

# Barth Syndrome Journal

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Fall 2012

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

## Impressive Progress Revealed at 2012 BSF Conference ~ My Room with a View

By Matthew J. Toth, PhD, BSF Science Director



Matthew J. Toth, PhD  
BSF Science Director  
(Photo courtesy of BSF ~ 2010)

The Barth Syndrome Foundation's (BSF) 2012 Conference was the fourth I have attended and the third I have helped to plan. Several BSF volunteers and I focused on making the Science and Medicine (SciMed) sessions of the Conference (Thursday and Friday, June 28<sup>th</sup>-29<sup>th</sup>) the best possible. I think that we can claim success. Many scientists and physicians who attended the sessions were honestly enthusiastic about the experience, and they are a group that is not easy to impress. This year, many family members attended these SciMed talks and the Poster Session, which can be considered "dry material" for the non-scientist. It is this material that I want to talk about for those who may not have had the opportunity to listen to the speakers or may have overlooked some of the truly remarkable insights the speakers provided. Thankfully, the audio recordings of almost all of the speakers' presentations are available on BSF's website (International Conferences >> 2012 International Scientific, Medical & Family Conference) along with their slides that were shown. I encourage you to take some time to review them and see/hear for yourself.

For 2012, we had the privilege of devoting an entire morning to talks about the clinical nature of Barth syndrome. *(Cont'd on page 4)*

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Barth Syndrome Foundation 2012 Conference attendees gathered in St. Pete Beach, FL to learn/share the latest research findings of Barth syndrome. *(Photo courtesy of Amanda Clark ~ 2012)*



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**Matthew J. Toth, PhD - *ex-officio***  
Science Director, Barth Syndrome  
Foundation, Inc.  
E-mail: matthew.toth@barthsyndrome.org  
Skype Name: matt\_bsf

# Letter from the Executive Director

## A Recipe for Success

By Lindsay B. Groff, MBA, Executive Director, Barth Syndrome Foundation



Lindsay and Charlotte  
(Photo courtesy of BSF ~ 2011)

In my family, spending time together centers around food. American Thanksgiving offers the perfect example of coming together to enjoy each other's company, as well as a delicious feast. As the host, proper planning is critical. Coordinating the menu, shopping for ingredients, and properly timing the side dishes requires strategy. In our household, we honor traditional recipes with the turkey, mashed potatoes, stuffing, and my grandmother's famous coleslaw. We also allow room for new recipes like butternut squash soup or garlic roasted Brussels sprouts. Sure, we endure minor problems along the way — the biscuits get burned, or the fancy tablecloth goes missing. In the heat of cooking this elaborate meal, I often wonder why I volunteered for such a large undertaking.

However, once our family sits down together, we smile at the beautiful spread, comforted by the familiarity and intrigued by the new additions. We share with each other our gratitude before we dive into our meal.

Creating a strategic plan is quite similar to preparing a Thanksgiving feast. Proper planning is imperative in both cases. In fact, the more planning on the front-end, the smoother the process goes. Like coordinating the menu, Barth Syndrome Foundation (BSF) must coordinate the resources within our community to pull the plan together. Similar to the use of traditional recipes at Thanksgiving, we consider BSF's rich history before making drastic changes; however, new ideas are encouraged. We certainly anticipate a few bumps in the road during the process, but just like we refuse to allow those burned biscuits to ruin the holiday; a few minor setbacks won't thwart a well devised plan. In the end, strategic planning involves teamwork, coordination, and an eye toward the future. After all, we have to plan to be sure we have room for dessert!

Thank you,

Lindsay Groff, MBA  
Executive Director

Note: BSF will host a strategic planning meeting this January and will share the results with the entire community in early 2013.

### Why Should Organizations Plan<sup>1</sup>?

Strategic planning provides enormous benefits. It can:

- Bring clarity and agreement on mission and vision
- Help organizations prepare for the future
- Help organizations anticipate and manage change
- Improve the decision-making processes
- Promote effective stewardship
- Align the board and staff
- Provide an opportunity to recommit to the cause
- Educate participants about institutional history
- Identify existing strengths in the organization
- Provide an opportunity to analyze the organization's systems and processes
- Reinforce the need to commit to continuous improvement

<sup>1</sup>Driving Strategic Planning: A Nonprofit Executive's Guide  
By Sue Waechter [Board Source]

# Barth Registry & Repository 2.0

## A Work in Progress!

By Lindsay B. Groff, MBA, Executive Director, Barth Syndrome Foundation

Much progress has been made during the transition of the **Barth Registry & Repository (BRR)** from the first version into the newest iteration, affectionately called BRR 2.0. As announced in June, Barth Syndrome Foundation (BSF) has been selected to participate in a two-year pilot program of the National Institutes of Health called the Global Rare Disease Registry and Data Repository (GRDR). The GRDR program will collect anonymized patient health information from participating registries established by the individual rare disease organizations in a way that will allow analyses of data across many rare diseases as well as facilitation of clinical trials and other studies.

Those of you who attended the 2012 Conference may have met with Vanessa Rangel Miller of PatientCrossroads. Vanessa explained the benefits of the GRDR, how it will function, and BSF's integral role within the program. Since the announcement of our acceptance into this important and innovative program, the BRR Committee has accomplished several key items including the updating of the questions that will capture the data for the registry (including some questions used in the Severe Chronic Neutropenia International Registry [SCNIR] to make collaborative investigations on neutropenia easier) and the transferring of Institutional Review Board oversight from the University of Florida and Boston Children's Hospital to the Western Institutional Review Board (WIRB).



To add to the excitement, just as the BRR 1.0 winds down, a new publication has been released entitled, **"The Barth Syndrome Registry: Distinguishing disease characteristics and growth data from a longitudinal study."** (Am J Med Genet A. 2012 Oct 8. doi: 10.1002/ajmg.a.35609. [Epub ahead of print]) This paper, co-authored by Amy E. Roberts, Connie Nixon, Colin G. Steward, Kimberly Gauvreau, Melissa Maisenbacher, Matthew Fletcher, Judith Geva, Barry J. Byrne and Carolyn T. Spencer, is an important addition to the field of Barth syndrome clinical knowledge. As the title states, the data collected by the BRR are presented for all to reference. Publications like this allow researchers to use the information collected in the BRR to advance our understanding and to help find specific treatments for Barth syndrome.

### Barth Syndrome Foundation, Inc.

PO Box 618  
Larchmont, New York 10538  
Telephone: (850) 273-6947  
Fax: (518) 213-4061  
E-mail: bsinfo@barthsyndrome.org  
Website: www.barthsyndrome.org

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### AFFILIATES

**Barth Syndrome Trust (UK & Europe)**  
1 The Vikings  
Romsey  
Hampshire  
SO51 5RG  
United Kingdom  
Telephone: +44(0)1794 518785  
E-mail: info@barthsyndrome.org.uk  
Website: www.barthsyndrome.org.uk

### Trustees

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Nigel Moore, Trustee  
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**Barth Syndrome Foundation of Canada**  
1550 Kingston Road, Suite 1429  
Pickering, ON L1V 6W9  
Canada  
Telephone: (905) 426-9126  
E-mail: inquiries@barthsyndrome.ca  
Website: www.barthsyndrome.ca

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### Barth Trust of South Africa

49 Abelia Road  
Kloof, Pinetown  
3610 Natal  
South Africa  
Telephone: 082-465-1965  
jeannette.thorpe@barthsyndrome.org/  
South\_Africa.html

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12, rue Lalo  
75116 Paris  
France  
Telephone: +33 1 45 00 86 12  
E-mail: associationbarthfrance@orange.fr  
Website: http://www.barthfrance.com

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**"We are excited about the GRDR pilot program and delighted that BSF is on the leading edge as a participant."** ~ Yaffa Rubinstein

PhD, Director of Patient Resources for Clinical and Translational Research, Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS), National Institutes of Health.

For more information, go to Global Rare Diseases Patient Registry and Data Repository ([grdr.info](http://grdr.info)).

# Impressive Progress Revealed at 2012 BSF Conference ~ My Room with a View

(Cont'd from page 1)

## Clinical Aspects & Epidemiology of Barth Syndrome

On Thursday morning, **Dr. Richard Kelley** (Kennedy Krieger Institute and Johns Hopkins University, Baltimore, MD) spoke about the biochemical dysfunction which he believes to be the cause of many of the symptoms of Barth syndrome. Though I communicate with Dr. Kelley frequently, and I understand his ideas (I think!), he always teaches me something new and remarkable every time he speaks. Dr. Kelley's lecture is packed with insights that come from spending a long medical career caring for special people. **Dr. Colin Steward** (Bristol Royal Hospital for Children, Bristol, UK) reported about his experience with the National Health Service Barth Syndrome Clinic in Bristol, England. I am a bit envious (in a good way) of the support that Dr. Steward and colleagues working with the Barth Syndrome Trust have garnered to establish this important service. Probably the most strident voice telling the medical world that physicians should be considering the diagnosis of Barth syndrome more often, Dr. Steward has put together a team that shows what can be done by a few dedicated clinicians. Establishing routine medical evaluations at the Bristol Barth Syndrome Clinic allows new and better information to be collected about Barth syndrome, advances clinical care for the group as a whole, and of course, directly addresses the needs of Barth syndrome individuals and their families. **Dr. W. Todd Cade** (Washington University School of Medicine, St. Louis, MO) presented results of clinical studies he has been doing and provided a preview of his latest NIH-sponsored study — a study for which the BSF community volunteered its support in recruiting participants. Dr. Cade is one of the few researchers performing clinical studies or clinical trials with Barth syndrome individuals. This is precisely the sort of research that the BSF community needs to keep supporting because true medical advancements are made this way. **Dr. John Lynn Jefferies** (Cincinnati Children's Medical Center, Cincinnati, OH) joined us on Friday morning when he discussed the serious cardiology problems faced by our boys/young men, particularly left ventricular non-compaction. He presented the recent experience of a critically ill Barth syndrome individual who needed the mechanical circulatory support of the Berlin Heart (read the latest article in the news: Ft. Wright baby fights for life and new heart). Cardiology is always a "front line" issue for our community, and we are very fortunate to rely on Dr. Jefferies and the healthcare group at Cincinnati Children's Medical Center with its outstanding Pediatric Cardiology Department.

Less familiar to the nonscientist but critically important to our better understanding of Barth syndrome, were the biochemical talks on Thursday afternoon. These presentations dealt with *tafazzin* — the gene whose dysfunction leads to Barth syndrome — and the other genes, proteins, and lipids that affect or are affected by *tafazzin*.

## Biological Function of *Tafazzin* and Cardiolipin

**Dr. Steven Claypool** (Johns Hopkins University, Baltimore, MD) detailed the perplexing position of the *tafazzin* protein in the mitochondria when one tries to explain how it modifies cardiolipin — the lipid biochemically linked with Barth syndrome. **Dr. Miriam Greenberg** (Wayne State University, Detroit, MI) showed how cardiolipin defects in yeast affect iron biosynthesis and how this may impact energy metabolism. **Dr. Frédéric Vaz** (Academic Medical Center, Amsterdam, The Netherlands) described another lipid disease with 3-methylglutaconic aciduria called MEGDEL syndrome. **Dr. Michael Schlame** (New York University, New York, NY) presented a revolutionary hypothesis for explaining the enzymatic mechanism of *tafazzin*. This mechanism postulates that the *tafazzin* enzyme only works in areas of the mitochondrial inner membrane that are in very tight curvature (non-bilayer) — something that is rarely present. This amazing idea (yes, very dry stuff but if you are into this sort of thing, very exciting) explains why/how we need the *tafazzin* enzyme in the first place. **Dr. Christopher McMaster** (Dalhousie University, Halifax, Canada) showed that two sets of genes, one involving mitochondrial recycling and the other arginine production, also impact *tafazzin* function. As Dr. Kelley pointed out in his presentation, arginine levels are low in Barth syndrome individuals, so here is an example of the linkage between yeast and the human disorder at a very basic level.

## Mouse Model of Barth Syndrome

The Friday morning presentations focused on what we have learned from the mouse model of Barth syndrome. BSF members should already know that this mouse model was provided by BSF and is available to any interested researcher via Jackson Laboratories. I was especially excited when **Dr. Colin Phoon** (New York University, New York, NY) showed the parallels of this mouse model to the pathologies we see in Barth syndrome individuals. Dr. Phoon showed that these mice can develop a non-compaction in their hearts at the earliest stages of life which is very similar to the human condition, and he provided data implicating reactive oxygen species as a factor. Animal models of human diseases are not usually this similar to the human condition, and Dr. Phoon's work demonstrated the scientific/medical value of this mouse model. **Dr. Zaza Khuchua** (Children's Hospital Medical Center, Cincinnati, OH) revealed how the mouse model resembles Barth syndrome in the way the body uses food as fuel. This is a very striking parallel to what Dr. Cade and others have shown through their exercise studies with the boys/young men. Dr. Khuchua explained that the mouse model shows signs of fatty acid oxidation impairment. **Meghan Soustek** (University of Florida, Gainesville, FL) who will obtain her PhD very soon, spoke about how the mouse model displays defects in its mitochondrial function, and she explored the intriguing idea of using gene therapy to correct the disorder in the mouse model and eventually in humans. **Dr. Michael Kiebish** (Berg Diagnostics, Omics Division, Natick, MA) showed that by breeding the Barth syndrome mouse model with mouse models of other genetic diseases, we can better understand how lipids like cardiolipin play a role in mitochondrial dysfunction. **Dr. Adam Chicco** (Colorado State University, Fort Collins, CO) explained that dietary linoleic acid supplementation or thyroxine treatment of the mouse model improved the cardiolipin abnormality but did not remedy its mitochondrial problems. These mitochondrial problems seem to concern energy utilization rather than respiratory chain dysfunction.

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In attendance at BSF's 2012 Conference were a record number of boys/young men diagnosed with Barth syndrome. They enjoyed spending an entire week together building on friendships and just being "normal." (Photo courtesy of Amanda Clark ~ 2012)

### Mitochondrial Physiology of Barth Syndrome

Friday afternoon provided another chance to hear about some spectacular advances in how we look at Barth syndrome at the cellular level. **Dr. Mindong Ren** (New York University School of Medicine, New York, NY) proposed the use of a pharmaceutical that has the interesting property of reversing some of the cardiolipin defects that are the unique sign of Barth syndrome. This is an exciting development and represents the first pharmaceutical approach towards treating Barth syndrome. **Dr. Grant Hatch** (University of Manitoba, Winnipeg, Canada) spoke about how a gene involved with fat-burning may compensate for *tafazzin* deficiency. **Dr. William Pu** (Boston Children's Hospital, Boston, MA) presented on the unique energetics of Barth syndrome cells using state-of-the-art technology. In addition, Dr. Pu showed how he developed two induced pluripotent stem cell lines (iPS cells) from the skin cells of two Barth syndrome individuals and how he treated these cells with chemical compounds to determine any positive effects. The iPS cell technology is an exciting and powerful technology whose inventors were awarded the recent 2012 Nobel Prize in Medicine. **Dr. Anton de Kroon** (Utrecht University, Utrecht, The Netherlands) spoke about how the *tafazzin* enzyme uses certain lipids to remodel cardiolipin. **Dr. Yuguang (Roger) Shi** (Penn State University College of Medicine, Hershey, PA) spoke about how cardiolipin damage in the mitochondria can signal the problems associated with aging or diabetes and how genes similar to *tafazzin* can alter these processes.

In addition, there was an extensive and well-attended Poster Session on Thursday evening, and, for the first time, two poster presenters were selected to speak at the meeting on Friday afternoon. The lectures and poster presentations were exciting because of the new information they provided. Very revealing were the Question and Answer periods at the end of the lectures as well as the brainstorming or summary periods. The **Brainstorming and Conference Wrap-Up** discussion on Friday afternoon was remarkable and lasted over an hour. The discussions and collegial interactions among the participants and the families are an important part of this international Conference.

### Future Exciting Directions

On Saturday morning, BSF's International Scientific and Medical Advisory Board met to discuss many important issues including how to develop Dr. Ren's idea of a pharmaceutical treatment for Barth syndrome. There are many take-home messages from BSF's 2012 Conference, but the one I want to leave you with is that **we are constantly moving towards a better understanding of and treatments for Barth syndrome**. Poignantly, the SciMed sessions closed with a video clip of Ben Thorpe from the 2010 Conference, thanking the scientists and physicians for their hard work and encouraging them to keep on trying. I think Ben would have been pleased.



Photo of Matt Toth introducing Ben Thorpe at BSF's 2010 International Conference. (Photo courtesy of BSF ~ 2010)

# Barth Clinic at Kennedy Krieger Institute (KKI)

By Hilary Vernon, MD, PhD, Johns Hopkins University School of Medicine, Baltimore, MD

"I am delighted and honored for the opportunity to participate in the new Barth Syndrome Clinic. Neutropenia is a condition that we see frequently in our Pediatric Hematology Clinic, but the appropriate guidelines for management may depend on many factors including the underlying cause and individual risks. We know that neutropenia is common in people with Barth syndrome, and we look forward to working with families to provide guidance and information and also learn from their experiences." ~ Cliff Takemoto, MD



Dr. Hilary Vernon and Dr. Cliff Takemoto at the first-ever Barth Syndrome Clinic at Kennedy Krieger Institute, Baltimore, MD. (Photos courtesy of BSF ~ 2012)

The first **Barth Syndrome Clinic at KKI** was held on Tuesday, September 4, 2012. Two families attended the interdisciplinary clinic. The medical team consisted of Dr. Richard Kelley and Dr. Hilary Vernon (Metabolism), Rebecca McClellan (Genetic Counseling), Dr. Reid Thompson (Cardiology), Dr. Cliff Takemoto (Hematology), Brittany DeCroes, PT, DPT (Physical Therapy), Jackie Krick, MSN, RD, LDN and Patricia Miller, MS, RD, CSP, LDN (Nutrition). These practitioners are all dedicated to helping those affected with Barth syndrome. From the clinicians' perspectives, the first clinic was a success, and each Barth family has since received a full summary letter detailing their visit. The upcoming December 4, 2012 clinic will offer a full schedule with five families in attendance. In addition to the clinic itself, the participating families, medical practitioners and BSF staff plan to enjoy dinner and social time together the night before.

For more information about the clinics, call (443) 923-2783 and ask for Rebecca McClellan or email [mcclellan@kennedykrieger.org](mailto:mcclellan@kennedykrieger.org). Future clinics are planned for March 5, 2013 and June 4, 2013. Space is limited, but still available.

## Exciting Expansion of our International Scientific and Medical Advisory Board

By Kate McCurdy, BSF Board Member; ex-officio member of BSF Scientific & Medical Advisory Board



The Barth Syndrome Foundation's (BSF) international Scientific and Medical Advisory Board (SMAB) is a wonderful and dedicated team of researchers and physicians who generously donate their time and expertise to our mission. Without them, we would not be able to review grant applications with multi-dimensional expertise or write medically-approved educational materials about Barth syndrome (BTHS), a very complicated disorder. These eminent scientists and doctors are central to our goals and our operation. We try hard not to burden them with projects that we can manage ourselves, but we rely on them heavily.

The SMAB really is a team. Many of its members have worked collaboratively on BTHS projects and fully understand the tremendous benefits of what can be gained when several different perspectives and/or fields of expertise are focused simultaneously on a given issue or a brainstorming discussion. Our SMAB leads the way for many of our researchers who see the rewards that come from comparing notes between animal models or from varied approaches to a common puzzle.

For a number of years now, we have kept this group at about 13 voting members, and it has worked very well. We sense that we are on the precipice of some really exciting new chapters in the story of developing treatments and a cure for BTHS. So we have decided to add some new members to expand our expertise. I am incredibly pleased to introduce them to you here (though most of these highly respected experts need very little introduction to many reading this newsletter). Our new SMAB members, who each have agreed to serve for a four-year term, include:

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(Cont'd from page 6)



**W. Todd Cade, PT, PhD, Assistant Professor of Physical Therapy and Internal Medicine, Washington University School of Medicine, St. Louis, MO, USA**

Dr. Cade's research interests include mechanisms and treatments for whole-body and myocardial nutrient metabolism abnormalities in various metabolic diseases. He has been involved with BSF since 2006 when he first attended our international conference to participate in the clinical consultation sessions with Dr. Barry Byrne and Dr. Carolyn Spencer. Dr. Cade is very important to BSF, since his background in exercise biology makes him uniquely qualified to perform certain clinical studies with BTHS individuals. Dr. Cade received a prestigious National Institutes of Health (NIH) R01 award in 2012 and also has been awarded three BSF grants for projects pertaining to exercise training and nutrient metabolism.

Dr. Cade earned a PhD in Rehabilitation Science/Applied Physiology from the University of Maryland, a BSc in Kinesiological Science from the University of Maryland, an MSc in Physical Therapy from the University of Miami, Florida and is a licensed physical therapist. He completed a post-doctoral fellowship at Washington University School of Medicine, and holds an NIH-funded Career Development Award from the NIH. Dr. Cade is a forward-looking researcher who has been, and will be, an important player in our efforts to perform clinical studies with BTHS individuals. His wife, Ashley, has become a strong BSF volunteer as well.



**William T. Pu, MD, Associate Professor, Harvard Medical School; Department of Cardiology, Boston Children's Hospital; Principal Faculty, Harvard Stem Cell Institute, Boston, MA, USA**

Dr. Pu is a pediatric cardiologist who focuses on basic research to understand the regulation of heart development and function. Dr. Pu also uses induced pluripotent stem cell (iPS) technology to understand how gene mutations can prevent the heart from growing normally, causing congenital heart defects. His lab is seeking to reprogram skin cells from patients with specific heart malformations to create iPS cells which then can be used to create heart muscle cells. This way, researchers can observe how heart diseases develop, from beginning to end and better understand how certain conditions started as well as how they may be able to be treated. At the 2012 BSF biennial conference, Dr. Pu showed his success in developing iPS cells from two BTHS individuals — unique achievements that have been submitted for publication. Importantly, he is in the process of donating these very valuable cell lines to the Barth Registry and Repository (see pg. 3) so that any researcher can access them.

Dr. Pu is Board Certified in Pediatrics and Pediatric Cardiology. He has been awarded two research grants from BSF for projects involving iPS cells and metabolite abnormalities in cardiomyocytes. In addition, he is well-funded from the NIH, receiving two concurrent R01 awards, part of an RC1 award, and an R21 award. Dr. Pu's clinical background and his performance at the highest levels of biological research make him a wonderful addition to the SMAB.



**Mindong Ren, PhD, Associate Professor, Department of Cell Biology, New York University School of Medicine, New York, NY, USA**

Dr. Ren came to BSF as a scientist with a background in the fruit-fly genetic model system and cellular biology. His seminal paper on the fruit-fly model of BTHS provided BSF with increased exposure to many varied scientific disciplines — an important step in expanding BTHS research. Dr. Ren's research interests include the role of cardiolipin in health and disease and the pathogenic mechanism of BTHS. Dr. Ren and colleagues at New York University School of Medicine recently characterized the inducible *tafazzin*-knockdown transgenic mouse model and showed that it recapitulates all the salient features of the cardiomyopathy in BTHS (Phoon CKL, Acehan D, Schlame M, Stokes DL, Edelman-Novemsky I, Yu D, Xu Y, Viswanathan N, Ren M. **Tafazzin knockdown in mice leads to a developmental cardiomyopathy with early diastolic dysfunction preceding myocardial noncompaction.** J Am Heart Assoc 2012, 1:jah3-e000455). Now, he is carrying out systematic investigations of the pathogenic mechanism of BTHS-associated cardiomyopathy in this model. These will be significant because new understanding gained will not only elucidate the pathogenesis of BTHS but also shed light on the mechanisms of common diseases such as diabetic cardiomyopathy and heart failure, where cardiolipin deficiency has been implicated as an important contributing factor.

Dr. Ren's research on BTHS has been funded by the United Mitochondrial Disease Foundation and the NIH. In addition, Dr. Ren has been awarded three BSF research grants for work on the BTHS drosophila model, genetic suppressors and potential drug repositioning. He has attended our international conferences since 2006. He brought his two sons along with him in 2012, so that they could gain some first-hand knowledge about the important work their father does.



**Arnold W. Strauss, MD, BK Rachford Professor and Chair, Department of Pediatrics, University of Cincinnati College of Medicine; Director, Cincinnati Children's Research Foundation; Chief Medical Officer, Cincinnati Children's Hospital Medical Center; Professor, UC Department of Pediatrics, Cincinnati Children's Hospital, Cincinnati, OH, USA**

Dr. Strauss is a distinguished pediatric cardiologist, scientist, educator and leader. Prior to his arrival at Cincinnati, he was the Chairman of the Department of Pediatrics at the Vanderbilt University School of Medicine and Medical Director of the Monroe Carell Jr. Children's Hospital at Vanderbilt, a position he held from 2000 to 2007. Under his leadership, the university built and opened a new hospital for children, expanded its pediatric faculty, and increased grant funding for pediatric research. Earlier, from 1981 to 2000, Dr. Strauss was Director of the Division of Pediatric Cardiology at Washington University/St. Louis Children's Hospital.

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# Exciting Expansion of our International Scientific and Medical Advisory Board

(Cont'd from page 7)

A respected scientist, Dr. Strauss's research focuses on understanding the molecular basis of disorders of mitochondrial fatty acid oxidation and genetic causes of congenital heart disease and cardiomyopathies. He is the recipient of two of the most prestigious awards in research: the American Heart Association's Basic Science Research Award for groundbreaking work that led to finding genetic defects that can cause heart failure and sudden death in infants and children and the E. Mead Johnson Award for Excellence in Pediatric Research. Dr. Strauss has been a supporter of BSF since its formation, both as a physician and as a researcher. His wife, Patricia, also has become a strong advocate for us. Dr. Strauss has received two grants from BSF for early work toward creating a mammalian model of BTHS, and he has attended several of our conferences. His vast experience, wide-ranging expertise and wise insights will add greatly to our international SMAB.

In addition, I am delighted to announce that both **Iris L. Gonzalez, PhD** and **Richard I. Kelley, MD, PhD** have accepted an additional four-year term on our SMAB and that Michael Schlame, MD has agreed to continue as the SMAB Chairman for another four years. Also, **Barry J. Byrne, MD PhD**, who has served so ably on our SMAB since 2002, has unfortunately decided to step down from our SMAB given his many other current responsibilities. Dr. Byrne fully expects to remain involved in BTHS research and treating BTHS patients.

Please join me in thanking all of the members of this terrific group for their dedication to our boys and young men and to our cause. Their focused attention and hard work are truly making a difference!

## Heartfelt Thanks to Barry J. Byrne, MD, PhD

By Kate McCurdy, BSF Board Member; ex-officio member of BSF Scientific & Medical Advisory Board



Barry J. Byrne, MD, PhD  
(Photo courtesy of BSF ~ 2010)

**A**fter a decade of dedicated service on BSF's Scientific and Medical Advisory Board (SMAB), Barry J. Byrne, MD, PhD (Professor, Pediatrics and Molecular Genetics & Microbiology; Associate Chair, Pediatrics, University of Florida; Director, UF Powell Gene Therapy Center; Gainesville, FL) has decided to step down at the end of his current term, given all of his other responsibilities. Dr. Byrne has been a vital member of this group since 2002 and has been integral to many of its most important initiatives in both advancing scientific understanding of Barth syndrome (BTHS) and improving clinical treatment of BTHS patients. He has been the cardiologist for a number of BTHS patients cared for at the University of Florida (UF) in Gainesville and was instrumental in establishing our Barth Registry and Repository (BRR) that began there. He and others from UF came to our conferences, collected longitudinal cardiac data and contributed those data to the BRR. Furthermore, his lab also has done important work that potentially may lead eventually to gene therapy for BTHS. On a personal note, Dr. Byrne is known by many of the boys and young men with BTHS as one

of the best dancers at our conferences and the most awesome doctor at Paul Newman's Boggy Creek Camp in Florida. He is a great friend of BSF and of the Barth syndrome community. We all thank him from the bottom of our hearts for all he has done for us, and we look forward to continuing to work with him on many ongoing and new projects and to seeing him at our conferences in the future.



Drs. Barry Byrne and Randy Bryant  
at BSF's 2010 International Conference. (Photo courtesy of BSF ~ 2010)

# Bryn and Rhys A Story of Hope after Tragedy

By Kate, Mother of Affected Individual, New Hampshire, USA



Kate, Sandt and Bryn, age 10 months  
(Photo courtesy of Kate and Sandt ~ 2012)

Our first son, Rhys, died very unexpectedly of dilated cardiomyopathy on February 10, 2011. The autopsy revealed no indication of bacterial or viral infection, so a genetic cause was suspected. The pediatric hospitalist who delivered the report to us suspected Barth syndrome (BTHS) and encouraged us to see a genetic counselor. We had already made the difficult decision to get pregnant again by this time so she pulled some strings to get us seen right away. The counselor we saw went over our entire family history with us and came up with a short list of possibilities. BTHS was at the top of the list, however it was deemed only moderately likely as there had been no sign of skeletal muscle abnormalities in Rhys' autopsy report. She told us to let her know if we wanted to send tissue samples to Amsterdam (Emma Children's Hospital/Academic Medical Center) for analysis.

About a month later, we learned we were pregnant again, and I was put into the high-risk category and scheduled for many fetal echos. After the first two came back normal, we felt we might be able to breathe again, but I kept having nagging doubts and decided it was time to rule out BTHS. In September, I asked that tissue samples be sent to Amsterdam for genetic testing. Months passed with no news. Evidently there was some sort of logistical issue with getting the samples flown over. We were told it would happen but would be a waiting game.

The first indication of trouble with the pregnancy showed on the last scheduled fetal echo. All of the measurements were normal but the heart "looked thick." A repeat echo ended with the same frustrating result, so an echo was suggested during the first week of life. Bryn was born on December 15, 2011, and, although he was tiny, he was deemed

"gestationally appropriate" and large enough not to be put on any watch lists. He also scored a 9-9 Apgar! He latched well and was a sweet, quiet, ever-watchful baby. We were hopeful that the echo would prove he was healthy — for how can a child with a cardiac problem score a 9-9?! However, the echo showed a decline in his heart function from "normal" in the fetal echo to 33% ejection fraction (EF) at day two. He also suddenly started showing signs of lethargy, and tests revealed thickened blood and a possible kidney infection. He was transferred to the NICU at Dartmouth, New Hampshire where he spent the next 11 days.

Our world was rocked again. The heart issue appeared different, but how could both our boys have different cardiac problems? Because BTHS was still a suspect, Bryn was referred to Dr. Gerald Cox at Boston Children's immediately. A cardiomyopathy genetic panel was drawn, and the waiting game began again. Biochemical urine tests were done and roughly one year after Rhys' passing, we got the call from our genetic counselor. The lab results from Amsterdam were back and they indicated BTHS for Rhys, urine tests for Bryn indicated the same. They were 99.9% sure...the *tafazzin (TAZ)* mutation was really just a formality. We got the final genetic results in early April.

Rhys' and Bryn's godparents's sprang into action after the biochemical diagnosis. They discovered the Barth Syndrome Foundation (BSF) and even invited themselves to dinner with the McCurdys! I lurked on the listserv for a month or so before finally getting the courage to introduce myself. We found out about the BSF conference that was happening in just a few short months, and my parents helped us with plane tickets.

We had been struggling with the issue that, while Bryn looked so good, he had a disease that had claimed his brother. Friends and family alike could not fully comprehend our anxiety and would, understandably, get frustrated with our reclusiveness. But here was an entire community of people who not only got the "chronic illness" thing but understood BTHS as well. Our experience at BSF's 2012 Conference was nothing short of amazing. To be able to interact with other families and meet affected boys and men showed us that these boys could thrive. Meeting the people who are treating and researching BTHS helped us to understand what Bryn was going through. The Foundation and the conference gave us the hope we needed — that Bryn would not have to follow in his brother's footsteps — that he could grow up.

# "No Worries" with Support of BSF and the Barth Community

By Mel, Mother of Affected Individual, Melbourne, Australia



(L-R) Sebastian, Mel, Aiden (age 1), and Luke  
(Photo courtesy of BSF ~ 2012)

In 1997, my entire family was genetically screened for Barth syndrome (BTHS). I was 19 years old, and results identified my grandmother, my mum and myself were carriers of BTHS. At the time, the results didn't have any effect on me; I was in 2<sup>nd</sup> year university, and having children was the LAST thing I was concerned about! The results did impact my parents though. They had lost their first-born son in 1970, my brother, Craig. He was three months, three weeks and three days old when he passed away, and the reason given was "heart complications." Conclusions have now been made that he suffered from Barth syndrome, and this was the reason his young life was taken.

What was most significant from the family's results was discovering that my Uncle Greg (mum's brother) had the BTHS gene. He was 45 years old at the time, and today is 61! The main symptom he shows is cardiomyopathy, and to this day he has never been hospitalised by illness. He is a true "Barth Wonder Story" and provides my husband Luke and me with real inspiration and hope for Aiden's future.

Because we knew I was a carrier of Barth syndrome, I had a Chorionic Villus Sampling (CVS) test performed at 12 weeks with both pregnancies. A CVS is a test that checks the pregnancy for genetic abnormalities. For our first pregnancy, the results came back "negative" for Barth syndrome, and gave us our eldest son, Sebastian. For our second pregnancy, the results came back "positive" for Barth syndrome, and gave us our second son, Aiden, who was born on May 26, 2011.

Due to his prenatal diagnosis of Barth syndrome, we had a full team of medical specialists attending Aiden's birth. As soon as he arrived, Aiden was taken straight to NICU, and apart from

needing to be on continuous positive airway pressure (CPAP) resuscitation for two hours, his doctors were very happy with his condition. Initial echos showed an obvious spongiform thickening of the left ventricle with an ejection fraction (EF) of 26%, and contractility at the lower end of normal. He was started on Carvedilol on day five, and because of his better-than-expected condition, we got to take him home with us on day 10!

Life at home was fantastic! That is, until Aiden was three weeks old. It was a Tuesday, and I noticed that he wasn't feeding well at all, was quite breathless, and had a blue tinge around his mouth and between his eyebrows. We took him straight to emergency at the Melbourne Royal Children's Hospital, and he was admitted into the critical care unit in the cardiac ward later that evening, in severe heart failure. Our world crashed!

Aiden was in a terrible state, with his EF down to 16%. It was at this time we first met Dr. Andreas Pflaumer, Aiden's cardiologist. Dr. Pflaumer was then, and still is, someone who will remain in the highest regard within our entire family. Together with Aiden's other medical team, Dr. Lexie Frydenberg (pediatrician) and Dr. Joy Lee (metabolics), they have made an unbelievable effort in getting to know and understand the complex disorder and management of Barth syndrome.

During Aiden's stay in hospital, we first contacted the Barth Syndrome Foundation. The immediate welcome and support from the entire community was quite overwhelming. Living in Australia meant we were geographically disconnected from most, but the emotional connection we felt was very much instant. It was during these first few contacts when we were persuaded by everyone's recommendation to attend BSF's 2012 Conference.

Attending the Conference was a huge learning curve for Luke and myself. While it was great to have individual discussions with Dr. Kelley, Dr. Steward, and the rest of the Barth medical specialists, the highlight for us was seeing and talking with all the Barth boys and men. Personally, my favourite memory was Amanda Clark taking the photo of all the "Barth Brothers" in the Grand Ballroom. Seeing them all standing together was a moment I'll never forget. On our return home, we felt empowered with knowledge about Barth syndrome and supported by all the life-long friendships that were made.

We feel very blessed to have Aiden in such a stable, mild condition at the moment. His latest cardio check showed that his heart function is within "normal" ranges, and his EF is now up to 61%! Even more exciting is he has just started walking at 16 months of age! Aiden is such a delightfully, happy young boy. People are always commenting on his bright smiles and magnetic personality. Who knows what the future will bring, but by having the support of the Foundation and the Barth community behind us, we feel confident we can take this journey on together!

# In Loving Memory ~ Our Angel, Miguel

By Rocio and Javier Socias, Parents of Miguel, Spain



Miguel with his best friend, Miguelón  
(Photo courtesy of Socias Family ~ 2012)

**M**y son is an angel! Although I am sure that any parent would say that about their children, Miguel not only is an angel but has also earned the right to be one.

He is one, and not only for having spent the last three and a half months of his life in a hospital, waiting for the miracle of life. It was a miracle that did not come despite all he had to endure during that time — several surgeries, hemorrhages, chylothorax, pneumothorax, ... But it was the human warmth that he had even though he was just five years old.

He is an angel because of his way of being... happy, curious, pleasant, friendly, and well educated. He had this "something" since he was very little, when I would take him for a walk in the double stroller as Javi's twin, when people would always say, "This one is more handsome (Javi), but this other one has such a nice face, he is so..., so...." All who knew him remember him, be it for something he might have said or something he did. Miguel was never unnoticed. As his aunt "O" said, "He had so much light within himself that he just had to finally go to his true destiny." However, knowing all this does not make his daily absence less painful.

Just today, one of my co-workers that I had not seen for a long time asked about him. He wanted to know how everything went. I had barely enough voice to tell him that he had died. Every time that I am in a similar situation, I am surprised when I can bring myself to give that answer without falling apart at that precise moment. Why do mothers or fathers have to live through such an experience? It is so unfair, especially for him. He still had so much to live for, like his sixth birthday; he was so much looking forward to this

celebration. Since May, he talked about this subject with the doctors and nurses. He told them how it would be, that his brother would be with him, that the room would be decorated with balloons, that there would be presents for both.

He was an example for all of us during his entire hospital stay. He showed a strength beyond that of such a small child. He lived the situation as if it was natural, always tried to have fun and to enjoy it to the most. I remember one day when he was still in the hospital and the family came to visit, and all of us ended up playing blind man's bluff ("gallinita ciega," blind hen) even though he had a medication line.

I don't know if all kids in this situation are the same. Miguel was always like this, he liked everything. He enjoyed a ball point pen as much as a remote control car. He was not bored, or rather, he did not want to be bored, and to this purpose he used all resources within his reach, the main one being his imagination. Together with his best friend "Miguelón" ("Big Mike"), his horse-zebra stuffed animal, his adopted uncle José, his father or myself, he played everything imaginable. He invented fantastic characters, like the wizard Magón who lived in the magic forest and whose mission was to help all the children, a tall gentleman with a white beard and a magic wand, who even had a phone number 666 666666.

When they had to change his dressings while he had the Berlin Heart, they gave him a bolus to make him sleep a little so he would not suffer since the dressings hurt him so much; then, when he woke, we would tell him that the wizard Magón had come to visit him. He would smile and full of hope he would ask when would he be able to see him. Fortunately, and thanks to one of the health workers, Juan Carlos who disguised himself, Miguel was able to meet him just the day before the heart arrived. Javier told me that he had such a fun afternoon; I could not be there, but his father was with him all the time and "cared for him like a mother would."

With all my heart I thank all who were able to make him laugh during all those days. It was unbelievable how he was treated by the doctors, nurses, and other personnel of the Gregorio Marañón Hospital. They did all they could for him, as if he was their own little kid brother, their child, "Miguel I of the ICU," where he ruled for three months. They loaded him with gifts, even a remote controlled ICU helicopter, but the most important gift was the enormous doses of the best medicine that a mother can see being given to her child: the gift of LOVE.

Thinking back on this, with the hindsight we now have, it is as if he knew that he did not have much time left, so he did not want to waste it on being sad or angry. He so much wanted to live.

(Cont'd on page 12)

# In Loving Memory ~ Our Angel, Miguel

(Cont'd from page 11)

Miguel accepted the situation from the beginning. He knew that he was sick and would have to stay in the hospital until they cured him, far from his beloved brother from whom he was separated at the time of admission, far from his friends, and from his daily life. But I never saw him cry because of this, he just waited for the moment when he could get out of there and back to enjoying all that the future might bring, like bathing in his grandfather's pool, playing with his cousins at the beach, returning to school, losing his first tooth, kissing a girl for the first time ...

But none of this will happen now—he left on a beautiful summer morning, after fighting like a wild boar for 18 days to get his new heart to work and the rest of his organs along with it. He almost succeeded, but his lungs failed just when we thought that he had made it. His father and I think that he got fed up with it, that it was no longer fun, that he was tired of fighting. That he preferred to be in heaven with his grandfather Antonio, his aunt María Sol, and all those that were waiting to care for him and to pamper him as he deserved.

Three months have passed since that day, three months without him, and I have the same feeling of disbelief I had when the boys were born. The first time I saw them it seemed incredible that those little beings had come from my body, that they would be persons with their own identity, and that they were ours forever. I am sure that this is what parents will think when they see their children for the first time, but it is one thing what we wish for, what we expect, and something else that we get. Call it God, fate, or whatever, your life is at His disposal, giving and taking at will.

It seems like only a couple of days ago I worried that he was not eating enough or about how he was doing in school, but



Miguel and his friends!  
(Photo courtesy of Socias Family ~ 2012)

at the same time I feel like he left a century ago. Every time that I think he is not here, it seems incredible that I will no longer hear him laughing, his voice asking about everything with that tone that sounded like singing instead of speaking. I will no longer be able to caress him, or stroke his beautiful blond hair, or kiss him until he complains and calls me a pest. When I am conscious of this, I feel so much grief that I cannot breathe. At those moments, I would want to close my eyes and vanish. I miss him so much that I don't know how I can bear it. Every day I ask myself "Why? Why has all this happened? Why is he not here?" There is no answer now, but Javier, his father, tells me that we will know some day. Some day we will discover the reason and the purpose for so much suffering, and we will have the knowledge that his death was not in vain, that everything makes sense, that what has happened served a good end.

I then look around myself and see my son Javi, my husband, all of my family, and then I know I have to go forward for them and for Miguel. I must live for him and must get to enjoy him through his brother.

He will always be our child, we still love him like crazy. Even though we did not succeed, we fought tooth and nail to keep him with us. Even up to the last second, we kept up our hopes that everything would end well. I must understand that the time he spent with us was a gift, and that it is infinitely better to have had the opportunity to know and love him than not to ever have had him.

More than once he asked me when would he be able to fly. I know that now he can do so. He is up there, flying with his friend the wizard Magón, with Miguelón in one hand and his magic wand in the other, and with it he will help all children who need it, showing them a magic world to escape the pain and suffering of their existence, and he will help them to be as happy as he was in this world. And I know that he will always and forever remain in our hearts.



(Back row) Javier and Rocio  
(Front row) Javi and Miguel  
(Photo courtesy of Socias Family ~ 2012)

# 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 **67** articles have been published on BTHS research conducted with the support of BSF and/or BSF affiliate funding (denoted below with †) and publications that acknowledge biological samples and/or information from Barth families, the Barth Syndrome Registry and Repository, and/or BSF affiliates (denoted below with ▼). Listed below are all 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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2. Gawrisch K. **Lipids: Tafazzin senses curvature.** Nat Chem Biol. 2012 Oct;8(10):811-2. doi: 10.1038/nchembio.1068. No abstract available. (PubMed)
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4. Whited K, Baile MG, Currier P, Claypool SM. **Seven Functional Classes of Barth Syndrome Mutation.** Hum Mol Genet. First published online October 24, 2012. (Abstract)
5. Roberts AE, Nixon C, Steward CG, Gauvreau K, Maisenbacher M, Fletcher M, Geva J, Byrne BJ, Spencer CT. **The Barth Syndrome Registry: Distinguishing disease characteristics and growth data from a longitudinal study.** Am J Med Genet A. 2012 Oct 8. doi: 10.1002/ajmg.a.35609. [Epub ahead of print] (PubMed Abstract)\*▼
6. Finsterer JA, Marlies FB. **Haematological features in Barth syndrome. [Review]** Published Ahead of Print, Post Author Corrections, 4 October 2012. Curr Opin Hematol Volume 19(5) pgs. vii-vii,349-420 September 2012. (Abstract)
7. Ojala T, Polinati P, Manninen T, Hiippala A, Rajantie J, Karikoski R, Suomalainen A, Tyni T. **New mutation of mitochondrial DNAJC19 causing dilated and noncompaction cardiomyopathy, anemia, ataxia, and male genital anomalies.** Pediatr Res. 2012 Oct;72(4):432-7. doi: 10.1038/pr.2012.92. Epub 2012 Jul 13. (PubMed Abstract)
8. Kelly DP, Strauss AW. **Inherited cardiomyopathies.** N Engl J Med. 1994 Mar 31;330(13):913-9. No abstract available. (PubMed)
9. Carman GM. **An unusual phosphatidylethanolamine-utilizing cardiolipin synthase is discovered in bacteria.** Proc Natl Acad Sci U S A. 2012 Sep 25. [Epub ahead of print] No abstract available. (PubMed)
10. Zhuravleva E, Gut H, Hynx D, Marcellin D, Bleck CK, Genoud C, Cron P, Keusch JJ, Dummler B, Esposti MD, Hemmings BA. **Acylo coenzyme A thioesterase Them5/Acot15 is involved in cardiolipin remodeling and fatty liver development.** Mol Cell Biol. 2012 Jul;32(14):2685-97. Epub 2012 May 14. (PubMed Abstract)
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12. Tan BK, Bogdanov M, Zhao J, Dowhan W, Raetz CR, Guan Z. **Discovery of a cardiolipin synthase utilizing phosphatidylethanolamine and phosphatidylglycerol as substrates.** Proc Natl Acad Sci U S A. 2012 Sep 17. [Epub ahead of print] (PubMed Abstract)
13. Ronvelia D, Greenwood J, Platt J, Hakim S, Zaragoza MV. **Intrafamilial variability for novel TAZ gene mutation: Barth syndrome with dilated cardiomyopathy and heart failure in an infant and left ventricular noncompaction in his great-uncle.** Mol Genet Metab. 2012 Sep 18. pii: S1096-7192(12)00354-X. [Epub ahead of print] (PubMed Abstract)
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15. Schlame M, Acehan D, Berno B, Xu Y, Valvo S, Ren M, Stokes DL, Epand RM. **The physical state of lipid substrates provides transacylation specificity for tafazzin.** Nat Chem Biol. 2012 Sep 2. [Epub ahead of print]\* (PubMed Abstract)
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19. Galvao T, Daneault C, Des Rosiers C, Polster BM, Hoppel CL, Stanley WC. **Improved mitochondrial function with diet-induced increase in either docosahexaenoic acid or arachidonic acid in membrane phospholipids.** PLoS One. 2012;7(3):e34402. Epub 2012 Mar 30. (PubMed Abstract)
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23. Cade WT, Spencer CT, Reeds DN, Waggoner AD, O'Connor R, Maisenbacher M, Crowley JR, Byrne BJ, Peterson LR. **Substrate metabolism during basal and hyperinsulinemic conditions in adolescents and young-adults with Barth syndrome.** J Inher Metab Dis. 2012 May 12. [Epub ahead of print] (PubMed Abstract)\*▼

# Funding Opportunities Relevant to Barth Syndrome Research

The following ongoing research initiatives at organizations other than BSF are particularly relevant to Barth syndrome:

## National Institutes of Health (NIH)

### **Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R01)** Funding Opportunity Announcement (FOA) Number: PAR-11-288

Open Date (Earliest Submission Date): September 5, 2011  
Letter of Intent Due Date: 30 days prior to applicable receipt date  
Expiration Date: September 8, 2014  
<http://grants.nih.gov/grants/guide/pa-files/PAR-11-288.html>

Purpose: This Funding Opportunity Announcement (FOA) encourages Research Project Grant (R01) applications that will improve our understanding of how patterns of physical activity and dietary choice affect the health and fitness of children with physical disabilities.

Proposed research should account for the functional limitations of children with disabilities and their nutritional needs, as well as the physiological, psychosocial, and environmental factors that play a role in determining the health of this population.

### **Discovery of Genetic Basis of Mendelian or Monogenic Heart, Lung, and Blood Disorders (X01)**

Funding Opportunity Announcement (FOA) Number: PAR-11-307

Open Date (Earliest Submission Date): September 18, 2011  
Letter of Intent Due Date: September 19, 2011; April 16, 2012; April 15, 2013; and April 14, 2014  
Application Due Date(s): October 18, 2011; May 14, 2012; May 14, 2013; and May 14, 2014  
Expiration Date: May 15, 2014  
<http://grants.nih.gov/grants/guide/pa-files/PAR-11-307.html>

Purpose: To stimulate discoveries of the genetic basis of Mendelian or monogenic disorders that significantly affect heart, lung, and blood (HLB) systems, the NHLBI invites X01 to use the genome-wide sequencing capacity of the Mendelian Disorders Genome Centers which are funded under the HG-10-016.

### **Health Promotion for Children With Physical Disabilities Through Physical Activity and Diet: Developing An Evidence Base (R21)**

Funding Opportunity Announcement (FOA) Number: PAR-11-284

Open Date (Earliest Submission Date): September 16, 2011  
Letter of Intent Due Date: 30 days prior to applicable receipt date  
Expiration Date: September 8, 2014  
[http://www.grants.gov/search/search.  
do?mode=VIEW&oppId=110713](http://www.grants.gov/search/search.do?mode=VIEW&oppId=110713)

Purpose: This Funding Opportunity Announcement (FOA) encourages Exploratory/Developmental Research Grant (R21) applications that will improve our understanding of how patterns of physical activity and dietary choice affect the health and fitness of children with physical disabilities.

Proposed research should account for the functional limitations of children with disabilities and their nutritional needs, as well as the physiological, psychosocial, and environmental factors that play a role in determining the health of this population.

(Photos courtesy of Amanda Clark ~ BSF 2012 Conference)

(Cont'd on page 15)



Brie and Milosh (age 2)



Christopher (age 4) and Chris



Tiffini and Henry (age 4)



Wyatt (age 5) and Tim

# Funding Opportunities Relevant to Barth Syndrome Research

(Cont'd from page 14)

## National Institutes of Health (NIH)

### Innovative Therapies and Tools for Screenable Disorders in Newborns (R01)

Program Announcement (PA) Number: PAR-10-230

Opening Date: September 5, 2010

Letters of Intent Receipt Date: 30 days prior to application due date

Application Due Date: See <http://grants1.nih.gov/grants/funding/submissionschedule.htm>

Expiration Date: September 8, 2013

<http://grants.nih.gov/grants/guide/pa-files/PAR-10-230.html>

**Purpose:** This FOA, issued by the Eunice Kennedy Shriver National Institute of Child Health and Human Development, the National Institute of Diabetes and Digestive and Kidney Disease, the National Institute of Neurological Disorders and Stroke, and the National Institute on Deafness and Other Communication Disorders encourages Research Project Grant applications from institutions/ organizations that propose research relevant to the basic understanding and development of therapeutic interventions for currently screened conditions and "high priority" genetic conditions for which screening could be possible in the near future. In this FOA, a "high priority" condition is one for which the development of an efficacious therapy would make the condition amenable to newborn screening.

## American Society of Hematology Patient Group Research Grant Opportunities

To draw together the multitude of hematology-related research grant opportunities that are available through patient groups, the Society has created a section on the ASH Web site that simplifies your search for requests for blood and blood disease research topics. (<http://www.hematology.org/Research/2874.aspx>)

## Children's Cardiomyopathy Foundation

The Children's Cardiomyopathy Foundation (CCF) offers two annual grant programs to support innovative basic, clinical, population, or translational studies relevant to the cause, diagnosis, or treatment of cardiomyopathy (Dilated, Hypertrophic, Restrictive, Left Ventricular Non-Compaction, or Arrhythmogenic Right Ventricular Cardiomyopathy) in children under the age of 18 years. The goal of CCF's grant programs is to advance medical knowledge of the basic mechanism of the disease and to develop more accurate diagnostic methods and improved therapies for children affected by cardiomyopathy. (<http://www.childrenscardiomyopathy.org/site/grants.php>)

## United Mitochondrial Disease Foundation

The United Mitochondrial Disease Foundation (UMDF) Research Grant Program began in 1997 out of a desire to fund research toward diagnoses, treatments, and cures for mitochondrial disease. ([http://www.umdf.org/site/c.dnJEKLNqFoG/b.3790285/k.6CE6/Research\\_Grant\\_Program.htm](http://www.umdf.org/site/c.dnJEKLNqFoG/b.3790285/k.6CE6/Research_Grant_Program.htm))

### UMDF Clinical Research Fellowship Training Award

Statement of Intent Receipt Date: October 31, 2012 (no later than 5:00 PM EST)

Application Due Date: February 1, 2013

Statement of Intent Application Form

The UMDF clinical fellowship award is a one or two year award designed to support the training of physician scientists who plan to practice clinical management of patients with mitochondrial disorders AND to conduct clinically (patient) oriented research in the field of mitochondrial medicine.

**Purpose:** The primary goal of the UMDF clinical research fellowship training award is to expand the number of clinicians and physician scientists practicing clinical management of patients with mitochondrial disorders AND conducting clinically (patient) oriented research in the field of mitochondrial medicine.

## The North American Mitochondrial Disease Consortium (NAMDC) Fellowship

**Purpose:** NIH-funded 1-year fellowship will provide a unique clinical trials training opportunity for a senior fellow who will move on to practice mitochondrial medicine at an attending level. The NAMDC fellowship is open to clinicians, senior post-doctoral non-MD scientists, junior faculty, and established investigators wishing to develop or re-focus their careers on clinical research on mitochondrial disease.

Funding is at an NIH scale PGY6 level. Three single one year fellowships will be funded. The second fellowship which is now available can begin as early as October 1, 2012. Click here for more information.

# Thankful for our Growing Support We Couldn't Do it Without You!

By Stephen B. McCurdy, Chairman, Barth Syndrome Foundation

**A**s I write this, the remnants of Hurricane Sandy are still evident here in the Northeast. Some homes still do not have power; New York City tunnels are still flooded; families are still in shelters; and the aftermath of high winds and unprecedented floods still litter the landscape. Once again, the American people, with many of our friends overseas, have opened their wallets, while others have contributed their time and even their homes to help those in need. In the end, like other natural disasters before her, eventually Sandy will pass into memory.

Barth syndrome is different. While Sandy may have been the storm of a lifetime — Barth syndrome can be a lifetime of storms for those it touches. It may not strike as many people, but for those affected it can be just as devastating. Barth syndrome rarely grabs the headlines; far fewer people are even aware of the need to help. While rebuilding and repairing after Sandy will take \$billions, we know what we need to do. For Barth syndrome, the answers are still hidden behind doors that only further investments in research can unlock. For Sandy, the resources will come from insurance companies, Federal Emergency Management Agency (FEMA), state and local governments and individual savings. We in the Barth Syndrome Foundation (BSF) are fortunate to have you — our legion of loyal donors and fundraisers!

Every dollar that you raise and contribute to BSF moves us closer to our goal of a cure and/or effective treatments for Barth syndrome. Earlier in 2012, with the guidance of our Scientific and Medical Advisory Board (and support of our international affiliates), BSF awarded nine new research grants for a total of US \$341,002, each of which moves us closer to our goal. In June, we held an inspiring and very exciting International Barth Syndrome Scientific, Medical and Family Conference, the sixth in a series of increasingly successful and well-attended conferences that invariably give birth to a host of new

research ideas and closer cooperation among our scientists. Because of your donations, BSF is able to find and provide a "virtual home" as well as a robust and caring community for affected families. Our programs are described more fully throughout this newsletter, but none of them could happen without your support, for which we are eternally grateful!



Devin (age 8) and Henry (age 4) at the Hey, Henry Henry Rooftop Baseball Game  
(Photo courtesy of Tiffini Allen ~ 2012)

## Tiffini = Fundraising Phenomenon!

Tiffini Allen is our Fundraising Phenom! She has created at least four fundraising events or activities over the past year including the TAZ Piggy Bank, the Barth Pajama Party, the Barth Bowling Party, the Hey, Hey Henry Rooftop Baseball Game, and the much talked about Hey, Hey Henry Cards that everyone attending BSF's 2012 Conference got a chance to see first-hand. Tiffini is showing us all a way to turn fun things you and your family like to do into fundraising and awareness events! More about the Henry Cards later...

## Eliza's Ice Cream Social

Eliza McCurdy and her good friend Drew Crowley are both active in their church youth groups at the local Episcopal and Roman Catholic Churches, respectively. This last summer, they held an ecumenical fundraiser for BSF, inviting the youth groups from both churches, as well as their families and friends to an Ice Cream Social for fun and fundraising, with all net proceeds going to BSF. Not surprisingly, the event attracted a big crowd despite a heavy rain from beginning to end. Quickly adjourned to the church parish hall, everybody had fun, ate ice cream, and raised money for BSF. A simple idea, a couple of happily willing groups of kids and a hot summer day turned out to be a winning combination for Barth!



10 of the 11 members of Team Will who competed in the 2012 Westchester Triathlon, along with Will himself.  
(Photo courtesy of Eliza McCurdy ~ 2012)

## Team Will Races Again!

For the 9<sup>th</sup> year in a row, intrepid members of Team Will (this year there were 11 of them) threw themselves into the cool waters of Long Island Sound at 7am on September 23<sup>rd</sup> to begin the Jardin Westchester Olympic-length

(Cont'd on page 17)

(Cont'd from page 16)



Members of Team Will and their families  
(+ the McCurdys)  
(Photo courtesy of Eliza McCurdy ~ 2012)



Randy, Andrew (age 23) and Brenton at the  
BSF/JDRF 2012 Golf Tournament  
(Photo courtesy of BSF ~ 2012)



Cherie and Lattigo (05/01/2000 — 1/25/2002)  
(Photo courtesy of Cherie Schrader)



Brian and friends substituted poker chips for  
volleyballs in support of BSF!

Triathlon (0.9 Mile Swim; 25 Mile Bike and 6.2 Mile Run). Each proudly wore his/her easily recognizable Barth shirts during the bike and run portions raising money and awareness for BSF. Once again, Gary Rodbell acted as coach and led the way. Joining Gary was his 17 year old daughter, Julia, who had recently completed a half Iron-man Triathlon. The next generation of Team Will is stepping up to take their places in the ranks! In case Team Will got too hot, the local fire department volunteers showed up with their 90+ year old antique engines to support Team Will and BSF. This event just keeps getting better and better! Look for Team Will again in the Panama City, FL full Ironman race on November 2, 2013!

### The Buddemeyer Golf Tournament Lives on!

Randy Buddemeyer and Tim Rivers, his partner at the real estate firm Newmark Grubb Knight Frank, held their annual golf tournament again to raise money for BSF and JDRF (Tim's son has Juvenile Diabetes). Over 120 golfers teed off on October 1<sup>st</sup>, a sunny Florida morning, followed by awards, festivities and a talk by Andrew Buddemeyer about BSF and his family's experience with the disorder. The event was hosted by Lauren Thompson from the Golf Channel Network who was so taken by the event that she has agreed to become a regular donor and emcee for next year's tournament. In addition to Tim and Randy's own firm, premier sponsors included: Redcoats, Slay's Restorations and Yardi/SiteStuff. The pictures of the Buddemeyer boys putting their putting skills to the test show clearly how much fun there can be in fundraising!

### Cherie Schrader Mother's Day Mini Photo Session

Cherie Schrader hosted A Mother's Day Mini photo session at the LaPorte Fairgrounds Pioneer Village....with live music, photos, food, and cotton candy. Families gave Mom a beautiful family experience, with all proceeds going to Barth Syndrome Foundation.

### Drake moves from Volleyball to Poker!

Last year, Brian Drake held a well-attended Barth Syndrome Volleyball Tournament as a way of bringing attention to and raising money for BSF. This year, he moved the event inside substituting cards and chips for volleyballs and nets! Apparently, Brian's friends are up for any event that Brian throws, as everyone had a great time. Once again, the proceeds benefited BSF. We can't wait to see what the event will be next year, Brian!

### Conference Fundraising Breaks Records!

By all accounts, the 2012 BSF Conference was a roaring success, as you can see by articles throughout this newsletter; fundraising was no exception. Through the efforts of Dr. Matt Toth, BSF Science Director, BSF was able to secure funds from the National Institutes of Health (NIH) — The Office of Rare Disorders (ORD) and the National Heart Lung and Blood Institute, specifically. Recognition by the NIH, the world's leading health research funding organization, of the importance of our Conference to the advancement of science is a real accolade. Matt deserves much credit for winning their support and for securing Dr. Stephen C. Groft, Director of the Office of Rare Diseases Research (ORDR), NIH as our keynote speaker! John Wilkins, and several families including the Addingtons, Penas and Allen-Dollards, as well as our Barth Affiliates, also used the Conference to highlight BSF's need for funding. Together with our Executive Director, Lindsay Groff, and Matt Toth, we raised more money for the 2012 Barth Conference than ever before!

(Cont'd on page 18)

# Thankful for our Growing Support

## We Couldn't Do it Without You!

(Cont'd from page 17)

### A Chance for Everyone to Help

Like most charities, BSF struggles to balance its books each year and relies on year-end gift giving to raise almost 50% of the money we spend all year. Conference years are a special challenge for BSF, and this year is no exception. As you can see by the many families mentioned above, we depend on everyone to pitch in and help ensure that BSF does not run a deficit. This year, a number of our volunteers will be calling our Barth families across the US to ask them to help out by sending holiday cards to their friends and relatives asking for donations to BSF. Modeled after Tiffini's very successful "Henry Cards," these customized cards include a family picture and a holiday note that encourages the recipient to consider a donation to BSF. BSF and its many programs are so important to all of us, they deserve our active support. Our friends and families are constantly asking what they can do to help — here is a perfect way to let them know! Please say "yes" when the BSF volunteer calls to explain this simple program. It couldn't be easier... and it couldn't be more important!

#### Donations Received in Honor of:

Allen-Dollard, Henry  
Addington, Devin  
Baffa, Kevin  
Buddemeyer, Andrew  
Derusha-Mackey, Nicole & Sarah  
Drake, Abram  
Fairchild, Jake  
Greenberg, Rebecca  
Higgins, Jack  
Holly, Benjamin  
Hopp, Wallace  
Kalapasev, Milosh  
Langdon, Dr. Edward  
Mann, Benjamin  
Marchessault, Michael  
McCurdy, Will  
McCormack, Susan  
Osnos, Evan & Sarabeth  
Pena, Christopher  
Randell, Jay & Amer  
Sernel, Ryan  
Stewart-Groff, Charlotte  
Thorpe, Thomas  
Van Dusen III, George  
Wilkins, John  
Woodward, Connor

#### Donations Received in Loving Memory of:

All Barth Boys We've Lost  
Anderson, Esther  
Bowen, Michael  
Cook, Lattigo  
Drake, Rachel  
Frysiak, Frances  
Holly, Caleb  
Johnson, Lois  
Lee, Ada  
Moreland, Sandra  
Percival, Gloria  
Pittack, Esther  
Poe, Gertrude  
Russell Sherer, Carroll  
Telles, Michael  
Thorpe, Benjamin  
Verona, Evelyn



Greyson (age 4)



Jake (age 14)



R.J. (age 14)



Robert (age 26)

# Barth Syndrome Trust

## Message from the Chair

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

This year has seen us hard at work completing a few major projects for the Barth syndrome community.

### Website

Our newly revamped **website** ([www.barthsyndrome.org.uk](http://www.barthsyndrome.org.uk)) is now up and running. It is easy to navigate and filled with lots of information, stories, and pictures. Please visit the site and tell us what you think.

### Barth Syndrome Education Guide

In a joint project with the NHS Barth Syndrome Service in Bristol, we have compiled a comprehensive Barth Syndrome Education Guide. This Guide is an essential document for all families and educators who have a child or young person with Barth syndrome. It explains how to manage Barth syndrome within the UK education system and gives parents, affected young people and teachers the tools they need to make school life successful. We also have a Short Education Summary which is a handy day-to-day reference sheet for teachers.

Although these Education Guides have been written primarily for UK families, they contain a wealth of information pertinent to *all* families, regardless of where you live. Please contact the Barth Syndrome Trust on [info@barthsyndrome.org.uk](mailto:info@barthsyndrome.org.uk) if you would like your free printed or electronic copy of the Guide(s).

### Plans for 2013

Next year will see us preparing new resources for older children with Barth syndrome working closely with the Bristol Team and the affected boys and siblings. These resources will help young people understand Barth syndrome. We hope that this will be useful for all families, and, in particular, for those young people who are becoming more independent and more involved in their own care. We will also be creating a resource pack for families needing information about starting or growing their family when there is a risk of having a son with Barth syndrome or a daughter who may be a carrier of the Barth gene.

### Update on Isaiah

In the Fundraising section below, you'll read about Joshua and his team who cycled 1,000 miles in support of Barth syndrome. The team raised an incredible £1260 for the Barth Syndrome Trust. Joshua is the uncle of young Isaiah who is currently in hospital in Newcastle. After a sudden cardiac decline, Isaiah was put on a Berlin heart while he awaits a transplant. This is such a difficult time for the family who are far away from their home and face such an uncertain time of waiting and worrying. We wish Isaiah a speedy recovery.



Isaiah, age 18 months  
(Photo courtesy of BST ~ 2012)

### Congratulations to Three of our Families

In previous editions of the newsletter, we all grieved as we featured memorials for three of our beloved young boys — Philip Brown (22nd May 05 – 29th Sep 09), Oscar Stobart-Hook (26th Oct 10 – 21st June 11) and Jack Reddin (22nd Dec 09 – 30th Dec 09). It gives us great joy to report that these three families have recently had some good news to share...

Congratulations to **Gemma and Adam Reddin** on their recent marriage. This was a lovely celebration enjoyed by the Frost, Reddin, and Bull families and all their friends.



Alice

(Photo courtesy of BST ~ 2012)

**From Alan Brown & Maike Lange:** We are thrilled to introduce Alice, our new addition to our family. Alice was born on 12 July 2012 weighing 2.82kg (6lbs 3oz). We are a little family once again! The anniversary of the death of our son, Philip, was on 29<sup>th</sup> September, but Alice made it so much easier for us to handle it this time.

We are very happy to announce the safe arrival of a son for **Liz and Guy Stobart-Hook**: Jensen Oscar was born on 1 October 2012 at 8.57am weighing 2.94 kg. Liz and Jensen were allowed home on the 3<sup>rd</sup> October and have settled in reasonably well.



Jensen Oscar

(Photo courtesy of BST ~ 2012)

# Our experience of the Bristol Barth Syndrome Clinic, UK

By Veerle Van Langendonck, Belgium

Being a mom of two Barth boys, I know, just like many of you, the worries of the threatening "what if it worsens?" Yet, the big difference is that my boys don't have the same issues of keeping up with their peers in daily life, especially not with regard to the muscle problems. They do have other issues which they have to cope with. For years we have been having their blood taken to help research. I am happy to tell you our story of our visit to the Bristol Clinic in September which was, like BSF's 2012 Conference in Florida, a fantastic experience.

While we thought it would be just a quick blood sampling and a little talk, it was so much more!! A whole team, extremely motivated, was waiting for all of the families and we felt so welcomed! Physical tests, echos, holters, ECG, blood samplings, a nutritional educational game for the boys, a presentation about medical records, talks with the cardiologist, nutritionist, dentist, and physical therapist. Last, but absolutely not least, the driving force, Dr. Steward, who was in charge of these splendid clinics and who really gave so much time for us. I want to stress we were so impressed by their dynamism and their sincere interest in trying to do something about this life-threatening disease. Also, many thanks to Debbie Riddiford for her magnificent planning.

After these amazing clinics, there was the fantastic social event organized by the Barth Syndrome Trust. The bowling and the pizza event, led by those two terrific ladies Michaela and Annick, was just a joy from the first minute to the last: we talked, laughed, and had a great time together with all of the families and the whole Bristol clinic team!

Our boys still talk about the weekend in England, "which was so much fun!" even though it included a clinic, blood samplings and holters. What a positive, motivated team! What an approachable person Dr. Steward is and how lucky the Barth syndrome community is to have this man in their midst. I am really convinced that he is one of those people who can make a big difference in research, who has his heart with the Barth families, and who is determined to do something about it, as he has already shown extensively in the past decade! Our sincere congratulations for what he, his team, and the Barth Syndrome Trust have already accomplished and will certainly accomplish more in the future! Well done and so many thanks to all of you!



Family Gathering - October 2012 (Photo by Samantha Holmes ~ 2012)

## Save the Date

**Barth Syndrome NHS Clinic  
Thursday 11<sup>th</sup> and Friday 12<sup>th</sup> April 2013  
Bristol Royal Hospital for Children  
Followed by a Family Gathering on Saturday 13<sup>th</sup> April 2013  
Venue TBD**

# Barth Syndrome Trust

## Fundraising events in UK and Europe

Thank you to everyone from all over the world who contributed to Barth Syndrome Trust (BST) in any way — through fundraising, challenges, supporting our brave cyclists and runners, standing orders or direct donations. Special mention goes to Suzy Green and friends in Cambridgeshire, who have raised over £1000 at several recent events. Also to Eleanor Crawford whose participation in the Great Scottish Run raised £550.

### Overton Sheep Fair — a 'ewe-nique' event

In July 2012, we were all whisked back in time as the Hampshire village of Overton was transformed to the 1940's. The BST Pancake Stall was the place to be! Our volunteers, some dressed in period costume, worked for two days making delicious pancakes, running a tombola and generally entertaining the passers-by. Special thanks go to **Annick Manton** who organised the fundraising event, **Greg and Kerry Ann Manton** for the catering and all the volunteers and family members who turned up to help. Our aprons were suitably adorned

with hearts or stretched logos to catch the spirit of the sheep fair. Thanks to everyone's hard work and the support of the Overton community, we raised over £900 from the event.



Logo designed by Candice Bristow



(L-R) Dominique and Kerry-Ann at the Sheep Fair making pancakes. (Photo by Samantha Holmes ~ 2012)

### The Ultimate UK Cycle Challenge ~ A 1000 mile journey the length of Great Britain

This year we were fortunate enough to have not one but two teams of cyclists successfully complete this challenge. Their stories share a common theme of triumph against the most momentous challenges of weather, injuries and accidents but their individual strength and determination, and the support of their teams meant the difference between success and failure. Read their inspiring stories below.

#### John O'Groats to Land's End ~11th August – 18th August 2012

By Joshua Haycock (*Uncle of Isaiah*)

Cyclists: Joshua Haycock; Dom Rawlinson-Millichap; Rachael Dawes; Louis Stinchcombe; Michelle Stinchcombe; Peter Rawlinson-Millichap (who rode a few miles each day even though he was support)

Everything happened in that week with crashes, punctures, and injuries. "Blood sweat and tears" is the saying which I think describes the whole trip. We had some really hot weather and some wet and cold but we still managed to enjoy every day as we knew it was for a great cause. We camped out most nights and were on the road for 7:30am. We rode for 8–10 hours

a day and had a rest every 15–25 miles. We climbed over 26,000 feet through the eight days. Our highest point was the Summit of Shap, which is 1,407 feet above sea level with great views of the Scottish Highlands and much of the United Kingdom.

Everyone will remember something that happened each day — something which made everyone laugh, or a great descent or climb or the views of the sea or the countryside. Throughout the eight days the group just got stronger and stronger, and we bonded like family. Everybody brought something special to the team and we kept each other going. We are thinking of doing it again because we enjoyed it so much.

What an achievement! Thank you to Joshua and team, the support crew, those who helped with accommodation, and all donors for a total of £1260.



Josh's team's start point

(Photo courtesy of Joshua Haycock ~ 2012)

(Cont'd on page 22)

# Barth Syndrome Trust

## Fundraising events in UK and Europe

Land's End to John O'Groats ~ 22nd August – 4th September 2012

Cyclists: James Kirkpatrick, Joe Hall, Matt Bidault, Alec Raeside, and Tom Thorpe ("5 Skinny Men")  
Tom is the brother of Benjamin Thorpe (02 Sep 94 — 17 Nov 2010)

This team effort raised £4000 for various worthwhile charities, with over £1600 for the Barth Syndrome Trust. They had to battle relentless rain, wind and mist, punctures, injuries and total exhaustion but their determination kept them going. What was their strategy? In Tom's words, it was summed up quite simply:

**Step 1:** Moan as you approach a big hill

**Step 2:** Make it up that hill by any means necessary

**Step 3:** Feel an overwhelming sense of euphoria as you realize you now have a huge downhill waiting

**Step 4:** Waste all energy racing down the hill in an attempt to reach a top speed before realizing you've just cycled into a valley and have another beast of a hill waiting

**Repeat steps 1 to 4 for 1000 miles!!**

The team set up a great blog so that we could all witness their progress, including the additional "forfeits" they set for themselves as fundraising targets. At one point, the "5 Skinny Men" turned into "5 Hairy Men" and later into "5 Shaved Men." As they finally reached the tip of Scotland, they seemed to suddenly lose most of their clothing and became "5 Painted Men!" All for a great cause guys, and we salute you all.

Tom's cycle ride finished on what would have been his younger brother Benjy's 18<sup>th</sup> birthday. As Tom so aptly states: "The cycle challenge was focused on all those who still face life's physical challenges." Benjy was known and loved by many in our Barth community (September 2, 1994 – November 17, 2010). We love Benjy's family, and this seemed a fitting tribute to his life.



"5 Skinny Men" complete their journey from Land's End to John O'Groats! (Photos courtesy of Tom Thorpe ~ 2012)

### In Memory of Peggy New

By Annick Manton, Volunteer, Barth Syndrome Trust



Margaret "Peggy" New  
(Photo courtesy of BSTrust ~ 2012)

It was with great sadness that I heard of the death of Mrs Peggy New of Overton, Hampshire. Peggy joined the ranks of our volunteers when we started our fundraising efforts in the village. She was a staunch supporter of the Barth community and always showed a deep interest in all our boys, especially my grandson, Nicholas. Behind the scenes, Peggy supported all our Christmas Bazaars. She loved to knit and used to make dolls and their outfits that we sold for Barth Syndrome Trust (BST). Over the years, she also donated many prizes for our raffles. Peggy was my friend and I shall miss her warmth, her kindness and her eternal optimism. We offer our deepest sympathy and love to her daughter, Terri Allison, BST's grass roots Fundraising Coordinator.

Terri Allison would like to thank everyone who sent condolences: "I was overwhelmed and very moved with the messages of sympathy sent to me from so many connected with the Trust, including some I had never met. It was lovely that they showed they cared. Mum was such a special lady."

# Barth Syndrome Foundation of Canada

## President's Report — Brainstorming for Barth

By Lynn Elwood, President, Barth Syndrome Foundation of Canada



(L-R) Carol Wilks, Lois Galbraith, Cathy Ritter, Lynn Elwood, Susan Hone and Chris Hope  
(Photo courtesy of BSFCa ~ 2012)

It was a beautiful fall weekend when the Barth Syndrome Foundation of Canada (BSFCa) Board and Executive gathered on Lake Kasshabog to review 2012 and plan 2013 for the organization. Friday afternoon was warm, and we started the weekend with lunch on the porch among the falling leaves. From there, we went into a full weekend of creative thinking and discussion, working through our budget and program plans for the 2013 year.

In reviewing 2012, there are a number of areas of success that we plan to replicate in 2013. First is more focus and direct interaction with our affected individuals. This helped to direct our priorities such as the family outreaches and the wallet cards (point form description of BTHS and personal health information) which were well received. We continue to have strong volunteer support and in 2013 will plan to further utilize their talent. We were able to participate in funding research and gained a closer working relationship with some of the Canadian researchers.

Our financial review showed that we were very accurate in our 2012 budget even though, as expected, our revenue was down from the previous year. While our financial health was still strong, our bank balances were less than they have been in years. We needed to continue to be creative in raising funds and thrifty in our program plans in order to plan for a balanced budget in 2013.

We spent a considerable amount of time working through funding plans. The result is that we will seek grants from key groups, continue with the Executive-driven fundraisers, and work to increase the number of grassroots fundraisers. Several grassroots funding ideas were discussed and some are planned, while other ideas will be shared with our volunteers.

Key program decisions included the plan for an adventure event for the affected individuals as identified in the Needs Assessment; a Western Canada outreach and hospital awareness visit; BSFCa website improvements, and more close contact with our volunteers and donors. On the administration side, we will be making some adjustments in response to changing Canadian legislation. We will work through these in the coming months and will review them with members at our Annual General meeting in spring of 2013.

As in the past, the Canadian planning weekend was a focused and hard working few days but also a chance to spend time with a great group of friends. We left the weekend recharged, well fed, and excited to complete this year and get going on the 2013 programs.



(L-R) Lynn Elwood, Les Morris, Cathy Ritter and Chris Hope  
(Photo courtesy of BSFCa ~ 2012)



BSFCa bears ~ a great way to raise funds and awareness!  
(Photo courtesy of BSFCa ~ 2012)

# From the Heart! Getting the Message Out There

By Harry and Helen Hope, Grandparents of an Affected Individual, Canada



Helen Hope  
(Photo courtesy of BSFCA ~ 2012)

As grandparents of grandsons who have been diagnosed with Barth syndrome, we have seen the first-hand effects of this genetic anomaly in our family. Everyday living, while becoming a challenge for the affected individual and siblings, is met with courage and tenacity by our involved family, just as every Barth family must.

Harry's involvement with Rotary has enabled us to visit areas where the ordinary tourist does not normally go. As well, we have met many people that one would not ordinarily meet. We have spoken to people professionally involved in medicine and tried to explain what this disease is and how it affects our grandson. Our Barth shoulder travel bag has become an item of interest, easily drawing fellow travellers into a conversation which we hope will be of great interest to them and that they might recall later and pass on to others.



Harry Hope  
(Photo courtesy of BSFCA ~ 2012)

Our travels with Rotary International allowed Helen to visit the B.M. Birla Heart Research Centre in Kolkata, India and meet with paediatric surgeons to tell them about Barth syndrome. Perhaps in searching out candidates for their life-giving surgery at their Healing Little Hearts project, they may meet a boy with Barth-like symptoms and know what it is. As we all know, Barth syndrome knows no economic or cultural boundaries.

After attending the annual general meetings of the Barth Syndrome Foundation of Canada (BSFCA) and seeing the dedication of those attending, we cannot help but think of them when an opportunity arises to carry on their enthusiasm. The BSFCA Golf Tournament is an example of well-executed planning and dedication.

By getting the word out and informing as many people as possible, one never knows who it may touch. It just might be another (Barth) parent with an obviously sick little boy, who is not getting any answers. We will do our best to be as informative as possible and pass on all information to anyone who is interested and will listen.

## Volunteer Corner

By Lois Galbraith, Volunteer, Barth Syndrome Foundation of Canada



Les's talented woodworking!  
These scissor caddies were given  
to Barth families at BSF's 2012 Conference!  
(Photo courtesy of BSFCA ~ 2012)

We would like to thank all our volunteers and recognize a few in this issue. **Celia McGuinness's** Barth teddy bears were a selling sensation at the conference in Florida this June. **AnnMarie Daley and the nurses in the Heart Cath Lab of Peterborough Regional Health Centre** donated \$100 to the BSFCA in return for caps sewn for them by **Lois Galbraith**. As well, **Natalie Sisson** spent numerous hours preparing "golfer envelopes" for our September 10th annual golf tournament. Kudos to **Lynn Elwood** for her work on the BSFCA website ([www.barthsyndrome.ca](http://www.barthsyndrome.ca)). **Les Morris** made us fifty-five Canadian scissor caddies that we distributed to Barth families at BSF's June conference. The talents and generosity of all of our volunteers are welcomed and truly appreciated.

# BSFCa Golf Tournament 2012

## A Recipe for Fun and Success!

By Co-Chefs Lois Galbraith and Cathy Ritter, Barth Syndrome Foundation of Canada

### Meal preparation and golf have much in common....



(Back) Robert  
(L-R) Adam, Lois and Josh  
(Photo courtesy of BSFCa ~ 21012)

- Begin with a beautiful, sunny Ontario fall day
- Add the pristine, sprawling Legends Course at Woodington Lake Golf Course
- Garnish with the world's best and experienced volunteers
- Sprinkle with our very special Canadian guys – Robert and Adam
- Mix in twenty-eight foursomes of eager and dedicated golfers
- Heat up the fun with awesome golf prizes and giveaways
- Blend in the international support of our Floridian golfers Jan, Sharon and Joanie and our generous hole sponsor, the Anderson family, from Scotland
- Combine these ingredients with a delicious evening meal and a silent auction for more fun, and you have a successful, stimulating and scrumptious day of great golf and fundraising.

Monday September 10, 2012 was the BSF of Canada's 8<sup>th</sup> Annual Golf Classic, "Driving for the Cure." We once again were very

pleased with the golf day and we raised C\$17,600. We can now look forward to Monday September 11, 2013 and another wonderful recipe to share with friends and family.

### Boogie for Barth 3.0

For the third year in a row, Susan and Bob McJannett planned, sponsored and ran a great rhythm and blues dance featuring George Olliver and his band "Gangbuster" to benefit the Barth Syndrome Foundation of Canada. September 29<sup>th</sup> saw the Toronto Humber Yacht Club rocking with great sounds from this legendary band. "Gangbuster" consisting of Tony Padalino on keyboards, Mike Sloski on drums, Eli Eisenberg on bass, and ably led by George Olliver "the Blue Eyed Prince of Soul." These men are legends in the Toronto music scene.

**Tony Padalino** played keys with Grant Smith and the Power, was a member of the Club Bluenote house band and part of the Canadian Idol TV band. **Mike Sloski** played drums with the **Cameo Blues Band** and served as drummer for many of the famous U.S. artists who played in Ontario. **Eli Eisenberg** is an amazing bass player, the youngest of the group, he too plays with all the touring artists. **George Olliver**, leader and singer, was the front man for the legendary Canadian group

"Mandella." Put these four men together and you are guaranteed a great evening of high energy R&B. Highlight as always was when George got the ladies up to sing "Mustang Sally" with him. The dance floor was filled all evening. **Frank Malfara and Linda Upshall** sold 50/50 tickets during the evening.

As well, we had a silent auction with items to fit almost all tastes. As the evening wound down, **Lois Galbraith and Carol Wilks** collected the auction slips and delivered the items to the winning bidders.

When the dust settled, C\$3,100 was raised for this deserving charity. "This year's event was harder than in years past," noted Bob, and whether or not we do it again, all of us who have contributed have had a wonderful experience and have raised well over C\$12,000 for the Canadian Barth organization."



A silent auction was held during Boogie for Barth 3.0  
(Photo courtesy of BSFCa ~ 2012)

# Barth France — One Cause a Year (AGAIN)

By Stéphane Lemaire, Barth France Member



Stéphane Lemaire, Barth France Member  
(Photo courtesy of Barth France ~ 2012)

See page 27/29 for the French translation of this article.

**M**y story with the Barth France is common, an everyday one. I'm a chief executive officer and my associate is the grandfather of Raphaël (who suffers from Barth syndrome). As a real-life witness to the illness, I got to know it by observing first hand the joys and despairs of my associate.

Barth syndrome was not my fight. Raised as a product of the state system's care for children, I was bound to become a defender of children's rights and projects that would enable them to build themselves up.

There are many great causes to defend, and I will never be able to say 'yes' to all of them or give enough time. So, a few years ago, I made a "One Year, One Help" decision: one cause a year. Initially, I wanted to share with my employees a common project chosen by the team. A firm is full of different sensitivities, and fighting for others was a way to encourage team spirit, by growing together.

Therefore, after having "clothed" a few villages in Morocco, supported a few French orphanages, joined a few Christmas events for children of the homeless, and done fundraising for Emmaus,

Barth syndrome has a special meaning to me. An orphan illness... A fight against the unseen, unknown, unperceivable. The term orphan says it all!

My first "One Year, One Help" for the Barth France was easy...I wrote a cheque! I naturally helped out in a marketing operation which didn't make any difference to my financial situation. After this first step, meeting Raphael for the first time and being faced with families whose children were sick was a real turning point, so, what strength and hope were they going to give me? Sixteen months later, I had achieved two half-marathons, two triathlons and my first half Ironman under the colors of the Foundation. Don't ask me if I like to run!! You wouldn't believe me! I only practised martial arts, my swimming skills were very poor, and a motorbike accident had crushed my hopes of running. At that point, the challenge became one... As it simply wasn't for me.

I remember reading a testimony of an athlete who ran for Barth. He made the effort more conceivable by comparing it to a "permanent Ironman" that a Barth kid fights daily. That idea spoke to me, and that's most of the reason I joined the team! As for the rest, I feel it important to mention the Mannes family (Barth France) who works whole-heartedly not only for us athletes, but also to raise awareness of Barth syndrome.

I am truly proud in 2012 to have raised 9435€ on the Half Ironman of Aix-en-Provence and a little over 2000€ for the rest of the races. I would feel very full of myself if I could give you a perfect recipe for fundraising. Sadly, I don't have one! Fundraising is as difficult as recovering after having put so much effort into it! I challenged myself for it, as I did for the first Half Ironman...feverishly! My goal for 2013 is first to change my famous catch phrase "**One Year, One Help**" into "**One Year, One Help, AGAIN!!**" Then training for Ironbask, a "long distance" triathlon for Barth France. As for fundraising... That's my secret! But I promise it will be great!



## Barth France, Not Only Sport...

Because not everybody runs, and because not everybody lives as an "Ironman," this year Barth France has created new events to fundraise and spread information about Barth syndrome ...new events to reach out to additional donors, to establish new contacts... Therefore, last July, 70 golfers competed in the second edition of the Barth France Golf Trophy. At the start, each contestant was given a perfume donated by Guerlain, and, in the end 20 prizes were awarded, thanks to numerous sponsors. The entrance fee was 15€, all of which was donated to the association by Golf de La Bretesche, and additional funds also were collected from generous donors. All in all, over 4000€ were collected that day.

During a dinner, one of our friends, as a joke, said Barth France should organize a poker tournament as a change from triathlons and marathons. We all had a good laugh; however, a few days later, the idea matured and all that was left was to organize it. This was no easy task. Finding a venue, dealers, tokens, prizes, making leaflets, getting official permissions. It was a huge amount of work and effort, nearly as much as training for a marathon, but we do hope this new event will be a great success!

Because Barth France will always be Barth France, we couldn't resist the pleasure of showing you a few of our sportsmen, among whom, Xavier, who completed Ironman in Barcelona in just over 12 hours under the Barth France colors; and Marc, Phil, Jerome, Frederic, Didier, Arnaud, and Jean-Phi, who competed for Barth France in the Half Ironman of Aix-en-Provence.

We have many projects for 2013, such as the Paris Marathon, Ironbask, the world championships of long distance triathlon in Belfort, and so many others. New people join the team regularly, bringing their different experiences.

(Cont'd on page 28)

# Barth France — Une année, Un soutien

By Stéphane Lemaire, Membre de Barth France



Stéphane Lemaire, membre de Barth France  
(Photo courtesy of Barth France ~ 2012)

Voir les pages 26/28 pour la traduction anglaise de cet article.

**J**e suis chef d'entreprise et mon associé est le grand père de Raphaël (atteint du syndrome de Barth). Sensibilisé par proximité, j'ai pris le temps de comprendre le syndrome en témoin ponctuel des joies et des peines de mon associé. Le syndrome de Barth n'était pas mon combat. Enfant du social, j'étais prédisposé à défendre l'enfant, la cause de l'enfant et les projets qui permettent à celui-ci de devenir grand en toute intégrité.

Il existe beaucoup de belles causes à défendre et je n'aurai jamais assez de "Oui" disponibles ou de temps pour m'engager dans tous ces projets. J'ai donc opté depuis plusieurs années pour le "One Year, a Help," une cause à défendre par an. A l'origine, je souhaitais partager avec mes salariés un projet "élu" par l'équipe. L'entreprise est un vaste champ de sensibilités, se battre pour les idées des autres c'est mobiliser l'esprit d'équipe, l'enrichir en œuvrant dans le bon sens.

Alors, après avoir "habillé" quelques villages au Maroc, quelques orphelinats en France, après avoir participé à des opérations "Noël" pour des enfants de sans abri et récolter pour "Emmaüs," Barth est arrivé avec une résonance toute particulière. Maladie Orpheline... Un combat contre l'invisible... Peu connu, peu perceptible. Quand on dit Orpheline, finalement on dit tout !

Mon premier "One Year, a Help," pour Barth France a été facile... Faire un chèque! J'ai donc naturellement participé à une petite opération "marketing" qui, financièrement, n'altérait en rien ma trésorerie. Après cette première action, c'est la première rencontre avec Raphaël et le face à face avec sa famille qui ont été décisifs. En résumé, quelle force et quelle conviction allaient-ils me transmettre? 16 mois après, j'ai bouclé deux semi-marathons, deux triathlons et mon premier Half Ironman aux couleurs de Barth France. Ne me demandez pas si j'aime courir!! Vous allez être surpris! Je ne pratiquais que les Arts Martiaux, je nageais mal et un accident de moto avait amoindri toutes idées de course à pied. C'est là que le défi est devenu logique... Car pas fait pour moi!

Je me souviens avoir lu un témoignage d'un athlète courant pour Barth. Celui-ci relativisait l'effort éphémère qu'il venait de faire en le comparant à "l'Ironman permanent" que vit un Enfant "Barth" jour après jour. C'est cette idée m'a plu et je crois que je me suis engagé, aussi, pour ça! Pour le reste, il faut quand même parler quelques instants de la Famille Mannes (Barth France) qui œuvre dans la simplicité, tant pour nous, sportifs engagés, que pour faire connaître le syndrome de Barth.

Je suis réellement fier, en 2012, d'avoir collecté 9435€ sur le Half Ironman d'Aix et un peu plus de 2000€ sur le reste des courses que j'ai réalisées. Par vanité, je serais heureux de vous confier une formidable recette de collecte, mais je n'en ai pas ! La collecte est aussi difficile que la récupération de celle-ci! Je me suis simplement challengé pour elle, comme je me suis challengé pour ce premier Half Ironman... avec fièvre! L'enjeu de la saison 2013 est, dans un premier temps, de modifier le fameux slogan "One year, a help," en "One Year, a Help, AGAIN!!!" Puis de me préparer pour l'IronBask, un triathlon "longue distance" aux couleurs de Barth France. Quant au montant de la collecte... C'est un secret! Mais je vous promets qu'elle sera belle!



## Barth France, du sport... mais pas seulement

Parce que tout le monde ne court pas, et parce que tout le monde ne vit pas forcément "Ironman," Barth France a souhaité cette année proposer de nouveaux événements, toujours pour faire connaître le Syndrome de Barth, et pour collecter des fonds...de nouveaux événements pour toucher de nouveaux donateurs, pour établir de nouveaux contacts.... Ainsi, en juillet dernier, 70 golfeurs ont pris le départ de la 2ème édition du Trophée de Golf Barth France. Chaque participant s'est vu remettre, au départ, un parfum offert par la Maison Guerlain, et 20 d'entre eux ont été récompensés lors de la remise de prix, très généreusement dotée grâce à de nombreux partenaires. Le droit d'entrée a été, traditionnellement, fixé à 15€, reversés intégralement à l'association par le Golf de la Bretesche, chaque participant étant par ailleurs libre de faire un don à l'association. Au total, ce sont plus de 4.000€ qui ont été collectés par Barth France au cours de cette journée.

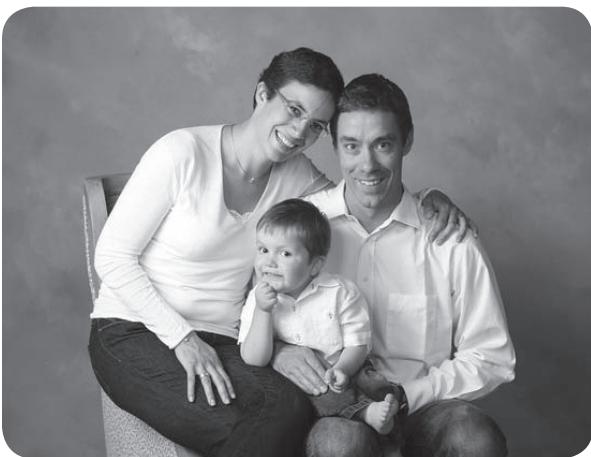
Lorsqu'au cours d'un dîner, un ami nous lance, comme une boutade, que Barth France pourrait organiser un tournoi de Poker, histoire, de temps en temps de parler d'autre chose que de Triathlon ou de Marathon, nous prenons sa proposition à la rigolade....mais quelques jours après, l'idée avait fait son chemin....il ne restait plus qu'à s'occuper de l'organisation....ce qui n'est pas une simple affaire : trouver la salle, les croupiers, les jetons, les lots pour les gagnants, réaliser les affiches, obtenir toutes les autorisations nécessaires....beaucoup beaucoup de travail et d'effort....presqu'autant qu'un entraînement pour un Marathon! Mais nous espérons bien que ce nouvel événement sera une réussite!!

Mais, parce que Barth France reste Barth France, nous ne pouvons résister au plaisir de vous mettre quelques photos de sportifs qui se bougent pour Barth....entre autres, Xavier, qui a terminé l'Ironman de Barcelone en un peu plus de 12h, avec le drapeau de la Barth Syndrome Foundation, et Marc, Phil, Jérôme, Frédéric, Didier, Arnaud, Jean-Phi qui ont participé, toujours sous les couleurs de Barth France, à l'Half Ironman d'Aix en Provence....

Pour 2013, les projets sont nombreux, parmi lesquels le Marathon de Paris, l'Ironbask, le championnat du Monde de triathlon longue distance, à Belfort, et tant d'autres!....L'équipe s'étoffe et accueille de nouveaux coureurs, triathlètes expérimentés ou néophytes. (Suite à la page 29)

# Barth France, Sports, But Not Only...

(Cont'd from page 26)



Florence, Philippe and Raphaël  
(Photo courtesy of Amanda Clark ~ 2012)

## A Clinic a la Mode de Paris

We were lucky to go to the Bristol Clinic in March 2011, and to go twice to Florida, for the BSF's 2010 and 2012 international conferences. Every time, meeting doctors who are familiar with, not only the illness from a specialist's point of view (dilated cardiomyopathy, neutropenia...), but also Barth syndrome as a whole, was a great source of knowledge. Spending a few days, talking exclusively about Barth syndrome and discussing with doctors enabled us to better understand specific symptoms and feel less apprehensive about daily life. During that time, we met other families, other children, who went through roughly what we are going through and with whom the communication was so much easier than with any other family. The feeling of being understood can only be found in such a place, despite the language barrier.

Many French-speaking families suffer from Barth syndrome in Europe, and we would like to give them the opportunity once a year to get the most specialized treatment for the disease, where there would be a day of consultations with specialists in this disease, such as a cardiologist, hematologist, nutritionist, geneticist, and possibly other experts who would be willing to join us. They will also have the opportunity to meet other families so they can share and create new friendships. We hope this will lighten their burden by making them more informed and offering support.

Our project for the fall 2013 (spring would have been ideal but seems too soon) would be to organise two days in Paris. The first one at Necker Hospital, the reference center for rare illnesses, where there would be a day of medical appointments with specialists of this disease, such as a cardiologist, hematologist, nutritionist, geneticist, and any other doctor who would be willing to join. The second day would involve a place where families could interact. Having been to the Bristol Clinic, we know how challenging such a project is but also how rewarding it can be. We very fondly hope it will soon kick-off in France and that many families will join us.

## Barth Listserv in French: Knowledge is Power!

By Valerie Lallemand, Belgium

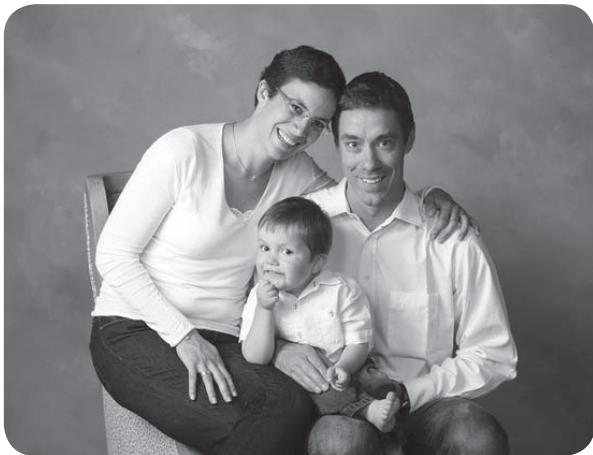
From attending the Barth Clinic in Bristol last March and by talking to another francophone family, we had the idea of writing a French summary of the different topics of the anglophone Listserv which we receive daily on our email. I realised how lucky I was to have studied English. There is a huge advantage to understanding this language as I am able to access so much information on the Family Listserv, one of the Barth Syndrome Foundation's (BSF) exchange and discussion platforms. As they say in English, "Knowledge is Power." Many times during medical appointments with my son, Jules, information I read on Listserv has influenced my conversation with doctors, bringing a positive outcome. I tried to imagine how frustrating and isolating it would be for someone who didn't understand English, to lack information and support because of a language barrier. Being the mother of a sick child, I wanted to contribute to changing things as best I could.

Therefore, we decided with BSF in the US and Barth France to enable French families to access a monthly summary in French of the Listserv's topics. The subjects are divided into several sections: **Nouvelles de la Communauté Barth** (Barth Community's News) where parents of a newly diagnosed child can introduce themselves, one can find news about hospitalized children and good news such as sports results, children's school grades and birthdays. In the **Santé** (Health) section, subjects such as scientific research and medical questions are dealt with. We would like to stress the necessity of having medical advice from a child's own doctor before changing the child's treatment, as the news we translate is only informative. In the **Vie Quotidienne** (Daily Life) section, we discuss topics such as nutrition, specialised equipment and support the children can have at school... In the **Couverture Media** (Media Coverage) section, we publish links to recent articles about the syndrome, and TV shows that have talked about the illness. The **Lever des fonds** (Fundraising) section tells us about the different ways to raise funds.

This summary is sent monthly to French families. We also offer an English translation of their questions so that they can be published on the English Listserv, in their name. We then translate the resulting commentaries and suggestions back in to French for the French families to read. If you also wish to receive our monthly summary, please send us your email address ([valerie@barthfrance.com](mailto:valerie@barthfrance.com)) or your home address if you don't have internet (Barth France – 12 rue Lalo – 75116 Paris – France).

# Barth France, du sport... mais pas seulement

(Suite de la page 27)



Florence, Philippe and Raphaël  
(Photo courtesy of Amanda Clark ~ 2012)

## UNE CLINIC A LA MODE DE PARIS

Nous avons eu la chance de participer, en mars 2011, à la Clinic de Bristol, et d'aller, à deux reprises, en Floride, lors de la conférence de la BSF. A chaque fois, rencontrer des médecins qui connaissent, non seulement la pathologie qui dépend de leur spécialité (cardiomyopathie dilatée, neutropénie, ...), mais également le Syndrome de Barth dans son ensemble, a été pour nous source de nombreux apprentissage; le fait de venir passer une ou plusieurs journée, spécifiquement pour parler du Syndrome de Barth, échanger avec les médecins, nous a permis également de mieux comprendre certains symptômes, et d'aborder le quotidien un peu plus sereinement. Lors de ces journées, nous avons également pu rencontrer d'autres familles, d'autres enfants, qui traversaient, à peu de choses près, la même chose que nous, et avec qui le dialogue était tellement plus facile qu'avec n'importe quelle autre famille... Cette sensation d'être compris, nous ne la retrouvons que là bas, en dépit de la barrière de la langue.

Un certain nombre de familles francophones sont touchées, en Europe, par le Syndrome de Barth, et nous souhaiterions pouvoir leur offrir la possibilité de profiter d'un suivi médical propre au Syndrome de Barth, lors d'une journée de consultations spécifique, ainsi que de la possibilité de rencontrer, pour celle qui le souhaitent, d'autres familles, pour échanger, partager, et créer des liens, pour que la différence, une fois dans l'année, soit moins lourde à porter.

Notre projet, vraisemblablement pour l'automne 2013 (nous aimerais pouvoir organiser cela dès le printemps, mais les délais risquent d'être un peu courts), est d'organiser 2 journées à Paris. La première au sein de l'hôpital Necker, centre de référence des maladies rares, et où exercent plusieurs médecins qui connaissent bien le Syndrome, pour une journée de consultations, avec un cardiologue, un hématologue, un nutritionniste, un généticien, ainsi qu'éventuellement d'autres spécialistes qui pourraient se joindre à nous. La deuxième journée serait davantage orientée vers l'échange entre les familles, en offrant aux familles la possibilité de se retrouver dans un endroit convivial. Pour avoir assisté à la clinic de Bristol, nous savons que ce projet est un véritable challenge, et nous espérons de tout cœur qu'il va voir le jour rapidement, et que de nombreuses familles pourront y participer.

## Barth Listserv en Français: Le savoir c'est du pouvoir!

By Valerie Lallemand, Belgium

C'est en participant à la Clinique Barth à Bristol en mars dernier et en discutant avec une autre famille francophone que nous est venue l'idée de faire un résumé en français des différents sujets de discussion de la Listserv anglophone qui jour après jour arrivent dans nos boîtes de réception de courrier électronique. Ayant étudié l'anglais pendant mes études, je me suis rendu compte combien c'était une chance et un énorme avantage de comprendre et pratiquer cette langue et de pouvoir ainsi avoir accès à cette mine d'informations qu'est la Listserv, ce forum d'échanges et de discussions de la Barth Foundation. Comme dit l'expression anglaise, être informé c'est avoir du pouvoir (Knowledge is Power). Combien de fois, lors de rendez-vous médicaux avec Jules, n'ai-je pas orienté la discussion avec les médecins en fonction de tel ou tel élément que j'avais lu sur la listserv et souvent avec des résultats tout à fait bénéfiques. J'ai essayé de m'imaginer dans la peau de quelqu'un ne comprenant pas l'anglais et me suis rendu compte combien cela devait être frustrant et isolant de ne pas pouvoir disposer de cette source d'informations et de support mutuel pour une simple question de langue. Etant maman d'un petit garçon atteint, j'avais également envie d'apporter ma petite pierre à l'édifice en fonction de mes capacités.

Il fut donc décidé, d'un commun accord avec la Fondation Barth aux Etats Unis et avec L'Association Barth France, de mettre à disposition des familles francophones un résumé mensuel des sujets de la Listserv. Les sujets traduits sont classés selon diverses rubriques: Nouvelles de la Communauté Barth, où se présentent les familles d'un enfant récemment diagnostiquée, les nouvelles au sujet des enfants hospitalisés ou également les bonnes nouvelles comme les résultats sportifs ou scolaires des garçons affectés, les anniversaires. Dans la rubrique Santé, sont regroupés les sujets relatifs aux recherches scientifiques, les questions d'ordre médical que se posent les familles. Nous attirons néanmoins l'attention des lecteurs francophones sur l'aspect informatif des traductions et sur la nécessité de toujours demander un avis médical avant de considérer un changement de traitement. La partie Vie Quotidienne rassemble les sujets sur l'alimentation, la vie à l'école avec les équipements et soutiens spécialisés dont peuvent bénéficier les enfants, etc.. Dans la section Couverture média, nous mentionnons les références des récents articles publiés sur le syndrome ou les liens internet vers les émissions de télévision dont le syndrome a fait l'objet. La rubrique Lever des fonds informe des nombreuses initiatives qui permettent de récolter des fonds (sportives ou autres).

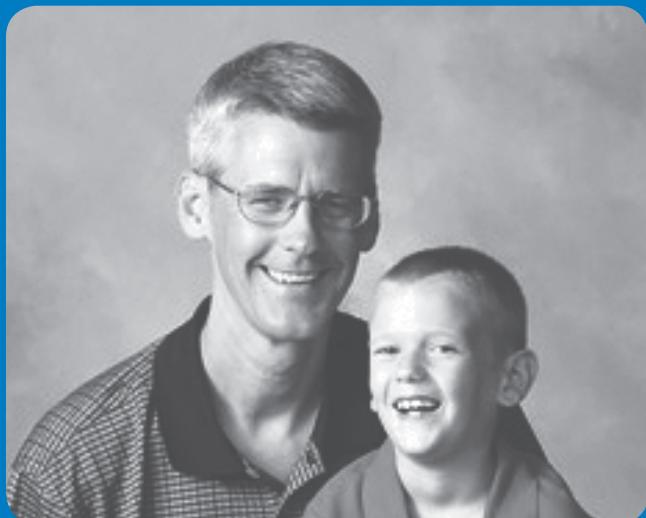
Ce résumé est envoyé mensuellement aux familles francophones qui le souhaitent. Nous proposons également la traduction en anglais des questions que pourraient se poser ces familles avec publication sur la Listserv anglophone en leur nom. Nous assurons ensuite le suivi avec la traduction vers le français des réactions et commentaires reçus. Si vous souhaitez, vous aussi, recevoir ce résumé mensuel, vous pouvez nous communiquer votre adresse mail (valerie@barthfrance.com), voire votre adresse postale si vous ne possédez pas d'accès à internet (écrire dans ce cas à Barth France – 12 rue Lalo – 75116 Paris – France)

**Barth families, have you moved lately?  
Please help us keep your information current.**

In the past, the Post Office notified us of address changes. However, with so few actual mail pieces being sent during the year, we will not know you have moved unless you tell us. If your telephone number and/or email address has changed, please let us know. If we do not have your email address, please go online to add it to your contact information.

Barth families, if you think any information on your family might be incorrect, please be sure to update us.

Visit BSF's website and complete the **Contact Information** form that can be found under *Families >> Update Contact Information* (<http://www.barthsyndrome.org/english/View.asp?x=1568>). Thanks in advance for helping us "keep house."



Greg and Benjamin (age 8)



Tracy and Bly (age 8)

*(Photos courtesy of Amanda Clark ~ 2012)*

## Donations Made Easier

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

**Donate online:** You can donate to BSF or any of the international affiliates by going to our website, [www.barthsyndrome.org](http://www.barthsyndrome.org), and clicking on the "Support BSF" link on our home page, or through Network for Good (<https://www.networkforgood.org/donation/ExpressDonation.aspx?ORGID2=22-3755704>) or through Paypal ([https://www.paypal.com/cgi-bin/webscr?cmd=\\_s-xclick&hosted\\_button\\_id=8XRHKG52LB7L4](https://www.paypal.com/cgi-bin/webscr?cmd=_s-xclick&hosted_button_id=8XRHKG52LB7L4)).

**Donate through Causes on Facebook:** Join us on our online social network ([http://www.causes.com/causes/46297-the-barth-syndrome-foundation?q=barth+syndrome+foundation&rank=0&utm\\_campaign=search](http://www.causes.com/causes/46297-the-barth-syndrome-foundation?q=barth+syndrome+foundation&rank=0&utm_campaign=search)).

**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.



# Power of Kindness



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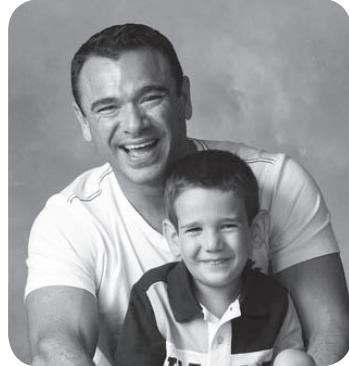
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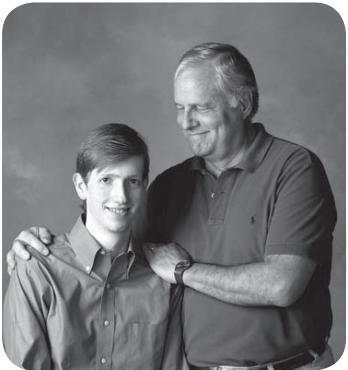
Shi, Dr. Yuguang (Roger)  
Shih, Dr. Renata  
Shindou, Dr. Hideo  
Shirley, Alan & Denise  
Shum, Bill & Ginny  
Slaugenhoupt, Dr. Susan A.  
Smith, Deborah  
Smithson, Sarah  
Smolski, Ed  
Smoot, Dr. Leslie  
Snyder, Floyd  
Somerharju, Dr. Penti  
Soustek, Meghan S.  
Sparagna, Dr. Genevieve  
Spencer, Dr. Carolyn T.  
Stanford, RN, Dianne  
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Steinberg, Jack  
Steward, Dr. Colin G.  
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Strauss, Dr. Arnold W.  
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Taylor, Damani  
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Thorpe, Jeannette  
Titorenko, Dr. Vladimir  
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Tunguz, Stefan  
Tweed, Scott  
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van Ommen, Dr. Gert-Jan B.  
van Raam, Dr. Bram J.  
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Vaz, Dr. Frédéric M.  
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Wilkins, Sue  
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Williams, Kathy  
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Wroe, Martha  
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Wu, Dr. Sean M.  
Xiao, Dr. Xiao  
Xu, Dr. Yang  
Ye, Dr. Zhaohui  
Zaragoza, Dr. Michael  
Zhang, Dr. Ji  
Zhang, Shali  
Zhou, Dr. Bin



Lynda and Derek (age 19) 2012



Kevin (age 23) 2012



Will (age 26) and Steve 2012



John (age 30) and Sue 2012

# Power of Kindness



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Anderson, Tommy & Allanna  
Andrews, Michelle  
Armstrong Jones, Peregrine  
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Basingstoke Voluntary Services  
Bat and Ball Club  
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Bearman, Matt  
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Blue Merle  
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Bristol, Gemma  
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Challenger, Paul & Margaret  
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Charles Family  
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Cleaner, Linda

Clements, Claire  
Cline, Tony & Sue  
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Coleman, William  
Commons, Jon  
Condilffe, Cynthia & David  
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Hornby, Anthony  
Horwood, Jane, Catfish Web Design  
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Hughes, Branwen  
Hunt, Tim  
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Jones, Cynthia  
Jones, Helena  
Jones, Patrick  
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Olden, Lee  
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Owen, Sarah  
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Payne, Jason  
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Potter, Rosalind  
Prest, Simon  
Price, Julie  
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Rawlinson-Millichap, Peter & Dom  
Raynor, Henrietta  
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Reddin, Mick  
Rees, Gaynor  
Reimar, Abi, Norina, Biggi & Michael  
Rennie, Trudi  
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Richardson, Jeanette  
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Riseborough, Kate  
Robbins, Matthew  
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Robinson, Sarah  
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Roseberry Sports and Community College  
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Rudd, Matthew  
Rudd, Timothy  
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Salmon, Gwen  
Salmon, Lawrence  
Salmon, Robertina  
Saywood, Lila  
Schlapak, Gregor & Sonja  
Schrader, Pascale  
Scott-Flegg, Susanna  
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Williams, Pat  
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Winn, Meliese  
Wites, Agnieszka & Wojciech  
Witzani, Horst & Linde  
Woodward, Tracy & Ian  
Woollen, Kate  
Woollen, Rob  
Woolley, Scott & Julie  
Wright, Jess  
Xuen Quartet



Raphaël (age 3) and Philippe 2012



Joe (age 4) and Rachel 2010



Allanna and Alfie (age 12) 2010



Nick (age 14) and Pam 2012

# Power of Kindness



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 Adams, Lynda & Phillip  
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 Allison, Vickie  
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 Chalmers, Stephanie & Shawn  
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 Cherniak, Miriam  
 Cherniak, Morris & Evelyn  
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 Christie, Steve & Jill  
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 Cowper, Margaret  
 Cox, Patty & Gerald  
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Daly, Annamarie  
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 Davies, Diane  
 Davison, Marilyn & John  
 Deforest, Wilmer & Ina  
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 Demick, Diane  
 Dickson, Bel  
 DiDiodato, Giulio  
 Dilland, Darren  
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 Dolenc, Julia  
 Dolenc, Marko  
 Douse, Dave  
 Dove, Judy & Randy  
 Downard, Jason  
 Duclos, Bev  
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 Ebata, Lyle  
 Ebata, Jim  
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 Elton, David ~ Max Bell Foundation  
 Elwood, Adam  
 Elwood, Bryan & Susan  
 Elwood, Lynn & Rick  
 Ernst, Gabriele & Kevin  
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 Francis, Mark  
 Fink, Penny  
 Flanagan, Mae  
 Fraser, Wendy  
 Freeland, Dagmar  
 Galbraith, Lois & Les Morris  
 Gallacher, Betty  
 Garment, Linda & Philip  
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 Gilmour, John & Christine  
 Gilmour, Paul ~ Precision Aero  
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 Gordon, Travis  
 Gordon, Ann  
 Gregorio, John ~ Strictly Excavations Inc.  
 Haggert, John & Judy  
 Halifax, Carol & Guy  
 Hamilton, Reg  
 Hill, John  
 Hill, Richard  
 Hill, Robert  
 Hintze, Audrey  
 Hocevar, Helen & Frank  
 Hone, Barbara  
 Hone, Chris & Susan  
 Hone, Josh  
 Hope, Harry & Helen  
 Hope, Michael & Christine  
 Hope, Robert  
 Hope, Terry & Ruth  
 Hope Aero,

Howard, Sharon  
 Hubner, Malca  
 Hughes, Ron & Cathy  
 Humphries, Jack & Jan  
 Irwin, Kevin  
 Jardine, Ward  
 Jarvis, Peggy  
 Jayavendra, Paul  
 Jeffery, Elaine  
 Kadoguchi, Lilly  
 Kahn, Stephen  
 Kahn, Vera & Dave  
 Kemp, Bill  
 Koll, Tom  
 Kralvic, Andrew  
 Kritschgau, Rick ~ Vision 2000  
 Krueger, Rudy  
 Kugelmann, Jan  
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 Lamorie, Bruce & Sharon Weller  
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 Lindsay, Sue  
 Lister, Verna  
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 Lyall, Diane & Doug  
 Lyall, Judy  
 Lyall, Susan  
 MacDonald, Carol & Fred  
 Madigan, Jim  
 Malfara, Frank  
 Malick, Mel & Suzanna  
 Magennis, Lylem  
 Magennis, Jasmine  
 Magennis, Debra  
 Mandar, Scott  
 Mann, Ruth & James  
 Marcucci, Gabe  
 Marchesin, Elvy  
 Masterson, Moira  
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 McGarry, David  
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 McGlaughlin, Larry & Jackie  
 McGuiness, Celia  
 McJannett, Susan & Bob  
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 Melen, Pat  
 Mill, Dawn & Marvin  
 Millar, Ronald & Margaret  
 Miller, Nancy  
 Miller, Pickard Law  
 Milne, Peter  
 Miloff, Maury  
 Moore, Nigel  
 Morenz, Caitlyn & Dave  
 Morenz, Sharon & Terry

Morris, Erik  
 Morris, Elsie  
 Morris, Ian ~ Jones Deslauriers  
 Moss, Don  
 Nagel, Siegfried & Erna  
 Nagel, Thomas  
 Newfeld, Jeff & Beth  
 Noddle, Bev & Norm  
 Olson, Sharon  
 Paczkowski, John & Vicki Perkins  
 Paul, Bill  
 Patrick, Barbara  
 Pearce, William & Bev  
 Perkins, Phyllis  
 Pickup, Jacob  
 Pitkethly, Maureen  
 Poitras, Mike & Sandi  
 Primus, Christopher  
 Prodan, Magda  
 Quackenbush, Earl & Carolyn  
 Raymond, David & Lila  
 RBC Foundation  
 Ritter, Ryan  
 Ritter, Cathy & Chris  
 Roberts, Marlowe, Jackson, Jackson & Assoc.  
 Robinson, Jim  
 Rodgers, James & Tina  
 Ross, Barbara  
 Sawh, Harold & Ruby  
 Schillaci, Paul  
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 Weaver, Joan  
 Webb, Lindsay ~ Lindsay Webb Financial Inc.  
 Weeks, Ken & Jane  
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 Wild, Andy  
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 Wilks, Helen  
 Willis, Dave  
 Wood, Gale & Peter  
 Woodcock, Sharon & Roy  
 Worsley, Dorothy  
 Wright, Jessica & Cameron  
 Wuthrich, Grace  
 Young, Ron & Lenora  
 Zavitz, Peter

Note: All photos featured on pages 31-35 are courtesy of Amanda Clark and taken at BSF's 2010 and 2012 international conferences.



Karen and Travis (age 17) 2012



Jared (age 18) and Chris 2012



Cathy and Ryan (age 21) 2012



Lynn and Adam (age 22) 2012



Barth Syndrome Foundation

PO Box 618

Larchmont, NY 10538

Phone: 850-273-6947

Facsimile: (518) 213-4061

Email: [bsinfo@barthsyndrome.org](mailto:bsinfo@barthsyndrome.org)

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



Jack (age 2)



Noah (age 11)

## 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 includes 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**

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