctor dance off at Friday Night Social at BSF 2016 conference

Barth Syndrome Foundation 2017 Research Grant Awardees

By Matthew J. Toth, PhD, Science Director, Barth Syndrome Foundation

The 2017 cycle of the Barth Syndrome Foundation (BSF) Research Grant Program marks the 16th year of this highly successful plan to increase our understanding of Barth syndrome (BTHS) so that specific therapies may be developed. Many of the therapies discussed at the BSF International Scientific, Medical & Family conferences have their origins or have been supported by this BSF program. Bezafibrate therapy, gene therapy, enzyme replacement therapy, exercise therapy, and lipid replacement therapy can all be tracked to the early support that BSF research grants have provided over the years.

The Barth Syndrome Foundation (BSF) now has awarded 104 grants to 60 different investigators for a total of over US \$4.5 million in funding. Importantly, these investigators and others have garnered over US \$19 million in additional funding from the National Institutes of Health (NIH) and other major funding agencies over the years (see graph). However, it takes more than funding to make scientific and medical advances — it takes dedicated and hard-working researchers, along with Barth syndrome (BTHS) individuals who have partnered with the researchers over the years. Below is my brief summary of the 2017 awardees.

Abbreviations, acronyms, and definitions:

- apoptosis—programmed cell death that is normal for the body
- BTHS—Barth syndrome
- CRISPR—a useful technique to easily alter the gene of a cell
- CL—cardiolipin; the lipid known to be reduced/alterd in BTHS
- MLCL—monolysocardiolipin (cardiolipin missing one fatty acid chain); elevated in BTHS
- KD—knockdown mouse model of Barth syndrome
- KO—knockout mouse model of Barth syndrome developed by Dr. Strathdee
- MICOS—subcellular organelle important for proper mitochondrial function (mitochondrial contact site and cristae organizing system)
- NETosis—a form of cell death that is non-apoptotic (neutrophil extracellular trap)
- OXPHOS—the major energy producing activity of the mitochondria (oxidative phosphorylation reactions)
- PDH—the enzyme pyruvate dehydrogenase which is the metabolic branch point between glycolysis (outside the mitochondria) and OXPHOS (inside the mitochondria)
- *tafazzin*—the gene that codes for TAZ which is defective in BTHS
- TAZ—the enzyme that when defective causes BTHS



Borko Amulic, PhD, Lecturer (Assistant Professor), University of Bristol, Bristol, UK

“Neutrophil dysfunction in Barth syndrome”

Award: US \$49,967 over 2-year period

**Partial funding for this award was provided by Barth Syndrome Trust*

Using blood samples from BTHS individuals to uncover the reasons for neutropenia. Neutropenia is one of the most misunderstood symptoms of BTHS, and it is just as much a threat to life as heart problems. The reasons that this major class of white blood cells is reduced in BTHS is unknown even given all that we now know about the *tafazzin* gene. Dr. Amulic along with Dr. Colin Steward, will use blood samples from BTHS individuals attending the UK clinic to investigate: 1) the metabolism of neutrophils; 2) whether non-apoptotic cell death (NETosis) is involved with neutropenia; and 3) whether neutrophils from BTHS individuals lack the ability to destroy bacteria as they ought. Neutrophils will be isolated from individuals and then examined for subcellular structural integrity/functionality by confocal microscopy and for biochemical reactions with the Seahorse™ metabolism detection system. In addition, the researchers will determine whether NETosis, a non-apoptotic form of cell death on which Dr. Amulic is a leading authority, plays any role in BTHS. Finally, Dr. Amulic will examine the bacteria-killing abilities of these isolated neutrophils by measuring bacterial destruction, cytokine release, and the granule release that normally accompanies neutrophil bactericidal or bacteria-killing function. Understanding the defects in neutrophils and the dysfunction of neutropenia will provide therapeutic insights for BTHS and other neutropenia-associated diseases.

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Barth Syndrome Foundation 2017 Research Grant Awardees

(Cont'd from page 11)



Martin van der Laan, PhD, Professor, Saarland University, Homburg, Germany

“Mutual connections of mitochondrial membrane architecture and cardiolipin homeostasis in Barth syndrome”

Award: US \$48,906 over 1.5 year period

**Funding for this award was provided by Association Syndrome de Barth France*

What is the impact of *tafazzin* dysfunction on the sub-organelle structure of the mitochondria called MICOS? MICOS is a structure inside the mitochondria (subcellular and sub-organellar) where many of the biochemical functions of the mitochondria are associated, especially the OXPHOS functions. Dr. van der Laan performed the groundbreaking studies that contributed greatly to our understanding of MICOS. MICOS is involved with the complex folds of the inner mitochondrial membrane called cristae and can be visualized by electron microscopy. When the proteins that make up the MICOS structure are altered, the mitochondria resemble (using electron microscopy) what is observed in BTHS and other diseases (like 3-methylglutaconic aciduria). We know that cardiolipin or CL is a vital component of the mitochondria, and Dr. van der Laan will use genetic and biochemical studies to investigate how *tafazzin* gene dysfunction affects MICOS. By using genetic mutants of MICOS in combination *tafazzin* deletions (in the human HEK293 cell line using CRISPR), Dr. van der Laan will determine the effects on MICOS' assembly, stability, and functionality. He will use electrophoretic, immunological, ultrastructural, and genetic techniques to study MICOS' role in BTHS cell lines and determine if overexpression of certain proteins can ameliorate the BTHS cellular phenotype in a manner similar to what Dr. van der Laan has found in other systems. Altering the MICOS machinery either genetically or pharmacologically may be a novel way to treat BTHS by attacking the basic pathophysiology of this disease.



Miriam Greenberg, PhD, Professor, Wayne State University, Detroit, MI

“Cardiolipin activates pyruvate dehydrogenase (PDH) — a potential new target for treatment of Barth syndrome”

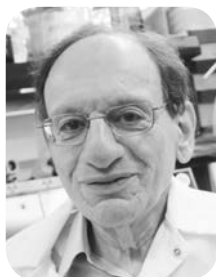
Award: US \$50,000 over 1-year period

Would increasing PDH activity be therapeutic for BTHS? Through radioactive labeling experiments, Dr. Greenberg has found that one crucial enzyme in cellular metabolism, PDH or pyruvate dehydrogenase, is lowered in a BTHS-like cell line that she has constructed (mouse myoblast C2C12 line that is deleted for the *tafazzin* gene). Interestingly, she also has observed that adding cardiolipin or CL increases this PDH enzyme activity back to normal levels. PDH controls metabolism, and the enzyme itself is controlled in numerous ways (transcriptional, phosphorylation, and dephosphorylation). The chemical compounds dichloroacetic acid and phenyl butyrate have been shown to alter the PDH activity of the cell and are being tested as therapeutics for other rare metabolic diseases. Could these compounds or similar PDH-affecting compounds be therapeutic for BTHS? Dr. Greenberg will examine which subunit(s) of PDH interact with CL (or MLCL or other phospholipids) and cause the enzyme activity decrease she observes, and whether the phosphorylation status of PDH is affected. As a second aim, she will identify whether augmenting PDH activity with chemicals will correct the deficiencies she observes. The hope is that this focus on a major metabolic pathway enzyme will be therapeutic for BTHS and other mitochondrial diseases.

(Cont'd on page 13)

Barth Syndrome Foundation 2017 Research Grant Awardees

(Cont'd from page 12)



Richard Eband, PhD, Professor, McMaster University, Hamilton, Ontario, Canada

“The cause and consequences of plasmalogen depletion in Barth syndrome”

Award: US \$50,000 over 1-year period

**Partial funding for this award was provided by Barth Syndrome Foundation of Canada*

Plasmalogens, which are lipids that are similar to cardiolipin, are greatly reduced in BTHS, and dietary supplements with plasmalogen precursors may be useful in treating BTHS. Dr. Eband has discovered that plasmalogens, which are lipids with a certain unique linkage (ether bond) in their chemical structure, are significantly reduced in BTHS cells and in the hearts of the KD mouse model of BTHS —reduced just like cardiolipin (CL) levels are reduced in BTHS but even more pronounced than CL reduction. The role of plasmalogens in the cell are unknown and this class of lipids has been termed the “forgotten lipids” which underscores our ignorance of them. Plasmalogens are synthesized in separate organelles or subcellular compartments called peroxisomes, and these plasmalogens make up more of the total phospholipids in the mouse heart than CL does (5x more). Dr. Eband will investigate the distribution of plasmalogens in the different subcellular organelles of BTHS lymphoblasts, inhibit a degrading enzyme of plasmalogens called iPLA2 with bromoenol lactone to see if this restores plasmalogen levels like it did with CL (note the earlier connection with Dr. Ren’s fruit-fly work with bromenol lactone), and determine if adding alkylglycerols improves plasmalogen deficiency in BTHS cells. Dr. Eband will use subcellular fractionation, immunoblotting techniques, and nuclear magnetic resonance to examine this cellular dysfunction. Adding alkylglycerols like O-hexadecylglycerol to the diet (in a manner similar to linoleic acid supplementation which has been tried for BTHS) was found to be useful in treating a mouse model of a peroxisomal disorder disease, and it may have a similar therapeutic value for BTHS treatment.



Colin Phoon, MPhil, MD, Associate Professor, New York University School of Medicine, New York, NY

“Prenatal cardiac phenotype as a platform for testing Barth syndrome therapies”

Award: US \$70,000 over 2-year period

**Partial funding for this award was provided by the Paula and Woody Varner Fund*

Using the KO mouse model to develop a platform for testing prenatal therapies. Previous work with the KD mouse model of BTHS has shown the value of prenatal chemical treatments of n-acetylcysteine and MitoQ10 in reducing cardiomyopathy and fetal deaths. With the new KO mouse model developed by Dr. Strathdee (and supported by BSF), a more consistent and streamlined platform for investigating prenatal demise due to *tafazzin* gene dysfunction is possible. Dr. Phoon will investigate the characteristics of the KO mouse model by focusing on prenatal markers and after treatment with n-acetylcysteine, MitoQ10, and melatonin, which are compounds of interest demonstrated with the KD mouse model. In addition, Dr. Phoon and colleagues will make a KO mouse model in which the *tafazzin* gene has been deleted in the heart and compare it to the mouse model where the gene is deleted in all tissues of the body. An additional focus of his work will be to study the gastrointestinal tract, which is not well understood and which has been implicated clinically in BTHS pathology. Dr. Phoon hypothesizes that there is a developmental cardiomyopathy of BTHS (exemplified by the high incidence of neo-natal demise in KD mice) and an adult-onset cardiomyopathy (mild in the case of the KD mice) which are distinguishable and perhaps are treatable in separate ways. Recognizing BTHS individuals very early in life or in utero and being able to treat them *in utero* may hold great advantages for BTHS individuals and their families, especially as genetic testing becomes more prevalent.

The Serendipitous Discovery of Elamipretide

By Hazel Szeto, MD, PhD, Director of Research, Social Profit Network, White Plains, NY

“The chance discovery of this tiny molecule has completely changed my life. It has taught me the importance of bioenergetics in healthy living and to develop a more holistic view of health and disease. I hope it will also impact on the lives of many people in the BTHS community.” ~ Hazel Szeto, MD, PhD



Dr. Hazel Szeto
(Photo courtesy of Dr. Szeto 2018)

Stealth Biotherapeutics just announced that they have completed enrollment for TAZPOWER, a clinical trial to evaluate elamipretide for Barth syndrome (BTHS). Lynda Sedefian and Matt Toth asked me to share the story behind the discovery of elamipretide and my hopes that it will help BTHS patients.

Let me first say that elamipretide (first called SS-31) was discovered entirely by serendipity. I had been on the faculty at Weill Cornell Medical College since 1979. Until 2004, I had never done research on mitochondria and was entirely unaware of rare mitochondrial diseases like BTHS. I had been working in a totally different field of research, with a goal to develop better opioid analgesic drugs. I had found some really excellent candidates. They were small peptides named “SS peptides” for “Szeto-Schiller,” with Peter Schiller being the peptide chemist behind the design and synthesis of these peptides. We thought these compounds would never reach the brain because they are highly water-soluble. But we were wrong. These peptides not only got into cells, they very selectively localized to mitochondria. That was a stunning surprise and the beginning of a new adventure for me.

At that time, I knew almost nothing about mitochondria other than they make ATP and were considered the cell’s powerhouses. I then read that mitochondria produce reactive oxygen species (ROS) and facilitate apoptotic cell death. It was also known that many drugs that target mitochondria are toxic. I was therefore worried that the SS peptides might be toxic to mitochondria. However, study after study showed that the SS peptides protect mitochondria from all kinds of stress and mitochondrial toxins. SS-31 was our first attempt at developing a peptide analog that has all the mitochondrial protective properties but lack opioid activity.

SS-31 has been studied extensively by academic researchers around the world. They all confirmed our initial observation that SS-31 targets mitochondria and helps protect their ability to make ATP and reduce ROS production. In animal studies, SS-31 minimized tissue damage caused by a lack of blood flow to the heart, kidney, and brain. Other studies showed that SS-31 can help in heart failure and neurodegeneration, including Parkinson’s Disease, Alzheimer’s Disease, and amyotrophic lateral sclerosis.

These results were compelling enough that I thought SS-31 should go into clinical development. Development of a new drug takes a long time and is very costly and highly risky. A company was formed in 2007 and initial funding was provided by Morningside Ventures Investment. Peptides are generally not considered to be good drug candidates because they do not cross membranes well. The SS peptides may be viewed as “cloaked” or “stealth” as they can evade cellular membranes. I therefore named the company Stealth Peptides (renamed Stealth BioTherapeutics in 2014).

Clinical trials were started with SS-31 in 2010, and it was found to have excellent safety profile in humans. There are now multiple clinical trials evaluating the efficacy of SS-31 for primary mitochondrial diseases caused by genetic mutations as well as age-related chronic diseases associated with mitochondrial dysfunction. Elamipretide is the official generic drug name for SS-31 granted by the World Health Organization in 2016.

In the meantime, I kept trying to understand how SS-31 works. Our early work showed that SS-31 penetrates cells and gets through the outer mitochondrial membrane (OMM) but is then stuck on the inner mitochondrial membrane (IMM). This meant that either SS-31 cannot penetrate the IMM or it binds tightly to something on the IMM. We speculated that SS-31 must interact avidly with cardiolipin,

(Cont’d on page 15)

The Serendipitous Discovery of Elamipretide

(Cont'd from page 14)

“Elamipretide represents a paradigm shift in targeting the fundamental cause of cellular energy failure rather than correcting specific genes or proteins for individual diseases.” ~ Hazel Szeto, MD, PhD

a phospholipid that is only found on the IMM. It took us another eight years to demonstrate this convincingly using a wide range of experimental approaches. This was an important finding as no other drug has been shown to elicit its action by targeting a membrane lipid.

Cardiolipin has a conical shape that favors membrane curvatures and the formation of cristae membranes. Cells that require a lot of ATP, such as the heart, have high cardiolipin content and tightly folded cristae in order to maximize surface area for the respiratory protein complexes. Loss of this tight cristae structure reduces ATP production. We found that the binding of SS-31 to cardiolipin helps bring cardiolipin molecules together to form curvatures. SS-31 helps tighten up cristae curvatures, speed up electron transfer, and promote ATP production.

ATP provides energy for all cellular functions. Without adequate ATP, the tissues with high energy demand will not function properly. This includes the heart, skeletal muscle, brain, retina and kidney. By promoting ATP production, SS-31 increases contractile function in the heart and skeletal muscle and improves vision and cognitive function in many animal disease models. In disease states, prolonged treatment with SS-31 can even repair damaged mitochondria and restore mitochondrial structure. Restoring cellular ATP then allows the cell to repair its structure and regain function.

Promising clinical results with elamipretide for muscle weakness and heart failure led Stealth Biotherapeutics to propose that elamipretide may help BTHS patients. The mutation in the *tafazzin* gene in BTHS mitochondria results in the failure to make adequate amounts of cardiolipin and results in the accumulation of monolysocardiolipin. Monolysocardiolipin does not have a conical shape and is therefore not capable of supporting cristae curvatures. Can elamipretide have any effect on BTHS mitochondria with limited cardiolipin content? Perhaps elamipretide can interact with MLCL and alter membrane structure?

I am grateful to Matt Toth who invited me to the 2016 Barth Syndrome Foundation international conference. At that meeting, I met Nathan Alder, and I realized that he has the expertise and tools to answer these questions. With generous funding provided by the Social Profit Network, Nathan has determined that SS-31 binds to monolysocardiolipin with the same affinity as cardiolipin, and that binding to monolysocardiolipin increases lipid packing in the membrane to promote curvatures. SS-31 might be able to improve cristae curvatures in BTHS mitochondria and improve ATP production. These results are very encouraging, and we are hopeful that elamipretide can benefit BTHS patients.

Elamipretide represents a paradigm shift in targeting the fundamental cause of cellular energy failure rather than correcting specific genes or proteins for individual diseases. Mitochondrial production of ATP requires the concerted effort of many proteins in the electron transport chain. Rather than target a single protein, these peptides target cardiolipin to induce structural change in the membrane and modify the spatial organization of many mitochondrial proteins to optimize electron transfer. Such a mechanism of drug action has never been described before.

The chance discovery of this tiny molecule has completely changed my life. It has taught me the importance of bioenergetics in healthy living and to develop a more holistic view of health and disease. I hope it will also impact the lives of many people in the Barth community.

Scientific and Medical Advisory Board Changes

By Kate McCurdy, Scientific & Medical Advisory Board, Emerita, Barth Syndrome Foundation

Announcing changes in our Scientific and Medical Advisory Board (SMAB) membership is always a bittersweet exercise, as it involves both thanking those who have served so ably and welcoming those who are joining anew. Dr. Jeffrey Towbin and Dr. Mark Tarnopolsky have each decided that, though they have found serving on our SMAB both enjoyable and rewarding, they are no longer able to dedicate the time that this position deserves due to other commitments, and so they are stepping down. At the same time, we are honored and excited to welcome two new wonderful members to the SMAB – Dr. Brian Feingold and Dr. Michio Hirano.

Those leaving our SMAB:



Jeffrey A. Towbin, MD

Dr. Towbin was a founding member of our SMAB and has served since the committee was formed back in 2001. He has done much to help put us “on the map,” as he is a pediatric cardiologist who is highly regarded around the world. His specialties include heart failure and cardiomyopathy, and he has been extremely helpful over the years to BSF and to our families, both at the policy and the individual patient levels. We will miss him tremendously but know that Barth syndrome will always hold a special place for him.



Mark Tarnopolsky, MD, PhD

Dr. Tarnopolsky joined our SMAB in 2013 as a well-known expert in mitochondrial and neuromuscular diseases. His insight has been very valuable as we have addressed the neuromuscular aspects of our disease in clinical research and entered the new realm (for us) of clinical trials. He has lent his expertise in ways that have been very valuable, and we have appreciated his input into all of our scientific and medical programs.

Those joining our SMAB:



Brian Feingold, MD, MS

Dr. Feingold is an Associate Professor of Pediatrics and Clinical and Translational Science at the University of Pittsburgh School of Medicine (UPMC) and is the Medical Director of the Pediatric Heart Failure and Transplantation Programs at the Children’s Hospital of Pittsburgh of UPMC. His research interests include pediatric transplantation outcomes and pediatric heart failure, including various forms of cardiomyopathy such as non-compaction. He leads a clinical team that cares for a number of boys with Barth syndrome and has attended our International Conference in the past.



Michio Hirano, MD

Dr. Hirano is internationally known as an expert in neuromuscular, metabolic and mitochondrial diseases. He is a Professor of Neurology at the Columbia University Medical Center where he has both clinical and research responsibilities. He is Chief of the Division of Neuromuscular Disorders, Director of the H. Houston Merritt Center for Muscular Dystrophy and Related Diseases, and Medical Director of the Laboratory of Molecular Genetics/Laboratory of Metabolic and Mitochondrial Disease. His specialties include treating patients with mitochondrial diseases, myopathies and other neuromuscular disorders, and he also conducts research in order to advance what is known about them.

Please join me in thanking Drs. Towbin and Tarnopolsky; their service has made a real difference to our foundation, and we will miss them. We hope they will not be strangers as time goes on, as they always will have a home with our Barth family. Also, please help me welcome Drs. Feingold and Hirano to our SMAB; we know that their input into our scientific and medical programs will be extremely valuable, and we are very grateful for their willingness to become official BSF advisors. We have no doubt that they will add to our already strong SMAB, as we continue our quest together for treatments for this life-threatening disorder.

Meet BSF's Executive Committee

The Barth Syndrome Foundation (BSF) continues to grow every year: we find new families, we build our networks with other organizations, and we work hard to extend our reach into the science and medical communities. As a virtual organization, we want to be sure that our members know who serves on our Board and on the Executive Committee, made up of Board officers who meet monthly on the phone to discuss issues and decisions that come up between our quarterly Board meetings. To help you get acquainted with these officers, who are already familiar to many of you, we designed a simple questionnaire and we are running the answers below.



(L-R) Ken, Ronly, Dilen and Susan
(Photo courtesy of Susan McCormack 2018)

Susan McCormack, Chair

Where do you live? Wellesley, MA, USA (a town just west of Boston)

How old are you? 53

Who's in your family? My husband, Ken, me, my two daughters, Dilen (14) and Ronly (12) and our two rescue dogs, Magic and Gwen

What's your connection to Barth syndrome? My daughter, Ronly, was diagnosed as a carrier in utero. We don't know Dilen's carrier status.

How long have you been involved with BSF? I first contacted BSF when we were pregnant with Ronly in 2005. Like many who call BSF "out of the blue," I had never heard of Barth syndrome and was in shock that our family was affected. I had so many questions.

The Foundation was an invaluable source of information and support for us both back then and in the years since.

Why do you serve on the Board? I believe wholeheartedly in BSF's mission, goals and values and I want to do whatever I can to end the suffering that Barth syndrome causes. Board membership requires time, talent and dedication. I decided that I wanted to use my skills to help drive BSF toward future treatments and — one day — a cure for Barth syndrome.

Before you served on the Board, did you do any volunteer work for BSF? I joined the Board relatively soon after connecting with the Foundation. However, over the past 13 years, I've conducted fundraisers, helped with conference planning and execution, participated in volunteer retreats and served as Secretary of the Board. Given my somewhat unique position as the mother of a carrier — and not an individual with Barth syndrome — I've championed the inclusion of carrier issues in our strategic plans.

Favorite BSF conference memory? I have so many that it's hard to pick just one: When we lost power at the Don Cesar and we all huddled in the lobby with flashlights.....Watching the kids build sand castles together at the Hilton.....Meeting folks I had only known through Facebook or the list serv and giving them a hug.....Hearing about exciting new research directly from the scientists conducting it.....

What does the Foundation mean to you? BSF is a lifeline. We know that in times of need, the Foundation will be there for us.

(Cont'd on page 18)

"The Barth Syndrome Foundation does it all: serves as a tight-knit community for affected patients and families; advocates for their community; provides a vast array of educational materials; and helps fund critical research into this rare mitochondrial disease." ~ Colin Steward, PhD, FRCP, FRCPC

Meet BSF's Executive Committee

(Cont'd from page 17)



John Wilkins
(Photo courtesy of John Wilkins 2018)

John Wilkins, Secretary

Where do you live? Lincoln, Nebraska

How old are you? 36 years young

Who's in your family? Mike and Sue Wilkins are my parents. I have a sister, Jessica, brother-in-law, Mark, and niece, Anna.

What's your connection to Barth syndrome? I was born with Barth syndrome before it got its name. For nine years, it was called X linked cardiomyopathy.

How long have you been involved with BSF? Since the beginning, as my mother is one of the founders of the organization.

Why do you serve on the Board? I serve on the board because I like to be involved. I was honored to be selected to join this wonderful group of people to help lead the Foundation.

Before you served on the Board, did you do any volunteer work for BSF? Yes. I performed many odd jobs in the beginning. Then served on the 2010, 2012, 2014 and 2016 conference planning committee.

Favorite BSF conference memory? There are too many to mention! What I like best about the conference is the chance to be with my Barth family. A week to laugh and learn with people I enjoy being with.

What does the BSF Secretary do? The Secretary is responsible for keeping minutes of our board meetings as well as any committee meetings should that be requested. Thankfully, I can delegate this task to the wonderful Lynda Sedefian, our Executive Assistant, but I am ultimately responsible.

What does the Foundation mean to you? The Foundation means so very much to me. It is a large second family. It is hope, funding research and searching for a cure. The Foundation inspires me. It has shown me what a small group of people can do in a short amount of time.



(L-R) Kevin, Connor, Stacey and Ryan
(Photo courtesy of Kevin Woodward 2018)

Kevin Woodward, Treasurer

Where do you live? Phoenix, Maryland (20 miles north of downtown Baltimore)

How old are you? Old enough to be Connor and Ryan's dad

Who's in your family? My wife, Stacey, and two sons, Connor (age 8, BTHS), Ryan (age 4.5), and our dog Jack (age 12, Retriever mix).

What's your connection to Barth syndrome? My older son, Connor, has Barth syndrome.

How long have you been involved with BSF? I have been involved with the BSF since 2012, about six years.

(Cont'd on page 19)

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(Cont'd on page d)

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Wynne McGee, Krystal
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Woodward, Stacey
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Wright, Jessica

Meet BSF's Executive Committee

(Cont'd from page 18)

Why do you serve on the Board? I'm on the Board because I am passionate about the ongoing success of the Foundation and its mission. I quickly figured out that being a member of the foundation wasn't enough for me personally. I wanted to play a larger role in the direction, management and leadership of the BSF.

Before you served on the Board, did you do any volunteer work for BSF? Yes. Stacey and I began hosting quarterly dinners for out-of-town KKI clinic attendees. I sat on the Steering Committee for the 2014 BSF conference. We organized several family fundraisers as well.

Favorite BSF conference memory? Just being with other people who understand and appreciate all the challenges of BTHS.

What does the BSF Treasurer do? The BSF Treasurer holds responsibility for all financial aspects of the Corporation. I am also responsible for the proper disbursement of funds of the Corporation, as ordered by the Board and staff, and I provide regular financial statement updates to the Board at its regular meetings. The BSF Treasurer also sits on the Executive Committee of the Board of Directors, which is responsible for senior leadership and foundational guidance.

What does the Foundation mean to you? Support. Community. Hope.

Rare Disease Day 2018

Two members of our Barth syndrome community, Kate and Susan, recently participated in events marking Rare Disease Day. Kate and Susan were asked by Rare New England (RNE) to be part of its Speaker Series this year, helping to educate members of the medical community about the issues encountered by individuals affected by rare diseases such as Barth syndrome, as well as their families. Kate spoke to a group of doctors and medical students on February 26th at Dartmouth-Hitchcock Medical Center in Lebanon, NH, USA. Kate's presentation, titled "Barth Syndrome: Facing the Dragon," focused on her experiences finding a diagnosis and treatments. Kate and her husband, Sandt, lost their first son, Rhys, to Barth syndrome. Because of early diagnosis, their second child, Bryn, has received the care he needs and is a delightful six-year-old. Susan spoke at the Brandeis University Genetic Counseling Program in Waltham, MA, USA, on May 21st to a group of about 20 graduate students. She centered her talk on the unique circumstances surrounding her family's connection to Barth syndrome, including issues faced by carrier females and their parents. Both Kate and Susan were thankful to have the opportunity to increase awareness of Barth syndrome and to help teach the medical community about the condition.



Kate speaks at Dartmouth-Hitchcock Medical Center
(Photo courtesy of Kate 2018)



(L-R) Julie Gortze (RNE), Dr. Mark Korson (VMP Genetics)
and Susan *(Photo courtesy of Susan 2018)*

"As mother to a son with Barth syndrome this foundation is a total life line, and I mean that literally! This condition is rare and complex, so to know that there is a forum to share wisdom and expertise not only from other parents but also with medical and scientific specialists and experts in their fields and from around the world is nothing short of remarkable, you can't imagine how supportive that is and how there can never be enough thanks given!" ~ Parent of Affected Individual

Be Like Ryan

By Marc Sernel, Father of Affected Individual, Illinois



Ryan Sernel

Life can change in an instant. I thought I would be writing, as my tenure as Board Chairman comes to an end, about the Foundation and all of the great things we've done together during my nine years on the Board. That is all true, but hard to focus on at the moment. As many of you know, our family's life changed on the most horrible day of our lives in March. There's only one thing I can bring myself to write about at this time. I need to tell you about the most loving and amazing boy I have ever had the pleasure of knowing – my son Ryan.

Ryan was truly a parent's dream. He was a great kid who did all the right things – kind, polite, respectful, helpful, a model student, and just so much fun to be around. He was a joy to parent, but that's not to say it wasn't a lot of work. As any parent of a Barth child knows, it can be immensely challenging at times. We worked really hard to try to keep Ryan healthy, and constantly worried about his well-being. And while it required much more effort than parenting a "normal" child, we absolutely loved taking care of Ryan and he was doing so well. Ryan knew that he had something called Barth syndrome, but he never asked about his condition or seemed to have any interest in knowing more. He just lived his life. He went to his many doctor appointments and dutifully took his medicines and supplements. But he didn't let those inconveniences distract him from living life and having fun. This terrible disease never defined Ryan or got him down.

Ryan was a really happy and successful sixth grader with tons of friends. Ryan dreamed big dreams and never let his condition limit his ambitions. He had big plans for this spring and summer, as well as his teenage, college, and adult life. Ryan lived his life to the fullest, with a smile on his face and joy in his heart, for his entire time on Earth. And we loved every single minute of his time here with us. We have so many amazing memories and experiences; the "good times" were nearly all the time, and it's how we remember him. It is both a curse and a blessing that his life ended so suddenly — there was no prolonged hospital stay or suffering. We know other Barth families that have lost their children have not been as fortunate.



Golfing in Wisconsin

I remember hearing early on that kids with Barth syndrome might have trouble with math or other cognitive thinking. Not Ryan. He was always good at math, even getting an A+ in his accelerated math class this past trimester that was two years ahead doing 8th grade algebra. Ryan was indeed a straight "A" student, a bright boy who always strived to do his best. His teachers referred to him as a "rockstar", a "leader in the classroom", "someone that the kids looked up to", "well-liked by all of his classmates" among many other superlatives. Even in some elective classes that he was somewhat nervous about, like Technology and Music, he turned out to be a superstar. He consistently received the highest scores on his technology tests and received great praise from his teacher for his acting and singing skills in taking on the role of Miss Hannigan (from Annie), a performance we will never forget.

Ryan was a genuinely good person that touched the people he came across in special and unique ways. He knew not only how to talk to people but also to listen, and care about their interests and feelings. Everyone who knew Ryan felt their own special connection to him, felt his love, and loved him. There are countless stories from others that only confirmed what we already knew about Ryan's unique way of connecting with people. The story from a mother about when her son transferred into Ryan's school in third grade, and how the transition was difficult until he came home one day and told his mom: "I made my first new friend at school. His name is Ryan." Or the mother that told us that her daughter did not like any of the boys at their school... except Ryan. Or the teacher that told us that none of the kids in her class would get and laugh at her subtle jokes... except Ryan. Or the kids that told us that Ryan would help them with their work in class, even if he was not completely done with his. Or kids from his school that had moved away, but still felt connected to Ryan even though separated by hundreds of miles. Or teenagers, even adults, that met and developed a friendship with Ryan when catching Pokemon. The anecdotes are endless.

We always felt so proud of him when hearing these stories and compliments, but it didn't really surprise us. He truly brightened the day of those around him, every day and all the time.

(Cont'd on page 21)

Be Like Ryan

(Cont'd from page 20)



(L-R) Ryan, Catherine & Doug

come up with ideas for fun too. On one of our Final Four basketball trips to Houston, Ryan suggested that we go to a Houston Rockets game on our “day off” from basketball. “More basketball?” we asked him. Ryan assured us it would be a blast, and he was right. He also frequently suggested going out to one of his many favorite restaurants. Many times it was all of us, but sometimes it was just Tracy or I who would take him. We will so miss these countless 1-on-1 meals with Ryan — quality time at its very best.

Ryan also loved traveling, including our yearly trips to Florida but also new places we visited and explored. I’m so glad that we were fortunate to go on so many fantastic trips with him. Those experiences and memories are priceless. Summer weekend trips to his grandparents’ house in Wisconsin were a favorite of his too. He loved our usual activities up there — golf, tennis, pickle ball, the playground, the beach, backyard games, and going to some of his favorite restaurants. But I think what Ryan most enjoyed is just the amount of time we were all gathered together — talking, laughing, having a good time no matter what we were doing — without the distractions of daily life. That was what really mattered to Ryan, and he was at the center of all of it. We will forever cherish all of our great times as a family with him.



(L-R) Ryan and Doug

No one felt Ryan’s love more than us, his family. We all had our very own special and unique relationship with him, including his grandparents, aunts, uncles, cousins, and of course his brother, sister, mom and me. It was a treat spending time with Ryan — he was so much fun to be around and talk with. He just made you feel good. All you had to do was look at him and it brought a smile to your face. Not only was he a cute kid, but he was usually beaming with a smile himself that could melt your heart. And more than just smile, Ryan made us laugh every day. If it was a quizzical look, a one-liner, or just a funny reaction to something — he had wit and good humor beyond his years. I find myself wanting to text or call him with some inside joke, or story from the past, that only Ryan would understand and find funny. Each one of us in our family felt like the luckiest people in the world to spend so much time with Ryan. He was our special gift.

Ryan packed a lot into his twelve years here with us. He was always game for anything. If anyone wanted to do something or go somewhere, he was up for it. And usually once Ryan joined in, the rest of us wanted to be there as well. He was often the one to

Ryan could not physically handle the rigors of most sports, but he had great hand eye coordination and was a true competitor. He could hit a whiffle ball with the best of them. He enjoyed shooting baskets, and playing tennis and pickle ball. He could beat his friends at ping pong. He was a good golfer that, although he could not hit the ball particularly far, had a smooth swing and a great short game. He loved playing on a dodge ball team with his brother and their friends. And he loved doing just about everything in gym. Even running, as he would give it his all in the 50 yard dash, which was as much an endurance race as a sprint for him. And he was a huge sports fan — basketball, baseball, football, hockey, you name it. He knew the players and their stats, and loved playing his favorite players on video games. He joined fantasy leagues with his friends. He loved wearing sports jerseys to school. It was a tough decision which of his sports jerseys he should wear as his final outfit on Earth; we ultimately decided he would wear James Harden’s red Rockets jersey.

(Cont'd on page 22)

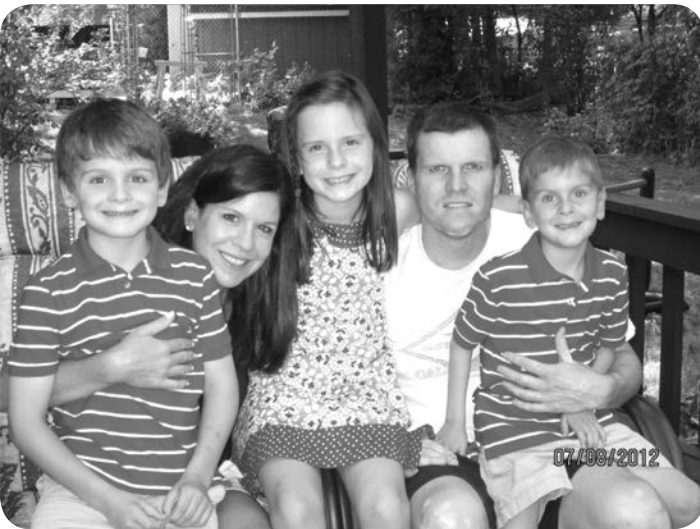
Be Like Ryan

(Cont'd from page 21)



Chess Champion

he couldn't participate in. But Ryan never, ever asked "why" he was unable to play, or bemoaned his physical inability to participate. Ryan's positive spirit never seemed dampened by his physical limitations. I recall one time when we had just finished watching one of his brother's baseball games, and because his brother had pitched well to win the game his teammates were chanting his name "Dougie, Dougie..." I looked over to Ryan, feeling crestfallen that he would likely never be able to have that feeling of being the hero for a sports team. And Ryan's reaction both reassured me that he was ok, and made me laugh..."what nerds" he said with perfect timing and not a care in the world.



Ryan's Family

Where Ryan was really starting to progress was in the sport of the mind — chess. He loved the competition. He loved winning and competing for trophies. He went to a week-long chess camp in Minnesota last summer and loved it, and planned on doing not one but two camps this summer. He had goals of how he wanted to increase his chess rating and be the top player on the high school team when he got there. Ryan had found an activity in which he could compete with all others on a level playing field, and he wanted to be the best chess player he could be. He had improved so much that he could even beat his dad more often than not. I'm sad we won't be able to see how good he might have become, but I'll be practicing to challenge him to a rematch someday in Heaven.

Although Ryan had difficulty keeping up with the physical endeavors of other kids, more often than not Ryan found his way into the middle of the fun. His friends loved him and wanted him involved in all they were doing. We sometimes felt bad that Ryan could not play team sports like his brother or sister, or that there were certain physically rigorous activities his friends would do that

We always knew there was a chance that Ryan might not be with us for our entire time here on Earth, and for that reason we made sure to always be grateful for him and shower him with love every day. Our family is devastated, but one thing we don't have is any regrets in terms of how much love we showed Ryan throughout his life. And we in turn received all that and more from him. Because if there is one word that defines Ryan, it is...LOVE. Love just poured out of Ryan. He loved each of his family members so much. You could feel it as he would look at you with those beautiful hazel eyes and adorable face, smile at you with that precious smile that would melt you, touch your hand, his big hugs, his sweet kiss, just the way he would talk to you. He was the person in our family who could sense if you were feeling down and boost your spirits. He would say "I love you, Mom" without prompting. He would give a hug to his sister when she needed it. He would talk and laugh with his brother about anything and everything. Ryan really was the best friend and constant companion of each one of us. And that of course makes it so hard. We miss his love every day and being able to love him back. But we know he is in Heaven continuing to love us every day and we will love him forever. Through our faith we

know that one day we will all be reunited with him, never to be separated again, and that is what keeps us going every day.

It is truly impossible for any one of us to fill the void of all of the positivity that Ryan brought to this world. Most of us that have lived decades longer haven't impacted as many as Ryan did in his 12 years here. We had a real life angel in our life. We so wish we could have had more time with him on Earth, but that angel taught us so much and we will continue to carry all of it with us. It is now our job, all of our jobs, to be a little bit more like Ryan. Make someone feel loved. Bring smiles to others' faces. Help those in need. Show empathy and compassion. Don't take things, or yourself, too seriously. Don't dwell on your limitations, and do what you are able to the best of your ability. In short, be like Ryan. We will love our sweet boy forever while trying our best to live our lives here like he did.

New York Islanders Host #BarthNight

By Nicole Derusha-Mackey, Board Member, Barth Syndrome Foundation

On February 9th, the New York Islanders hosted their second “Barth Night” at the Barclay Center in New York City, giving hockey fans an opportunity to learn about Barth syndrome and how they can help. Thirteen year-old, Devin, was the Islanders' guest of honor for their game against the Detroit Red Wings. In addition to riding the Zamboni (ice resurfacing machine) and dropping the ceremonial puck, Devin was also featured in a video shown on the scoreboard during the game.

But raising awareness for the Barth Syndrome Foundation (BSF) didn't stop there. Steve McCurdy, BSF Board Chairman *Emeritus* and Barth parent, gave a wonderful TV interview during intermission. On the concourse, almost two dozen #TeamWill volunteers raised awareness while collecting over \$3,000 in cash donations during the first two periods of the game. And in the owner's box, led by the Islanders' owner Jon Ledecky, a long-time BSF supporter, donors opened their wallets as well. In total, nearly \$100,000 was raised for BSF during the NY Islanders #BarthNight.

Devin had an experience of a lifetime during the New York Islanders' #BarthNight. It was one of the happiest nights of his life, and he will talk about it for years to come. His favorite part of #BarthNight, of course, was getting to meet all the Islanders players. Although Devin is typically a fan of the Red Wings, he was happy to be an Islanders' fan for the night and celebrated the Islanders' win. Many thanks to the McCurdy family, Islanders' owner Jon Ledecky, and the NY Islanders Organization for making this amazing night possible.

Back in Michigan, our family and friends hosted a watch party at a local sports bar. I sincerely appreciate all who attended, sponsored, and supported our fourth Devin's League of Superheroes event, raising over \$2,000 for BSF. More than 40 superheroes braved Michigan's blizzard conditions to attend the #BarthNight Watch Party, and a great time was had by all. Special superhero shout-outs to Laura & Gary Derusha and Shelly Horkey for hosting the watch party.



Jon Ledecky presents check to Steve and Kate McCurdy at NY Islanders' #BarthNight (Photo courtesy of Steve McCurdy 2018)



Devin is guest of honor at NY Islanders' #BarthNight (Photo courtesy of NY Islanders 2018)

You Can Make A Difference

Donate by check: Make check payable to **Barth Syndrome Foundation, PO Box 419264, Boston, MA 02241**

Donate online: You can donate to BSF by going to our website, www.barthsyndrome.org, and clicking on the "DONATE" link on our home page.

Employer Matching Gift Programs: Many donors are now taking advantage of a “Matching Gift Program” offered by their employer. The employer matches the funds donated by the employee to a charity and provides a convenient method for the employee to donate to a charity of his/her choice.

Securities: Securities can be gifted to the Barth Syndrome Foundation (BSF) in two ways: either via electronic transfer (in which your broker transfers shares using the BSF DTC number) or by physically mailing the paper certificates to BSF. Either method is acceptable, but it is necessary for you to contact your broker to initiate the transfer.

Planned Giving: One of the best ways to support our continued efforts is to remember BSF (or its affiliates) in your estate planning. Talk to your lawyer or estate planning professional about including BSF (or its affiliates) in your will.



Awareness of Barth Syndrome Continues to Grow

Many Barth syndrome (BTHS) related peer-reviewed journal articles are now being published. To date, a total of **147** articles have been published on BTHS research conducted with the support of BSF and/or BSF affiliate funding (denoted below with *****) and/or acknowledge biological samples and/or information from Barth families, the Barth Syndrome Registry and Repository, and/or BSF affiliates (denoted below with **Δ**). Listed below are articles relevant to BTHS that have been added to BSF's library since the last issue of the Barth Syndrome Journal. To view the complete bibliography on BTHS, please visit www.barthsyndrome.org.

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You are receiving this newsletter because of your interest in Barth syndrome. We would like to keep updating you with our latest news. If you wish to unsubscribe or change the format in which you read it — you may prefer to receive it by email for example — then please contact us and we will do so right away.

Barth Syndrome Trust

By Michaela Damin, Board of Trustees, Barth Syndrome Trust

News from the UK...

Data Protection

In May 2018, the new European Union General Data Protection Regulation (GDPR) came into effect. We have been busy reviewing our policies and procedures to make sure we are conducting ourselves to the highest standard when it comes to collecting, processing, sharing and storing your data.

If you currently receive this newsletter by email, you should already have received an Opt-in email from us and, unless you give us your consent to continue sending you our news via email, we will no longer be able to send you the newsletter.

For member families, you should have received an updated Consent to Share Information form, either by email or by post. Please ensure that you return this to us so that we can record your preferences and act accordingly.

We have updated our Privacy policy, which can be found on our website at www.barthsyndrome.org.uk.

Lastly, if you would like to unsubscribe from this newsletter at any time, please send an email to news@barthsyndrome.org.uk or contact us by telephone or by post (contact details are on page 21).

Funding Research

We are delighted to be funding half of Dr Amulic's research project, with BSF funding the other half. We wish the team well and look forward to hearing all about their research in due course.

Using blood samples from BTHS individuals to uncover the reasons for neutropenia. Neutropenia is one of the most misunderstood symptoms of BTHS, and it is just as much a threat to life as heart problems. The reasons for why this major class of white blood cells is reduced in BTHS is unknown even given all that we now know about the tafazzin gene. Dr. Amulic along with Dr. Colin Steward will use blood samples from BTHS individuals attending the UK clinic to investigate: 1) the metabolism of neutrophils; 2) whether non-apoptotic cell death (NETosis) is involved with neutropenia; and 3) whether neutrophils from BTHS individuals lack the ability to destroy bacteria as they ought. Neutrophils will be isolated from individuals and then examined for subcellular structural integrity/functionality by confocal microscopy and for biochemical reactions with the Seahorse™ metabolism detection system. In addition, they will determine whether NETosis, which is a non-apoptotic form of cell death that Dr. Amulic is a leading authority upon, plays any role in BTHS. Finally, Dr. Amulic will examine the bacteria-killing abilities of these isolated neutrophils by measuring bacterial destruction, cytokine release, and the granule release that normally accompanies neutrophil bactericidal or bacteria-killing function. Understanding the defects in neutrophils and the dysfunction of neutropenia will provide therapeutic insights for BTHS and other neutropenia-associated diseases.

Board News

It was with much sadness that we said farewell to Nigel Moore who stepped off the Board as Treasurer at the end of February, after serving a mammoth four consecutive terms. People from all around the world wrote in to thank Nigel for his service:

"He has been so dedicated to our mission on a global front."

"Thank you so much for your commitment, time, and talents that you provided to BST while serving on their board over the past 12 years."

"He is one of my favorite people."

Nigel and his wife, Lorna, also serve on our Publications Committee and are very involved in creating, editing and sending out the BSF newsletter, BST fundraising newsletter, brochures and conference programs. We're very grateful for all his efforts over the years. Thankfully Nigel and Lorna will continue helping out on our Publications Team!



Nigel Moore

(Cont'd on page 27)

Barth Syndrome Trust

(Cont'd from page 26)



Cheryl & Geoff Parish

We welcome Geoff Parish to our Board! Geoff has taken over the role of Treasurer. His background is in business-focused IT and brings a commercial focus to delivering IT strategies closely aligned to business strategy, delivering projects and programs of all sizes to enable business improvement or transformation, and enhancing customer service through effective delivery of operations and support. He is an excellent strategist and change agent, able to see the "big picture", with a calm and methodical approach, and an all-round nice guy who would like to serve our families going forward in his role as Treasurer and board member.

Other Board News

On 7th February, Michaela Damin stepped down as Chairperson, and this role has been taken up by Cheryl Parish. Michaela remains on the Board of Trustees and continues in her usual role of family support and day-to-day running of the organisation. Cheryl brings her considerable experience in chairing meetings to assist Michaela by focusing on the support and development of the Board. *(Photos courtesy of BST 2018)*

News from Bristol

We welcome consultants Drs. Germaine Pierre and Effie Chronopoulou as new joint Leads for the Bristol Barth Syndrome NHS Service. They took over from Prof. Colin Steward when he retired at the end of 2017. Due to time and space constraints, we will feature Germaine and Effie in the next newsletter. We look forward to hearing from them about their exciting plans for the Barth syndrome Service going forward.

News from the NHS Barth Syndrome Service, Bristol

By Hayley Smith, Specialist Nurse

Farewell to Colin, Debbie, Nicol and Livvy...

We have had much change over the last six months with the team. Prof. Colin Steward and Debbie Riddiford retiring has triggered a period of change. Nicol Clayton, our expert dietician is also moving on to new opportunities, but she will remain with us for one day a week for a period of time and attend clinic where possible until the new team member has gained some experience.

The service is in the midst of interviews for this post as well as for a permanent physio. Over the last year or so, that post has been filled admirably by Olivia Berry, but this is on a rotational basis and we are keen to have a permanent member of the team to build expertise and provide consistency. As soon as the successful candidates are announced, we will introduce new members and provide contact details.

I have now been in post (officially) for almost a year and continue to thoroughly enjoy the role. (Who wouldn't with the great families who make the post so much fun!)

Creating an Adult Barth Clinic

The latest, and in some ways most challenging aspect of my role since Christmas, has been to develop an adult clinic for those over 18. Being able to organise the clinic in an appropriate setting is important to ensure that we maximise the experience and minimise the

(Cont'd on page 28)

News from the NHS Barth Syndrome Service, Bristol

(Cont'd from page 27)

difficulty to promote independence for each young person within their healthcare provision without diminishing the experience. We are making progress and hope to be able to invite our first group of young men in the near future. I have also sent letters to each of these individuals explaining the changes and delays in more detail.

Paediatric Clinics

The paediatric clinic has also evolved over the last six months and is now more streamlined. Whilst the clinics are not as big as before (fewer patients at each) I have worked hard to ensure that families have a "lunch break" to allow time for a meal and for socialising. We also aim to finish a little earlier and enable families to leave Bristol before the worst of rush hour!!

Clinics will now always occur on the first Friday of a month, unless there are exceptional circumstances. I have also developed a telephone tool that will facilitate a more individualised clinic – this means that I call families several weeks before clinic to discuss what issues are most relevant to their care at that time and use this to facilitate appropriate meetings with professionals to address concerns and queries.

Need to Get in Touch with Us?

With the new data protection law coming into force in late May, I also wish to take the opportunity to make parents/families aware that the hospital email addresses are NOT secure. You are welcome to contact us via these, but if you wish to share sensitive information, this may not be the best route. We are working on how we can improve contact.

We do have PKB – Patients Know Best, which is secure and a good way of contacting relevant professionals directly.

With the personnel changes that are taking place at the moment, I have only listed my contact details as I know these will not change. I hope by the time of conference/next newsletter to have a formatted and comprehensive list of names, numbers and email that I can share with all.

Contact Details

Alongside data protection rules, all uhbristol.nhs.uk email addresses are not secure. Please be aware of this if you wish to share personal information.

Miss Hayley Smith, Nurse Specialist

Hayley.smith@uhbristol.nhs.uk (not secure)

Hayley.smith32@nhs.net (secure)

0117 3421693 (no voicemail)

07795 507902 (with voicemail)



Barth Syndrome Foundation of Canada President's Report

By Susan Hone, President, Barth Syndrome Foundation of Canada



(L-R) Board members: Susan, Chris, Cathy & Lynn

After the winter that never wanted to end, it is finally spring in my part of the world. With spring comes a renewed energy. I, along with several other members of Barth Syndrome Foundation of Canada (BSFCa), am busy volunteering on a number of projects for the upcoming International Scientific, Medical and Family Conference. I am eagerly awaiting seeing my Barth family, both old and new.

BSFCa is proud to contribute \$2,500 to this year's conference. In addition, we will be sponsoring two Canadian researchers to attend the conference.

This year BSFCa is also contributing \$10,000.00 towards the annual grant process sponsored by Barth Syndrome Foundation. We are sponsoring Richard Epan, PhD, Professor Biochemistry and Biomedical Sciences, McMaster University, on "The cause and consequences of plasmalogen depletion in Barth syndrome." (See pages 11-13)

The chart below shows BSFCa's Financial Statement for the year ended December 2017.

Barth Syndrome Foundation of Canada ~ 2017 Financial Statement	
Revenue	\$ 39,602
Operating Expenses	12,676
Research Grant Funding	32,343
Net Revenue	(\$ 5,417)*

**Note: The Board made the conscious decision to have a negative net revenue.*

The articles below are all from members of BSFCa and are very personal in nature. Thank you to all the contributors for opening up their hearts and expressing their emotions in describing their experiences living with/or with someone who has Barth syndrome. Their stories motivate us to continue "Enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome".

My Tubie Family

By Jasmine, Mother of Affected Individual, British Columbia

The purpose of this article is to focus on feeding tube awareness, the truth of tube feeding. This is my family's truth/story of how we became a Tubie family and the experiences that go along with that.

My son, Caleb, was born a year and a half ago. Within 32 hours, he went into severe heart failure and ended up on life support. A heart and lung machine kept him alive for the first two weeks of his life. After being removed from life support because of fear of stroke, he fought and overcame many battles to stay alive. He was given a diagnosis on day 21 of his life, a diagnosis of Barth syndrome which forever changed our lives.

After roughly six months of being in the hospital, Caleb was discharged with a feeding tube in his nose that ran down to his stomach (called a nasogastric tube or NG-tube for short). Due to months of being intubated so that he could breathe, Caleb had developed an oral aversion and would not take nearly enough milk by mouth for him to thrive. He would also vomit regularly as we tried to find a volume of milk that his body would tolerate. Caleb's oral aversion intensified when he caught a cold that had him coughing up mucus and causing him to vomit more frequently. He went from orally taking half of each feed to taking nothing by mouth.

(Cont'd on page 30)

My Tubie Family

(Cont'd from page 29)

After 13 months of having an NG tube, Caleb underwent gastrostomy surgery and had a gastric tube (G-tube for short) placed in his stomach. An occupational therapist had made sure that Caleb had the ability to swallow, and the G-tube became a better long-term solution for us than continuing with the NG-tube which he was pulling out on a daily basis.

The following days were a blur of surgery, learning how to use his low-profile version of the tube (called a MIC-KEY button), how much formula to push and how to use the pump. We tried slowing the rate down, continuous feeds, night feeds. It was a struggle to find the right balance so that Caleb would tolerate his feeds and be hungry enough in between to motivate him to eat orally. Eventually, with the help of some other Tubie families and Caleb's dietician, we found a good rhythm and Caleb has slowly (he's under the 3rd percentile for his age in weight) started to gain weight.

Along with having tubes down their throats for so many months, boys with Barth syndrome can also have very strong oral aversions to food. Feeding tubes in the Barth community are not uncommon, and some boys have them for many years. There is one man who still has his and he is in his 30s. Caleb loves food, he will eat some incredibly strange things for a child his age (pickles, olives, feta cheese, raw onion). I use the term eat loosely as he still doesn't actually swallow anything. He will chew everything until it is mush and then spit it out or gags on it. Our hope is that one day Caleb will eat enough orally to give him the nutrients he needs to grow and thrive, but like most things with Caleb, it will happen on his time when he decides he's ready.

There are many reasons why he needs the feeding tube, and we have come to develop a love/ hate relationship with it. It definitely comes with its fair share of struggles. Caleb may have his button for a year, or he may have it for many years. We don't know and have come to accept that. What we do know is that it is giving him what he needs, allowing him to be a kid who can crawl around, go swimming, and even wrestle with his big brother. For now, we are a Tubie family and we are proud to be!



Caleb
(Photo courtesy of Jasmine 2018)

Life with Barth Syndrome

By Ryan, Affected Individual (age 26), Ontario



Jessica & Ryan
(Photo courtesy of BSFCa 2018)

I always find it kind of ironic and funny when people ask me what it's like living with Barth syndrome, whether it was as a kid or now as an adult living with my girlfriend, Jess, and working full time. I don't have anything personal to compare it to, since I've had it my whole life. I've never known anything different. It's like asking someone to compare two different meals. One they have had all their life and quite enjoy versus one they can only see. It's always seemed kind of a fruitless endeavour to me. "Ah look how much better that apparently is; just because I can't have it and it's not just like mine." It makes no sense.

When Susan first asked me to write this I had no idea what to say. So, I spent a week watching the people around me and talking to them about their experiences. I saw people worrying and stressing to make ends meet, trying to make it through the day, but having fun and goofing around with their friends and co-workers while doing it, wishing they could spend more time with family. In short, I watched them do and experience all the things I do on a day to day basis. Yes, there are things I'll never be able to do, and that is something I have come to accept from an early age. I would say, however, that that is true of just about everyone.

(Cont'd on page 31)

Life with Barth Syndrome

(Cont'd from page 30)

There are many things I can still do. I went to college, I can live on my own and pay my own bills. I hang out with friends and work full time. In short, I would say that, in my experience, there is no difference between living with Barth syndrome and living without it. True, there are more concerns and risks. But there are in everything. That's why it's called "life" and not "hiding." I feel like the more appropriate question would be: "How do I let Barth syndrome affect me and my life?" And the truth is... I decide not to let it. I do everything I can and want to. There are days when I get home from work and am exhausted and days I feel like garbage. There's no avoiding it. What I have to keep in mind is that tomorrow is a new day and I need to make the most of it. If it ends up being the last thing I do... then screw it. I refuse to let Barth syndrome hold me back from the life I want to live, I will not regret the chances I didn't take.

Barth Syndrome Foundation Saved My Son's Life

By Lynn, Mother of Affected Individual, Ontario

You just never know when a crisis will occur and you may need help. The good news is that whenever you do face that situation — the Barth Syndrome Foundation (BSF), its affiliates and the families are there to help.

In late April last year, I got one of those phone calls you hope never to get. My father called to tell me my son, Adam, was in a serious ATV accident. He would be okay but was "banged up pretty badly" and was being airlifted from the local hospital near our cottage to the best trauma hospital in Toronto.

We sprang out of bed and into action. First, I grabbed my Barth go bag — which has been packed and by the bedroom door for many years (thank you BSF!). Next, I picked up the huge binder of Adam's medical records (thank you Kate McCurdy and Shelley Bowen who impressed upon me the need to have this together). Next, I went to the website and printed off several of the FACT sheets. Then we headed off to the hospital to wait for the helicopter.



*(L-R) Les & Adam (age 28) enjoy some quality time
(Photo courtesy of BSFCa 2018)*

As we waited for Adam to arrive, we shared a pamphlet and some information sheets with the trauma physician on call. They asked some questions and took it all very seriously as they prepared for his care upon arrival. This was the start of sharing information from BSF with dozens of physicians and healthcare professionals as they worked on Adam. We were extremely impressed by the level of attention they paid to the material and to our guidance — and we believe it saved Adam's life.

Once the initial assessment was done by the trauma team in emergency, it became clear that Adam had fractured his C1 (top of the spine) and broken his jaw and several bones in his face. Serious injuries indeed and at least one surgery required. Fortunately, his neck fracture did not require a permanent rod in his neck, nor did it affect his mobility or neurological function. He was very lucky. Instead he needed to have a "halo," essentially a cage with pins into the skull and a stability vest that held his head and neck stable for several weeks until the neck fracture could heal. Adam did need surgery to put plates in his face and wire his jaw shut until it healed, then a minor procedure to remove the jaw wires.

From the very first, the Barth Syndrome Foundation, the parents and clinicians were there helping in so many ways. Susan Hone and Cathy Ritter gave me advice and the words I needed to convince the doctors and nurses to ensure he had glucose in his IV and especially before surgery. Shelley gave me contacts and advice, and Michaela put me in touch with Dr. Steward who provided the much needed guidance to the hematologists so they would be proactive with neupogen and keep it in his system until the halo was removed and the pin sites healed. Nicol Clayton, dietician in the UK, sent information on Adam's required daily nutrition amount, which allowed them to remove the feeding tube when his jaw was wired shut. Whenever there was something we needed to advocate for, there was someone or a written article to back us up so we could get the best possible care for Adam.

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Barth Syndrome Foundation Saved My Son's Life

(Cont'd from page 31)

Of course, it was a tough time for Adam, but he was an inspiration. He treated people with kindness and humour, even when he was suffering. The hospital staff told me on more than one occasion that they enjoyed working with him. They invited him to speak with high school students that tour the hospital, and he did that several times, including his grandpa and mother, and impressed us all. We had more quality time together than normal over those months and there were some moments we will cherish.

Throughout the weeks as we nursed Adam back to health, our extended family were active partners – visiting often, ensuring he was getting calories (he even gained a few pounds while his jaw was wired shut), advocating with all the practitioners, bringing us laughter and sharing hugs. And our Barth family was there – only an email or a phone call away whenever we needed them - and understanding when we were too exhausted or consumed to be in touch. We are so very lucky to have such a wonderful family – those related by blood and marriage, and those we have gained through BSF and affiliates. Thanks to all of you and our close friends, my wonderful son is well again. We thank you with all of our hearts!

The McJanett Un-Birthday Fundraiser

By Lynn Elwood, Board Director, Barth Syndrome Foundation of Canada, Ontario

Susan and Bob McJanett are wonderful people who are fun to be with. They have done a number of fundraising efforts for Barth Syndrome Foundation of Canada (BSFCa), and this year they chose the occasion of Susan's birthday as the focus for a celebration. Well not really her birthday – her "un-birthday". Susan's actual birthday is in November and that isn't a great time for a backyard party in Toronto, so they held the celebration in May and dubbed it the "un-birthday party".

It was a wonderful afternoon. The McJanett's live in a beautiful old part of Toronto and have a sloping back yard with lots of room and a gazebo near the back. They hired a band and invited friends, colleagues and neighbours to join for an afternoon of refreshments, music and friendship. Instead of birthday gifts, donations to BSFCa were encouraged. We laughed, danced, and at the end of the day, between the party goes and an "anonymous" donation, they raised \$5,000 for BSFCa. Those funds go a long way towards helping us to fund research, sponsor parts of the BSF International Scientific, Medical and Family Conference, and help the Canadian families affected by Barth syndrome. Thank you so very much Bob and Susan! Thank you also to Lois and Les for the connection to such great people and all the help at the event.



Celebrating the Un-Birthday
(Photo courtesy of BSFCa 2018)

Barth Italia Onlus

From Basic Research to Therapy: The Story of Barth Syndrome

By Daniela Toniolo, PhD, San Raffaele Scientific Institute, and Angela Corcelli, PhD, University of Bari Aldo Moro, Bari, Italy

"I am a scientist who has dedicated her entire life to basic research.... we aimed to open the way to future studies and to therapies for rare disorders. But we were not convinced that this would happen. At least in the case of Barth syndrome what we were hoping truly happened." ~ Daniela Toniolo, PhD



Dr. Daniela Toniolo
(Photo courtesy of Paolo 2018)

It was the summer of 1995. My lab was sequencing candidate genes in patients affected by X-linked disorders mapped to the long arm of the X chromosome. No human genome sequence was available at that time and the physical map of the X chromosome was under construction. Very few genes were known. We had developed an original approach to identify genes of the X chromosome where several rare genetic diseases had been mapped, and we were able to sequence the more appropriate candidates in selected patients. This was a very exciting period of my scientific life: with very primitive tools compared to what we have today and a lot of manual work and time spent to run wet experiments we could identify several disease genes and give the first clue to the understanding of the cause of many rare disorders. We hoped to open the way to future studies and possibly to therapies. But we were not sure.

One of the disorders we studied was Barth syndrome, a rare genetic disorder first described by Dr. Peter Barth. Peter, who was following most patients around the world, gave us samples of DNA and cell lines. We had few candidate genes to test, one was called G4.5. The gene was difficult to sequence and it encoded many different possible proteins. It had not been easy to understand its structure, but eventually we did and could sequence G4.5 in Barth syndrome patients. A new undergraduate student had joined my lab. Patrizia D'Adamo was not an expert in reading a sequencing gel, but she realized that in the DNA of one of the probands there was a different base that may cause a change in the protein. When she came to me and to her more experienced colleague, Silvia Bione, we immediately realized that we may have

found the gene responsible for Barth syndrome!

This was indeed the case, and the very complex gene that had produced so many nightmares to the lab was eventually called after a masochistic comic character very popular in Italy at the time, Tafazzi. Our paper was out in April of 1996: Bione S, D'Adamo P, Maestri E, Gedeon AK, Bolhuis PA, Toniolo D. A novel X-linked gene, G4.5. is responsible for Barth syndrome. *Nat Genet.* 1996 12(4):385-9.

In line with the Tafazzi character, it was suggested that the *TAZ* gene may be involved in lipid metabolism: the *TAZ* gene had sequence homologies with a family of established acyl-transferases involved in phospholipid biosynthesis and/or remodeling. Pursuing this was far beyond our technical possibilities and we gave up studying Barth syndrome. We had passed the sequence of the gene in patients to Amelia Morrone, head of the metabolic diseases diagnostic lab of the Meyer Hospital in Florence.

I did not follow the research on Barth syndrome until many years later, in 2009, when, in the journal of the Barth Syndrome Foundation, I saw the picture and the obituary of Michel Bowen. I knew him by name, as he was one of the patients we sequenced when we identified the gene. I wrote to his mother, Shelley, recalling the courage of those families who travelled a very long way to try to understand what was wrong with their children and their confidence in medicine and science. She invited me to the next meeting of the Barth Syndrome Foundation. When I arrived in Orlando, I realized what the Barth Syndrome Foundation has done to help families and to fund research and how much the knowledge of Barth syndrome had progressed since the late 90s.

It was fantastic, and I was very proud to represent the beginning of all that. In 2000, we had left the field. Fortunately, somebody else took the lead and tried to understand what *tafazzin* was doing in the cell, particularly in muscle and in the heart. Between 2000 and 2003, the groups of Dr. Peter Barth in the Netherlands and of Dr. Michel Schlame in New York showed that the metabolism of cardiolipin (CL), the main phospholipid of the mitochondrial membrane, was affected by *tafazzin* mutations and that the problem in Barth syndrome patients resulted from a reduction of the CL and in an altered composition of CL in mitochondria membranes in all tissues tested. They eventually showed the absence of tetralinoleoyl-cardiolipin, a major molecular species in several control tissues,

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Barth Italia Onlus

From Basic Research to Therapy: The Story of Barth Syndrome

(Cont'd from page 33)

"When I arrived in Orlando I realized what the Barth Syndrome Foundation has done to help families and to fund research and how much the knowledge of Barth syndrome had progressed since the late 90s. It was fantastic and I was very proud to represent the beginning of all that." ~ Daniela Toniolo, PhD

and the presence of higher levels of monolysocardiolipin compared to controls. In 2009, a very specific and simple diagnostic test, able to distinguish Barth syndrome from other infantile cardiomyopathies was developed based on high-performance liquid chromatography-mass spectrometry of blood. The work done by the biochemists was a big step forward. Ten years had passed since the identification of *TAZ*, and science had slowly but steadily moved onward; the biological mechanisms altered in patients had been clarified and a diagnostic test had been developed.

Now close to another 10 years have passed. A patient cell bank, constantly increasing in the number of samples, and mouse mutants, eventually constructed, provide models to better define disease mechanism and to test new drugs and gene therapy. Drugs for mitochondrial disorders are identified, and gene therapy is slowly progressing with new approaches being developed. Research in human disorders has profoundly changed since 1995, and I would say in a positive way: we have greatly increased our knowledge of Barth syndrome, and particularly new and more efficient tools are now available. What was started in 1995 was a shot in the dark and just the beginning of a story that has progressed slowly but steadily for more than 20 years of research by different groups with different expertise and now we hope may have a happy ending. I think that the story of Barth syndrome represents a good example of what research can achieve when good projects are funded and tools are available.

When, in 2014, Paola Cazzaniga, the mother of an Italian patient, with a dramatic story, wrote to me looking for help to start a Barth Association in Italy, I was ready to go back to Barth syndrome. With the help of the Florence group, particularly Alice Donati and Amelia Morrone, we organized the first meeting of what became the Barth Italia Onlus.

I am very happy to be involved with Barth Italia and their new project to improve diagnosis. In Italy, too few patients are diagnosed, and still too often we hear of the diagnostic odyssey of patients, going to multiple consultations and tests. As with most mitochondrial diseases, Barth syndrome is difficult to diagnose reliably because of the wide clinical heterogeneity and requires a detailed medical history and extensive knowledge and expertise of the diagnosing physician. Misdiagnoses are not uncommon, even in large hospitals that should know the disease!

In Italy, considering the good public health system and the availability of large clinical reference centers for infantile dilated cardiomyopathies, Barth Italia has considered that a screening could be a feasible task and planned the screening of CL in the blood of male pediatric age patients presenting with dilated cardiomyopathy. The screening should identify patients with altered CL, that could be confirmed as Barth syndrome by sequence of the *TAZ* gene. We plan to screen patients in all centers through Italy, and the project is now recruiting centers and clinicians. I am quite sure that eventually they all will agree to participate. The number of patients that we expect to identify will be very small, but it will be indeed a great success to diagnose new patients that may be followed properly and may receive new drugs and other treatments that are being developed or will be developed in the near future.

I am a scientist who has dedicated her entire life to basic research. I always considered a success to be when I was able to understand biological mechanisms. Now I am almost surprised to see that our work goes toward the cure of a disease. We wrote in many grant applications that we aimed to open the way to future studies and to therapies for rare disorders, but we were not convinced that this would happen. At least in the case of Barth syndrome, what we were hoping truly happened.



Barth Italia Onlus Report

December 2017 — April 2018

By Paolo Muller, Father of Affected Individual, Italy



(L-R) Dr. Daniela Toniolo & Paola
(Photo courtesy of Paolo 2018)

From November 2017 to now, Barth Italia has carried out various fundraising and education outreach initiatives.

The events confirmed that in just a few years we have built relationships that today have become very solid and that in the future will allow us to increase our initiatives.

In December, we organized the usual Christmas Charity Market which has now become a regular event for our city. Elena and Catalin, another Italian family with two sweet Barth boys, also promoted a similar initiative for the first time in their city, with excellent results.

In collaboration with another group, during the Christmas period we organized two bingo games that were greatly appreciated by grandparents and children.

On the occasion of the World Rare Diseases Day, we had two main events. The first was the annual Barth Dinner with friends and supporters that was also a great success. In particular, we have been able to educate all those who love and support us about the syndrome, our initiatives, ongoing research projects and future Barth Italia programs.

The second one, called “Fai la cosa...buona” (“Do the right thing.”), was realized in collaboration with some restaurants in Verona, Monza and Milan. They added an extra charge to the account that was then donated to Barth Italia.

It is a very interesting initiative that we hope to improve and repeat next year. On the occasion of the Verona event we met an actor, very well known and loved in Italy, who was photographed with a Barth bracelet and spoke about Barth syndrome during one of his shows. We hope that he will become an important ambassador for Barth Italia.

In the next months, we are preparing for two initiatives in particular. We are working on Barth jewelry that we have been selling for a year, sharing information about the syndrome with a lot of people. We are working on casual wear and leather bags too, with our beautiful Barth logo.

We are very happy to be bringing the collections to the Barth Syndrome Foundation’s 2018 conference in Clearwater Beach, Florida, to share them with the big Barth family, donating together and contributing to the conference.

The second one is a party at the beginning of June at the Tennis Club in our town where, between music and good Italian food, we will present a preview of the collections to Italian friends.

Thanks to all these efforts, this year at the conference there will be three families and ten doctors/specialists from Italy.

And, above all, we hope we can promote and finance a project to increase diagnosis in Italy. That is very close to our hearts. Thanks to this project, all the most important pediatric hospitals in Italy will test all children with dilated cardiomyopathy for Barth syndrome.





Barth Syndrome Foundation

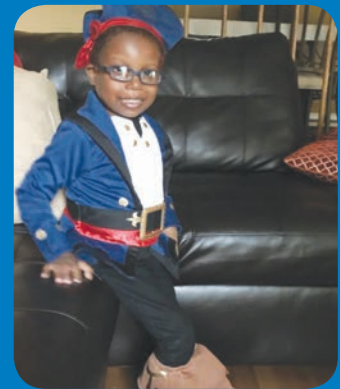
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Barth syndrome (BTHS; OMIM #302060) (ICD-10: E78.71)

A rare, life-threatening genetic disorder primarily affecting males around the world. It is caused by a mutation in the *tafazzin* gene (*TAZ*, also called G4.5), resulting in an inborn error of phospholipid metabolism.

Though not always present, cardinal characteristics of this multi-system disorder often include combinations and varying degrees of:

- **Cardiomyopathy** (*usually dilated with variable myocardial hypertrophy, sometimes with left ventricular noncompaction and/or endocardial fibroelastosis*)
- **Neutropenia** (*can be chronic, intermittent, cyclic, or not present*)
- **Low muscle mass and muscle weakness**
- **Growth delay** (*short stature in the early years, followed by accelerated growth in mid- to late puberty*)
- **Exercise intolerance** due to early fatigue
- **Feeding problems** (*e.g., difficulty sucking, swallowing, or chewing; aversion to some food textures; selective or picky eating; frequent vomiting*)
- **Cardiolipin abnormalities**
- **3-methylglutaconic aciduria** (*variable but typically a 5- to 20-fold increase*)



Quentin (age 6)
(Photo courtesy of Maurita 2019)



Ben (age 13)
(Photo courtesy of Amanda Clark 2016)

The personal opinions expressed in this newsletter are those of the authors of each article and do not necessarily reflect the views of the Barth Syndrome Foundation.