



Barth Syndrome
Foundation
www.barthsyndrome.org

Barth Syndrome Journal

Volume 16, Issue 1 ~ Spring/Summer 2016

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

Volunteer Participation in Rare Disease Research is Critical for All of Us

By Dominic Reeds, MD, Division of Endocrinology, Metabolism and Lipid Research, Washington University School of Medicine, St. Louis, MO

“Research into rare diseases has often produced critical discoveries that have revolutionized our understanding of human physiology and led to new treatments of both rare and common diseases. ... We thank you for your willingness to get involved and volunteer to help find a cure.”

As a clinical researcher, I am fortunate to be able to not only care for patients, but also to perform patient-oriented studies to clarify the causes and ultimately the best treatments for their conditions. With two inquisitive eight-year old sons at home, our family dinner conversation naturally turns to a discussion of the patients I have seen that day. On one such day, Dr. Todd Cade and I had performed an exercise study on a nine year old boy with Barth syndrome and early signs of heart failure. Both of my children wanted to know why the boy was in the hospital and then wanted to know if they could do anything to help. “Volunteer for the study,” I replied. They both wanted to know what the study entailed, and, after hearing about it, Nick asked if he could do it. I asked him why he wanted to sign up.



Dominic Reeds, MD

“It’s not fair that that boy had heart trouble. He is only a boy. I want to help these people. I want their lives to be better. I really want them to get better.”

(Cont’d on page 4)

| Inside this Issue | |
|--|-------|
| Volunteer Participation in Rare Disease Research | 1/4 |
| Researchers and Physicians, Come to Clearwater | 1/5 |
| #TeamBarth | 2 |
| Letter from Chairman | 3 |
| Barth Conference Keynote Speaker | 6 |
| Newly Revised Healthcare Professional Brochure | 6 |
| BSF Conference - A United Front and Strong Voice | 7 |
| 2015 Resesarch Grant Awardees | 8-9 |
| BTHS Clinic at KKI | 10 |
| Australian Family Outreach | 11 |
| BSF Development | 12-13 |
| Journal Articles Relevant to Barth Syndrome | 14 |
| Power of Kindness | a-d |
| BSF Affiliates & Rosters | 19 |
| Barth Syndrome Trust UK | 20-22 |
| BSF of Canada | 23-24 |
| Association Barth France | 24-26 |
| Association Barth Italy | 27 |
| Description of Barth syndrome (BTHS) | 28 |

Researchers and Physicians, Come to Clearwater!

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

It is a model of how disease research can be done and maybe should be done.

My first major interaction with the Barth Syndrome Foundation (BSF) was attending their 2006 International Scientific, Medical & Family Conference on Barth syndrome which was held that year in Orlando, Florida. Up to that time, my experience with rare diseases was limited to working for a biotechnology company that sought to commercialize individual chemical chaperones for treating several rare diseases. In Orlando, I realized that BSF had a disease in search of a treatment and not a treatment in search of a disease — a different perspective from what I was used to.

In 2006, there were few, if any, drug discovery plans for Barth syndrome. In the years leading up to that 2006 Conference, BSF began a Research Grant Program to encourage investigators and to generate more research into this ultra-rare genetic disease. Until BSF arrived in 2000, there were precious few researchers working in this area at all. Ideas about potential treatments had to take a backseat to first

(Cont’d on page 5)

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#TeamBarth

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

As you read through this newsletter, particularly the featured articles on the cover, I hope you'll feel as inspired to be part of this group as I do. Dr. Reeds's story (see page 1) about how his eight-year-old son eagerly signed up to be a control subject for Dr. Cade's study shows how much people care. We are grateful to Dr. Reeds for encouraging his son to help make a difference. His selflessness will make an impact in the research, as well as in the lives of our boys and men... people he is unlikely to meet. And, of course, we're thankful to Dr. Cade, and all of our researchers, for pushing hard to find answers about Barth syndrome.

When I read Dr. Matt Toth's (BSF Science Director) article asking all of our researchers and physicians to attend the 2016 Conference, I was also moved (see page 1). He speaks about how, just ten short years ago, when he joined BSF, there were no treatments in sight. And now, at our 2016 Conference, we have several, possible therapies to discuss. Wow!

Both articles underscore the importance of us coming together, all of us, as Team Barth. Bringing the concept of therapies, specific to Barth syndrome, from the laboratory to the patient will take tremendous team work. Together, we are Team Barth, and we all need to step up.

At the Conference, many of us will be together to hear about all of the exciting developments in science and medicine. We understand not everyone can make it. There will be plenty of opportunities to participate. Leading up to the Conference, during our time in Florida, and certainly afterward, you will hear more.

Specifically for our families: You will soon learn about clinical trials and their importance in finding treatments to help our boys and men. Along with the excitement, you will likely have questions. Rest assured, we will provide everyone with the tools and information needed to make a decision that is best for them.

Just as Dr. Reeds stated, I want to thank you, in advance, for your willingness to get involved and volunteer to help find a cure. We simply cannot do it without you.

I am as excited as Matt to join our families, researchers, and physicians this year in Clearwater, Florida! As always, Matt is available to each and every one of you to talk about any of these topics. Feel free to call or email him to talk about anything research related. In fact, all four of us on staff at BSF are here for you.

Let's go, Team Barth!

Lindsay B. Groff

Lindsay B. Groff
Executive Director



We hope to announce details about a clinical trial specially for Barth syndrome in the near future. Here are some topics we plan to cover:

- What is a clinical trial?
- How does a clinical trial work?
- Why should you participate in a clinical trial?

On the Brink of Something Special

By Marc Sernel, Chairman, Barth Syndrome Foundation

I generally don't like surprises. If I'm surprised in my line of work that usually means I haven't done a good enough job anticipating all possible developments or outcomes. I'm not even a big fan of "good surprises" either. Surprise parties are not really my thing, and I'm not one to say "surprise me" when ordering something. I like to know what I'm getting into. I prefer stability and predictability — some might call it boring — over lots of highs and lows.

A life affected by Barth syndrome is a life full of surprises, highs and lows. It started for my family with the biggest surprise of all — my son has one of the rarest disorders, something even the top pediatric specialists in Chicago have never heard of? The expectations of "normal" childhood development and milestones are replaced by uncertainty about what surprise might come next. Barth syndrome is a roller-coaster ride. Just when you think you know what to expect you learn to expect the unexpected, there's another dip or turn or loop-to-loop. Many times these are turns for the worse, but there can also be turns for the better as well. Cardiologists have been surprised at how the hearts of our boys have been able to recover from what seemed like the most dire of circumstances. But unfortunately this is a roller-coaster that also can fly off the tracks and lead to the most terrible of outcomes. Those in our community suffer the lowest of lows — the loss of a son or brother or grandchild — and other extreme hardships (e.g. extended stays in the hospital, heart transplants, mechanical "Berlin hearts") that most people don't even know or think about. There are always surprises in life — some good, some bad — but the range of possibilities expands in unthinkable ways when dealt the Barth card.

One of the many functions of BSF is to help those affected with Barth syndrome learn what to expect, to prepare families for what "surprises" might be in their future. If you know what to expect and can prepare for it, and even take action to avoid a serious problem, it can have life-saving ramifications for our boys and men. I can say from personal experience that learning from other parents in our organization, and knowing what to look out for, has been extraordinarily helpful to my family in keeping my son as healthy as possible.

In my role as Chairman of the BSF Board, I work with my fellow Board members to try to keep the organization on a steady course and, hopefully, avoid negative surprises. We have strategic planning sessions to establish our organizational objectives and plans to achieve those objectives. We attempt to identify and then mitigate potential risks for the organization. We look to other organizations for their lessons learned, and maybe mistakes they have made, to try to anticipate any surprises and make the best possible choices for BSF.

As much as I generally prefer to avoid surprises, the Foundation is on the precipice of new things, bringing both uncertainty but also great excitement about what the future holds. We are now at a real inflection point where our investment in research has brought us to consideration of therapeutic options for the first time ever. It's an important milestone and one worth pausing and considering, and thanking all those whose contributions of money and in many cases our boys own skin, blood and tissue samples have enabled. But as we approach the new frontier of clinical trials, we enter a domain that is as unpredictable as Barth syndrome itself. It will be with some uncertainty and anxiety that we will engage in these clinical trials, and those of us with affected boys or men will need to say "yes" despite the uncertainty, because there's no one else to do it for us. Once again, we will all be asked to step up ... to answer the call once more. Knowing this organization, knowing all of you, I have confidence that each family will make the decision to help the doctors and scientists find the best, safest treatments for our kids. In this I know I won't be surprised!

More than ever before, I believe we are on the brink of something special. I'm not quite sure what therapeutic option is going to be the one that makes a huge difference to our affected boys and men. We may be surprised as to what works or what doesn't. We have come a long way, and with your participation we will get to the promised land of a breakthrough therapy. I look forward to your continued support as we strive for the exciting surprise that awaits us.

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Volunteer Participation in Rare Disease Research is Critical for All of Us

(Cont'd from page 1)

"...research into rare diseases has often produced critical discoveries that have revolutionized our understanding of human physiology and led to new treatments of both rare and common diseases. ... It is only through the willingness and generosity of patients to participate in these clinical trials that science will advance."

There are many moments in life when you are proud of whom your children have become. I cannot recall a prouder moment than when he volunteered to participate in a study to hopefully help a group of people suffering from a "rare" disease.

As an endocrinologist who practices clinical nutrition, I often deal with patients with obesity and type 2 diabetes, two diseases that are at epidemic proportions. Barth syndrome is not a common disease, but just because a disease is rare does not mean that it is unimportant. Indeed, research into rare diseases has often produced critical discoveries that have revolutionized our understanding of human physiology and led to new treatments of both rare and common diseases.

A classic example of the interaction between rare and common conditions is in the unexpected relationship between two gene mutations that can cause extreme obesity on one hand or a congenital absence of fat with severe diabetes, congenital lipodystrophy, on the other. In 1994, researchers found that a group of congenitally obese, "ob/ob" mice had a mutation in the ob gene. This gene was found to be most active in fat tissue and encoded a protein that was later identified and is now known as leptin. Leptin was thought to inform the brain of energy status and fat stores but is now known to play surprisingly important roles in immune function, glucose and fat metabolism, fertility, and pubertal development. The discovery of the defect in leptin production in ob mice led investigators to examine leptin production in patients with congenital lipodystrophy. They found that not only did these patients not produce any leptin but that when they were administered synthetic leptin their diabetes went into near complete remission. Similarly, a group of people with a rare disease that causes early onset, severe obesity were found to be resistant to the effects of leptin, leading them to consistently overeat. The two rare diseases, a genetic form of obesity and a lack of fat, shared similarities and led to the discovery of the role of leptin. Prior to this, adipose tissue was considered to function merely as a place to store fat. However, it soon became clear that fat tissue produces a variety of hormones that are important in human health. Research into these two "rare" conditions caused a paradigm shift in the scientific viewpoint of the role of fat and led to a new view of the myriad roles that fat plays in our health.



Nick participates in exercise study

Barth syndrome is similar in that it is a rare disease but appears to be caused by a mutation that affects crucial steps in the function of mitochondria; components of the cell that burn fat and produce most of our energy — something critically important to human physiology in general. Important early studies by Dr. Todd Cade have now described the cardiac and metabolic changes seen in Barth syndrome — specifically a loss of the ability to burn fat and a shift to using glucose drives — probably worsens muscle protein breakdown and reduces skeletal and cardiac muscle mass. These basic, but critical studies, were the first to actually describe the metabolic effects of Barth syndrome and led to a better understanding of

HOW the changes in fat, protein and glucose metabolism could cause some of the complications seen in people with Barth syndrome and implicated crucial components of the mitochondria that may drive the disease process.

The findings from these studies have now allowed this group to collaborate with an adjacent laboratory that had no prior experience with Barth syndrome but that has expertise in mitochondrial metabolism. Even now, through a grant from the Barth Syndrome Foundation, this group is beginning to probe the actual changes that occur in the mitochondria leading to heart failure, loss of muscle mass, and easy fatigability. In yet another parallel between a "rare" disease and a common disease, heart failure due to advanced age shares similarities with Barth syndrome. As heart failure progresses, the heart and skeletal muscles have a reduced ability to burn fat, a greater need for glucose causing early fatigue and loss of muscle mass.

These diseases share another important link; the need for human studies and volunteers. It is only through the willingness and generosity of patients to participate in these clinical trials that science will advance. While the studies can be arduous and time-consuming they have already led to important changes in our understanding of the causes of the symptoms in Barth syndrome and hopefully will lead to new therapies not only for Barth syndrome but for other more common conditions. I have been truly fortunate to not only study the patients in our research unit but also get to know them and their families and become part of the community. We thank you for your willingness to get involved and volunteer to help find a cure. (Photos courtesy of Dr. Reeds ²⁰¹⁵)

Researchers and Physicians, Come to Clearwater!

(Cont'd from page 1)



Dr. Matt Toth at BSF's 2014 Conference
(Photo courtesy of BSF 2014)

knowing more about the *tafazzin* gene (whose dysfunction causes Barth syndrome), the unique lipid it affects (cardiolipin), and the pathological mechanism of this disease with its varied and seemingly unrelated symptoms (cardiomyopathy, neutropenia, growth delay, easy fatigue). At the upcoming 2016 Barth Syndrome International Scientific, Medical & Family Conference, we still cannot say that we fully understand Barth syndrome, but we absolutely can say that we have made great progress scientifically, and that now, hopefully, we are poised for therapeutic advances.

The science behind Barth syndrome is fascinating. How a single small gene can be responsible for the varied and life-threatening symptoms provides medical science with a model to understand more about the basic biochemistry or life, specifically mitochondrial function. At the 2016 Conference, researchers from all over the world will tell us what they know. Much of what we have learned about this disease can be traced to the influence of BSF through its Research Grant Program and through these biennial conferences which feature several clinical studies to take advantage of this unique gathering. Now, with the growth of medical clinics specifically for Barth

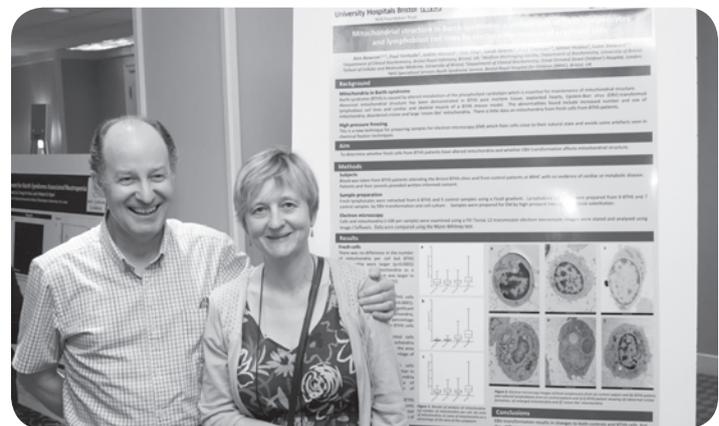
syndrome in both the UK and the US, knowledge about Barth syndrome and the best treatments for it take on a sustainable and professional healthcare status that benefits the worldwide BSF community, including researchers.

Before I came to BSF, most of my drug discovery experience was in traditional pharmaceutical research which involved screening for active chemical compounds in biochemical assays that we thought could alter crucial and controllable points in the pathophysiology of a disease — usually a common disease affecting millions of people. At the Scientific and Medical sessions of the 2016 Conference, we will hear about a traditional pharmaceutical treatment idea as well as the exciting opportunities in gene therapy, enzyme replacement therapy, lipid replacement therapy, exercise therapy, nutritional therapy, and other ideas. These last ten years have been productive, and there is a lot more to discuss and to prepare our community for as we enter the realm of clinical trials.

These biennial BSF conferences have become so popular that we eventually had to limit the number of speakers at the Scientific and Medical sessions due to time constraints. By providing poster presenter travel stipends, BSF has been able to greatly expand the Poster Session. The Poster Session is where researchers (especially young researchers), physicians, Barth syndrome individuals and their families can meet and where each group can inspire the other. These BSF conferences build on that unique interaction so that real and rapid progress can occur. Every two years, scientific and clinical researchers tell us about their published and unpublished work. Physicians and healthcare workers who are treating individuals with this rare disease get to meet with each other, encouraged by BSF through its health professional scholarship program. In addition, this largest gathering of Barth syndrome individuals under one roof allows IRB-approved studies to be conducted and therefore add to the knowledge base about the disease. It transforms the rarity of this disease into the seemingly commonplace, making it more human for each of us, especially for those who suffer from it. Many collaborations are begun or are expanded at the conference. It is a model of how disease research can be done and maybe should be done. If you have attended the conference before, you already know this, but if you have not, you should come and experience it.



Yoonjeong Lim at BSF's 2014 Poster Session
(Photo courtesy of BSF 2014)



Prof. Colin Steward & Ann Bowron at BSF's 2014 Poster Session
(Photo courtesy of BSF 2014)

Barth Conference Keynote Speaker



Ron Bartek and Lindsay Groff
at World Orphan Drug Congress
(Photo courtesy of BSF 2016)

Mr. Ronald J. Bartek is Friedreich's Ataxia Research Alliance's (FARA) Co-founder and President. He also is Chairman of the Board of the National Organization for Rare Disorders (NORD); a 4-year member, National Institutes of Health (NIH) National Advisory Neurological Disorders and Stroke (NINDS) Council; and former partner and president of a business and technology development, consulting and government affairs firm.

Friedreich's Ataxia is a genetic disorder with some important clinical similarities to Barth syndrome. FARA is an organization that communicates (in collaboration with other organizations) with government agencies at both state and federal levels in pursuit of policies and decisions intended to advance therapeutic development for the disorder. Ron is well-respected and trusted by peer patient advocacy leaders, policy makers in Washington, DC, leaders of numerous divisions of the NIH and by people he serves through FARA. He is known by all as a man who is thoughtful, who cares deeply and who delivers. Maybe most importantly, he is a father who got involved to help his son. Though Keith unfortunately died in January 2010, Ron has continued to work for treatments for all those who have Friedreich's Ataxia or who suffer from a rare disease. Ron has been a friend and advisor to the Barth Syndrome Foundation (BSF) from the very beginning. He and FARA have traveled down a very similar road to BSF to

translate scientific advances into actual therapies. In his speech, he will offer us all some first-hand insights into this process and will emphasize patients' critical roles in this process.

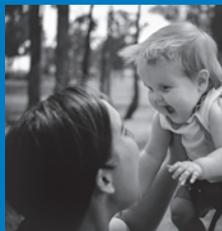
Ron graduated from the United States Military Academy at West Point and then spent four years as an Army officer, serving as a company commander in Korea and an Infantry and Military Intelligence officer in Vietnam. He also earned a Master's Degree in Russian Area Studies from Georgetown University. His professional experience includes twenty years of federal executive branch and legislative branch service in various defense, foreign policy and intelligence positions.

From all of his work in the rare disease field, Ron has a great deal of experience and expertise in topics very relevant to all of us involved in BSF. He and his organization are considered by many to be pioneers in the rare disease arena, and they have won many awards. Among them are recognition from the Pharmaceutical Research and Manufacturers of America and a Lifetime Achievement Award from Rare Disease Legislative Advocates. Most recently, on May 17, 2016, the National Organization for Rare Disorders (NORD)[®] announced that Friedreich's Ataxia Research Alliance, a passionate patient organization working to make a difference for everyone affected by rare diseases, will be one of the esteemed Rare Impact Award recipients this year.

We are excited for our attendees to hear Ron's inspiring message of hope and triumph, as told by someone who has many years of first-hand experience.

Newly Revised Healthcare Professional Brochure

Do You Know About
Barth Syndrome?



What's Inside

- Description of Barth syndrome
- Important clinical problems
- How to diagnose
- Inheritance
- Selected highlights of clinical knowledge
- Resources for physicians and families

Our lives depend on it!

The Barth Syndrome Foundation (BSF) has revised an informative brochure about Barth syndrome, written by people at BSF and reviewed by the clinical members of our international Scientific and Medical Advisory Board (SMAB). It provides a good overview of this complicated syndrome from a number of angles. One important section lists a summary of some of the unusual clinical complexities that can arise (sometimes very quickly) as a result of the multi-system nature of this disorder.

These two pages can be particularly useful for treating physicians and for patients during first visits with new doctors. Additionally, it can be vitally important in an Emergency Room when a physician unfamiliar with Barth syndrome is suddenly asked to care for a patient with the disorder.

There is also a section highlighting published journal articles that detail much of the current clinical knowledge about the syndrome. Physicians who would like to know more about a specific aspect of the disorder, or scientists trying to understand how far research has taken us to date, will find this of great interest.

If anyone — family, physician, scientist or donor — would like some additional hard copies, please contact Lynda Sedefian at lynda.sedefian@barthsyndrome.org. The brochure is also available on the BSF website at www.barthsyndrome.org. We have received very favorable comments about the usefulness of this brochure, and we think all will agree that this is an extremely valuable resource.

Barth Syndrome Conference – A United Front and Strong Voice for the Barth Community

By Emily, Mother of Affected Individual, Alabama

"Because of the information and connections made during our time at the conference, we have been able to make a solid game plan for Luke."

My name is Emily. My husband, Jason, and I live in Florence, Alabama with our three year old son, Luke. Luke was diagnosed with dilated cardiomyopathy in March of 2013. By May, his diagnosis of Barth syndrome was confirmed. In July of 2013, Luke's health continued to decline, and he was listed for a heart transplant.

After spending almost six months on a Berlin Heart, Luke received his life-saving gift of a heart transplant on January 28, 2014. By the spring of 2014, our lives had finally started normalizing, and we made the decision to attend the Barth syndrome conference that summer. Thankfully, with the help of Shelley Bowen (Director, BSF Family Services & Awareness), we registered just a few weeks before the conference and were able to attend. We braved the 12 hour car drive down to Clearwater, and we were excited to see what that week of the conference would hold for us.

Attending the conference for the first time was slightly intimidating and scary. I was nervous where we could fit in. With Luke being a heart transplant Barth boy, I knew that put us into an even smaller group within the Barth community, but we were greeted with open arms. It was wonderful to finally get to meet a group of people who could completely understand what we had gone through. It gave me so much hope to hear all the different stories from all the Barth boys, but it also became overwhelming at times to hear about all of the sadness this small group has had to endure. Because of that sadness though, a passion just as strong, grows within this group. The hope that one day there will be a cure for Barth syndrome is such a driving factor for all involved.

Throughout that week, we met a wide variety of specialists and gathered more information than I knew what to do with. At that time, Luke had just turned two years old. We were still recovering from his extensive hospital stay and heart transplant, but I gained immense knowledge and information that would be extremely helpful in the months to come.

Luke has thankfully had a smooth recovery from his heart transplant. His strength and stamina have increased over time, but we are now seeing where his recovery from his transplant has stopped and where we truly stand with his Barth syndrome. Because of the information and connections made during our time at the conference, we have been able to make a solid game plan for Luke. We have since visited the Barth Syndrome Interdisciplinary Clinic at Kennedy Krieger Institute in Baltimore, MD, and we are so pleased with our overall management of Luke's Barth syndrome. Sadly, we will not be able to attend this year's conference, but we look forward to the future when we can hopefully go again. It's a wonderful thing that our families and health care providers can come together at the Barth Syndrome conference as a united front and be a strong voice for the Barth community.



Luke (age 2) & Emily
(Photo courtesy of Amanda Clark 2014)



Luke (age 3)
(Photos courtesy of Emily 2016)



The 2015 BSF Research Grant Awardees

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

The 2015 Barth Syndrome Foundation (BSF) Research Grant Program attracted a large number of high-quality applications, half of whom were from outside the United States. With the completion of the 2015 research grant cycle, 14 annual award cycles have committed a total of US \$4.0 million to this important work through 95 research grants to 56 principal investigators around the world. BSF, with the advice of its international Scientific and Medical Advisory Board and some outside reviewers and with the support from its international affiliates, awarded funds to 6 research projects totaling US \$350,000 for the 2015 grant cycle. BSF is proud to be able to support the following grant recipients. As with all BSF grant cycles, the 2015 applications were submitted in October, 2015 and those that were accepted are actually awarded in the following year(s). Thus, the 2015 cycle awards are included in 2016 yearly expenses.

Abbreviations:

BTHS—Barth syndrome

KD—knockdown mouse model of Barth syndrome

TAZ—the *tafazzin* gene, which when mutated, leads to Barth syndrome

CL—cardiolipin; the lipid associated with Barth syndrome



Eric Ortlund, PhD, Associate Professor, Emory University, Atlanta, GA

Structural studies of human tafazzin

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

Obtaining the crystal structure of human tafazzin — the gene responsible for BTHS. The actual structure of an enzyme in three dimensions is valuable

in understanding its function; when it is mutated like in BTHS, the modified structure can help explain how it is dysfunctional. Dr. Ortlund will use crystallography methods to determine the structure of the human *tafazzin* protein, often abbreviated as TAZ. By mapping the known *tafazzin* mutations from BTHS individuals upon its 3-dimensional structure, researchers may be able to explain where the enzymatically vital part(s) of TAZ are located and hence may explain the pathomechanism of the disease. More importantly, the 3-dimensional structure of a disease target protein like TAZ may allow researchers to model chemical compounds that could interact with the mutated TAZ protein and correct its dysfunction. The crystal structure of proteins like TAZ expands the number of investigators that can work on BTHS research because it provides another tool to understand how the symptoms of BTHS are manifested, which, in turn, may lead to new therapies. Developing tools for different types of research and researchers has led to important advances in BTHS research, just like the KD mouse model has provided and continues to provide.



George Schweitzer, PhD, Postdoctoral Research Associate, Washington University School of Medicine, St. Louis, MO

Metabolic adaption in Barth syndrome

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

**Funding provided by the Will McCurdy Fund for the Advancement of Therapies for Barth Syndrome*

Using non-radioactive tracer studies in the KD mouse model and in BTHS individuals to identify the exact metabolic abnormalities of BTHS. BTHS individuals have an abnormal and inefficient way of extracting energy from the foods they eat. This dysfunction probably leads to the extreme fatigue they experience with even moderate physical exertion. One explanation for this fatigue is that BTHS individuals fail to utilize all the potential energy in the glucose or glucose equivalents (carbohydrates) of their food. BTHS individuals do not appear to “oxidize” or “burn” glucose like unaffected individuals (in fact, they do not “burn” glucose normally because they apparently do not use the oxygen in their blood properly). This abnormality was discovered by Dr. Todd Cade at the BSF conference clinics and was subsequently published. Dr. Schweitzer, along with Dr. Cade and other distinguished colleagues, will investigate the exact biochemical processes that are involved using isotopomer tracer analysis. (Isotopomers are non-radioactive compounds that can be used to follow the exact atoms that are changed in the biochemical pathways of metabolism.) By injecting trace amounts of isotopomer versions of foods like glucose, alanine, and arginine, Dr. Schweitzer will be able to see how much energy is derived from each of these foods in the KD mouse and in BTHS individuals. Understanding how BTHS individuals metabolize different food sources will provide explanations of how the symptoms of BTHS arise, will confirm existing nutritional therapies and may also suggest new therapies. Dr. Schweitzer will utilize and build upon the existing resources already assembled by Dr. Cade’s laboratory to perform this work.

The 2015 BSF Research Grant Awardees

(Cont'd from page 8)



Yuguang (Roger) Shi, PhD, Professor, University of Texas Health Sciences Center in San Antonio, San Antonio, TX
Molecular mechanisms underlying a causative role ALCAT1 in the pathogenesis of Barth syndrome
Award: US \$100,000 over 3-year period

How does inhibiting the ALCAT1 gene reverse the phenotype of the KD mouse and can pharmacological inhibition of ALCAT1 be therapeutic for BTHS? The ALCAT1 enzyme is similar to the TAZ enzyme because it modifies cardiolipin (CL). Dr. Shi discovered that deleting the gene for ALCAT1 reversed the cardiomyopathy of the KD mouse model of BTHS. Interestingly, this ALCAT1-KO + TAZ-KD double-mutant mouse still retains the cardiolipin abnormalities associated with BTHS. Dr. Shi will investigate how the rescue of the cardiomyopathy in the double-mutant mouse model occurs by examining the changes in its phospholipids. Building on this extraordinary result, Dr. Shi will also screen over 100,000 chemical compounds to find inhibitors of the ALCAT1 enzyme which may be therapeutic for BTHS by having the same reversing effect on cardiomyopathy in BTHS as the ALCAT-1 knockout does in the double-mutant mouse. Dr. Shi's work has revealed an alternate explanation for understanding the pathology of BTHS, and he has developed a strategy to perhaps find a novel therapy.



Miriam Greenberg, PhD, Professor, Wayne State University, Detroit, MI
Cardiolipin is required for mitochondrial protein processing
Award: US \$50,000 over 1-year period

Defects in protein processing as proteins are transported into the mitochondria may be responsible for the pathology of BTHS. Dr. Greenberg has uncovered evidence that when cardiolipin (CL) is altered in a cell, mitochondrial proteins do not get to where they are supposed to be — a subcellular series of events called mitochondrial protein processing. One of these altered mitochondrial proteins is called *frataxin* protein, and when the *frataxin* gene is mutated, it causes Friedreich's Ataxia — a metabolic disease with similarities to BTHS. Dr. Greenberg has altered a well-known mouse skeletal muscle cell line (C2C12) so that the *tafazzin* gene is deleted. This new cell line will allow direct comparisons of her extensive yeast data to human data. By using yeast cells with CL mutations and this new *tafazzin*-deficient mammalian skeletal muscle cell line, Dr. Greenberg will explore how mitochondrial protein processing is disrupted in BTHS by focusing on mitochondrial proteins like the *frataxin* protein. Understanding how this defective mitochondrial protein processing leads to the symptoms endured by BTHS individuals could provide a different way to understand and to treat this disease.



Robin Duncan, PhD, Assistant Professor, University of Waterloo, Waterloo, Ontario, Canada
A new enzyme and pathway in cardiolipin synthesis
Award: US \$50,000 over 2-year period
**Partial funding provided by the Barth Syndrome Foundation of Canada*

Investigating a newly discovered cardiolipin modification pathway to see how it can impact BTHS biochemistry. Dr. Duncan discovered that a certain gene, HRASLS1, which was first associated with inhibiting cell proliferation of cancer genes, also has the capacity to increase cardiolipin (CL) when overexpressed. Dr. Duncan will investigate how this enzyme functions and what properties/pathways this new enzyme alters in the context of BTHS. There are three enzymes known to directly modify CL: *tafazzin* (whose dysfunction causes BTHS), *ALCAT1* (see Yuguang (Roger) Shi abstract), and *MLCAT1*. This new, fourth, CL-modifying enzyme adds to our understanding of CL metabolism, and this may allow researchers to consider alternate ways to therapeutically impact BTHS.



Doron Rapaport, PhD, Professor, University of Tuebingen, Tuebingen, Bavaria, Germany
Use of high throughput screens in yeast to investigate the pathomechanism of Barth syndrome
Award: US \$50,000 over 1-year period
**Partial funding provided by the Association Barth France*

Using tafazzin-deleted yeast cells to visualize intracellular protein changes and to screen for overexpressed genes that may suggest therapeutic pathways to exploit. Dr. Rapaport will use yeast as a tool to discover what biochemical pathways and protein changes are dysfunctional in BTHS. Dr. Rapaport will use a sophisticated high-throughput assay to visualize at the subcellular level what proteins are altered or mis-directed in *tafazzin* deficient yeast cells by looking at each protein one at a time. In addition, Dr. Rapaport will use molecular genetic techniques to find suppressor genes that when overexpressed, reverse the phenotype of *tafazzin* deficient yeast cells. This information will provide a better understanding of which metabolic pathways are altered in BTHS and may suggest ways that the disorder can be treated by manipulating these pathways.

Editor's Note: A complete list of all grant awardees over the years can be found on BSF's website at www.barthsyndrome.org.)

Barth Clinic at Kennedy Krieger Institute

By Hilary Vernon, MD, PhD, Assistant Professor, Institute of Genetic Medicine, Johns Hopkins University and the Kennedy Krieger Institute, Baltimore, MD



Dr. Hilary Vernon

The Barth Syndrome Interdisciplinary Clinic at the Kennedy Krieger Institute (KKI) in Baltimore, Maryland has been dedicated to serving the needs of the Barth syndrome community since 2012. The interdisciplinary clinic, which includes cardiology, hematology, genetics, nutrition, physical therapy, diagnostic imaging, and laboratory evaluations is designed to offer “total care” within a one-day visit. The clinic is held four times a year, and has served more than 25 patients with Barth syndrome ranging in age from infancy to adulthood (each of whom has had one or more visits).

Local, national and international patients and families have all been seen at the clinic, and patient care is tailored to suit individual needs. The clinic’s doctors and therapists have the ability to provide regularly scheduled care to local patients, or to provide long-distance consultation care to patients who live too far away to be seen regularly.

In addition to medical care, the clinic is also dedicated to contributing to research in Barth syndrome. Since 2015, the clinic has included an ongoing natural history study, with the goal of understanding how Barth syndrome affects individuals as they get older and to recognize unmet clinical challenges.

The Barth Syndrome Interdisciplinary Clinic also offers a great opportunity for families and medical personnel to meet and socialize. The evening before the clinic, a dinner organized by one of our local BSF families is attended by both the families and medical personnel. Additionally, on the day of the clinic, there is a designated lunch hour for the families and medical personnel to have some “break” time together.

On a more personal note, the clinic has been a real joy for me to direct. I have met so many wonderful clinicians and families and am continually amazed by all of the things I am learning. I am looking forward to sharing these points at this summer’s conference.



John (age 34) and Dr. Reid Thompson



Brian, Jack (age 5) and Rachel
(Photos courtesy of BSF 2016)



Families and clinicians gather for lunch
at Barth Syndrome Clinic at KKI

Keeping Our Sons as Happy and Healthy as They Can Be

By Melanie, Mother of Affected Individual, Australia



Families gather at Aussie Outreach

T.S. Eliot once famously said, “**Only those who will risk going too far can possibly find out how far one can go.**” In March 2016, a small turnout of Barth families came together for the first Australian outreach gathering in Melbourne. Each family arrived with mixed questions and uncertainty about the realities we could achieve as a collective group regarding Barth syndrome in Australia. Where do we start? Who do we need? What do we want to achieve? How can we achieve it?

Amongst the blur of all these questions, there was one that instantly united us: How can I keep my son as happy and healthy as he can be? How far can we go; how far will we go; to ensure we get this answer right.

This article is in no way trying to convince you that we came up with all the answers! To be completely honest, the weekend raised more questions than were answered. The main objective was certainly obtained, and that was to simply start talking to one another — to share stories, compare stories, laugh at stories, and cry over some too.

Our first afternoon together was spent beside the pool, with families from Boronia, Victoria; Warrandyte, Victoria; and Schofields (New South Wales) sitting under the mild sunshine, and of course enjoying a few beers as we got to know one another a little better. Later that evening, a family from Eatons Hill, Queensland joined us, and for the first time we had our “Aussie Barth Boys’ all together — Cam (age 17), Aiden (age 4), and Eli (age 2).

It is important to note that there were other Australian families who would have loved to have joined us for this first gathering, but for a range of reasons this wasn’t possible for everyone who was interested. It is certainly our aim to move the location of future Australian gatherings to other cities. This will ensure all families have an equal chance to participate and enjoy the benefits a weekend such as this provides.

To start the following day, we had an online meeting with Matt Toth (BSF Science Director) and Shelley Bowen (BSF Director of Family Services & Awareness). The purpose of this meeting was for Matt to step through where the Barth Syndrome Foundation is in its current research towards advanced therapies and treatments and gave us ample opportunity to ask questions about the what/why/how. Matt was very generous with his time, and even contributed ideas about ways we could reach out to Australian doctors and hopefully get someone from Australia involved with the Foundation.

The end of this session left us feeling enthused and encouraged and certainly with a sense of “we can do this” mentality. Our first goal is to use the connections we have within Australian medicine and attempt to find an Aussie doctor who is willing to get involved and is passionate about supporting us to “keep our sons as happy and healthy as they can be.”

After the online meeting, we headed out to lunch at the top of Mount Dandenong, which normally offers beautiful views over Melbourne. Unfortunately, Melbourne’s weather lived up to its reputation, and it was a very overcast and cloudy day, so no views could be seen! Nonetheless, after lunch we challenged ourselves through the garden maze before celebrating with some ice cream to end an enjoyable afternoon. Later that evening, many more laughs were shared, and lifelong friendships cemented towards that common goal; “how can we keep our sons as happy and healthy as they can be?”

On our final morning together, we were fortunate to have breakfast with Pam Holmes, who is Michaela Damin’s aunt. (Michaela is Chair of the Barth Syndrome Trust {UK & Europe}.) Pam does a lot of fundraising work for Barth Syndrome Trust, and was very eager to meet the Barth boys and contribute to our plans moving forward as a team.

So what do those plans look like? We have our common goal; we have a supportive base; we have the support of the Barth syndrome worldwide family. Our big objective for the gathering was simple: strengthen friendships. That objective was certainly achieved and will continue to be achieved as we move towards an exciting phase of Barth syndrome awareness within Australia. We look forward to the follow-up gathering in 2017!



(L-R) Cam (age 17), Aiden (age 4), & Sebastian (Photos courtesy of Melanie 2016)

From Small Seeds ... A Fundraising Story

By Lindsay Groff, Executive Director, Barth Syndrome Foundation

When I was invited to join a local networking group called, *Women of Wit and Wisdom*, I was unsure of how I would fit in. Most of the women own and operate businesses in the Greater Philadelphia Area. Representing a nonprofit, I felt uncertain of how we could network to help each other.

At one of the meetings, we were asked about our ideal customer. Since that isn't applicable to my work at the Barth Syndrome Foundation (BSF), I told the group about our great programs and, instead of a customer profile, I talked about our donors. What I didn't realize at the moment was that I had planted a seed.

A few days later, I received an email from a fellow "Woman of Wit and Wisdom" who encouraged me to contact her colleague, Angela, at Investors Bank. She explained that this bank gives grants to nonprofits, and I should find out if they would be willing to donate to BSF.

To be honest, I did not believe that BSF would qualify for a grant, since we do not have an office in the bank's community. However, I contacted Angela. After telling her about BSF and our work,



(L-R) Lindsay is presented with check from Investors Bank (Photo courtesy of BSF 2016)

she said that we certainly did qualify and that I should apply. It turns out that, as the Executive Director for BSF, my home office met their guidelines for giving back locally!

Of course, the deadline for applications happened to be THAT DAY! I dropped everything and spent the next couple hours writing the grant application, filling out paperwork, and driving to make the last FedEx shipment. A few months later, I received the news that we had received

a \$1,000 grant from Investors Bank and a matching grant from Roma Bank. A simple conversation resulted in \$2,000 to BSF.

Angela invited me to the bank to present the checks. Here is a photo of that moment. I encourage all of you to consider stepping out of your comfort zone and asking people in your community to donate to BSF. You might be surprised to find an opportunity like this! I learned a few things from this experience that I'd like to share with all of you: (1) It never hurts to ask; (2) People genuinely want to help; (3) Businesses want to give; and (4) Don't rely on assumptions.



#GivingTuesday 2015

We did it. In ONE DAY, BSF raised \$35,217. We did it together, and we did it with LOVE. Thank you all for pushing hard to make a difference in the lives of these boys and men affected by this horrible disorder. That's thanks to you, our beautiful BSF family. Imagine the possibilities in what we'll tackle next!



Henry (age 4)

Happy Heart Week 2016

Happy Heart Week is a campaign centered around Henry's birthday, May 2nd. Birthdays are always such a sweet celebration. But, even more now, birthdays remind Henry's family, in a very literal way, of our gratefulness for each breath every day. For Henry's 4th birthday, we celebrated his beautiful life; his challenges with Barth syndrome; the other boys and men suffering from this disease; and the great work the Foundation is currently doing. This virtual celebration took place the whole week of Henry's birthday, Sunday May 1st through Saturday May 7th. Through an email and social media campaign, we surpassed our goal and raised money to help fight Barth syndrome.

(Photo courtesy of Henry's family 2016)



Ben (age 11)

A Great Expression of Love and Support

Some of Ben's friends came up with the idea of raising funds for BSF — calling it "Ben's Barth Bucks". Ben's teacher organized it, and the classes are competing to see who can raise the most money and win a "pickle party". Every day for four weeks, interesting facts about Barth syndrome are being announced to the students during their morning announcements, so the awareness is a huge part of it. The kids are bringing in donations every day, and have been creative in their giving. A couple of them had brought money for souvenirs on a field trip, but then decided to donate it instead! And, since Ben is a huge baseball fan but can't play, others are doing "Bases for Ben" and donating a dollar for each base they get in baseball and softball. Still others are involved in making signs and posters, and making the announcements with the Barth facts. A good group of kids, and a GREAT teacher to encourage it! (Photo courtesy of Amanda Clark 2014)

(Cont'd on page 13)

Power of Kindness

(Donor categories are based upon the past 18 months of cumulative giving from 10/1/2014–4/19/2016)

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Kirk, Becky
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(Cont'd on page b)

Power of Kindness

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 Segal, Mark & Heather
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 Sherer, Tony
 Shtull, Ora
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 Sims, Mac & Joanne
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 Singer, Scott & Mary Jo Romano
 Sleeper, Emmy Lou
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 Vail, Clayton
 Van Valen, Colleen
 Van Valen, Ed & Jeanne
 Van Valen, Kera
 Varner, Judy
 Viebranz, Elaine
 Vourlekis, Melina
 Wallerstein, Diana
 Waterman, Chris & Marcy Wilkov
 Waters, Marty & Helen
 Watt, Richard & Gill
 Waugh, Emily
 Wayne, Sharon
 Weissman, Alan
 Werner-O'Brien, Claire
 Weston, Garrett
 White, Bill & Marian
 Wilkins, Sue & Dr. Mike
 Wilkins Burnham, Joanne
 Williams, Kathleen & Suzi Anderson
 Wofford, Daniel
 Wolff, Patricia
 Xu, Dr. Yang
 Yi Lee, Ching
 Young, Lisa
 Zehner, Jon & Carlyn
 Zelin, Joy
 Zierk, Holly
 Zierk, Tom & Gail

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 American Express Philanthropy
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 Blumenthal, Matthew
 Blumenthal, Sen. Richard & Cynthia
 Branagh, Bill & Nancy
 Branagh, John & Megan
 Buddemeyer, Randy
 Cantor Fitzgerald Relief Fund
 Lummis, William & Dossy
 Malkin, Scott & Laura
 McCurdy, Steve & Kate
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 The Lebensfeld Foundation
 Wood, Loren
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 American Express Foundation
 American Express PAC Match
 Avanti Polar Lipids, Inc.
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 Barth Syndrome Foundation of
 Canada

Bauer, Jon
 Bellig, John
 Bellig, Mike & Sara
 Bercovich, Bruce
 Branagh, Thomas & Diane
 Brody, D.W. & Tracy
 Colgate
 Crozier, Nancy
 Drake, Bryan
 Evoy, Larry & Sally Houstoun
 Fogarty, Ghotay
 Susan & David Fowler Charitable
 Gift Foundation
 Gonzalez, Dr. Iris
 Hagar, Rodney & Elissa
 Hales, Bryan
 Hechinger, Nancy
 Heineman, Ben & Cris
 Tutwiler, Temple & Lucy
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 Malkin, Louisa
 John Marshall Family Foundation
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 Mann, Allen & Rosa
 McCormack, Susan & Ken Marra
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(\$50 - \$999)

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 Ameriprise Employee Gift
 Matching Program
 Ameriprise Financial Employee
 Giving Campaign
 Amlicke, Tom & Susan
 Ammann, Sandra
 Anderle, Eric & Rachel
 Anderson, Jeff
 Anderson, Tommy & Allana
 Arnosti, Carol
 Arovas, Gregory
 Asaff, Ernie & Colette
 Austin, Susan
 Axelrod, Dr. David & Jessica
 Azar-Kuhn, Laura
 Baer, Susie & Ben Chan
 Baffa, Matt
 Baffa, Ted & Rosemary
 Baker, Keith & Laurel
 Baker, Timothy
 Ballard, Stacey & Frank
 Barab, Stuart & Anne
 Baron, John & Christine
 Barth, Dr. Peter
 Bater, Jennifer
 Bates, David
 Bauer, Steve
 Beckett, Robert
 Becton, Dickinson and Company
 Behnke, Eugene & Margaret
 Bell, Jenny
 Bennett, Judith
 Berens, Wayne & Harriet
 Berland, Jonathan & Leslie
 Berman, Dr. Harris
 Bernal, Francisco
 Berry, Rick & Carla
 Berry, Shane
 Beshar, Peter & Sarah
 Bettenhausen, Bob & Joanne
 Betz, Megan
 Bialo, Ken & Kate
 Bierbaum, Frederick & Dionna
 Biscoe, Laurie
 Bittel, Rhonda
 Blauvelt, Frank & Ilah
 Blumenthal, Dr. David & Dr. Ellen
 Bodary, Michael & Lisa
 Bogert, Amy & Robert Baldwin
 Bogert, Carroll
 Bohnert, Kaye
 Bosak, John & Anita
 Bosak, Mary
 Botto, Patrice
 Bougger, Annie
 Bowen, Shelley & Michael
 Bowerman, Margaret
 Bowman, Ron & Nancy
 Branagh, Diane
 Branagh, Geoffrey
 Branagh, Katie
 Branagh, Kimberly
 Branagh, Jack & Lyn
 Brandt, Douglas
 Bravelets
 Brennan Jones, Mary Augusta
 Brewster, Brinks & Laura
 Britt, Peggy
 Britton, Lori & Jimmy
 Brody, David & Jeanne
 Brooks, Cameron & Tawnya
 Bruno, Ellen
 Bryant, Bobby & Pamela
 Buddemeyer, David
 Buddemeyer, Donna
 Buddemeyer, Leslie
 Burdgick, Lindsey
 Burdine Auto Body, Inc.
 Burgan, Francine
 Burgher, Bedford
 Burke, Agnes
 Burmeister, Marita & Chuck
 Burns, James
 Burr, Richard
 Butera, Jacque & Donald Kyle
 Cade, William
 Calvert, Elaine
 Campbell, Tonja
 Canova Jr., Frank
 Capasso, Ruth Ann
 Cappello, Frances
 Carpenter, Susan
 Carroll, Terrence & Ellen
 Carvalho, Marlene
 Casale, Betty
 Chandler, Peggy
 Chandler, Robert & Darla
 Chapin, Steve & Debbie

Cheatham, Joyce
 Cherniak, Miriam
 Citta, Laura
 Clarady, Carrie
 Clark, Rebecca
 Cleek, Robin
 Cohune, Vicky
 Coleman, Charlie
 Collazzi, V.C.
 Condon, Jane
 Condon, Mark
 Congdon, Margot
 Connolly, Bradford & Dorothy
 Consos, Joey
 Con-Way Inc.
 Conway, April
 Coombe, Amy & Burt
 Cotters, George & Shelley
 Couch, Arthur
 Couch, Rick
 Courtney, Shirley
 Cowan, Esther
 Cox, Dr. Gerald
 Cracchiolo, James & Marilyn
 Crawford, Michael & Eleanor
 Cripps, Tia
 Crowley, Chuck
 Crowley, Peter & Gretchen
 Crozier, Kathryn
 Cunniffe, Sister Margaret
 Dannels, Dick & Terry
 Bodary, Michael & Lisa
 Darnell, Laurin
 Dastous, Brock
 Jack D. Davis, Inc.
 DeAngelo, Suzanne
 DeFazio, Diane
 Demman, John & Florence
 Derusha, Laura
 Derusha, Rachel & Eric Anderle
 Derusha-Mackey, Nicole
 Derusha-Mackey, Sarah
 Despins, Suzanne
 Deutscher, William & Lisa
 Develle, BJ
 Devin's League of Super Heroes
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 Doebler, Richard & Mary
 Dollar, Michelle
 Dollard, John & Janet
 Donnalley, Jen
 Drake, Frank & Teresa
 Drake, Marcia
 Draper, Steve
 Dugan, Adam
 Dugan, Patricia
 Dunn, Robert & Helen
 Duplantis, Lizzie & Seth
 Durando, Courtney
 Eaton, Thomas & Gretchen
 Edgemon, Matthew
 Egueur, Madeleine
 Fidazzo, Linda
 Elliston, Ann
 Engberg, Marc
 Engberg, Renee
 Enterline, Carol
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 Epstein, Paul & Leah
 Erdhaus, John & Sharron
 Eslava, Cynthia
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 Fenwick-Smith, Felice
 Ferdina, Elizabeth

(Cont'd on page c)

Power of Kindness

(Donor categories are based upon the past 18 months of cumulative giving from 10/1/2014–4/19/2016)

(Cont'd from page b)

Fifer, Scott
 Firestone, Jim & Ann
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 Fisher, Anthony
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 Flatt, Charles
 Fletcher, Keith
 Flint Area School Employees
 Florez, Michelle
 Floyd, Edmond & Julie
 Floyd, Jane
 Flynn, Clare
 Ford, Earl & Merry
 Forsyth, John & Linnet Tse
 Forte, John & Gertrude
 Frasier, James
 Friar, Ellen
 Frondorf, Ed
 Fuller, Barbara
 Gaehle, Benjamin
 Galbraith, Lois
 Galli, Erin
 Ganote, Felicia
 Garcia, Linda
 Garcia, Rudy & Lin
 Garry, Bobby & Leigh
 Garver, James
 Gausepohl, John
 Generali, Joan
 Gerhart, Lucinda
 Gewitz, Dr. Michael & Judy
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 Gittelman, Ann
 Godbey, Steven & Kathleen
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 Goolsby, Elaine & Richard
 Goulet, Mike & Vickie
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 Gray, Dusty & Melany
 Green, Chris
 Green, Frances
 Green, Mitch & Susan Yamaguchi
 Greenberg, Dr. Miriam & Dr. Shifra Epstein
 Greene, Rev. Dorothy
 Gripp, Christine
 Groff, Lindsay
 Grose, Madison & Nora
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 Gungor, Umut
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 Hallam, Ken & Theresa
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 Harman, James & Merlene
 Harris, Mark
 Haviland, Jordan
 Hazen, Ned & Liz
 Heilmann, Kathy
 Hensley, Charlotte
 Hensley, Christopher & Deborah
 Heslin, Frank & Linda
 Hiden, Bob & Ann
 Higgins, James & Julia
 Higgins, John & Liz
 Higgins, Kelsey
 Highland, Jason
 Hill, Dr. Carter & Dr. Winnie Mann
 Hille, Mary
 Hinkle, Jennifer
 Hinkle, Russell
 Ho, Jasper & Winifred
 Hobbins, Antonia
 Holbrook, Frank

Holly, Barry
 Holly, Craig
 Holly, Kristi
 Holly, Peggy
 Holly, Ron
 Horkey, Shelly
 Horluc, Dr. Todd & Jennifer
 Horton, Nancy
 Howell, David & Patricia
 Hubbell, Frederick & Linda
 Hubby, Pam
 Huppert, Betty
 Ingersoll, Chris
 Ingersoll, Jared & Tina
 Irving, John & Emily
 Ison, Ann
 Jaimes, Eneida
 Jarecha, Paul
 Jefferies, Dr. John & Shari
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 Johnson, Jennifer
 Kalapasev, Ned & Brie
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 Karp, Joseph
 Kaufman, Jeff & Wendy
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 Kelly, Seth
 Kennedy, Holly
 Kensek, Paul & Florence
 Kim, Jenni
 Kimball, Philip & Eleanor
 Kimmey, Joseph
 King, Charlie
 King, Ernest
 King, John & Allis
 Kintzer, Donald & Karen
 Kinzell, Holly
 Kirk, Becky
 Kirkbride, Brandon & Kayla
 Klampe, Patrick & Pamela
 Klimchak, Michael
 Klockner, Daniel & Nancy
 Kop, Garrett & Dianne
 Kozora, Jamie
 Kroger, Fred & Robbin
 Kroger, Suzanne
 Kugelmann, Mike & Catherine
 Kurtenbach, Douglas
 Kutzer, Donna
 Kwittken, Aaron
 Lajoie, Judy
 Lampman, Ralph & Karen
 Langwith, Elizabeth
 Larson, Bruce
 Lascurettes, Nancy
 Lascurettes-Mangiapanne, Denise & Vince
 Lauer, Valerie
 Lawrence, Peter
 Lawson, Richard
 Le Jamtel, Jerome
 Leadbeater, Michael & Mary
 Lee, Richard
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 Levinsky, Michael & Rosario
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 Lind, Michael
 Lindquist, Douglas & Patti
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 Logan, Christie
 Long, David
 Lord, Sarah
 Lummis, Ghent & Ginger
 Lynch, Lilian
 Lyon, Janie
 Maca, Sarah
 MacKay, Douglas

Magnussen, Lindsay
 Mahoney, Sara
 Malkin, Elizabeth
 Mancino, Angelo & Rosemary
 Mann, David & Sheila
 Manwell, Emily
 Marcoux, Vicki
 Marra, Kim
 Marsico, Michael
 Martin, John
 Martin, Melanie
 Massie, Kaylan
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 McClellan, MGC, Rebecca
 McCormack, Helen
 McCormack, Dr. Timothy & Marcia
 McCracken, Paul
 McFee, Wendy & Jmel Wilson
 McGreevy, Annie
 McHugh, Laura
 McNair, Gail
 McNally, Mark
 McNeil, Thomas & Michelle
 McPhee, Kristina
 McTear, John & Donna
 Meighan, Paula
 Mellen, Ann
 Menkhaus, David
 Merrill, Allan
 Michaud, Steven & Yoko
 Michener, Sandt & Kathryn
 Mihalich, Jim & Jane
 Miller, Denise
 Miller, Jane & Clyde Rayner
 Millman, Paul & Susie
 Minnick, Gates & Daisy
 Minor, Chris & Marv Hubbell
 Mitchell, Joseph & Sabrina
 Mofidi, Shideh
 Moncure, John
 Montanaro, Louis & Theresa
 Moreland, Dr. Brian & Rachel
 Morey, Dale & Bonnie
 Morgan, Anne
 Morgan, Derek & Laura
 Morgenstern, Marc & Louise
 Morock, Kim
 Morris, Erin
 Morsberger, Raechel
 Moskowitz, Rebecca
 Mueller, Carl & Suzanne
 Murphy, Donald & Jane
 Myers, Douglas
 Myers, Edward & Anita
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 Neece, Michael
 Neece, Sue
 Nelson, Joanna
 Nelson, Scott & Teri
 Nelson, Wayne & Joyce
 Nemanic, Erik & Kathy
 Nevel, Bonnie
 Newacheck, Rick & Pamela
 Newton, Theodore & Audrey
 Nicoll, Rev. Tom & Mary
 Norman, Jon
 Nurkowski, John
 Oates, Jason
 O'Connell, Ryan & Janet
 O'Connor, Mary
 Odell, Heather
 Olsen, Steven & Nancy
 Olson, Amy
 Olson, Dick & Sharon
 Olson, Donald & Eunice
 Orso, John & Theresa
 O'Shaughnessy, Jim & Missy
 Osterhaus, Greta
 O'Sullivan, John
 Pacak, Dr. Christina
 Palmer, Kirk & Lori
 Paolizzi, Vincent & Celeste

Parlato, Carolyn
 Peck, Susan
 Pella Rolscreen Foundation
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 Percival, Cathy & Steve
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 Phillips, Mark & Katherine
 Phillips, Nancy & Tom
 Pickett, Lisa
 Pierson, Allene
 Pierson, Frank & Nancy
 Plumez, Jean & Dr. Jackie
 Powell, Brian
 Pratt, Berit
 Prendergast, Sheila
 Taegtmeier, Dr. Heinrich
 Puhr, Edward & Kathryn
 Purdy, Dr. Bill & Molly
 Radelat, Troy
 Rader, Stephanie
 Renbarger, Sam
 Rhodes, Alberta
 Rigsby, Robert & Deborah
 Rimler, Frank
 Rizedesign
 Roberts, John & Betty
 Robinson, Joe & Connie Sue
 Robinson, Kyle
 Robinson McFarland, Jane
 Rodamer, Bob
 Rodan, Amnon & Dr. Katie
 Rodbell, Michael & Margaret
 Rodbell, Mitchell & Liz
 Rogers, Catherine
 Rogers, Jim & Michelle
 Moreland, Dr. Brian & Rachel
 Rorschach, Bob & Susan
 Rosenfeld, Lisa
 Rosenshine, Jon
 Roth, David
 Rothman, Robert & Amy
 Rothschild, Adam & Kathy
 Rozbruch, Robert & Yonina
 Russell, Peter & Helen Im
 Rutledge, Elizabeth
 Ryan, James & Miriam
 Ryan, Patricia
 Ryan, Dr. Robert
 Sabin, Todd
 Sams, Louise
 Sanborn, Noel & Virginia Lee
 Sander, Mike
 Saporito, Gennaro & Alisa
 Schible, Laurie
 Schlame, Dr. Michael & Laili Moshirzadeh Moayedi
 Schlapak, Gregor & Sonja
 Schmitt, Robert
 Schoenherr, Dan & Heather
 Schreiber, Mary Ann & Ed Mooney
 Schumacher, Dean & Donna
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 Segal, Mark & Anne
 Sellet, Rebecca & David Campbell
 Senkel, Bill
 Shepherd, Greg
 Sherer, Peter & Marilu
 Sheridan, Melanie
 Shields, Jim
 Shiring, Kathleen
 Shirley, Janice
 Simon, Thomas & Nancy
 Singer, Scott & Mary Jo Romano
 Skinner, Adrienne

Slawson, Kirsten
 Smith, Dr. Herbert
 Smith, Toniaanne
 Snedeker, Rob & Fran
 Solomon, Lauren
 Sonderegger, Ted & Mary Ann
 Sorrento, Nora
 Spalla, Katie
 Sparks, Kara
 Spear, Laurinda
 Spencer, Louise
 Spotts, Dr. Jules
 St. Joseph Notre Dame High School
 Staudt, Dennis & Patty
 Stenson, Marie
 Stenson, Michael
 Stephens, John Mark
 Stevens, Sandra
 Stevenson, Robert & Sharon
 Stone, Jeff & Vicki
 Stone, Kimberly
 Stoner, David & Teresa
 Strohl, Linda
 Stromberg, Bob & Judy
 Stromberg, Robert & Lucielle
 Strong, Gary
 Stuhlfreyer, Tom & Kim
 Stuhlfreyer, William & June
 Sullivan, Barb
 Sullivan, Tim
 Sutherland, Laura
 Suzuki, Matthew
 Swabe, Carolyn
 Sweetman, Tina
 Tadeucci, Katie
 Taylor, Paul
 Teboul, Susie & Daniel
 Temby, Michelle
 Teodecki, Amanda
 Thach, Christy
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 Thompson, Nathan
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 Trolard, Ron
 Tunguz, Stefan & Julie Kinch
 Turull, Albina
 Unger, Karl
 United Way California Capital Region
 UnitedHealth Group
 Vail, Clayton
 Van Dehey, Walter & Susan
 VanBuren Brown, Missy
 VanRensselaer, Cheryl
 Vernon, Aaron
 Viebranz, Elaine
 Viegas, Martin
 Vogt, Jerre
 Wadsworth, Kimu
 Wagner, Doreen
 Walden, Leslie
 Waller, Scott
 Walsh, Liz
 Walters, Nancy
 Walton, Darren
 Warne, Linda
 Waterman, Chris & Marcy Wilkov
 Weigel, Larry & Patricia
 Weissman, Alan
 Wells, Anna
 Welsh, Bill & Denise
 Weltlich, Bob & Dodie

(Cont'd on page d)

Power of Kindness

(Donor categories are based upon the past 18 months of cumulative giving from 10/1/2014–4/19/2016)

(Cont'd from page c)

Weltlich, Mark
Weltlich, Steve & Sharon
Wendling Orthodontics
Werth, James & Kimberly
Wertlieb, Linda
West, Shilo
Wharton, Philip & Philippa

Wiederspan, Mark & Jess
Wilkins, John
Wilkins, Sue & Dr. Mike
Wilkins Burnham, Joanne
Willcoxon, Anne
Williams, Jennifer

Williams, Kathleen & Suzi
Anderson
Wilson, James
Wind, Kathleen
Winfrey, Ofelia
Withem, Angela

Wood, Kelli
Woodcox, Larry & Carolyn
Woodward, Steven
Wright, Marilyn
Wynn, Ken
Yon, Kirk & Judy

Young, Penny
Zangara, James & Marie
Zeitner, Eric
Zierk, Holly
Zoland, Yale & Wendy
Zorthian, Greg & Robin

“The Barth Syndrome Foundation is an incredible organization whose professional members — scientists, physicians, psychologists, nutritionists, physical therapists and others — freely interact with patients and their families, all focused on finding effective treatments for this rare disease. Though small, the Foundation manages to fund research (several projects every year!), host a biennial scientific and family conference that draws an international audience, and provide a vast support network for patients and families. The connectedness felt among all in the Foundation — from the sickest infants to the young men who have survived all challenges, to their parents and families, to those working towards a cure — is unlike anything I've witnessed in any other professional organization.” ~ Colin Steward, PhD, FRCP, FRCPCH, Bristol Royal Hospital for Children, United Kingdom

Power of Kindness — Time and Advice

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Adams, Dr. Sean
Alder, Dr. Nathan N.
Angelini, Dr. Roberto
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Association Barth Italy
Atwell, Joshua & Morgan
Axelrod, Dr. David
Azar-Kuhn, Laura
Bakovic, Dr. Marica
Baffa, Kevin
Barth Syndrome Foundation of
Canada
Barth Syndrome Trust
Baile, Dr. Matthew G.
Bankaitis, Dr. Vytyis
Barrientos, Dr. Antoni
Barth, Dr. Peter G.
Bastin, Dr. Jean
Blumenthal, Matthew
Bobbitt, Whitney
Bolyard, RN, BS, Audrey Anna
Boozer, Allanna
Bowen, Michael
Bowen, Shelley
Bowron, PhD, FRCPATH, Ann
Branagh, Megan & John
Braverman, Dr. Nancy
Broadridge
Brown, Dr. David A.
Bruno, Ellen
Bryant, Dr. Randall
Buddemeyer, Randy
Burrige, Dr. Paul
Byrne, Dr. Barry J.
Cade, Dr. W. Todd
Camadro, Dr. Jean-Michel
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Conway, Dr. Laura J.
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Cutting, Dr. Gary
Damin, Michaela
Dancis, Dr. Andrew
Das, Dr. Anibh
Daum, Dr. Guenther
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Derusha-Mackey, Nicole
Develle, B.J.
di Rago, Dr. Jean-Paul
Dinca, Ana
Donadieu, Dr. Jean
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Drake, Bryan
Dudek, Dr. Jan
Elstein, Dr. Deborah
Endo, Dr. Toshiya
Exil, Dr. Vernat J.
Feinberg, Dr. Mark W.
Ferreira, Dr. Carlos
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Fuchs, Dr. Beate
Funai, Dr. Katsu
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Garratt, DClinPsych, Vanessa
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Hatch, Dr. Grant M.
Hazen, Dr. Stanley
Henry, Dr. Susan
Higgins, Jack
Hijkata, Dr. Atushi
Hirano, Dr. Michio
Hollander, Dr. John
Hone, Susan
Hope, Chris
Hoppel, Dr. Charles

Horowitz, Dr. Jeffrey
Houten, Dr. Sander
Huffman, Dr. Kim M.
Huizing, Dr. Marjan
Jackson, Dr. Ian
Jain, Dr. Mukesh K.
James, Dr. Cynthia
Jarosz, Daniel
Jefferies, Dr. John Lynn
Jofre, Jaime
Kadenbach, Prof. Bernhard
Kagan, Dr. Valerian E.
Kalapasev, Brie
Kalapasev, Ned
Kelleher, Dr. Joanne
Kelley, Dr. Richard I.
Khuchua, Dr. Zaza
Kim, Dr. Junhwan
Kirkland and Ellis
Kugelmann, Lee
Kugelmann, Steve
Le, Dr. Catherine
Lenaers, Dr. Guy
Lesnefsky, Dr. Edward J.
Lever, Beverly
Li, Yiran
Lloret, Dr. Placido
Lorberboum-Galski, Prof. Haya
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Lu, Ya-Wen
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Mancino, Michael
Mann, Sheila
Mannes, Florence
Mannes, Philippe
Manton, Annick
Marshall, Dr. Bess
Mashek, Dr. Douglas
Mavroidis, Dr. Manolis
Mayr, Dr. Johannes
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Merill, Dr. Alfred

Metallo, Dr. Christian
Millay, Dr. Douglas
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Moore, Nigel
Morales, MS, LGC, Ana
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Porter, Dr. George
Potter, Dr. Paul
Pu, Dr. William T.
Raja, Vaishnavi
Ren, Dr. Mindong
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Ritter, RN, Catharine L.
Rodbell, Gary
Rodbell, Julia
Rossano, Dr. Joseph W.
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Witt, Dr. Stephan
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Woodward, Stacey
Ye, Cunqi
Yin, Dr. Huiyong
Yu, ScD, Xin
Zaragoza, Dr. Michael V.
Zeller, Anne
Zeller, Bill
Zhang, Dr. Ji
Zhang, Jun

In Remembrance and Celebration

(Cont'd from page 12)

Our extended community lost three incredible women recently. Each of them left a lasting legacy in many ways, including their requests for gifts to the Barth Syndrome Foundation (BSF), in lieu of flowers. Thanks to their final acts of generosity, their spirit lives on through the work of the Foundation. May they rest in peace.

Henry lost both of his great-grandmothers. His mom, Megan, told us this: "Both women were great supporters of us and would have done anything they possibly could to help relieve our family of any stress caring for and worrying about Henry. They loved him dearly!"

Juanita May King, Great-Grandmother of Henry



(L-R) Grandma Juanita, Grandpa Charlie, Megan, Henry, John & Grady

After a life of love, wonder, spirit, and adventure, Juanita May King (88), Grandma, passed away peacefully on July 27, 2015. Her vibrant life was testament to her belief that "dreams really do come true." A strong advocate for public education, Juanita taught math and computers at the local junior high school. Her example inspired her children and grandchildren to pursue

their own educations to the highest level. Juanita's life revolved around family. Everyone in the family will miss her fried chicken, Thanksgiving dinners, homemade jellies, and gifts of pecans shelled with care. She was always so excited to hear of what was going on in each of her grandkids and great-grandkids lives, and always in complete support. You knew she believed in you, without fail. Our family will forever cherish the time we got to spend with her in our home in California. We will miss her dearly!

Natalie Branagh, Great-Grandmother of Henry



(L-R) Grandma Nat & Henry

Natalie "Nat" Branagh was Henry's Great-Grandmother, and "Great" she was. Nat had such a tender heart, and cared so deeply for others. One of the ways Nat cared for others was praying for them. She prayed daily for her family, friends, and even strangers she had just met. Nat was one of Henry's most faithful

"prayer warriors" ever since Henry was admitted to hospital at three months old. Nat had friends all over the world, and she quickly put them to work praying for Henry. It was these prayers that made us feel so loved and comforted even in the darkest of times. Our family was so greatly loved by Nat, and we miss her from the very bottom of our hearts. *(Photos courtesy of Henry's family)*

In Remembrance of Carol Cook, Mother of Lattigo

Carol will always be remembered by friends and family for her thoughtful, kind-hearted spirit. She always thought of others. When someone with Barth syndrome was ill, Carol would call BSF to check-in on the child. And when a family grieved the loss of a child with Barth syndrome, Carol grieved. Carol knew the heartache of having a child with Barth syndrome and she knew the lasting grief of losing a child from Barth syndrome.

To know Carol, was to love her. She found joy in making friends with everyone, through everything she did. From art to animal care, she shared her knowledge and passion through caring for all of us. Carol's desire for donations to be made to BSF to benefit those with Barth syndrome is an act of love reflective of the legacy she leaves behind. We will all miss Carol and we thank you for your contribution to BSF in her memory.

(Photo courtesy of Carol's family 2015)

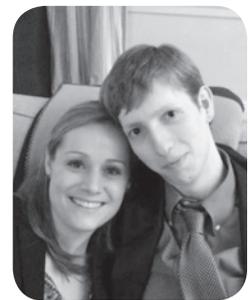


(L-R) Dakota, Carol & Bailee

In Celebration of the Marriage of Caroline Pierson and Garrett Ames-Ledbetter

BSF is particularly near to Caroline Pierson's heart. Caroline's aunt and uncle, Kate and Steve McCurdy, participated in the founding of BSF in support of their son, Caroline's cousin and dear friend, Will. Two of twelve first cousins, they grew up spending summers together in Lakeside, Michigan. Their relationship inspired Caroline to become a pediatric nurse practitioner and, although Will was three years older than Caroline, she earned the nickname "big sister" by nagging him to finish his G-tube feeds and take his medications on time. Will even taught high school-aged Caroline how to give a shot and was willingly her first victim! Thank you, Caroline and Garrett, for honoring Will by choosing BSF to receive gifts in celebration of your wedding.

(Photo courtesy of Caroline 2015)



(L-R) Caroline & Will

Awareness of Barth Syndrome Continues to Grow

There has been a significant increase in Barth syndrome (BTHS) related peer-reviewed journal articles published. To date, a total of **128** 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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Rare Disease Day 2016

2016 State Leadership Conference winners Jessyca Flores and Tayla Wibecam competed at Rare Disease Day 2016 with a display and talks about Barth syndrome. They represented Hodgson Vocational Technical High School, where they are in the Dental Assistant Program.

Photo (L-R): Jessyca Flores, Kevin (age 27), and Tayla Wibecam
(Photo courtesy of Dr. Iris Gonzalez)

Page 14 Barth Syndrome Journal ~ Volume 16, Issue 1



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Barth Syndrome Trust — Update from Chair

By Michaela Damin, Chair, Barth Syndrome Trust (United Kingdom & Europe)

The year so far has been a busy one for the Barth Syndrome Trust (BST), and we'd like to tell you a bit more about what we've been doing in our efforts to help the Barth families in our region.

Barth Syndrome NHS Service

In January, a clinic for the older boys and men was held in Bristol. They were put through their paces by Dani Goodman (Occupational Therapist) and Lucy Buckley (Physiotherapist) who had great suggestions and adaptive equipment to help with specific issues. For example, Nick finds that he can concentrate better if he has something to occupy his hands. No problem! Within minutes Dani had a selection of goodies in front of him to try, and he chose a Tangle Toy which has eighteen curved jointed sections to twist and manipulate into almost limitless contortions.



Tangle Toy

Then, it was on to a full cardiac check-up where we met Dr. Alison Hayes who is heading up the cardiology side.

There was also an opportunity to discuss any dietary and growth concerns with Nicol Clayton (Dietician), talk through any specific areas of concern confidentially with Ness Garratt (Psychologist), and chat with Prof. Colin Steward about general issues and the management of infections. This annual check-up is vital to make sure that detailed examinations are done and long-term data are gathered, and it is offered to each UK Barth family free of charge.

February saw me back at the Bristol Royal Hospital for Children, along with the team for the annual meeting with Dr. Edmund Jessop from NHS Highly Specialised Services. We rely on the provision of specialist funding from the NHS in order for Prof. Steward and his dedicated team to provide the wonderful array of services available to our Barth families, and we were delighted to hear that everyone is satisfied with the way that things are progressing. If you use the Barth service and have any suggestions of how it can be improved or if you have any feedback, please share this with us at any time.

March — We kicked off March with a Trustees' meeting during which we elected our new board member, Mrs. Cheryl Parish. I've known Cheryl for many years. She has wonderful problem solving skills. These are usually provided along with some amazing baking and a cup of tea. She is genuinely interested in all our families' welfare and is a great person to call on when you need some advice about how to plan for success.

From Cheryl....

"I have worked with young people all of my life, from running a preschool, to teaching primary children, to coaching adolescents and young adults. I am a mother to six children, three of whom have special educational needs requiring robust support with both their education and adult life. I have also experienced the sharp end of health issues as our eldest daughter spent her short life in a high dependency neonatal unit, and we had the subsequent challenge of bereavement following her death. I am an NLP (Neuro-Linguistic Programming) practitioner, and I am experienced in the practice of mindfulness; I use techniques from these as well as coaching skills to support my clients and their parents.

I believe education and life opportunities should be accessible to all, but I know that sometimes the rigidity of the system can make that harder than it needs to be. As a Parent Governor for my local secondary school, I find it very useful to be involved with the decision-making process in a large organisation. It offers me the opportunity to get a 360 degree perspective on the way educationalists strive to offer a broad and balanced curriculum. My background in education, my personal family experiences, my professional experience as well as my determination, make me a force to be reckoned with in my pursuit to help youngsters and their families create the happiest, most fulfilling life possible."

(Cont'd on page 17)



Barth Syndrome Trust — Update from Chair

(Cont'd from page 16)

Mother's Day

March 6th was Mother's Day here in the UK, and I was asked to contribute to the Jeans for Genes blog which you can read at <https://jeansforgenesukblog.org/>. This was a great opportunity to shout out to all the amazing "rare mums" I've met on this journey.

2nd UK Genetic Disorders Leadership Symposium

On 11th–12th March, I represented BST at the 2nd UK Genetic Disorders Leadership Symposium in London, presented in partnership with Global Genes (<http://www.geneticdisordersuk.org/>). This is a unique event, bringing together leaders from about 90 charities throughout the country. Talks ranged from gene editing techniques to making a film about your condition. Teenage years and transitions were also addressed in depth, as was creating and maintaining patient registries. Speaking of which... if you are a parent to someone with Barth syndrome or are an adult with Barth syndrome **have you made sure your data is in the Barth Syndrome Registry & Repository?** We need your participation to make sure that the data collected is accurate and complete (<https://barthsyndromeregistry.patientcrossroads.org/>).

What are We Working on Now?

At the moment, we are hard at work helping with the 2016 International Conference in Clearwater, USA. Together with Shelley Bowen, Nicol Clayton, Stacey Reynolds, Emily Burgess, Dani Goodman, Lucy Buckley, and Liz Perkins, we are scoping out the nutrition, occupational therapy, physiotherapy and transition to independence sections of the upcoming conference. Our aim is to find concrete ways in which to enhance the lives of all our families through practical advice for day-to-day life. This talented and dedicated team is a pleasure to work with, and we are grateful for their time and commitment to our families.

Focus on Our Families

This edition features highlights from three of our stars – Dillon (age 11), Alfie (age 15) and Alfie's brother, Jay (age 17). Dillon and Alfie both have Barth syndrome. Thank you mums, Julie and Allanna, who were brilliant interviewers.



Dillon (age 11)
(Photo courtesy of BST 2016)

Dillon

What are you doing at the moment? I'm at school full-time, and I'm not really enjoying it as I'd much rather be on the farm. My best subject is Maths. I am going to secondary school in September, and they are trying to get me an electric wheelchair to get around so watch out legs!

What are your favourite foods? Chocolate biscuits.

Hobbies? Farming and going to Young Farmers' Club. I love cows and sheep and now have 13 chickens (2 are cockerels), and I sell the eggs to family and friends. I love going stock judging at Young Farmers.

Holidays? Going to be a zoo keeper for the morning this weekend for my birthday present, so anything like that is good. Also going to the beach or where there is a pool.

Condition? I cope pretty well as I can't change it, but sometimes I get frustrated when I can't do what the other children can do and I can't keep up with them. I hate the needles too. The best thing about the condition is that I now have lifelong friends all over and get to see them at clinics.

I want to be a farmer when I grow up or anything that involves animals.

(Cont'd on page 18)



Barth Syndrome Trust — Update from Chair

(Cont'd from page 17)

Alfie

Alfie attends St. Matthew's Academy and loves it. He is currently studying for his National 5 Exams. He is on a reduced timetable to help him cope with his fatigue and uses his power wheelchair to get around the large school. He is studying to take five subject exams: Maths, English, Chemistry, Design & Manufacture and also Graphic Communication. His favourite subject is Maths.

Alfie would like to be a primary school teacher. He took part in a work experience placement recently doing this at his old primary school, and he loved it.

Favourite food? Noodles in any form!

Leisure activities? Alfie says he can get bothered that Barth syndrome limits what he wants to do sometimes, but these are some of the things that he loves doing... He attends a class in school called "Sports for All." This is an inclusive programme for pupils who have disabilities and also for pupils who are not affected by any disabilities (for example when the pupils are playing basketball then ALL pupils are in sports wheelchairs). This is a wonderful and well-attended programme organised by the senior pupils. Alfie also plays after-school badminton with his friends on a Friday. Even if he is too tired to play much this is very important to him to meet up socially with his friends.

He goes to St. John's Youth Group on Sunday evenings and again is not always able to fully participate in all activities but always enjoys the social gathering of catching up with his friends and he loves being part of the community.

He loves to build things and experiment and loves nothing more than pottering around with all his gadgets and tools. Mum says that they have lots of deliveries from eBay and Amazon with wires, lights, and solutions for all his experiments, and this is truly when Alfie is in his glory! He has a dresser full of all his bits and bobs and his own work bench and tools.

Alfie loves to go on caravan holidays, and his favourite memory is when the family toured around Scotland in a camper-van and he caught a rainbow trout. His dad cooked it on the shores of Loch Morlich in the Cairngorm Mountains. Closer to home, he loves going on rides on the back of his dad's motor bike.

Alfie is very close to all his cousins, his family, his friends and is very much a big part of his brother, Jay's group of friends...he is a very sociable boy.



(L-R) Jay & Alfie (age 15)

Jay

Jay also goes to St. Matthew's Academy and is currently studying Maths, English, Physics, Chemistry, and Design & Manufacture for his Higher Nationals exams.

Favourite subject? Maths.

Jay wants to stay in school for 6th year and pursue a career in medicine or chemical engineering.

Jay is attending a pre-entry programme at Glasgow University for a week this June for a taster in medicine.

He has a Saturday job at his local pharmacy, and he's enjoying it.

Jay loves football and plays for Tass Thistle Football Club.

Favourite food? Nandos...chicken is always a winner!

Hobbies? Playing guitar, listening to music, and hanging out with his brother, family, and friends.

Jay loves skiing holidays and has been to Austria twice and Italy once skiing. He says his best memory is his very first time out on the slopes.

Jay thinks Alfie is happy, fun, and positive. He is very proud of his brother and how he copes and deals with everything that goes on in his life.

Jay has written many times about Alfie, their relationship, and how much his brother inspires him. Alfie has also written about his relationship with Jay and how Jay inspires him and how he looks up to him. This makes me very proud that they can both still annoy one another as brothers do, but at the very forefront is their special bond of love and total dedication to each other.

One last big fact... Alfie and Jay share a HUGE love for all things Star Wars.



(L-R) Jay & Alfie
(Photos courtesy of BST)

Barth Syndrome Foundation of Canada President's Report

By Susan Hone, President, Barth Syndrome Foundation of Canada

Spring is quickly approaching, and I can't wait. As many of you know, my son, Jared, had major spine surgery this winter and is slowly recovering. Being cooped up in the hospital for most of the winter kept us warm, but now we can't wait to get outside and enjoy the nicer weather.

Thanks to ever improving technology, I was able to keep up with my duties as President of Barth Syndrome Foundation of Canada (BSFCa) while sitting in the hospital room. I can't imagine going back to the days of no phone or internet in the room. How barbaric that seems now!

The Board, Executive and volunteers of BSFCa have been busy with numerous projects over the fall and winter. Fundraising is always on the agenda. Lynn Elwood and family held a raffle of gorgeous handmade wooden items donated with love by her Dad, Les, and son, Adam. Tickets were sold internationally, and we had winners from one end of North America (British Columbia) to the other, (Florida). The draw took place at our annual BSFCa planning session in November. We embraced the Giving Tuesday trend and took it one step further, turning it into Giving December. We were very successful in raising a little over \$14,000.

With our fund raising efforts, we were able to contribute \$25,000 towards the annual grant process sponsored by Barth Syndrome Foundation. The Research Grant Program is a major event, and we are again pleased that we were able to be a part of the process and contribution. Also, we are excited to be donating \$1,000 to the The Biophysical Society of Canada's conference in June. This is the first medical conference in Canada, which we are aware of, where Barth syndrome is on the agenda. Dr. Richard Epand will be chairing a session related to Barth syndrome/cardiolipin and mitochondria. Among the speakers will be doctors Michael Schlame, Valerian Kagan, Grant Hatch, Miriam Greenberg, and Stephen Claypool.

This year is an International Barth Conference year, and I can't wait to see everyone! BSFCa Executives have been working throughout the fall and winter helping to make the conference the success it always is. Lois is an active participant on the Conference Steering Committee and is currently busy booking flights, accommodations, etc. for the physicians who will be presenting at the conference. Susan and Chris Hope have been busy assisting Shelley Bowen with planning the family portion of the conference. We are proud to say we will be sponsoring the photo booth, poster session, a breakfast at the conference, and three Canadian doctors to come to the conference.

On the home front, we are working on a family activity get together, hopefully in May. We have a volunteer scouring the internet looking for grants we can apply for to raise funds to help fund research, conferences, etc. One of our most important activities has been supporting two of our families who have individuals undergoing major health issues. Personally, the support, encouragement, and advice I have received over the last few months has been tremendous in keeping my spirits up and asking the doctors the right questions. I would be lost without the support of my Barth family.

If you would like to follow us more closely, please check out our website at www.barthsyndrome.ca which is regularly updated by Chris Hope. We now have a Facebook page as well!

BSFCa 2016 Planning Session

By Susan Hone, President, Barth Syndrome Foundation of Canada

The BSFCa planning weekend was held on November 20–22, 2015. The scenic and peaceful Lake Kashabog, Ontario was the perfect setting to hold the meeting (and it was free). Sitting in front of a roaring fire were Lynn Elwood, Cathy Ritter, Chris Hope, Susan Hone, Lois Galbraith, Carol Wilks, and our chefs/dishwashers, Les Morris (also Board advisor) and Adam.



[Back Row L-R] Adam (age 25) & Les
[Front Row L-R] Cathy, Lois, Lynn,
Susan, Chris & Carol

(Cont'd on page 20)

BSFCa 2016 Planning Session

(Cont'd from page 19)

Friday evening was spent reflecting on the past year and some updates by Cathy Ritter on the Barth Syndrome Foundation. Our focus continues to be on affected individuals and their families and what we can do to make living with Barth syndrome a little easier. We discussed many topics over the weekend, including how to prevent volunteer burnout, brainstorming new fundraising ideas, participating and funding of the upcoming 2016 International Scientific, Medical and Family Conference, preparing the 2016 budget, and increasing awareness of Barth syndrome in Canada. The weekend is never long enough but we managed to get through our lengthy agenda and still have some valuable time for socializing, taking a few pictures, and drawing the winners' names for a raffle.

Thank you once again to Les, Lois, Adam, and Carol for their hospitality and keeping us well nourished. They selflessly prepared, cooked, and cleaned all at no charge to keep our administrative costs at a minimum, so that every dollar we raise is directed to achieving our mission of "Enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome."



Multi-generational planning
(L-R) Cathy, Les, Adam & Lynn

Barth Syndrome Foundation of Canada (BSFCa) Volunteers: Mysteries in the Making

By Lois Galbraith, Volunteer, Barth Syndrome Foundation of Canada

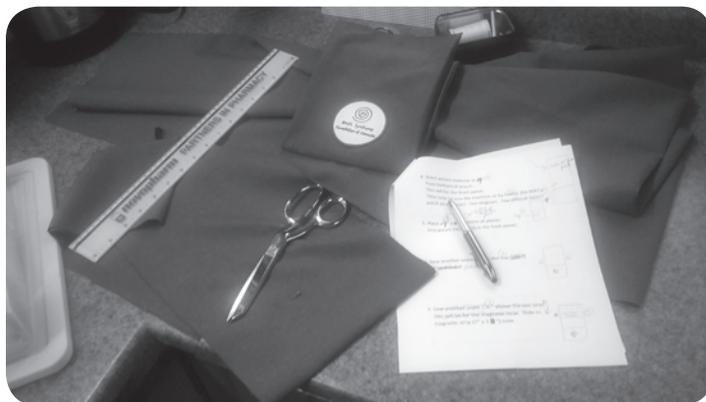
We love each and every opportunity we get to praise our fabulous volunteers. Led by the examples of our original volunteers Cathy, Lynn, Chris, Karen and Susan, our volunteers are always ready to pitch in, roll up their sleeves and get at it!

If in a monthly email update we mention a task or project needing doing, we have replies immediately offering help.

Besides the work of our six executive members, volunteers are presently engaged in the following: web-searching Canadian foundations to find grant-giving possibilities by Jeannine; editing of BSF print materials by Les; the knitting of Barth sweaters for our famous Barthur Bears by Celia, and the planning of a 'Dance' fundraiser once again by Bob and Susan.

There is sometimes intrigue and mystery involved as well. There is woodworking being worked on by Adam, Les and Lois and major sewing being undertaken by Carol, Jeannine, Cheryl, Audrey, Carol and Lois. The photos below may help you to solve the mystery — or not! The conference will bring clarity for families to these mysteries.

It is a known fact that 'volunteers live longer.' This is great news because we at the BSFCa cannot do without our vibrant volunteers. They are such an integral part of all we do and all that we are! We love all our volunteers dearly.



Mysteries in the making ~ clarity at the conference!
(Photos courtesy of BSFCa 2016)

Association Barth France

Patagonian Expedition Race—Racing for Barth Syndrome

By Anne-Marie Dunhill (Article from SleepMonsters.com - Adventure Sports Network)
Reprinted with permission from SleepMonsters.com



Olivier, Frédéric, Wilfried and Claire
before the start of the Patagonia Race
(Photo courtesy of Frédéric Decamps 2016)

The contrast between the setting in which this interview took place and the wilds of Patagonia couldn't have been more striking. SleepMonsters interviewed Olivier Renard, team captain of Patagonia4Barth, to find out more about the team as he was rushing through Paris on his way to a business meeting. The sounds of the French capital: traffic rushing, horns blaring, sirens wailing and raised French voices were clearly audible in the background yet Olivier Renard's voice was as calm and steady as if we'd been chatting in a café.

Presenting each member of his team, Olivier naturally began with himself. He was a professional tennis player for fifteen years who was ranked in the top 50 players in France in the 90's. He then transitioned to outdoor sports, completing several well-known ultra running races such as the Diagonal des Fous as well as an Ironman race, and starting his own successful business as an insurance broker.

When asked what had drawn him to adventure racing and the Patagonian Expedition Race in particular, Olivier said, "I like the challenge, doing different things. I want to know if I can get to the finish line, it really is the challenge that appeals to me. I decided to put together a team after seeing a television program here in France about the race. I'd never kayaked, so I learned. I'd never done mountain biking, so I learned."

Due to his solid connections and reputation in the business world, Olivier was able to secure sponsorship from four well-known corporations in France: SwissLife, DNCA Investments, Ciprés Assurances and Natixis Global Asset Management.

Olivier went on to explain that after conceiving of the project to compete in the "Last Wild Race", he approached a close friend, Eric Plantain, and they set about forming the team. Eric's only condition for being a part of the adventure was that the team race for a cause as he had one that was close to his heart: Barth syndrome.

According to the website, "Barth syndrome (BTHS) is an X-linked genetic condition." It is a rare genetic disease that primarily affects male children. When Eric Plantain had to withdraw from the team due to an injury, Olivier decided to keep the name and race for the cause.

After Eric withdrew, Frédéric Decamps stepped in. (Both Frédéric and Olivier are based in the Côte d'Azur region of France.) Frédéric is also a former professional tennis player who has competed in adventure races as well as having his own business as a fitness coach for high-level athletes. In 2014 he competed in the Embrun Ironman in France.

Continuing his explanation of how the team was formed, Olivier said that after recruiting Frederic, he contacted the French mountaineering federation in search of a female adventure racer. Claire Grosseuvre joined the team along with her husband, Wilfried. The Grosseuvre couple are the most adventure racing experienced members of the team and they discovered the magic of Patagonia Chile during a world tour in 2009 and vowed to return.

Parents of two young children, they are based in Annecy. Claire has extensive mountaineering and adventure racing experience which includes the race organized by Gérard Fusil, Raid Caneo Nature, which was the longest adventure race ever organized in France (The race was 11 days.)

As parents of two boys, Wilfried and Claire took turns travelling down to the Côte d'Azur to train with their new teammates whilst the other one watched the children at home. Wilfried wrote a message to SleepMonsters detailing their pre-thoughts. He said, "We know that PER will offer us all what we are expecting when you are looking for beautiful landscapes, wildlife fauna meetings, hard conditions to go ahead and an exceptional adventure with four human beings."

(Cont'd on page 22)

Patagonian Expedition Race—Racing for Barth Syndrome

(Cont'd from page 21)

Writing about racing for a cause, Wilfried added, “Those kids are fighting every day to stay alive so we can fight 10 days in the name of them. It is the opportunity to communicate about this association, and to help those kids dreaming with a team in a remote area, with lightness and craziness.”

Concluding the interview as the captain arrived on the doorstep of his business meeting Olivier said, “I like the fact that each one of us brings different skills to our team and we are quite complementary.”

When asked for a team photo to illustrate this article, Olivier laughed and said, “Actually, we don’t have one at the moment because this weekend is the first time that the four of us will be training all together as a team.”

Barth France Recent Events

2nd Charity Dinner

Our 2nd Charity Dinner was held on February 5, in Paris. Two hundred people attended in support of Barth France. Thanks to the generosity of all the guests, more than 30.000 Euros was raised. Everyone seemed to enjoy themselves thoroughly.



2nd Charity Dinner

5th Barth France Gospel Concert

On April 8, a Gospel Concert to benefit Association Barth France was held. The group Gospel Colors sang in the church Our Lady of the White Coats in support of the Association Barth France. Participation was free, and the evening was beautiful!



Gospel Concert

Barth France Paris Marathon Team

Every year, in honor of those fighting Barth syndrome daily, many runners proudly run different races among which is the famous Paris Marathon.



Barth France Paris Marathon Team
(Photos courtesy of Association Barth France 2015-2016)

Barth Italia: Work in Progress!

By Paola Cazzaniga, President, Association Barth Italia



(L-R) Paola & Pietro (age 14) at Rare Disease Day 2016

Following our first meeting Barth Italy has grown and is now involving many of our families. We've organized a range of activities aimed at widening awareness of Barth syndrome and at raising funds which will also enable at least some members of our scientific committee to attend the Barth Syndrome Conference in July in Clearwater, Florida.

In addition, a number of families are looking to participate independently, and we are all excited about the prospect of becoming part of the greater Barth community and of making our contribution to its work.

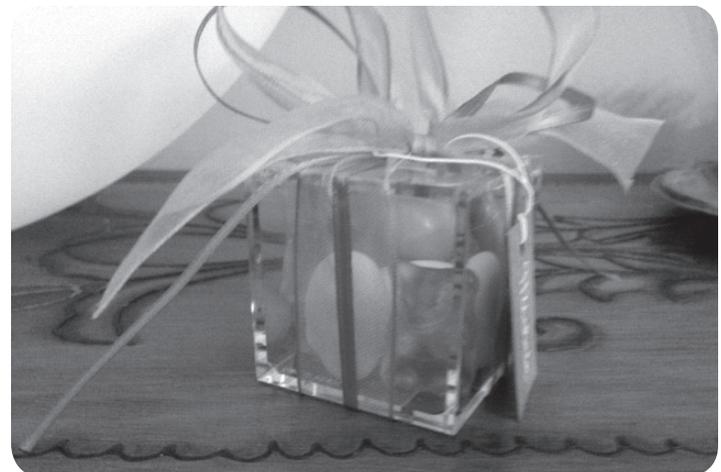
We would like to thank the parents of Ruben, who organized a concert of gospel music in support of the Foundation in Pisa on February 20, 2016. To mark the Rare Disease Day 2016, we organized a charity dinner in Monza, the historical city which is also the seat of Barth Italia.

Our many supportive friends filled the whole restaurant, and this encouragement helps us continue with our mission.

A big thank you goes to all the generous friends and volunteers who have helped start a permanent funding activity, "Barth Bomboniere." These pretty gifts are proving popular and are enabling us to foresee that we will soon be able to contribute to the research grants.



Pisa: Gospel for Barth Italia Dinner



"Barth Bomboniere" for permanent fundraising activity (Photos courtesy of Barth Italia 2016)



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Barth syndrome (BTHS; OMIM #302060)

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*)



Cole (age 6)
(Photo courtesy of BSF 2016)



Darryl (age 34)
(Photo courtesy of BSF 2014)