



**Barth Syndrome  
Foundation**

[www.barthsyndrome.org](http://www.barthsyndrome.org)

# Barth Syndrome Journal

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

## On the Edge of a New Frontier

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

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(L-R) Jack (age 22) and Dr. Matt Toth at the social event at BSF's 2014 Conference  
*(Photo courtesy of BSF<sup>2014</sup>)*

We have entered a new frontier in science and medicine by planning to test Barth syndrome (BTHS) specific therapies in humans. These specific therapies go beyond what has been done so far by taking advantage of the research knowledge gained over the years. We now have over seven different therapeutic ideas that may make a real difference in the lives of our boys and men. Many of these ideas are the direct result of the Barth Syndrome Foundation (BSF) Research Grant Program. Some will fall off the list over time and others likely will be added, but this is a very promising start.

The 2014 BSF Research Grant Program attracted the largest number and probably the most competitive set of applications in its history. By competitive, I mean that the high quality of the applications made it difficult to decide which ones would eventually receive funding — a good indication of the value of the program and an exciting challenge

for the judging committees. With the completion of the 2014 research grant cycle, 13 annual award cycles have committed a total of US \$3.6 million to this important effort through 89 research grants to 52 principal investigators around the world. BSF, with the advice of its international Scientific and Medical Advisory Board, and with support from international affiliates, awarded 10 research projects totalling US \$560,851 for the 2014 grant cycle. BSF is proud to be able to support the following grant recipients. (A complete list of all grant awardees over the years can be found on BSF's website at [www.barthsyndrome.org](http://www.barthsyndrome.org).)

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## Layman's Perspective on Therapy Development

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

As the Barth syndrome community — scientists, doctors, patients, families, and donors alike — enters a new and exciting therapy development phase of conquering Barth syndrome (BTHS), I have been asked to briefly give my layman's perspective on this new thrust — "why now?" and "how are we approaching it?"

When we created BSF in 2000, much less was known about BTHS than is known today. At that time, we were very fortunate that Dr. Peter Barth had described some of the clinical aspects of the disorder (1983) and that Dr. Daniela Toniolo and others had discovered the gene responsible for the disorder (1996). Dr. Richard Kelley also had realized that certain ranges of a "biomarker" called 3-MGA were associated with BTHS but that did not offer a definitive diagnosis (1991). And, just before we met as a group for the first time in Baltimore, Dr. Peter Vreken and others discovered that a phospholipid

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# Standing on the Shoulders of Giants

By Lindsay Groff, Executive Director, Barth Syndrome Foundation



Lindsay Groff  
(Photo courtesy of Lindsay<sup>2015</sup>)

**"If I have seen further than others, it is by standing upon the shoulders of giants." ~ Sir Isaac Newton**

Eliminating the suffering caused by Barth syndrome is a big job. None of us is strong enough, smart enough, or capable enough to do it alone. The founders of this organization knew that fact right from the beginning when they established the Foundation. Collaborating with others is deeply woven into the fabric of this organization. One of the ways we do this is by working with many other groups to understand their best practices, as well as learn from their mistakes. Standing on their shoulders allows us to see further.

In February, I attended the National Health Council's (NHC) Voluntary Health Leadership Conference with Board member, Kevin Woodward. BSF is one of the smallest members of the NHC, measured by number affected, budget size, and staff size. Meeting the standards of the NHC is no minor feat. Attendance at this meeting means that we have access to the other (larger) members as well as guidance on nonprofit governance. While there, Kevin and I learned much that will help BSF continue to move swiftly toward our mission. We heard about proposed government policies that could alter the landscape for groups like ours. We listened to pharmaceutical companies discuss patient-focused drug development, an increasingly relevant topic for us. Perhaps the most useful part of the meeting was the chance to brainstorm ideas with other nonprofit groups. No matter the size, each group was willing to help others. Our membership and participation in the NHC is one way in which we keep the collaborative spirit alive.

In April, I attended the World Orphan Drug Congress in Washington, DC. This was BSF's first time at this meeting. I was excited because it is the first time we now have our own "orphan drug" (bezafibrate) to consider! Although I was a first-time attendee, I was greeted by several people who knew BSF. Executive Directors and CEOs from other groups literally embraced me when they saw my nametag due to relationships that had previously been formed by both Shelley Bowen and Kate McCurdy. Representatives from the National Institutes of Health (NIH), the Food and Drug Administration (FDA), big pharma, biotech firms, and patient advocacy groups all joined together to talk about how to get treatments to those suffering in the fastest, safest way possible. At this meeting, as well as similar prior meetings, contacts were made, relationships were formed, and connections were rekindled. Navigating the FDA approval process is new to BSF. It will require education, strong partnerships, and good fortune. Sometimes, luck is a matter of preparation meeting opportunity. After attending the Orphan Drug Congress, I feel more prepared for what lies ahead and happy for the opportunity to explore new avenues of collaboration.

In June, I will join approximately 15,000 attendees at the Biotechnology Industry Organization (BIO) International Convention in Philadelphia, PA. As a patient advocacy group, we were granted a free booth in the exhibit hall, as well as complimentary attendance at all educational sessions. At this conference, I hope to gain further insight into the process of developing treatments for ultra rare diseases with small patient populations. Thankfully, there are specific educational tracks that will cover relevant topics. Again, the networking will likely be the best part of this event. I'm looking forward to representing you at the convention, to learn as much as I can from others.

At all of these meetings, we stand on the shoulders of others, so to speak, and at times, we serve as the shoulders on which others stand to further their mission. Working together as a community and with other groups is the only way that we can conquer Barth syndrome. The generosity of each and every one of you in this community allows us to stand on YOUR shoulders in order to see further. Thank you for lifting us up!

# Making a Difference

By Marc Sernel, Chairman, Barth Syndrome Foundation

Sometimes it can feel like nothing we do makes much of a real difference. We live in a big world and are faced with challenges that feel outside of our control. We wonder if our vote or our voice or our contributions matter in the grand scheme of things. We ask ourselves, "Is it worth the effort?" in our jobs and our lives. While we sometimes want to throw up our hands and succumb to doubt and cynicism, most of us still look for ways to put a positive imprint on something and make a difference.

As members of the greater Barth Syndrome Foundation (BSF) family, I want there to be no mistake: You are making a greater difference for BSF and those affected by Barth syndrome (and beyond) than you may appreciate. Our membership spans the globe but our numbers are minuscule compared to other organizations, and therefore every contribution moves the needle and can be felt in palpable ways. Every donation, large or small, makes a difference to our bottom line and ability to pursue our organizational objectives. Volunteer efforts make BSF the wonderful community and resource for affected families that it is. Researchers and doctors going the extra mile, not for the money or accolades, but just because they care, have helped to save lives, improve daily lives, and move us closer and closer to a treatment for Barth syndrome. The silver lining of being involved with a very small organization is that each contribution truly is impactful on our world.

The members of our Board are among those committed to making a difference for the betterment of BSF and the lives of those affected by Barth syndrome. No member in my tenure on the Board has taken his job more seriously, or cared more about doing the right thing, than Steve Kugelmann, whose multiple Board terms recently ended in April. Steve's refusal to accept the status quo, pushing the organization to strive for higher and higher standards of excellence, was instrumental in guiding BSF to the successes it has achieved over the past decade. Two other people that personify the make-a-difference ethos of this organization are our newest Board members, Florence Mannes and Matthew Blumenthal. In addition to being a mother of three (including an affected son) and a lawyer at a French bank, Florence started the French affiliate of BSF and has spearheaded fundraising and establishment of a Barth clinic in France. Matt's impressive accomplishments — including degrees from Harvard and Yale Law, and service as a commanding officer in the U.S. Marines — are coupled with his personal experience (through his lifelong friend, Will McCurdy) with the devastating impact of Barth syndrome and profound commitment to helping this organization achieve its goals. Welcome, Florence and Matt!

So how can you help make a difference for BSF? For starters, active participation in the organization, like coming to the conference and engaging in the organization's other efforts, is incredibly important to us. Affected individuals and families can contribute medical data to the registry and blood and tissue samples to the repository. Donating to the organization, and helping to fundraise from others, is another critical component that accelerates BSF forward in pursuit of its objectives. These efforts over the past 15 years have enabled us to make great strides in understanding the syndrome and placed us on the verge of having potential treatment options nearing readiness for clinical trials.

The possibility of a clinical trial in the near future may present us with another opportunity to make a difference. Those of us with affected children may be called on to seriously consider joining appropriate clinical trials that will help scientists and doctors evaluate a particular therapy. We'll need each other to take this important next step because any clinical trial will require a not insubstantial percentage of our small number of affected individuals to participate. BSF has always prided itself in "punching above its weight" and we'll need to continue to do so if we want see a successful clinical trial happen in the near future.

With apologies to Dr. Hilary Vernon for stealing her quote, "Barth syndrome may be a very rare disorder but it doesn't feel rare to the person or family that's affected; for those people and families it's their whole world." But just as the impact of Barth syndrome does not feel rare or trivial to those it affects, so too are all of your contributions to this small organization felt in a much more personal and amplified way. Working together, with all of us doing our part to make a difference, we can change the world for those suffering from Barth syndrome.

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# On the Edge of a New Frontier

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**"We now have over seven different therapeutic ideas that may make a real difference in the lives of our boys/men, much of which is the direct result of the BSF Research Grant Program. We now need the BSF community to step up and make the sacrifices needed to test these potential treatments."**

Abbreviations:	
BTHS—Barth syndrome	KD—knockdown mouse model of Barth syndrome
CL—cardiolipin and MLCL—monolysocardiolipin (cardiolipin missing one fatty acid chain)	ROS—reactive oxygen species (very reactive chemical compound)
iPS cell—induced pluripotent stem cell line (Bill Pu's work)	

As with all BSF grant cycles, the 2014 projects were submitted last October and those that were accepted by BSF were actually awarded the following year. Thus, they are included in 2015 fiscal year expenses.

	<p>Valerian Kagan, PhD, Professor and Vice-Chairman, University of Pittsburgh, Pittsburgh, PA <a href="#">"Mechanism and role of cardiolipin oxidation and hydrolysis in Barth syndrome"</a> Award—US \$100,000 over 3-year period <i>*Partial funding for this award was provided by the Barth Syndrome Foundation of Canada</i></p>
	<p><b>Understanding on a molecular level how the mitochondrial oxidation products observed in Barth syndrome models of human disease cause its distinctive pathophysiology.</b> Inappropriate oxidation of various biological molecules (like lipids and proteins), yielding increased Reactive Oxygen Species or ROS generated in the mitochondria, may be the root cause of the symptoms suffered by Barth syndrome (BTHS) individuals. This "oxidation" hypothesis has evolved through the work of several BSF grant recipients and provides a mechanistic basis for developing highly focused therapies. Dr. Kagan will concentrate on identifying: the types of cardiolipin (CL) oxidation products or byproducts that are the result of <i>tafazzin</i> dysfunction found in various cellular models of BTHS (yeast, fly, mouse, and iPS cells), how this leads to the accumulation of MLCL (the distinctive metabolite of BTHS), and how this MLCL accumulation or oxidized product production leads to mitochondrial dysfunction. Dr. Kagan will also employ specific oxidation inhibitors to determine if they can reverse the mitochondrial dysfunction of BTHS. Dr. Kagan proposes that by intervening at the early steps of CL oxidation, it may prevent or reverse the downstream effects that cause the symptoms of BTHS (reverse the pathophysiology).</p>
	<p>W. Todd Cade, PT, PhD, Associate Professor, Washington University School of Medicine, St. Louis, MO <a href="#">"Effects of resistance exercise training on cardiac, metabolic, and muscle function and quality of life in Barth syndrome: Part II"</a> Award—US \$45,313 plus \$13,530 in volunteer travel over 1-year period <i>*Funding for this award was provided by the Will McCurdy Fund for the Advancement of Therapies for Barth Syndrome</i></p>
	<p><b>Determining whether resistance exercise training with protein supplementation has clinical value.</b> Dr. Cade has published several clinical studies that demonstrate the physiological uniqueness of BTHS individuals. So far his research with three BTHS individuals has not shown the expected benefit in fatigue resistance with resistance exercise training in this small group (even though strength appeared to increase), but the research has also shown no harm (not a trivial matter). This surprising "negative" result is based on a few individuals, and Dr. Cade is seeking to strengthen the study by expanding the number of individuals tested. In this year's application, Dr. Cade has added nutritional monitoring and protein supplementation to the volunteer's diet during the training. The full benefits of the exercise training may require extra protein supplementation to the volunteer's diet to support an increase in muscle mass which may positively affect fatigue. Protein catabolism was previously shown by Dr. Cade to be adversely affected in BTHS individuals, and muscle mass is abnormally low for them as well. By expressly adding extra protein and monitoring the volunteer's diet, Dr. Cade expects to observe a more robust clinical improvement in fatigue and muscle mass. This 2014 award, along with Dr. Cade's other clinical studies, helps to lay a solid foundation of clinical and physiological data that are essential for BTHS clinical research, especially as it applies to any future drug/FDA study.</p>

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# On the Edge of a New Frontier

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	<p>Christina Pacak, PhD, Assistant Professor, University of Florida, Gainesville, FL <b>"Correction of mitochondrial dysfunction in Barth syndrome"</b> Award—US \$100,000 over 3-year period <i>*Partial funding for this award was provided by Association Barth France</i></p>
	<p><b>Preclinical experiments in mice to allow for gene therapy in humans.</b> Gene therapy is an obvious therapeutic possibility for any Mendelian disease like BTHS, however there are no FDA approved gene therapy products in the US, and there is only one approval in Europe. There is a sad legacy with US gene therapy, mostly unfair, which is only now becoming superseded with new vectors which are actually viruses that infect cells and correct the genetic defect by supplying a "good gene" to the nucleus. In collaboration with Dr. Barry Byrne and the Powell Gene Therapy Center at the University of Florida, Dr. Pacak will be constructing and testing the vectors that will ultimately be used to treat BTHS individuals. Dr. Pacak will be using a type of vector (serotype 9) of the adeno associated virus (AAV) class that is being developed in parallel with similar gene therapy efforts for Pompe disease and other disease projects at the Powell Gene Therapy Center. She will be testing three different types of gene promoters to see which works best in the laboratory before testing these vectors on the BTHS knockdown (KD) mouse. This application is intended to help determine how gene therapy could be performed for BTHS individuals.</p>
	<p>Cynthia James, PhD, Research Associate, Johns Hopkins University, Baltimore, MD <b>"How do women adapt to being a Barth syndrome carrier? A mixed methodological study of psychological adjustment and reproductive options"</b> Award—US \$48,563 over 2-year period</p>
	<p><b>Issues faced by carriers of Barth syndrome.</b> Dr. James, along with her collaborator, Rebecca McClellan, MGC, CGC, will be interviewing and gathering data that deal with the emotional and practical aspects of living as a female carrier of BTHS. This part of the BSF community was specifically engaged at the 2014 BSF Conference, and the need for more information and better awareness was apparent. This award is in response to a Request For Applications (RFA) that BSF had advertised for the 2014 cycle. The project will involve two phases. The first phase will collect information from carrier women using semi-structured phone interviews from three groups (dating/making reproductive plans, childbearing/rearing, parent/grandparent of adult child(ren)). The second phase has the goal of quantifying difficulties, predicting psychological risks, and testing themes identified in the first phase. This second phase will use the information collected from phase one to develop a test questionnaire that will tabulate how the groups adapt to the different psychological stresses. Moreover, because there is no study of this nature addressing carrier issues in conditions involving cardiac disease, the results of this study may have wider applicability to inform genetic counseling and promote research into issues faced by mutation carriers for other genetic conditions.</p>
	<p>John L. Jefferies, MD, MPH, FAAP, FACC, FAHA, Director, Advanced Heart Failure and Cardiomyopathy Services; Associate Professor, Pediatric Cardiology and Adult Cardiovascular Diseases, The Heart Institute; Associate Professor, Division of Human Genetics, Cincinnati Children's Hospital Medical Center, Cincinnati, OH <b>"Assessment of quality of life, anxiety, and depression in Barth syndrome: Expanding the scope of comprehensive care"</b> Award—US \$28,750 over 2-year period <i>*Partial funding for this award was provided by Barth Syndrome Trust</i></p>
	<p><b>Investigating depression among Barth syndrome individuals.</b> The 2014 BSF International Conference highlighted the large problem of depression and its consequences with many older BTHS individuals. This award is in response to a Request For Applications (RFA) that BSF had advertised for the 2014 cycle. Dr Jefferies is a pediatric cardiologist who really understands that this is an important aspect of a complex patient's health, so he is very interested in this. He and the staff at Cincinnati Children's Medical Center will gather data about the extent and the seriousness of this depression and suggest ways to mitigate it. Dr. Jefferies proposes two aims: the first is to characterize the prevalence of depression, anxiety, and of health-related quality of life effects, through patient reported outcomes; the second aim is to develop a Barth-specific checklist, identify gaps in clinical care, and use this to improve care delivery and information for families.</p>

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# On the Edge of a New Frontier

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	<p>Adam Chicco, PhD, Associate Professor, Colorado State University, Fort Collins, CO "Translating murine <i>Taz</i> deficiency to human Barth syndrome: Focus on impaired lipid oxidation" Award—US \$49,998 over 1-year period <i>*Partial funding for this award was provided by Barth Syndrome Trust</i></p>
<p><b>Following up on observations about Coenzyme A deficiency in BTHS knockdown (KD) mice and extending this into iPS cells.</b> Dr. Chicco used metabolic profiling to discover that <i>tafazzin</i> KD mice have low levels of Coenzyme A (CoA) in their heart and liver tissue—CoA is a vitamin B5 derivative that is a vital part of the biochemistry of the mitochondria. Dr. Chicco's hypothesis, which builds on the work of Dr. Cade with BTHS individuals, is that CoA synthesis in knockdown (KD) mice is down-regulated due to dysfunctional fatty acid oxidation — fatty acid oxidation also takes place in the mitochondria. In essence, some of the symptoms of BTHS may be the result of an imbalance in fatty acid oxidation which causes a low CoA level and a reliance on glucose for energy — exactly what Dr. Cade has observed in BTHS individuals. This hypothesis explains how BTHS individuals metabolize glucose almost exclusively and not fatty acids, and it may also explain why BTHS individuals have low muscle mass. Dr. Chicco wants to extend the observations he has made in the KD mouse into iPS cells and cell lines derived from BTHS individuals to confirm this hypothesis. If confirmed, then a therapy based on restoring fatty acid oxidation to normal levels would be the next step.</p>	
<i>(Photo courtesy of Dr. Chicco)</i>	
	<p>Colin Phoon, MD, MPhil, Associate Professor, New York University School of Medicine, New York, NY "Novel antioxidant therapies in a mouse model of Barth syndrome" Award—US \$50,000 over 2-year period <i>*Funding for this award was provided by the Paula &amp; Woody Varner Fund</i></p>
<i>(Photo courtesy of Dr. Phoon)</i>	
<p><b>Maximizing the value of the BTHS knockdown (KD) mouse by altering its induction of <i>tafazzin</i> deficiency and rescuing the symptoms with antioxidant treatments.</b> Dr. Phoon, along with Dr. Ren, had shown in earlier work supported by the BSF the importance of ROS in the heart of knockdown mouse model (KD) of BTHS. They also discovered that inducing <i>tafazzin</i> dysfunction early in the life of the KD mice was lethal, but they also found that treatment with a common clinical compound, N-acetyl cysteine (NAC), could rescue this lethality. This is a significant scientific result that shows the pathophysiology of BTHS may be positively altered during an individual's early development with a chemical therapy. In this application, Dr. Phoon seeks to test specific antioxidant compounds on the KD mouse model and determine if lethality can be rescued in a similar way to what was observed with NAC. It is unclear if the effect of NAC on KD mice is due to antioxidation or to some other property. In addition, in another aim Dr. Phoon seeks to alter the induction of <i>tafazzin</i> deficiency in the KD mouse model to obtain mice that more closely resemble BTHS individuals with symptoms between the extremes of lethality at birth and the cardiac problems seen only in later life. Modifying the induction protocol of the KD mice to make it resemble the human condition more closely is of great value for researchers.</p>	
	<p>Trudy M. Forte, PhD, Director of Research, Lypro Biosciences, Inc., Berkeley, CA "Reversal of cardiolipin deficiency in Barth syndrome mouse model" Award—US \$49,997 over 1-year period <i>*Funding for this award was provided by the Will McCurdy Fund for the Advancement of Therapies for Barth Syndrome</i></p>
<i>(Photo courtesy of Dr. Forte)</i>	
<p><b>Using nanodisks for lipid replacement therapy in KD mice.</b> Dr. Forte, in collaboration with Dr. Ryan who invented the nanodisk technology, will test the concept of lipid replacement therapy using the knockdown mouse model (KD) of BTHS. This work builds on this novel therapeutic idea that was first supported by BSF in the 2012 cycle with Dr. Ryan, where the possibility of supplying the missing cardiolipin in BTHS cells was first proposed. Cardiolipin is uniquely deficient in BTHS cells and so supplying it may be therapeutic. Dr. Forte has shown that nanodisks of CL can deliver CL into cultured cells. Nanodisks are protein-lipid particles that appear to supply lipid compounds and chemicals into cells and, presumably, into various organs of the body. Dr. Forte is attempting to do the same with the KD mouse model. Lypro Biosciences is a biotech company in the San Francisco Bay area. There are many unknowns with this novel treatment idea, but its potential to impact BTHS therapy is high.</p>	
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Connor (age 4) & Kevin  
*(Photo courtesy of BSF2014)*



Wyatt (age 7) & Amanda  
*(Photo courtesy of Amanda Clark2014)*



Mario (age 11)  
*(Photo courtesy of BSF2014)*



William (age 15)  
*(Photo courtesy of Amanda Clark2014)*



R.J. (age 17)  
*(Photo courtesy of BSF2014)*

# On the Edge of a New Frontier

(Cont'd from page 6)



Michael T. Chin, MD, PhD, Associate Professor, University of Washington, Seattle, WA

**"Enzyme replacement therapy in heart failure associated with *tafazzin* deficiency"**

Award—US \$50,000 over 1-year period

\*Funding for this award was provided by the Will McCurdy Fund for the Advancement of Therapies for Barth Syndrome

(Photo courtesy of Dr. Chin)

**Experiments both *ex vivo* and *in vivo* with BTHS knockdown (KD) mice to prove inappropriate ROS, hypertrophy, and apoptosis cause the symptoms of Barth syndrome which should be ameliorated by enzyme replacement therapy.** Dr. Chin has shown that he can produce recombinant modified human *tafazzin* protein and deliver it to the mitochondria of mouse muscle cells (C2C12) temporally made to be *tafazzin* deficient, and that this recombinant protein has the expected physiological effects. This work was supported by a BSF grant in the 2013 cycle as a step towards developing enzyme replacement therapy (ERT). Dr. Chin intends to confirm the usefulness of this recombinant engineered *tafazzin* protein by using cell culture systems including those from the KD mouse to show the importance of abnormal ROS production in the symptomology of BTHS. The ultimate goal is to show that this recombinant protein treatment rescues the biochemical and physiological abnormalities associated with *tafazzin* deficiency at the biochemical, cellular, muscular, and organismal levels. Dr. Chin will use a surgical manipulation of the heart (transverse aortic banding) in KD mice to produce a more severe form of heart failure, and then he will attempt to reverse this heart failure with enzyme replacement therapy. The therapeutic implications for BTHS individuals are obvious and significant.



Renata Goncalves, PhD, Postdoctoral Research Associate, Harvard T. H. Chan School of Public Health , Boston, MA

**"Correcting mitochondrial ROS production to decrease Barth syndrome symptoms"**

Award—US \$50,000 over 1-year period

(Photo courtesy of Dr. Goncalves)

**Determining ROS defect in BTHS knockdown (KD) mice and finding compounds that specifically suppress the defect.** The publications by Dr. Pu and collaborators (Wang et al., 2014), and by other BSF grant awardees (Drs. Greenberg, Phoon, Ren, and He), propose that increased reactive oxygen species, ROS, could be the mechanism that causes the symptoms of BTHS. Treatments that lower inappropriate oxidizers like ROS should be therapeutic, theoretically. One should also know that certain oxidized compounds, like lipids, are essential for the proper functioning of the cell, and that a total loss of oxidized products is expected to be lethal. General antioxidant treatments in BTHS have never been shown to be effective, and antioxidant therapy has shown limited benefit in other diseases where ROS has been implicated as a player. Dr. Goncalves proposes to identify the specific sites of ROS production and measure the rate of production in the KD mouse model of BTHS. She also proposes to intervene at the specific sites in mitochondrial metabolism that generate inappropriate levels of ROS — a novel concept compared to the general antioxidant therapies tried in other diseases. These specific ROS-generating inhibitors (unpublished) offer the promise of accomplishing the goal that general antioxidant treatment has not fulfilled.

Barth syndrome is and remains a life-threatening disease which causes our families to live in a cruel state of anxiety and worry. To reverse this situation, the BSF Research Grant Program was started in 2002, and it has been the major expense of BSF over the years. This Program is fulfilling its promise of nurturing science and researchers to make the world a place "in which Barth syndrome no longer causes suffering or loss of life." We now need the BSF community to generously step up and make the sacrifices needed to test these potential treatments. We need volunteers to submit themselves to clinical trials to test these ideas and determine their value for themselves and for their future brothers. We need researchers to intensify their efforts to develop therapies. We need donors to support all of this work. And we need collaborators to leverage our work and share in the burden and successes. We cannot make the world we want to live in until all of us come together as a community and make a contribution in whatever way we can.

(Photos courtesy of BSF<sup>2014</sup>)



English & Ben (age 18)



Oliver (age 23)



Ryan (age 24)  
& Aldo (age 23)



Peter (age 28)



Joshua (age 31)  
& Darryl (age 33)

# Layman's Perspective on Therapy Development

(Cont'd from page 1)

called cardiolipin was deficient in those with Barth syndrome as a result of problems with its changing from one form to another (2000). What we have spent the last 15 years working on together is filling in some of the blanks. We certainly can't explain everything yet (and so, happily, some really good researchers continue to try to solve more of the underlying mysteries of this complicated disorder), but we have made a great deal of progress.

Very simplistically, think of a chain of events that leads from a mutation in a single gene, through lots of different complex processes in a human body and ultimately results in the clinical manifestations that we see in the guys who have BTHS. We started by knowing something about the first link in this chain — the gene mutation — and we also were beginning to realize that there was a special and very important fat molecule called cardiolipin that was different in our guys, and we saw some of the physical and medical issues that those with BTHS displayed — the final link in the chain.

Thanks to the hard work of scientists and doctors around the world, patient participation in studies and data collection projects, and donors and funding institutions that have supported the work, we now know a lot more. In an extremely abbreviated and selective summary of BTHS scientific and medical knowledge, we now understand that:

A	A gene called <i>TAZ</i> is mutated in one of many different ways to cause the disorder
B	As a result, a protein called <i>tafazzin</i> is deficient
C	A critically important phospholipid called cardiolipin is scarce in its mature form in BTHS, though its precursor is over abundant (the ratio between the two is actually a distinctive signature of BTHS); one of cardiolipin's roles is to be a vital component of a certain part of the membrane of mitochondria (the energy centers of cells)
D	One result of this aberration is hypothesized to be an increase in the level of <u>Reactive Oxygen Species</u> (ROS) that is produced, which causes oxidative stress and is detrimental
E	Several amino acids (essential building blocks of proteins) are deficient, which leads to various significant metabolic issues
F	Among other clinical manifestations, guys with BTHS have underdeveloped skeletal muscles and severe exercise intolerance

One can imagine that if one could "fix" the problem in any one of these links in the chain, perhaps everything "downstream" would work better and the health of the guys with Barth syndrome would improve, maybe even dramatically. If so, then whatever that "fix" was would be a treatment for the disorder.



(L-R) Drs. Peter Barth & Richard Kelley  
at 2000 meeting on BTHS  
(Photo courtesy of BSF<sup>2000</sup>)

So, since we know these things now and have better ways of measuring outcomes, it is time to begin work on developing treatments! But before anything can be tested on a human being, we all need to know that there is good reason to believe that a particular treatment has real promise. Importantly, there now exist two very critical models of BTHS that possible treatments can be tried on beforehand. The first is a mammalian model of the disorder — in our case, a knockdown mouse developed in 2008 by a company that BSF (led by Dr. Matt Toth) paid to create. One can imagine how vital it is to research to be able to work with a complete animal that displays the full characteristics of BTHS. However, our BTHS mammal is a mouse not a human being, and as amazingly similar genetically as mice and humans are, clearly there are some key differences. Therefore, we are incredibly fortunate to have another model now available to offer further insights. It was first developed in 2014 by Dr. William Pu who took skin cells from BTHS patients, turned those back into iPS cells — induced pluripotent stem cells — and then grew human BTHS heart cells in a special way so that they work together to mimic a piece of heart tissue. While this model is not an entire animal and cannot shed light on interactions between various aspects of this complex

disorder (such as cardiomyopathy, neutropenia and skeletal muscle underdevelopment, to name a few), at least some aspects of one crucial organ system can be studied in actual human cells/tissue. Even as a layman, I can appreciate how wonderful it is to have these two models — a mammal with BTHS (albeit not a human) as well as a part of a human organ with BTHS (albeit not a complete being) to test various treatments on before approaching BTHS patients.

And so with all this background work done, we now stand at a really amazing, scary, exciting, nerve-wracking and potentially life-saving inflection point. We are *approaching* a time when we will be ready to test treatments — specifically for Barth syndrome — in patients.

(Cont'd on page 9)

# Layman's Perspective on Therapy Development

(Cont'd from page 8)

None of this will be instant, and much of it will not end up working; that is how science progresses. But right now, there are seven therapeutic approaches that show promise and are in various early stages of development.

Potential Therapeutic Approach	Might Help Solve BTHS Issue Listed on Previous Page
1. Gene therapy	A
2. Enzyme replacement therapy	B
3. Lipid replacement therapy	C
4. ROS scavenging therapy	D
5. Nutritional therapy	E
6. Exercise therapy	F
7. Repurposed drugs (of many different types)	The ones farthest along in this category address C and D

BSF is currently involved in some way in development work on each of these. Through this year's BSF grant program, BSF and our affiliates are supporting work by highly respected researchers to further advance many of these. Some of these paths take substances or approaches that already are known to work in humans and try to apply them to Barth syndrome, while others involve new approaches. With the help of our wonderful Scientific and Medical Advisory Board and other experts, we will continue to monitor progress on each of these. Over time, some will drop off the list and others will be added. We will all have to work together and do our parts in order to continue to push this incredible effort ahead, but it is an amazing milestone that we are now discussing possible treatments for Barth syndrome. It is an extremely exciting time!

## Results from 2012 Research Study: Decoding Mealtime: Feeding and Eating Behaviors

By Emily Burgess, OTS, Virginia Commonwealth University, Richmond, VA; Stacey Reynolds, PhD, OTR/L, Virginia Commonwealth University, Richmond, VA



Dr. Stacey Reynolds performs smell test (Photo courtesy of Dr. Reynolds<sup>2014</sup>)

It has long been noted within the Barth community that boys with Barth syndrome exhibit some unusual feeding and eating behaviors. Picky eating, difficulty swallowing, gagging, and outright refusal to eat are just some of the behaviors that can make mealtime stressful for boys with Barth syndrome and their families<sup>1</sup>. Since 2010, our team of occupational therapy researchers has sought to find out why these behaviors occur, with the long term goal of reducing the stress and participation limitations that these behaviors can cause. In 2012, we conducted a study at the Barth Syndrome Foundation (BSF) International Conference looking at chemical taste sensitivity, food preferences, and feeding behaviors in 24 boys with Barth syndrome ages 4-17 years old. We used chemical taste strips to test the boys' sensitivity to bitter (PTC) and salty (NaB) tastes. We also asked parents to complete a food inventory and Short Sensory Profile. A summary of our findings are outlined below and a full description of the study can be found in Volume 3, Issue 1 of the Journal of Rare Disorders<sup>2</sup>.

### Findings based upon 2012 study:

- Boys with Barth syndrome are more sensitive to bitter tasting compounds and less sensitive to salty tasting compounds, meaning only a small amount of bitter taste is needed for the taste to register, and a larger amount of a salty taste is needed for the taste to register.

(Cont'd on page 10)

# Results from 2012 Research Study: Decoding Mealtime: Feeding and Eating Behaviors

(Cont'd from page 9)

- Boys with Barth syndrome differ significantly from their typical peers in terms of taste and smell sensitivities but not in their overall tactile (touch) sensitivity.
- Boys with Barth syndrome exhibit significant differences in their feeding and eating behaviors, including greater incidences of: food refusal, difficulty swallowing, requesting separate meals, exhibiting eating habits that differ from the rest of the family, and decreased appetites.
- There is a statistically significant relationship between the food refusal behaviors of boys with Barth syndrome and their higher taste and smell sensitivities.

These results indicated that the differences in feeding and eating behaviors seen with boys with Barth syndrome are likely to be due, at least in part, to differences in chemical taste perception and in taste and smell sensitivities. To build on this research, our team was awarded a BSF Idea Grant in 2013, and returned to the International Scientific, Medical & Family Conference in 2014 to collect data and test some new methods. These methods included:

- A new taste test to examine sensitivity and hedonic responses to different salt concentrations in chicken soup.
- A standardized test (Sniffin' Sticks) to measure smell sensitivity.
- Tongue photographs, enlarged to count the size and density of taste receptors.
- A new (simplified) food inventory that examines frequency and enjoyment of foods from different food groups.
- Video analysis of boys with Barth syndrome participating in mealtime routines.

**While we are still in the process of analyzing the data from this round of collection, here is a first look at our findings:**

- Individuals with Barth syndrome were found to have smell thresholds within the normal range, though significantly more sensitivity than age-matched non-Barth syndrome peers. Over half (57%) of the individuals with Barth syndrome reported having an "increased" sense of smell (more sensitive than most people) as opposed to only 11% of typical peers.
- Perceptions of the saltiness of chicken soup did not differ between individuals with and without Barth syndrome (i.e., both groups were able to tell which soup samples had more or less salt). However, boys and young men with Barth syndrome tended to have a higher tolerance and enjoyment for the soups with higher salt concentrations.
- Individuals with Barth syndrome did not eat more salty or sweet foods than their age-matched peers, but they did eat fewer vegetables, fruits, grains and proteins.
- Common feeding problems identified in our video analysis study include difficulty with tongue lateralization in the mouth, difficulty forming a food bolus, extended periods of open mouth chewing, pushing food out of the mouth, and difficulty swallowing.

We hope that this data will continue to inform a line of research supporting an end goal of developing feeding supports for parents and interventions for individuals with Barth syndrome. We welcome comments and suggestions from the Barth community, and thank Shelley Bowen and the Feeding Advisory Board, which has also helped to inform this research.

## References

<sup>1</sup>Reynolds, S., Kreider, C., Bendixen, R.M. (2012). [A mixed-methods investigation of sensory response patterns in Barth syndrome: A clinical phenotype?](#) American Journal of Medical Genetics, Part A.

<sup>2</sup>Reynolds, S., Kreider, C.M., Meeley, L.E., & Bendixen, R.M. (2015). [Taste perception and sensory sensitivity: Relationship to feeding problems in boys with Barth syndrome.](#) The Journal of Rare Disorders, 3(1), 1-9.



Dr. Shelley Lane performs taste receptor test  
(Photo courtesy of Dr. Reynolds<sup>2014</sup>)

# Getting the Correct Diagnosis is Everything

By Michelle, Parent of Affected Individual, Ohio

**"I know for a fact that the diagnosis has changed him. There are so many people at the Barth Syndrome Foundation to talk to and get information. We are no longer alone. He knows where he belongs and there are other boys out there like him. He has been talking to the other boys and can't wait to meet them at the conference in 2016. He doesn't feel so scared not knowing what is going on with him. This diagnosis has also saved me. I was so depressed and such a mess mentally not knowing what was going on with my child. Having a support group with the Foundation was an actual life saver for my son and our family."**



Clint (age 15)  
(Photo courtesy of Clint's family<sup>2015</sup>)

Getting the correct diagnosis is everything. Our family was lost for 14 years, before we finally found the right road. Clint is now 15 years old and was just diagnosed with Barth syndrome last year.

During the first years of Clint's life, he fought multiple infections. The doctors never believed how sick he was. Every month Clint would get sick for about a week. The symptoms would start to ease by the time I could get him seen by a doctor. The doctors always gave us looks like I was making all this up and would just tell us that Clint would grow out of it. We went through countless doctors and ER visits, and Clint suffered through this every month until he was 10. We went to a new dentist trying to figure out why his mouth was in such bad shape. The dentist was dumbfounded as to how a 10-year old's mouth could have such severe ulcers and periodontal disease. The dentist called us that night after doing research all day and advised that Clint be tested for neutropenia. I just started crying. Finally a doctor was listening to me. We had Clint tested the next day, and he was neutropenic. Then we headed down the journey of finding a hematologist.

During the next year, we found out that Clint had cyclic neutropenia. He was started on Neupogen, and he started feeling a little better. Some of his symptoms did ease up. He was tested for the ELANE gene mutation, which came back negative. He had a lot of other symptoms, including muscle weakness, slow growth, a lot of GI problems, osteoporosis, and migraines. We saw many hematologists about these symptoms, because it still seemed that we were missing something. Not one of them even thought about doing more genetic testing. Clint was enrolled in the Severe Chronic Neutropenia International Registry. I just kept asking questions trying to get these doctors to listen to me.

After attending the Neutropenia Family Conference in 2014, I came back home with a mission. I made an appointment with Clint's hematologist and told him this had gone on long enough. I wanted to see a geneticist. I knew there was something else going on with Clint. After pushing for a month, we finally had a muscle biopsy done and the TAZ mutation was found. Finally, after 14 years, we knew what Clint had. Finally, we had a home with the Barth Syndrome Foundation and the registry.

I immediately contacted the Barth Syndrome Foundation (BSF) and talked to everyone that I could. After speaking with Shelley Bowen (Director, Family Services & Awareness) and Matt Toth, PhD (BSF Science Director), I actually now have resources to use to talk to the doctors. We also have the resources to find doctors who know how to treat Barth syndrome. We already have gone out of state to some of these doctors in order to get a treatment plan. There are so many people at the Barth Syndrome Foundation to talk to and get information. We are no longer alone.

We started different treatments, and Clint has been a totally different child since the diagnosis. I know for a fact that the diagnosis has changed him. He knows where he belongs and there are other boys out there like him. He has been talking to the other boys and can't wait to meet them at the conference in 2016. He doesn't feel so scared now that he knows what is going on. This diagnosis has also saved me. I was so depressed and such a mess mentally not knowing what was happening to my child. Having a support group with the Foundation was an actual life saver for my son and our family.



# The Will McCurdy Fund for the Advancement of Therapies for Barth Syndrome

By Steve McCurdy, Chairman Emeritus, Board of Directors, Barth Syndrome Foundation



(L-R) Will, Steve, Eliza and Kate  
*(Photo courtesy of McCurdy family)*

What was a dream fifteen years ago is coming to pass. Researchers are moving from basic research on Barth syndrome to exploration of therapies specifically designed for Barth patients. Therapy development takes time and money, and therapies must be tested on human patients before they can be widely deployed. But work now is planned or underway on seven different potential therapies that could help alleviate the symptoms or even cure Barth syndrome.

We are entering a new era in Barth syndrome research, and this new period will create new demands on the Barth Syndrome Foundation (BSF) and all of our constituencies. The two greatest challenges will be fundraising and the informed participation of those with Barth syndrome in therapy trials designed to test the safety and efficacy of proposed treatments.

It may be a decade or more before the first treatment providing significant improvement in the lives of those suffering from Barth syndrome is approved. With researchers, doctors, families, and BSF working closely together, however, this is a mountain that we must and we can climb!

Therapy development is much more expensive than basic research, primarily due to the cost of the human trials mentioned above. Often, it is pharmaceutical companies that fund the lion's share of therapy development and approval. They typically offset the high rate of failure by pursuing multiple trials targeted at larger markets. But, since Barth syndrome is a rare disease (and therefore a small potential market), BSF will have to bear a greater share of the burden, raise greater sums, and cultivate new partners to bring any successful therapies to common use. Lindsay Groff (BSF Executive Director), Dr. Matt Toth (BSF Science Director) and the Board of Directors are focused on these new challenges and are creating plans to address them.

To help accelerate this effort and to demonstrate that BSF is capable of meeting its part of the challenge, the McCurdy family and its friends and supporters have created a fund in Will McCurdy's memory that is unambiguous as to its purpose — the "Will McCurdy Fund for the Development of Therapies for Barth Syndrome". Initially funded by a one-time \$2 million donation from Kate, Steve, and Eliza McCurdy, Dr. Paul Russell, and Laura and Scott Malkin, and with the addition of approximately \$500,000 in donations given in Will's memory by other members of his family and many friends, the fund begins at \$2.5 million. Our goal is to seek donations to grow this fund to \$3 million before year-end 2015, and to continue to raise funds necessary to attract third-party donors and investors in therapy development in the years to come. Will's fund is not an endowment. Its principal and income will both be used as "seed" funding for therapy development. Though we now begin with this large amount of money dedicated to this purpose, significant additional donations will be needed to continue to complete our drug development work. Fundraising will need to be everyone's job if we are to reach our goal of effective treatments and a cure for Barth syndrome.

Sadly, Will McCurdy died before these treatments could become a reality for him, but he hoped deeply that progress that would help his "Barth brothers" would continue to be made. Will frequently donated biological samples and medical data for clinical advancement and participated in well-thought-out experimental procedures. Now, though it feels bitter sweet, it seems appropriate that Will's legacy also includes a memorial fund that will help accelerate BSF's therapy development efforts. He was proud that BSF consistently punches way above its weight.

## BSF Seed Funding Helps Create Medical Research Breakthrough "Heart on a Chip" Recognized for Innovation

With seed funding from the Barth Syndrome Foundation (BSF), Dr. Pu created a medical research breakthrough, "Heart on a Chip". Although more testing is needed, this innovation could help unlock answers about Barth syndrome. It could also lead to discoveries in other diseases. Through collaboration across multiple fields of biology and engineering, this work elevates organ-on-a-chip technology to a new level and sets a powerful precedent in medical research. This is at the forefront of personalized medicine, which is creating such a buzz these days. That is why this specific research was included in the American Heart Association's list of *Top Cardiovascular Disease Research Advances in 2014!*

# We Named Him Will

By Steve McCurdy, Chairman Emeritus, Board of Directors, Barth Syndrome Foundation



Will McCurdy  
1/23/1986–10/25/2014  
(All photos courtesy of McCurdy family)

We cried tears of happiness when he was born. We named him for his grandfathers, William after my father, William McCurdy, and Russell as his middle name after Kate's Dad, Dr. Paul Russell. William Russell McCurdy. It sounded like the name of a Supreme Court Justice, one nurse told us. We decided to call him Will before we had any idea just how apt that name would turn out to be. He indeed exuded willpower throughout his life.

Everyone has gifts. Some are innate and obvious from an early age. Others seem to develop in time and in response to life. Will was quietly determined to do things on his own schedule from before birth, refusing to make a timely entrance into this world despite many hours of encouragement by his mother and his obstetrician. Once born, he refused to sleep, driving an experienced baby nurse to distraction. Talking, walking, riding a bike, all waited until Will was ready and then, the speech was in full and articulate sentences, the walk was across the room unaided, and the bike ride was around the block. It was always when Will decided he was ready and never before.

His instinct was to observe closely and learn before making his move. In later years during dinner table conversations, this proved unnerving as he would listen, wait and then without fanfare, make a cogent point that inevitably turned the discussion in a new direction... or ended it! When conversations got more heated, it was usually his calm countenance that cooled the discourse. We all learned to be more thoughtful by engaging with Will.

Perhaps the most remarkable thing about Will was his inability to be self-centered. Despite the escalating toll paid in pain and confinement that the battle with Barth syndrome took on his body and his life, Will's focus was always outward. He would awaken from yet another procedure under anesthesia and his first groggy words were always directed to the nurses and doctors caring for him, to thank them for helping him. His relationships with people tended to be profound, and his conversations usually were of substance. He talked about real things with anyone who would discuss them with him, and he cared deeply about the people who connected with him. He was not a frequent contributor to the Barth families' listserv, but his wisdom and insight made his comments most welcome. When a Mom voiced her feeling of guilt about leaving her Barth child to attend her best friend's wedding, Will urged her to go saying "Since we can't get away from Barth, it's your responsibility to get away from it for a few days when you can... because we love you. And it's your responsibility to pack as much joy and fun and laughter as you possibly can into that time. You're doing it for [your son] – not in spite of him." It was the insight and permission we all needed.

Will's greatest love was his family and friends. They offered him their love in return, but they also gave him the intellectual stimulation that his fertile mind craved and a vital link to their experiences and the outside world that seemed to recede further and further from his reach as the years progressed. No one who met and engaged with Will left untouched. For the members of Team Will, his Barth family, his teachers and school friends, the doctors, nurses and hospital staff, his many adoring cousins, aunts and uncles, and especially his beloved sister Eliza, Will was a role model. He never understood why that was the case and would say that he never did anything that anyone else in his shoes would not have done. He didn't know it, but there were many college essays written about him and the nobility of his approach to his life's struggle. Most of us with far fewer challenges and far more time, fail to build lives as rich or a character as admirable as his. It is ironic that as Will's body gradually weakened and finally succumbed to the ravages of Barth syndrome, his character followed the motto found on our blue Barth bracelets, "Grow Stronger".

In the end, Will faced his impending death with grace, courage and a deep, abiding love for us. There was nothing fair about the hand he was dealt. But no one could have played it any better.



Will

# Watch

By Will McCurdy

**Editors' Note:** Portions of this essay that was written by Will in 2010 were read at the Celebration of his life on November 15, 2014. One can see why his family thought that it was appropriate to create the Will McCurdy Fund for the Advancement of Therapies for Barth Syndrome in his memory.



Will  
(Photo courtesy of Amanda Clark<sup>2012</sup>)

Watch this man. Watch this engineer. Watch him as he struggles to run his faulty old steam engine. Watch as this complex marvel of a machine rumbles and shudders down a lonely track whose map has long ago been lost. Or perhaps the day's cartographers never bothered to create a chart for this, such a rarely used track. Watch him, for even as the engineer curses his clanking rusted old machine, he takes a certain loving pride in it. This unwieldy behemoth is his and his alone. He knows that many a man had a hand in building his machine. The steelworkers. The electricians. The welders. They all rightfully claim part ownership and heritage, yet they cannot claim it to be theirs. While the engineer knows that his engine wouldn't exist without these men, their names and faces, once clear in his mind, now fade from his memory. Now more pressing thoughts take the more prominent of his cerebral seats. Even as the specifics of his engine's beginnings slowly blur, he does not and cannot forget his machine's past. He can never forget.

Watch the engineer as his machine approaches every grade. Watch him mouth a silent prayer that the squealing brakes will hold fast. Watch his faith that around the next bend the rails on which he and his engine run will continue to stretch out beneath them.

them. During this journey, the engineer's not alone on the train. But even as he pulls many cars behind him, he remains alone in his engine. He is always alone. Watch as his loneliness overcomes him. Watch as he needs to be needed. Watch the trailing cars fulfill that need. As much as the engine needs the coal car's fuel and the engineer needs the purpose provided by the passenger cars — they need him too. The cars give the engine purpose, and the engine gives the cars direction.

The engineer has spent so many years with his machine that he cannot remember his life without it. They know one another well. But even as well as the engineer knows his steel steed, his machine continues to surprise him. Every night the engineer diligently checks on the health of his engine. Watch in the growing darkness as his flashlight scans the blue hued exterior, methodically inspecting each and every inch. Wheel to stack, couple to couple, the engineer searches. On occasion, he discovers a new scratch in the paint or finds a hole where a previously secured bolt was dislodged and was lost during the day's run. There is, however, scant the engineer can do to repair his old friend. Every time he discovers a new hurt, whether it be a dent in the bowels of the coal stove or an extinguished running light, there is a sadness and pain that quietly wells up in the engineer that is at times almost unbearable. He realizes that he and his machine are but two parts of a whole. The engine cannot work without its engineer, and the engineer's life is meaningless without his engine. On the nights when an insult to his machine is found, as the engineer drifts off to sleep he cannot help but weep for his friend. His tears leave tracks through the coal dust on his cheeks, leaving a visible testament of his love for the engine. Invariably, when the next morning's sun peeks over the distant mountains, his piqued face begins to warm. He does not want to wake. He does not want to see his engine's wounds. He does not want to go on. He does not want to know what injurious obstacle his engine may face today, lurking around the next bend on the coming track. Perhaps the best remedy is to not wake up, dreams the engineer. To not start the machine again. Surely this is the best way. The engine will never again lose another bolt whilst jostling over the tracks. With no movement, the pebbles on the track can never fly up and create another nick in the beautiful paint. Yes, this is the best choice, lie here looking at the beautiful sunrise over the mountains and never again feel pain or sadness. Why didn't I do this before, dreamt the engineer? And then out of the beautiful blue sky a single lightning bolt struck a tree no more than six feet from the engine. This gave the engineer such a start that he was suddenly wide awake. No longer lulled by his dreams and drugged by sleep, he was angry, scared and confused. What had just happened? All of a sudden his mind seemed to have been wiped clean. He slowly began to realize that an engine ceases to be an engine if it never moves. It becomes a bird's nest or a rodent's burrow. While these are things of value, they are not an engine. And what of the coal and passenger cars? They all needed to be pulled. They all needed the engine. And if the cars needed the engine, that meant that they needed the engineer too. Reluctantly, the engineer rose, stretched, wiped his face clean, and coaxed his cursed old friend to life. Even though he didn't know where this track led and even though around the next bend there might not even be any track, the engineer spoke a quiet prayer and engaged the throttle. With a whoosh, the machine lurched alive again. Soon the sound of the squeaking wheels testified that they were again on their way.

(Cont'd on page 15)

# Watch

(Cont'd from page 14)

As they came around a bend, a long straightaway lay out for miles in front of them. The engineer encouraged the machine, and the engine began to pick up speed. Far off in the distance, the engineer saw a fork in the track. What's more, he saw that soon after the fork, one of the two tracks lay unfinished. The other track led up and around one of the tallest mountains that the engineer had ever seen. As he slowed the engine down and grew closer to the fork, he began to make out a building and several figures standing by the fork in the track. These men clearly weren't engineers, for there were no engines. Who could these men be? What were their intentions? As the engine and engineer drew closer, the engineer could just make out a dirt road leading to the building by the fork in the tracks. The engineer surmised that these men were here for his machine. What they wanted with his engine he didn't know. They had clearly traveled a great distance to arrive in the middle of nowhere, to wait by a fork in the railroad track. The engineer knew that without these men's help he and his engine could not safely make it onto the correct track. He had to put his trust in them. As they came closer still, the engineer could see the rusted out carcasses of trains that had run off the track. Seeing this pitiful sight instantly caused in the engineer a great dread that he had never before known. Had these men caused this atrocity? Again, he thought of stopping short of the switch and never moving again. But what would become of the engines that he knew were days behind him? The engineer knew his charge. To chart a new map of the track. He was tasked to send the new map back so no other engines would end up as rusted remains cast aside the rails. As the engineer pushed his machine forward, the machine seemed to anticipate the coming danger and protested by sputtering and belching out a column of thick black smoke from its stack. The engineer gently prodded his machine on. An unsettling calm washed over the engineer as he realized he had done everything he could do. It was now in the hands of the ever enlarging figures to complete the correct switch. A powerful mix of emotions swept over the engineer as the distance closed between the engine, engineer, and switching station. Panic, fear, sadness, distrust, trust, love, peace, and confusion, all pulsed equally through the engineer's veins. Watch as he begins to make out the faces of the men standing at the switching house. They are smiling. Watch as they wave the engineer, his engine, and his cars onward.

All of a sudden, I feel as though I am weightless and as though I am falling. I don't have the courage to look down to see what I am going to land on. Even though I don't know the quality of the earth below me, I feel as though my machine and I have done everything we can do. Ready or not, willing or not, I'm waiting to see whether the men at the house, my engine, and I are going to make the correct switch. I'm waiting to see where I'm going to land. Watch me.



Will  
*(All photos courtesy of McCurdy Family)*



Will and Dr. Peter Barth at the first-ever meeting on Barth syndrome in 2000



Will and Eliza

# Our Heroes Have Already Been Busy in 2015!

By Sandra Stevens, Fundraising Project Manager, Barth Syndrome Foundation

**"Whether you're giving your time, expertise, or money, you are responsible for every step we take.... Generosity is contagious, and people love to help. Their success is living proof that when people learn about someone in need, they welcome the chance to lend a hand."**

As you read through the wonderfully inspiring reports of progress in this journal, please remember that none of it would have been possible without you, our precious donors. Whether you're giving your time, expertise, or money, you are responsible for every step we take. Everything, from the breakthrough medical research, or support and advice for one more family, and even producing this great journal. You are making all of it possible and bringing us closer to a day when Barth syndrome will no longer cause suffering or loss of life.

Below is an update of the events organized so far this year, by our fantastic volunteers, to raise money to beat Barth syndrome. As usual, people have either taken a thing they love to do, or have ability to do, to inspire their friends and families to give. Generosity is contagious, and people love to help. Their success is living proof that when people learn about someone in need, they welcome the chance to lend a hand.



Henry (age 3)

## Happy Heart Walk, February 28, 2015

For the 3<sup>rd</sup> year running, Henry's parents and their friends organized this walk and picnic in the Bay Area, California, to raise awareness and dollars for BSF. Several local businesses donated food for around 200 people, plus greetings cards, t-shirts, prizes, and photography, free of charge. This year, there was a second fundraising event too, arranged by friends near their old home, in Fort Worth, Texas. This event just gets bigger and better every year. (*Photo courtesy of Henry's family<sup>2015</sup>*)



William (age 15)

## Rare Disease Day, St. Joseph Notre Dame High School

On Rare Disease Day, February 27<sup>th</sup>, Will in California, got together with his friends at St. Joseph Notre Dame High School to organize a bake sale to raise funds for BSF. Thanks to the support and baking skills of everyone involved, not only were donations raised, but even more people are aware of Barth syndrome. (*Photo courtesy of William's family<sup>2015</sup>*)



Eli (age 1)

## Rare Disease Day, Australia, March 5, 2015

Elissa, mother to Eli, in Sydney, Australia, told us all about a fundraising event that took place at the school where she teaches. The students wore jeans to school instead of their uniform, in exchange for a donation to benefit BSF. The children approached Elissa, offering her their lunch money to help her Eli. Many of them smashed their piggy banks and gave all their money. (*Photo courtesy of Eli's family<sup>2015</sup>*)



Zac runs for Barth

## Memorial Hermann IRONMAN, April 26, 2015

Zac, a good friend of Barth parents, and a new member of Team Will, participated in the Texas 70.3 half-IRONMAN to raise money and awareness for BSF. He was very touched by the overwhelming generosity of those who donated. "Your support is absolutely amazing. It provided inspiration to me as I struggled to put one foot in front of the other, and I know it continues to inspire Henry's family and others affected by Barth syndrome. This event was about the experience . . . about the experience of giving. I am so thankful that each of you joined Whitney and me in giving to this great cause." (*Photo courtesy of Zac<sup>2015</sup>*)

(Cont'd on page 17)

# Our Heroes Have Already Been Busy in 2015!

(Cont'd from page 16)



Team Will

## Team Will 2015

The other superhero members of Team Will are as active in Will's memory as they ever were, with many feats of strength and endurance planned for 2015. We'll be posting their events on the BSF website as we hear about them. Not every fundraising effort needs to be so physically challenging, and even the most modest walk-a-thon can raise precious dollars. (*Photo courtesy of Team Will<sup>2014</sup>*)



Michael breaks boards for Barth

## Breaking Barth 2015

In July 2014, Michael Neece, a dear friend of BSF, broke 500 boards with his hands and feet. This July, on Saturday 25<sup>th</sup>, Michael is repeating the event, but taking it to a whole new level. He's recruiting more students at his own local martial arts dojo, to break even more boards. The big news is that this time, he's also hoping to sign up dojos from across the US, and even internationally, to hold their own events. (*Photo courtesy of Michael Neece<sup>2014</sup>*)

## Fundraising Ideas

What activities do YOU enjoy? Whether it's cooking, crafting, or exercise, why not simply add a fundraising component to it? It's the perfect way to give back to a great cause while doing something you love. As soon as you have an idea for something you'd like to do, please contact me at [sandra.stevens@barthsyndrome.org](mailto:sandra.stevens@barthsyndrome.org) and we can talk about how you can start planning.

## Ideas to Get you Started

There are so many simple ways to make a big difference to boys and young men affected by Barth syndrome. Here are some ways you and your friends can make an impact:

Office Jeans Day	Pie Smash	Potluck	Game Night
Bake Sale	Craft Sale	Wishing Well	Trivia
Coin Drive	Movie Night	Cook Off	Lemonade Stand
Car Wash	Picture with Santa, Easter Bunny, etc.		

## If You're Feeling a Bit More Ambitious

Why not open up your fundraising efforts to a larger crowd? Organize a fun event with a truly meaningful mission. Here are some great ways to engage friends, neighborhood residents, co-workers, and more:

Bike-A-Thon	Variety Show	Car Show	Luncheon
Garage Sale	Holiday Gatherings	Fun Run	Read-A-Thon
Flea Market	Toy Drive with Monetary Donation	Fashion Show	Restaurant or Retail Promotion
Auction	Walk-A-Thon	Music Concert	School Fundraiser
Scavenger Hunt	Sports Tournament	Talent Competition	Church Fundraiser
Super Bowl Party	Black-Tie Event	Boat Racing	Dine Out for a Cause
Golf Outing		Dance-A-Thon	

# Involvement in Barth...Returns the Favor!

By Ghent Lummis, Volunteer, Barth Syndrome Foundation

"... The Barth boys and men may not look strong to you, but they carry me every single day of my life."



Ghent taking on the Memorial Hermann Ironman (May 2011) in Texas

saying, "I would love to, but I am going to go run up that mountain". Kate took one look at the mountain and said, "You need to join my team". I knew that her team must involve the Barth Syndrome Foundation, so I immediately agreed. Then I said, "So what are we doing?" Kate said, "Our target event is Ironman Arizona next year." As impossible as that seemed to me, recovery suggested that I undertake any task with faith, dedication, and hard work. With that, anything is possible. I realized I would be 50 years old at the start of this event, with just over a year to prepare — not much time for such an undertaking! Nevertheless, I committed to train as hard as I could, saying that if I could get to the starting line healthy, we would give this thing a try.

It was a very tough year, as I was sick and injured for most of it, but I persevered. The day before the event, I had the pleasure of meeting Will again, and I was impressed with his maturity, intellect, and straightforwardness. The Ironman was a wonderful experience, and I realized that my involvement would likely go a lot longer than one event. Will and his family being there to cheer my teammates and me on was just icing on the cake.



Ghent taking on the Ironman Lake Placid (July 2011)  
(Photos courtesy of Ghent Lummis<sup>2011</sup>)

**M**y involvement with the Barth Syndrome Foundation began with my cousin, Will McCurdy. I first met Will in 1988, in Boston. I was twenty-eight years old and was there to compete in the Boston Marathon. Over twenty years passed before I saw Will again. For all those years, I had no idea of the struggles that Will and his family faced on a daily basis.

One reason that I was oblivious to their situation was our geographic separation, as I lived in Florida and they were in New York. The other reason for my lack of involvement was my own struggle with alcohol and drug addictions, which ravaged my life and kept my world very, very small. In 2007, things got bad enough for me to take steps to face my demons and, as I did so, I made a commitment to get healthy. I took up trail running to facilitate prayer and meditation, and my fitness level improved greatly.

In October of 2008, my first cousins, including Kate McCurdy and her husband, Steve, rendezvoused in Santa Fe for a family reunion. I got up early one morning and headed out for a run, and quickly ran into Kate and her sisters going out for coffee. They invited me to join them, but I declined,

A year and a half later, my outlook had changed somewhat. I was beaten up from my third Ironman and was signed up for a fourth, but I was tired. I was tired of the pain, the financial demands, the lack of a social life, and simply tired of being tired. I decided that I would walk away from the sport of triathlon and mail a check to the Barth Syndrome Foundation for the amount I spent on the sport, and everyone one would be happier. My decision made, I went to retrieve my mail. In it was a letter from a mother of one of the Barth boys in Europe. She thanked me for my involvement, and said that her son followed Team Will over the internet, staying up overnight because of the time difference. She said he cheered whenever one of us crossed the finish line and that he could not understand why people he had never met were working so hard to keep him alive but that it meant the world to him.

I have not been burned out one moment since receiving that letter. I understand that my job is to participate for those who are unable to, and I gladly accept the pain and costs associated with my efforts in that regard. I am constantly grateful for the inspiration these boys and men provide and the way they have changed my life.

The nature of my contributions changes over time. My wife, Ginger, and I contribute time, effort, and finances to the extent that we can. After all, the Barth boys and men may not look strong to you, but they carry me every single day of my life. For that, I am grateful.

# A Grandmother Turns her Pain into a Precious Gift

By Sandra Stevens, Fundraising Project Manager, Barth Syndrome Foundation

**"I could no longer kiss the hurt and make it all better . . . we all grow wiser and I now know and recognize my limitations. And so I do what I can, when I can. And being part of this group is something that I can do."**



**Travis and Moira**  
*(Photo courtesy of Moira's family)*

In March 2013, the Barth Syndrome Foundation received a generous gift from the estate of beloved Moira, well-known member of our community, based in Ontario, Canada. In life, Moira was all too familiar with the terrible reality of Barth syndrome and its devastating effects. Before she passed, she made up her mind to make sure that the work to beat Barth syndrome would continue without her.

In 1958, her first-born son died suddenly, from something then diagnosed as "fibro-elastosis". She was told it was a one-in-a-million occurrence. It's possible that three years later, when her second son was delivered still-born a week before his due-date, she thought it was just another awful stroke of bad luck.

Over 30 years later, in 1995, when her daughter's eight-month-old son fell ill with the same symptoms, it was like a horrible, recurring nightmare. Finally, a geneticist referred to Barth syndrome for the first time. At that point, Moira started to search the internet for information.

Then, she found a posting online from a parent in Australia. At last, she and her family were no longer alone.

They put together their own information packet to show to doctors, most of whom never seen or heard of Barth syndrome.

A heart-wrenching story, all too familiar to many families affected by Barth syndrome.

Moira left a legacy to the Barth Syndrome Foundation, ensuring that families who receive a diagnosis have an easy source of up-to-date information available as well as people to reach out to. Her donation also supports the ongoing work done by researchers and clinicians, to search for treatments and eventually, to beat this horrible disease, once and for all.

Moira has made sure that, after her passing, she can continue to help her daughter and grandson. With her gift, it's possible that they won't need to experience the same pain and suffering that she did. That's a wonderful legacy.

## Why Not Make Your Own Lasting Gift?

It's very easy to make sure the Barth Syndrome Foundation is included in your will. And once that's done, you can rest assured that your gift will live on after you, helping to beat Barth syndrome for good.

For more information on how to make a lasting gift to the Barth Syndrome Foundation, please contact me at [sandra.stevens@barthsyndrome.org](mailto:sandra.stevens@barthsyndrome.org) or toll free 855-662-2784 or 855-NO-BARTH (local line: 914-303-6323) for more details. And you can download a document with simple tips, at [www.barthsyndrome.org/donate](http://www.barthsyndrome.org/donate).

In the words of Moira herself: *"I could no longer kiss the hurt and make it all better . . . we all grow wiser and I now know and recognize my limitations. And so I do what I can, when I can. And being part of this group is something that I can do."*

## You Can Make A Difference

**Donate by check:** Make check payable to Barth Syndrome Foundation, PO Box 582, Gretna, NE 68028

**Donate online:** You can donate to BSF by going to our website, [www.barthsyndrome.org](http://www.barthsyndrome.org), and clicking on the "**DONATE NOW**" link on our home page.

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

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



# In Loving Memory of Colin

Colin was a hero... He taught me so much and continues to do so. Not a day goes by that I don't think about and miss this courageous, strong, caring young man that I had the privilege of calling my baby brother.

Love, Lauren



Colin



Alison, Colin, and Ian



Lauren, Candice, and Colin

When I found out Col had passed away I was stunned and devastated. Then, almost instantly, I had a flash of memories rush in. There were smiles — so many smiles.

I would love to share them all, but that would become a book. So I will keep this short and sweet. Col was many things; happy, kind and gentle, with one hell of a sense of humour. He was a real joker and loved a good old tease!

Col was passionate and loved many things — WWF, gaming, skating, beer, his bearded dragon, animals, and his friends and family. Col and my Mum had an extraordinarily beautiful relationship, such love and devotion.

The best way for me to describe Col is as a shooting star; wonderful, rare, passing too quickly, but while it's here, it's simply spectacular as it lights up the sky.

I would like to end off with this poem, which sums up my feelings about our beautiful Barth Boys, past and present.

Col, my squirt, you will be in our hearts, always.

Love, Candi, your short sister xxx



i carry your heart with me (i carry it in my heart)

i am never without it (anywhere)

i go you go, my dear; and whatever is done

by only me is your doing, my darling)

i fear not fate (for you are my fate, my sweet)

i want no world (for beautiful you are my world, my true)

and it's you are whatever a moon has always meant

and whatever a sun will always sing is you

here is the deepest secret nobody knows  
(here is the root of the root and the bud of the bud  
and the sky of the sky of a tree called life; which grows

higher than soul can hope or mind can hide)

and this is the wonder that's keeping the stars apart

i carry your heart (i carry it in my heart)

E. E. Cummings

(Photos courtesy of Colin's family)

# Opportunity to Participate in Barth Syndrome Research

Dr. Cynthia James and colleagues at Johns Hopkins School of Medicine are conducting a clinical study entitled, "How do women adapt to being a Barth syndrome carrier? A mixed methodological study of psychological adjustment and reproductive options". This study is supported by the Barth Syndrome Foundation through a 2014 BSF Research Grant Award. (See pg. 5 for more details).

## Are You a Barth Syndrome Carrier?

**What is this research study?** As more boys and men are correctly diagnosed with Barth syndrome, more women are finding out they are Barth syndrome carriers. This telephone interview research study explores how Barth syndrome carriers navigate the family, reproductive, and psychological implications of being a carrier. We hope research study results will help both health care providers and patient organizations provide better care to carriers of X-linked conditions, especially Barth syndrome!

**Who can participate?** Any woman, age 18 or older, who is a Barth syndrome carrier.

**What will you ask me to do?** Be interviewed by telephone for about 45-60 minutes. The interviewer will be a health care provider who has worked with families affected by Barth syndrome.

**How do I find out more?** Please call or email us!

Study PI Cindy James at [cjames7@jhmi.edu](mailto:cjames7@jhmi.edu) or 443-287-5985

Study Genetic Counselor Rebecca McClellan at [rmcclel4@jhmi.edu](mailto:rmcclel4@jhmi.edu) or 410-502-2578

**There will be no monetary compensation for participating in this study.**

Dr. Cynthia James, Principal Investigator

Study number (IRB 00052976)

# Cardiolipin as Key Lipid of Mitochondria in Health and Disease — Mini-Meeting, Second Edition 2015

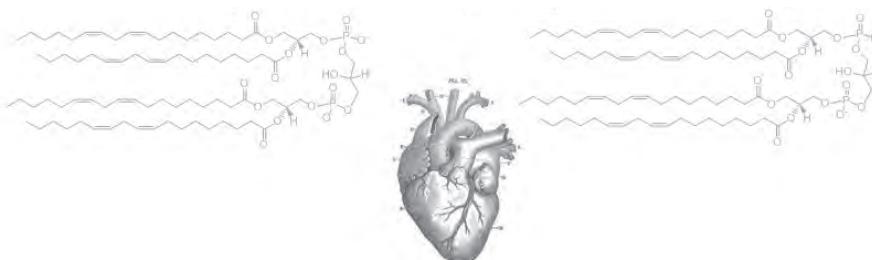
The lipid called cardiolipin is uniquely associated with Barth syndrome because research has shown that it is altered in our boys. Cardiolipin is found only in the powerhouse part of the cell—the mitochondria—and hence it is not unusual to understand that fatigue is one of the symptoms of Barth syndrome. As part of the large 13<sup>th</sup> Euro Fed Lipid Congress being held in Florence, Italy in September, several Barth syndrome researchers, including some of the Barth Syndrome Foundation's (BSF) Scientific and Medical Advisory Board members, will be attending a satellite meeting that focuses just on cardiolipin. BSF is helping to support this meeting. In addition, some of the family members of the Italian affiliate of BSF are also scheduled to be present. Just like the BSF biennial conferences, mixing researchers with family members will provide an enjoyable and rewarding experience for all.



## Cardiolipin as key lipid of mitochondria in health and disease

2<sup>nd</sup> edition, 30 September — 1 October 2015

as Satellite of the 13<sup>th</sup> Euro Fed Lipid Congress in Florence, Italy



# 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 **83** 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](http://www.barthsyndrome.org).

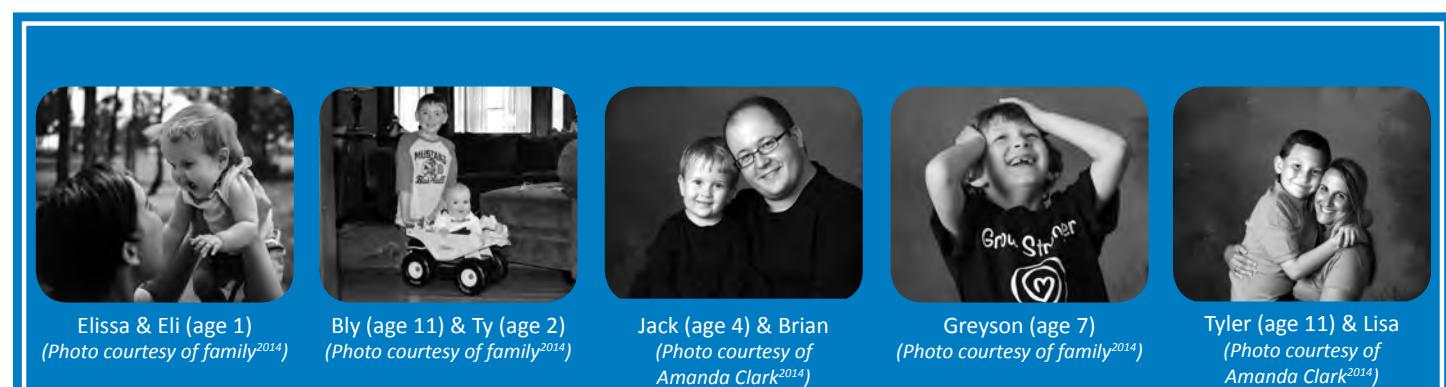
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# Awareness of Barth Syndrome Continues to Grow

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# Passing the Baton

By Michaela Damin, Chair, Barth Syndrome Trust

**"I have had the good fortune to witness from our very own group the wonderful way in which many of our parents have successfully managed the transition process, and I often catch myself thinking 'What would so and so do or say in this situation?' So thank you, Barth family, for your support and collective wisdom."**

**W**hen my boys were little and I was stressing about them, I was often told: Small children, small problems. Big children, big problems.

When you have a young child with Barth syndrome, then you probably have a child who is "small" in stature, but the problems certainly don't feel small. The love, hope, and joy you feel as a parent often seesaw against the counterbalances of anxiety, stress, and frustration. Parenting a child with Barth syndrome means you can never completely relax. You become so invested in your child, and you spend hours learning everything you can about his condition. You can seem overprotective or overbearing, often instinctively crouching into ninja mode at the sound of someone near him daring to sneeze aloud. You carry an arsenal of equipment to try and keep him safe, and you are always, always on duty.



Marco feeding Nick  
while in the ICU in 1998  
(Photos courtesy of Nick's family)

When my son with Barth syndrome was younger, all he needed was me to make everything okay in his world. What a wonderful gift of love and trust, and yet equally terrifying, as I felt that the weight of every decision fell squarely on my shoulders. We didn't always make the best decisions, but we did the best we could with the knowledge we had. As time went by, we eased into the everyday routine of trying to make the most of the complex and wonderful life we had been given. Our little boy had to deal with things we would never have chosen for him, but as long as we were there he was happy and all was well.

## Transition Time

And then our adorable little cherub started to grow up, and every single step he took toward adulthood was a cause for celebration, and a very necessary change started to happen. We were not the only ones he needed in his life to make everything all right. Nick is almost 17, and right now I feel like I'm in a surreal relay race — Nick is ahead of me holding out his hand waiting to grab that baton, and I'm running alongside him holding that baton out. In reality, I have such a firm grip on that baton that the poor guy has no hope of prising it out of my hand. Yet.

It is time to take everything I've learned in the past 16 years and somehow pass it onto my teenage son to give him the knowledge and skills he needs to manage his complex medical condition. We've put so much hard work into keeping him healthy and happy so far, and as he approaches adulthood, we want him to fight just as passionately and diligently for his life. His life, his choices, his needs, and his aspirations.



From cherub to college student

## Big Children — Big Problems

We are right in the middle of this transition to adulthood. We are learning quite a lot in this new phase, and we feel blessed to have come this far. We've learned that having Barth syndrome sometimes really just sucks. That's not quite the polite terminology we used when Nick and I sat in the dark in my car together and screamed and shouted and cried over the fact that, when he was feeling at his worst, he was too exhausted to even make it through watching his friends explode in a display of life, energy, and skill in their youth production of Grease. That was a horrible, horrible moment. A hug, although needed by both of us then, wasn't enough to make everything better. I wished I could give him my energy, my healthy immune system, my heart even. Trying to remind him of how lucky he was compared to some others was of no help to him whatsoever at that point.

## Changes for Both of Us

I am no longer that person who can make everything okay just by being there. My role as a mom has changed. When my kids were little, I tried to shield them from anything that might hurt them too much. I didn't always tell them about the loss of one of their Barth brothers if they were not emotionally ready to deal with it. When Nick and his brother, Matthew, were very young, we explained Nick's defibrillator in simple terms as a kind of ECG machine that looks at your heart beat and fixes it if it's not working properly. That was enough information at the time.

(Cont'd on page 25)

# Passing the Baton

(Cont'd from page 24)

Many years, later I overheard Matthew casually telling a friend that the defibrillator was there to shock Nick's heart if he fell down dead with a heart attack. The little kids grow up, and they know the underlying truth about everything. If you can't find a way to deal with the real issues surrounding their condition in a gentle but honest way, then it just means that, in their efforts to protect you, they are forced to deal with it alone. Dwell on the negative too much, and you risk tipping over into feeling powerless and overwhelmed.

## Asking for Help

At a doctor's appointment, Nick was brave enough to say that he needed some help, and so he had a short course of cognitive behavioural therapy. He was initially very sceptical about psychotherapy. He told me that he was feeling so bad because he had Barth syndrome, and talking about his feelings was not going to help him in any way and would, in fact, probably just make him feel worse. He had a good point. Psychotherapy alone was not going to be enough, but setting the time aside to actively deal with his issues meant that he was doing something. He was taking that first step towards identifying what he could control, and he could finally talk honestly to an objective third party. We also talked about possibly starting him on antidepressants, if needed.

We knew that we were going to have to get creative about rearranging his life to enable him to succeed. We had to radically change the way he participated in school and find ways to reduce his stress. We had seen first-hand how a life which contains stresses that seem to have no end wears you down and leads to a horrible cycle of anger, exhaustion and depression.



Exhibit A

## Seeking Balance

I wanted to find a solution that would let Nick do his chores. (*He snorted aloud while reading this and earmarked it as Exhibit A in his proof of my unusual cruelty*). Let's put things in perspective, here though. Apart from keeping his room "tidy" (*I use the term very loosely*), he only really has one daily chore, and that is to pack the dishwasher. To be honest, the actual dishwasher packing is not what's important. What's important is that I want him to be a functioning part of our family, learning the value of discipline and focus and the joy that comes from being useful. When he was feeling so terrible there was no energy left over to do a single chore.

I wanted him to be happy. Nick has a wonderful sense of humour which has seen him through the darkest of days and which continues to be a life force. For Nick, happiness meant that he needed the time and energy to spend on his friendships which had been eroded away by the demands of life in a high functioning secondary school.

## Further Education

Nick's support staff at school were our guardian angels. They gave him a few days off to rest while they spoke to his teachers, and they worked with us to come up with a plan. They reduced his school day from a 40-hour week (including transport) to a 16-hour week, all while still providing the subject tuition he needed to carry on with further education. Shorter lesson times each day also gave him the daily routine he needed.

Nick will be writing his final school exams next month, and I really hope he manages to get the marks he's hoping for. Missing a lot of school has its drawbacks, and trying to get the right balance is crucial. With hindsight, I think he could have managed a slightly longer school day, but the actual timetable didn't allow for it. He wants to go to college after his GCSE's and do his A levels, and then go onto university to study to be a primary teacher. He hopes to be able to teach part-time, if his condition permits it. If he changes his mind, he has the option of a vocational qualification or perhaps an apprenticeship of some kind, perhaps in the catering arena, as he still loves cooking and baking. He has found the right college for him, which is not the one that all his friends are going to attend; instead he has chosen a much smaller college with a friendly and warm ethos where he feels he will thrive. He has opted to do his A levels on a part-time basis to create a manageable work load. He might take a bit longer to get there, but taking things at his own pace is crucial.

Another major project has involved transitioning Nick from his Statement of Special Educational Needs, which provided him with the support he's needed for his education, to the new Education, Health and Care Plan (EHC). This new Plan will benefit Nick, as it extends help for young people like him to the age of 25, whereas the old Statements ended at the age of 18. The process involved some very careful mapping out of Nick's specific needs, as well as numerous assessments, and I think we have come out with what promises to be a very useful document.

(Cont'd on page 26)

# Passing the Baton

(Cont'd from page 25)



## Independent Living

Our latest challenge was to equip him with a new electric wheelchair, as he outgrew the one he got when he first started secondary school. It took us 10 months and many follow-up calls and emails, but this week he got his new, bigger, and faster chair.

Though Nick is in the middle of what could be seen as the more dangerous "growth spurt years", it is understandable that he also needs more independence. One of the more ingenious devices we managed to get for him was an Oysta mobile phone and alarm device (see picture). He wears this around his neck or keeps it in his pocket when he's out alone or with friends. It has a built-in falls detector should he become unconscious. It also has a button that can be pressed if there is an emergency, and it links straight into the call centre who can send out an ambulance, if needed. The device has built-in GPS to pinpoint his location, and it works anywhere within Europe.

## Further Possibilities

We recently met with a specialist financial advisor to help plan for Nick's future, as we're not sure of how financially independent he will be, especially since the recent changes to our welfare system has meant that he will no longer be eligible for his Disability Living Allowance (DLA). From what we understand, the move to the new Personal Independence Plan (PIP) may well mean that young people affected by conditions like Barth syndrome or chronic fatigue land up being excluded due to the new criteria. As one parent I know stated so well – "Just as they land up entering the most expensive stage of their lives, the financial help disappears and prevents them from being truly independent."



## Under New Management

Nick has started to take a much greater interest in managing his condition himself, which is wonderful to see. All his medication is set out in a pill box, and we can see at a glance if he has taken everything. He has become really good at remembering to take his medication, which lets me take another step back as he takes one forward. He now needs to get back into doing his own GCSF injections, which he stopped doing as he found his hand strength and steadiness were not quite up to the task. I still go with him to all his major hospital appointments, but am taking more of a back seat these days, which has encouraged him to speak up for himself. I have had the good fortune to witness from our very own group the wonderful way in which many of our parents have successfully managed the transition process, and I often catch myself thinking, "What would so and so do or say in this situation?" So thank you, Barth family, for your support and collective wisdom.

Nick displays his pill box and new mobile phone alarm device

I know that our boys and men are often very different in the ways in which they are affected by Barth syndrome, and ours is just one example of one path chosen. I would love to hear from all the other young (and older) men with Barth syndrome to find out about the path you have chosen and how it's working out. What are you able to do with your life, and what lessons have you learned along the way?



Thank you, Barth family, for your support and collective wisdom (Photo courtesy of Alex<sup>2015</sup>)

# Review of the Bristol Clinic and Family Day

By Helen, Mother of Will (age 26), Hampshire, United Kingdom



Making new friends:  
(L-R) Jules (age 6) & Alex (age 5)

The May NHS Barth Syndrome Service Clinic was very enjoyable and well organised. How lovely it was to see old friends and families and to meet and get to know our new ones from Europe and London. I am always amazed at the bond our boys have with each other, and how they seem to pick up from where they left off six months or even a year previously.

Our day started early, and the morning soon went by in a flurry of blood tests, dexa scans, and meetings with the doctors. Despite Debbie Riddiford's careful planning, the timetables were soon forgotten but, nevertheless, everyone was seen by lunchtime and we all regrouped in the playroom at 1pm. Thank you to the hospital catering team who supplied us with a mountain of sandwiches, crisps, and fruit.

With Will being nearly 26, we opted out of the talk on how to swallow pills given by Dr. Vanessa Garratt, but I was pleased that I heard Dr. Colin Steward's presentation on the bezafibrate trial coming up later in the year.

We all disbanded for a rest before dinner. Most of the boys were pretty tired, except Joe, who seemed to have gained his second, or even third wind, so after a quick meal, it was off to bed in readiness for the Family Day on Saturday.

I had never been to the Noah's Ark Zoo Farm before and was surprised at the variety of animals and various activities. The younger children particularly enjoyed all that was on offer, and I was worried that 13-year old Alex was going to be left behind in the Gibbon enclosure after his Mum signed him up to help set up activities for the primates! No doubt we'll hear in due course...

On the dot of one, our pre-booked pizzas arrived, following brilliant planning by Annick and Michaela, and thanks also to Sarah and Dave Bull for bringing along plenty of drinks. Alex (age 20) was our official photographer for the day, and we've included a few of the great photos he took, thank you Alex! (See page 38 for additional photos).

Thanks too, to everyone who contributed to the weekend, Colin, Debbie, and the Team at Bristol, and to Annick for organising the Zoo — here's to the next one!!



Fun for all ages: Joe (age 6) & Alex (age 13)



Dr. Colin Steward is part of our Barth family



Boys at the Family Day (ages 5-25)  
(Photos courtesy of Alex<sup>2015</sup>)

## A quick note from Annick Manton...

As always, it is a privilege to attend the Clinic and to help organise our Family Day. This is a special group of people who are devoted to their families and to each other. Every family who attends has to make special arrangements and it always involves effort, but the benefits more than make up for it. A very special mention goes to the family who drove over from Belgium for the 4<sup>th</sup> year and to the family from Spain who travelled so far and had to brave the language barrier on top of it all. It was heartening to meet a very special sibling, Miriam, who took on the enormous task of acting as an interpreter for her affected brother, Xavier, and her dad. This was a big task for a young woman and she did a great job! She and her family were made to feel welcome by another Barth mum, Ana, who speaks Spanish.

Dr. Steward and the Team at Bristol have become part of our Barth Family and these weekends make for good medicine, great relationships, and happy memories.

# BST Treasurer's Report on 2014

Our fundraising efforts in 2014 were very encouraging, growing by 37% on the previous year. This is a truly magnificent achievement, despite very challenging conditions, and a real tribute to our fundraisers and donors, families and friends; thank you so much for all your efforts and kindness. We love hearing your stories and sharing them with others who also find them inspiring (please see our separate BST *Fundraising News*). Memorial donations continue to add a sizable amount to the total income, keeping alive the memory of boys and loved ones, whilst leaving a legacy of help for others.

Year	31 Dec 2013	31 Dec 2014
Receipts	£23,620	£32,404
Payments	£ 4,462	£22,539
Net	£19,159	£ 9,865
Cash Funds Year End	£49,797	£59,662

We are delighted that as a result of these efforts the Trust can continue to fund a research project here in the UK. In 2013, as part of the BSF Research Grant Program, BST funded a research grant to Douglas Strathdee, PhD, Head of Transgenic Technology, Beatson Institute for Cancer Research, Glasgow, to develop a mouse model of Barth syndrome, Characterisation of a conditional knockout of *tafazzin* in the mouse – Award — US \$49,837 over 2-year period; BST funding of this is continuing.

In 2014, we co-funded two research projects with BSF:

John L. Jefferies, MD, MPH, Director of Advanced Heart Failure, Cincinnati Children's Medical Center, Cincinnati, Ohio, USA: [Assessment of quality of life, anxiety, and depression in Barth syndrome: Expanding the scope of comprehensive care](#) – Total Award—US \$28,749 over 2-year period

Adam Chicco, PhD, Associate Professor, Colorado State University, Fort Collins, Colorado, USA: [Translating murine Taz deficiency to human Barth syndrome: Focus on impaired lipid oxidation](#) – Total Award—US \$49,998 over 1-year period

The abstracts for these awards are listed on pages 5-6 within this journal.

## Bizarre Behaviour for Barth Syndrome

By Steve, Fundraiser, Barth Syndrome Trust



William (age 3) and Emily  
(Photo courtesy of Steve<sup>2015</sup>)

After dipping my toe into the marathon world in September 2013, I discovered a perverse yearning to do more – but how to disguise this bizarre behaviour? I know, I'll tell everyone I'm fundraising for the Barth Syndrome Trust, and I think I got away with it. Final tally for 2014 was 13 marathons for the Trust (plus one other for a prior commitment).

Thanks have to be said to some very generous sponsors, many of whom have chucked money in more than once and especially to my support team (Sharon, Emily and William) who have to put up with constant run chat, calendar fights, mountains of trainers, and some of the smelliest running gear you could imagine.

The support team have been known to appear at some of the races, but I notice they favour the warmer parts of the year and those races that benefit from cafes and beach fronts. William has attempted a couple of jog alongs, but he seems confused as to why Daddy keeps running away from him. Emily is close to being banned from running in with me at the end. She is just a bit too quick, and a 100-metre sprint is not what I need at that point.

Obviously, 2015 requires at least 15 marathons (I think 16 are booked up for so far), but we needed something to round the fundraising off, and I find myself signed up for 5 in 5 at the end of November. The ongoing challenge is to join the 100 Marathon Club and run one hundred 26.2 mile races for the Barth Syndrome Trust.

Goodbye toenails.

<https://www.justgiving.com/RunMore-Cotch-RunMore>

# Alasdair Gray Memorial Fund

By Laura, Mother of Alasdair Gray, Scotland

**"When all you have are memories it becomes very important to keep the memories alive. ... Gone but never forgotten, always in our hearts and on our minds."**



Alasdair

20/10/2013—23/07/2014

Alasdair Gray. Our son and brother to Cameron, Lewis, and Fraser. Alasdair was born 12 days late on the 20<sup>th</sup> of October 2013 weighing 8lb 4oz.

In the early days, Alasdair suffered from acid reflux. He had a couple of chest infections in his first 6 months and was struggling to gain weight. Alasdair was otherwise reaching the expected milestones. Initially, the poor weight gain was put down to not being well when he had the chest infections. The time came for weaning, and Alasdair didn't take to weaning like our other boys had; it was a struggle to get him to take most solids.

We began to think he had food allergies and were referred to the hospital for investigation. After a couple of nights in hospital, we came home pending further tests and results. After just one night at home, Alasdair just wasn't himself, and we decided to return to the hospital with him.

Things were quickly taken out of our hands, as we watched our beautiful baby boy go from sitting up looking at us to passing out and being rushed to theatre with an enlarged heart. Our whole world began to fall apart. Never had we imagined Alasdair was so poorly.

Alasdair had a cardiac arrest and was resuscitated. After hours in theatre he was stabilised enough to transfer him to the Royal Hospital for Sick Children in Glasgow, where he had another cardiac arrest. Despite intensive care, his heart wasn't strong enough, and we had to say goodbye to him. He was just nine months old.

We were initially told that it was possible that a virus had attacked Alasdair's heart. It wasn't until a few months later, results came back saying Alasdair had Barth syndrome. Something we had never heard of before.

When all you have are memories, it becomes very important to keep the memories alive. We want to raise money for charities, raise awareness of Barth syndrome, and most importantly, do something positive in Alasdair's name and keep his memory alive. We want people to know Alasdair was here and remember him.

Alasdair's Dad, Fraser, wanted to start the fundraising and decided he would shave his beard. Alasdair used to love rubbing his hands over his Dad's stubble, and Fraser hadn't been able to shave since he was taken from us in July. A raffle was also organised at Fraser's work. Next, we held a Messy Play day, another raffle, and a cake stall. BST was chosen by the classmates of his brother, Lewis, as one of their charities. The support shown has been amazing. With the help of all who have donated, we have so far made a fantastic £6,373. We have more plans for fundraising and hopefully by the time the next newsletter comes out, we will have more to report on what we are hoping will be an exciting and successful fundraiser. Watch this space!

**Gone but never forgotten, always in our hearts and on our minds.**

Alasdair Leonard Gray



(Photos courtesy of Alasdair's family)



# President's Report

## Barth Syndrome Foundation of Canada

By Susan Hone, President, Barth Syndrome Foundation of Canada



Susan Hone

**B**arth Syndrome Foundation of Canada (BSFCA) had a year of significant change in 2014. The Government of Canada introduced a new Canada Not-for-Profit Corporations Act which every non-profit corporation has to comply with. Highlights of the new Act included rewriting our by-laws and changes to board positions.

The members of the Board have not changed, however some of their roles have. I have been appointed President, and Chris Hope's title is now Secretary/Treasurer. Lynn Elwood and Cathy Ritter are Directors of the corporation.

I would like to take the opportunity to thank Lynn Elwood for her years of service to BSFCA as President. Lynn's knowledge and professionalism helped BSFCA to grow into the successful charity we are today. I am so grateful Lynn will still be on the Board and able to mentor me as I take on the role of President. Cathy Ritter and Chris Hope deserve accolades as well for their invaluable service. Even though titles have changed, the four of us are committed to our mission of "*enhancing the lives and outcomes of Canadian individuals and families affected by Barth syndrome.*"

After ten years of successful golf tournaments, we have decided to change direction in our major fundraising activity. A new fundraiser is in the planning stages and will still be held the second weekend of September. Keep an eye on our new website for details closer to the date.

A new initiative in 2014 was calling our donors to simply say thank you for their contributions to BSFCA. Lois Galbraith and Les Morris undertook to make all the calls, and the results were very positive. We have decided to continue this practice in 2015, with Lois and Les volunteering to make the calls again.

Our annual mail campaign has recently been sent to all our previous donors, families, and friends in Canada. Thanks to Chris Hope for crafting another marvelous letter and getting the letters in the mail.

We are also pleased to be partially funding a new research grant entitled "[Mechanism and role of cardiolipin oxidation and hydrolysis in Barth syndrome](#)". This three year grant is being conducted by Valerian Kagan, PhD, Professor and Vice-Chairman of the University of Pittsburgh. (See pg. 4 for more information.)

As we plan for 2015, I can't help but reflect on our previous years and marvel at how far BSFCA has come as an organization. We pride ourselves on being a 100% volunteer organization and appreciate every volunteer who has joined us in this journey to enhance the lives and outcomes of those affected by Barth syndrome.

## Annual Meeting—Sharing Information and Friendship

By Lynn Elwood, Director, Barth Syndrome Foundation of Canada

On Sunday, April 19, 2015, we held our annual meeting and family outreach. This year, we were able to use the new Jones DesLaurier offices which provided an excellent meeting venue and a comfortable cafeteria area for everyone. Thank you very much to Ian and family for the use of the office and for making us so comfortable and to Phyllis Perkins who brought the traditional best-ever butter tarts that we all enjoyed just before the meeting started.

The meeting was well attended, with 14 people joining in the updates and conversation. President Susan Hone started us off with an understanding of the charity legislation changes and the resulting changes to the BSFCA board and positions. We then had an in-depth presentation and discussion led by Cathy Ritter about the Science & Medicine progress that is so exciting. There were questions and good discussion about the conference, grants, research progress



BSFCA Associate Dinner  
(Photos courtesy of BSFCA 2015)

(Cont'd on page 31)

# Annual Meeting—Sharing Information and Friendship

(Cont'd from page 30)

and upcoming studies into treatment options such as gene therapy, enzyme replacement therapy, and drug therapy. This laid the groundwork for Chris to share our annual report pamphlet (thank you Chris and Paula) and our financial position which Chris demonstrated with the aid of chocolate coins. Lois shared a volunteer update.

Fundraising was a hot topic in this year's meeting. We have been very fortunate to have a donor offer to match donations made this year. This means that every dollar we have donated or fundraised becomes two dollars to the organization, up to \$5,000! This great news, together with enthusiastic volunteers raising ideas, fueled a great discussion on fundraising options for the group and individuals.

At the close of the meeting, the larger group (including six more family and friends) had a fun game of glow-in-the-dark mini golf. The young and less-young enjoyed the chance to laugh and play. There was even a group with three generations — and the true golfers schooled us on how to sink those putts. Dinner at Wendell Clark's followed and gave people a chance to talk and catch up.

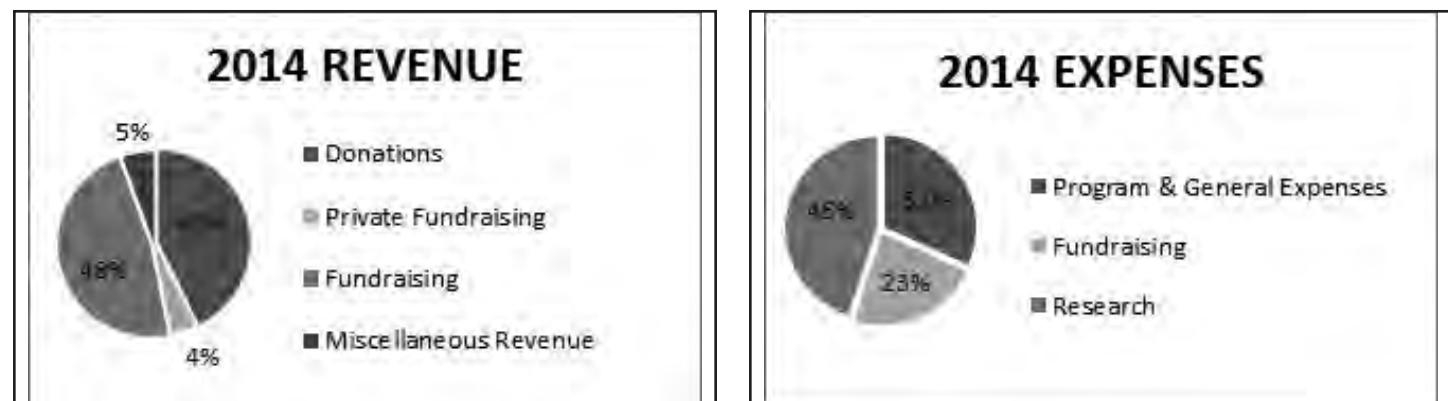
These family gatherings and annual meetings are such a great time for everyone to get together, and we all look forward to them. When Sheldon's Mom indicated they might not be able to come due to work, he promptly suggested that perhaps she should quit her job because this day is important to him. Sheldon and his family were able to get there, and we enjoyed seeing Sheldon's great progress and talking to Jin about some of the daily issues he faces. Robert spent his birthday with us and let us embarrass him a little with a set of balloons. Adam showed his Mom and Omi how to get the job done in mini golf and shared his routine with his Grandma and Grampa. We wish every Barth affected family in Canada could have joined the day, and we thought of you all as we enjoyed the updates, the food, and fun, and most of all, the people.

## Barth Syndrome Foundation of Canada 2014 Financial Report

By Chris Hope, Treasurer, Barth Syndrome Foundation of Canada

We are very happy to say that 2014 was another strong year financially. Thanks to Lois and her army of volunteers, our tenth golf tournament was very successful, and we continue to be fortunate to have wonderful donors who contribute to our Foundation on a regular basis. As usual, we keep a close eye on expenses, and our organization is run entirely by volunteers.

Summary of Financials for the Year Ended December 31, 2014	
Revenue	\$67,000
Operating & Program Expenses	\$33,000
Research Grant Funding	\$27,600
Net Revenue	\$ 5,500



# Focus on Families



## Les Completes 1 km Run

Les Morris (Grandfather of Adam) successfully completed the 1 km Run for Community Care Peterborough. He raised \$1,600 for Community Care "Meals on Wheels" and transportation of clients to appointments. These are both programs that he is involved with in his local community. Hats off to you, Les! (Photo courtesy of BSFCa)

Lynn, Les, & Adam (age 25)



## Sheldon Reunites with Friends

Sheldon thoroughly enjoys attending the annual meeting and outreach with his family and friends. He is fourteen years old and is looking forward to going to high school in the fall. He will be enrolled in an International Baccalaureate program. His quick smile is always a welcome sight! (Photo courtesy of BSFCa)

Sheldon (age 14)



Jessica & Ryan (age 24)

## Update from the Ritter Family

The summer of 2015 promises to be a busy time, full of planning and packing as "changes of addresses" are in the works. Cathy is already stocking up on Kleenex®; Chris is making plans for the empty bedrooms!

Ryan (24, BTHS) and his girlfriend, Jessica, will be moving to London, Ontario where Jess will be starting an undergraduate degree program. Ryan will use his chef skills to further his culinary adventures and expand his horizons. The two of them are busy making plans for their first home-away-from-home.

Ryan's older brother, Andrew, has accepted a position at an International Baccalaureate school in Dubai, UAE. He will be teaching History and World View to grades 7-10. Andrew will be able to continue his passion for adventure and travel... and has already broken the news to his parents that he won't be coming home for Christmas but instead, will be travelling to China to be with some of his university friends who are teaching there.

Thankfully, for grandma and grandpa, the only "change of address" in the works for Abbie and Noah (and their parents) is during trips to the cottage for adventures in swimming and exploring. One thing is for certain: we will all become proficient using Skype and Facetime! (Photo courtesy of Ritter family)

# First French Clinic

By Florence Mannes, Chair, Association Barth France



Brian, Raphaël, and Ethan have fun while getting to know each other



Q & A with the doctors during lunch time



Families enjoying dinner together on the River Seine  
(Photos courtesy of Barth France<sup>2015</sup>)

In 2010, we, as parents of a Barth boy, attended our very first conference in Florida. The next year, we also attended the NHS Barth Syndrome Service clinic in Bristol, England. As Barth France started to grow, we were happy to be able to fund research projects, but we felt that we could do more for French families.

In 2012, Charlotte Rigault and Jean Donadieu, two doctors with a high involvement with Barth France, published an article on the French Barth population, highlighting that there were 11 living French Barth boys.

In September 2013, the Bristol clinic team, together with Michaela Damin, came to Paris to share with the French doctors who are involved with Barth France their experience in organizing a clinic. We were convinced it would be useful for these 11 families to meet other families and doctors who know about Barth syndrome and to learn more about the disease.

At last a date was set for the French Barth Syndrome Clinic in Paris at the Hospital Necker for Sick Children on January 11, 2015.

Out of 11 boys, seven French families attended the Clinic, along with two Belgian French speaking families. It was more than we expected and more than the doctors were prepared for.

The morning was planned for private consultations. The boys could have an appointment with each of the doctors they felt they needed to meet. Appointments were set with a cardiologist, a hematologist, a geneticist, a doctor specializing in muscle function, and a doctor specializing in metabolic disease, each of them having a very good knowledge of Barth syndrome.

After a lunch shared with the families and the doctors, a nutritionist came to explain the main facts on nutrition for Barth boys. This was a start for Q and A and discussions on the different aspects of the disease.

Because we wanted this gathering to be more than the medical clinic, Barth France also invited the families to have dinner on the River Seine so that all the Barth boys could begin to know each other. It was great to see the teenagers sharing their experiences with younger ones, and it was fun to see the boys getting along.

It seems that the families will benefit from good medical follow-up on a regular basis. According to the doctors that saw the boys during the clinic and also from family feedback, what is needed is more education about the syndrome, more exchange with their peers, and more time to ask questions of the doctors.

In response to this feedback, it has been decided, together with the team of doctors of Necker Sick Children Hospital and Trousseau Hospital, that the next clinic will be planned for October 12<sup>th</sup>, with a slightly different format, allowing more time to educate boys and families. We really hope this will be even more successful than the first clinic.

We really have to thank the "Total Foundation" who funded this clinic (transportation, accommodation, lunch and dinner, medical examinations, ...).

# Charity Dinner to Support Barth France

By Florence Marnes, Chair, Association Barth France



Professor Damien Bonnet talking about Barth syndrome

In 2014, the Rotary Club of Orange (South of France) organized a charity dinner, the proceeds of which were given to Barth France and to another local charity. As this area in France is the leading producer of black truffles, the menu of the dinner was designed around the black truffle. More than 400 people attended this event. EUR 5000 were donated to Barth France.

On our way back home, we decided that it would be a great idea to host such an event in Paris, although we were aware that it would involve a lot of work. Organizing a charity dinner means dealing with several tasks, from finding a location, to hiring glasses and napkins, finding a cook and assistants, waiters, finding the truffles, the wine, ....

As the Chairman of Barth France, and having organized many events for the association, I was a bit afraid by the amount of work that the project would require...but my father-in-law was really enthusiastic about it and said he would take care of everything... which he did!

Thanks to the generosity of many of our friends and relatives, all the proceeds from the dinner were straight benefits, as many of the goods were donated to Barth France: the wine, the black truffles, the bread and dessert, and the hire of the location. Additionally, a very good friend of the family, who is a chef, also agreed to cook for this dinner and brought his team with him to help for the evening.

After the welcoming drink, the appetizer was served. It was a French paté, filled with black truffle. As the main course, our chef had prepared black truffle omelette, followed by a French cheese (Brie), also filled with black truffle. Chocolate cake was served for dessert. Barth France also organized an auction during this evening, selling some wine, Armagnac, and truffles that had been donated by friends.

Professor Damien Bonnet, who is a cardiologist with a very good knowledge of Barth syndrome, also came to talk about the disease.

In the end, 140 people attended the dinner, and the proceeds for Barth France were EUR 17.000.

Thanks to all our friends' generosity and dedication, this event was a real success, and we hope to be able to organize another charity dinner next year, in a bigger place, so that we can welcome more people.



Hervé, the Chef, leading his team preparing dinner



Guests socializing at charity dinner  
(Photos courtesy of Barth France<sup>2015</sup>)

# Power of Kindness



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(L-R) Hoi & Loong (age 2)



(L-R) Nick (age 16), Dillon (age 10), Ieuan (age 17) & Joe (age 6)



Nick, Tracy & Alex (age 20)

(Photos courtesy of Alex 2015)



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 and Shelley Bowen



Jared (age 21)



Ryan (age 24)



Adam (age 25)



Robert (age 29)



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## Barth syndrome (BTHS; OMIM #302060)

A rare, serious, genetic disorder primarily affecting males. It is found across different ethnicities and is caused by a mutation in the *tafazzin* gene (TAZ, also called G4.5), resulting in a complex inborn error of 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** (*chronic, cyclic, or intermittent*)
- **Underdeveloped skeletal musculature and muscle weakness**
- **Growth delay** (*growth pattern similar to but often more severe than constitutional growth delay*)
- **Exercise intolerance**
- **3-methylglutaconic aciduria** (*typically a 5- to 20-fold increase*)
- **Cardiolipin abnormalities**

## Save the Date!

### Barth Syndrome 8<sup>th</sup> International Scientific, Medical & Family Conference

July 18—23, 2016  
Hilton Clearwater Beach Resort  
Clearwater, FL



For more information, please visit Barth Syndrome Foundation's website:  
[www.barthsyndrome.org](http://www.barthsyndrome.org)